

# DIRECTORY OF GENETIC AND RARE DISEASE SUPPORT GROUPS & COMMUNITY SUPPORT ORGANISATIONS



Directory of  
Genetic and Rare Disease Support Groups and  
Community Support Organisations

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ISBN 0-9775358-0-0

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**As contact details may change, a regularly updated online version of the Directory is available at: [www.gardn.org.au/directory](http://www.gardn.org.au/directory)**

## Table of Contents

Introduction .....	7
The Genetic and Rare Disease Network .....	8
Acoustic Neuroma Association of WA .....	11
Albinism Fellowship of Australia .....	12
Alpha-1 Association of Australia .....	13
Androgen Insensitivity Syndrome Support Group Australia Inc.....	14
Angelman Syndrome Association of WA .....	15
Arthritis & Osteoporosis WA .....	16
Arthrogryposis Group (TAAG) Inc, The Australian .....	17
AusDoCC Inc (Australian Disorders of the Corpus Callosum).....	18
ausEE Inc.....	18
Australia Alopecia Areata Foundation.....	21
Australian Rare Chromosome Awareness Network (ARCAN) .....	22
Batten Disease Support and Research Association (Australian Chapter) .....	23
Bertolotti's Syndrome Support .....	24
Bowel Group for Kids (BGK) .....	25
Cardio Facio-Cutaneous (CFC) International.....	26
CDH (Congenital Diaphragmatic Hernia) Australia .....	27
Charcot-Marie-Tooth Association Australia – WA Division.....	29
CHARGE Syndrome Association of Australasia .....	30
Chiari and Syringomyelia Australia .....	31
Children's Tumour Foundation of Australia (NF Australia) .....	32
Chromosome 18 Registry and Research Society Australia.....	33
CJD(Creutzfeldt-Jakob disease) Support Group Network.....	34
CleftPALS WA .....	35
Coeliac Western Australia .....	36
ConnecTeD Foundation .....	38
Cornelia de Lange Syndrome Support Group .....	39
Cri du Chat Support Group of Australia .....	40
Cushing's Disease Support .....	41
Cystic Fibrosis .....	43
Cystinosis Support Group, Australian .....	44
Deafblind West Australians (Formerly Usher Syndrome Support Group of WA) .....	45
DEBRA Australia (Dystrophic Epidermolysis Bullosa Research Association) .....	46
Dementia Australia .....	47

Diabetes WA (DWA).....	48
Down Syndrome WA .....	49
Dyslexia SPELD Foundation WA Inc .....	50
Ectodermal Dysplasia Support Group (ozED).....	51
Epilepsy Association of WA Inc .....	52
Even Keel Bi-Polar Disorder Support Assoc. Inc .....	53
Fabry Australia .....	54
Face Up.....	56
Familial Hypercholesterolaemia Support Group of WA .....	57
Fragile X Association of Australia Inc .....	58
Friedreich Ataxia Network (FAN) .....	59
GAPPS Support .....	60
Gelastic Seizure Support Hub .....	61
HAE Australasia (Hereditary Angioedema).....	62
Haemochromatosis Australia.....	63
Haemophilia Foundation WA Inc .....	64
Heart Kids WA Inc. ....	65
HHT Alliance (Hereditary Hemorrhagic Telangiectasia).....	66
Homocystinuria (HCU) Network Australia .....	67
Huntington’s WA (Inc).....	68
Hypersomnolence Australia.....	69
Immune Deficiencies Foundation Australia (IDFA) .....	71
Kawasaki Disease Foundation Australia.....	72
Klinefelter’s Support Group WA.....	74
Learning and Attentional Disorders Society of WA (LADS).....	75
Leukodystrophy Support Group, Australian .....	76
Liver Foundation of Western Australia .....	77
LQTS Support Group of WA Inc .....	78
Lupus Group of WA Inc, The .....	79
Lyme Disease Association of Australia .....	80
Machado Joseph Disease (MJD) Foundation Ltd .....	81
Mental Illness Fellowship Western Australia Inc .....	82
Mitochondrial Disease Foundation, Australian (AMDF).....	83
Motor Neurone Disease Association WA (MNDAWA) .....	84
Mucopolysaccharide and Related Diseases Society.....	85
Multiple Sclerosis Society of WA Inc (MSWA) .....	86
Muscular Dystrophy WA .....	87
Myasthenic Association (The Australian, in NSW Inc) .....	88

Noonan Syndrome Awareness Association .....	89
Osteogenesis Imperfecta Society of Australia .....	90
Our Faces Support .....	91
Pallister-Hall Syndrome (PHS) Support Hub.....	92
Pallister-Killian Syndrome Foundation of Australia .....	93
Parenteral Nutrition Down Under (PNDU).....	94
Parkinson’s Western Australia .....	95
Pompe Association, Australian .....	96
Retina Australia (WA).....	97
Rett Syndrome Association of WA.....	99
Sanfilippo Children’s Foundation (SCF).....	100
Senses Australia .....	101
Short Statured People’s Association WA Branch.....	102
Red Nose WA (Formerly Sids and Kids WA) .....	103
Sjögren's Syndrome Association Inc, Australian .....	104
SOFTWA (Support Organisation for Trisomy and Related Disorders of WA).....	105
Spinocerebellar Ataxia Australia Inc .....	106
Syndromes Without A Name (SWAN) Australia .....	107
Tarlov Cyst Disease Society of Australia .....	108
Thalassaemia Australia Inc.....	109
Tourette Syndrome Support Group, Perth .....	110
Tuberous Sclerosis Australia .....	111
Turner Syndrome Association of Australia Ltd (WA).....	113
VCFS 22q11 Foundation .....	114
VHL (von Hippel-Lindau) Alliance .....	115
Williams Syndrome Association of Western Australia (WSAWA Inc) .....	116
XYY Syndrome Association of Australia Inc. ....	117
Support Organisations.....	118
Activ Foundation.....	118
Association for the Wellbeing of Children in Healthcare.....	120
Asthma Foundation of WA.....	121
Be Inspired Foundation .....	122
Cancer Council WA .....	123
Carers Association of WA (Inc).....	124
Centre for Genetics Education.....	125
Children and Young People with Disability Australia (CYDA).....	126
CLAN WA.....	127
ConnectGroups Support Groups Association WA Inc.....	128

Deaf Society Inc, Western Australian .....	129
Developmental Disability WA.....	130
Disability Services Commission.....	132
EDGE Employment Solutions.....	134
Ethnic Disability Advocacy Centre (EDAC) .....	135
Genetic Alliance Australia.....	136
Genetic Services of Western Australia .....	137
Health Consumers' Council (WA) Inc .....	138
HelpingMinds - Mental Health Carers and Friends Association .....	139
Immunodeficiency Research Centre.....	140
Independent Living Centre WA.....	141
Intelife .....	142
Ishar Multicultural Women's Health Centre.....	143
Kalparrin.....	144
Kidney Health Australia .....	145
Linkwest .....	146
Office of Population Health Genomics .....	147
People with Disabilities (WA) Inc (PWdWA) .....	148
Rare Voices Australia.....	149
Rocky Bay (and Spina Bifida and Hydrocephalus Association of WA).....	150
Self Help Queensland Inc (SHQ).....	151
SHOUT (Self Help Organisations United Together) (ACT) .....	152
Speak Easy Association Western Australia .....	153
TADWA (Technology Assisting Disability WA).....	154
The Neurological Council of WA Inc .....	155
Therapy Focus .....	156
A-Z Index of Synonyms of Genetic & Rare Conditions.....	157

## **Introduction**

The purpose of this Directory is to provide information on genetic and rare disease support services to health professionals, individuals, and genetic and rare disease support groups. The Directory is designed to assist with referral of individuals and families with either a known, newly diagnosed genetic and/or rare condition or predisposition to such a condition.

The groups listed in this Directory range from small groups of individuals with rare conditions to larger community organisations for people with more frequently occurring genetic conditions. These groups offer an important service to many Western Australians, by providing an opportunity to share information and experiences, as well as offering support through group activities and available services.

Genetic and rare disease support groups provide information and resource materials about the conditions that affect their members and families. The groups also endeavour to provide health professionals and the community with information about their group and the genetic and/or rare condition they represent.

The support organisations listed in the back of this Directory provide services that may be of additional support or assistance to individuals and families with either a known or newly diagnosed genetic and/or rare condition or predisposition.

Whilst the Genetic and Rare Disease Network (GaRDN) has provided information about all existing Genetic and Rare Disease Support Groups member organisations in Western Australia there may be some that are not included in this edition.

## **Acknowledgments**

GaRDN wishes to convey its appreciation to all the genetic and rare disease support groups, agencies and organisations who supplied the information included in this publication. GaRDN wish to acknowledge the Department of Health Western Australia, whose ongoing support makes this publication possible.

Every effort has been made to ensure that at the time of publication, the information contained in this Directory is free from error. The Genetic and Rare Disease Network can accept no responsibility for any claim that may arise from a person acting on information contained herein.

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## **The Genetic and Rare Disease Network**

The Genetic and Rare Disease Network (GaRDN) is a not for profit organisation based in Western Australia. It works to empower individuals and their families to achieve positive health outcomes. We inform health professionals and the wider community on the perspectives and experiences of those affected by genetic and rare diseases. We connect key stakeholders and service providers with people affected by genetic and rare diseases. GaRDN is funded by the Department of Health Western Australia as the peak body for genetic and rare disease support groups in this state.

Genetic and rare disease support groups can be a useful resource for individuals and families affected by a genetic and/or rare condition. Support groups can provide information about the condition and community resources, as well as understanding and an empathetic ear.

In Australasia, there are hundreds of such groups, many focussing on a specific genetic and/or rare condition. We can put you in contact with these groups or provide the support and guidance to help you establish a new support group.

GaRDN may also be able to provide information and support for conditions so rare that there is no specific local support group.

We maintain a database of genetic and rare disease support groups throughout Australia and with international links can provide up to date information on genetic and rare conditions — including the latest information on research developments.

GaRDN actively supports the ongoing development of genetic and rare disease support groups. Groups can publish articles and advertise and increase awareness of their support group's activities on the GaRDN website, social media and in our newsletters.

Staff can assist groups and individuals in researching information regarding particular genetic and rare conditions or predispositions.

Listing in this Directory is free to member groups. If you would like your group to be in a future Directory or on our website, please contact us.

### **What is a rare disease?**

As defined by International Conference on Rare Diseases and Orphan Drugs, rare diseases are any disorder or condition that is life-threatening or chronically debilitating which is statistically rare with an estimated prevalence of 5 in 10,000 people (stated in other WA publications as 1 in 2,000) or of similarly low prevalence and high level of complexity that special combined efforts are needed to address the disorder or condition. Although each disease is rare, collectively they will affect almost 1 in 10 people at some time in their life. This equates to affecting approximately 2 million people in Australia and 70 million worldwide. It is estimated there are up to 8,000 different types of rare diseases.

Due to their rarity, issues are often heightened in relation to diagnosis and treatment. Obtaining a diagnosis is can be difficult and many rare diseases may have no cure.



Eighty (80) percent of rare diseases are considered to have a genetic origin. Such diseases are present throughout a person's life, even if the symptoms are not always present. Rare diseases also include rare cancers as well as better known conditions such as Huntington's disease.

According to the Global Genes Project, approximately 50% of rare diseases do not have a disease specific foundation supporting or researching that disease.

## **What causes genetic conditions?**

There are three types of genetic conditions:

1. Heritable conditions: Due to a mutation in a single gene
2. Chromosomal conditions: Occurs when an individual is affected by a change in the number, size or structure of his or her chromosomes
3. Multifactorial conditions: Due to the interaction of the genetic information and environmental factors such as diet, chemical exposure and lifestyle.

## **What can be done about genetic conditions?**

### **Prevention**

Some people are more at risk than others for developing a condition that is due to the interaction of environmental factors with their inherited genetic information. They are "genetically predisposed" to develop these conditions. However, the presence of an environmental "trigger" is necessary for the person to be affected with the condition. In some cases, prevention of the condition can be achieved by the person avoiding being exposed to the particular environmental factor that will trigger the condition.

For example, it is possible to prevent about 70% of the cases of spina bifida (a neural tube defect) in babies if women who are more likely to have a baby affected with spina bifida take folic acid before pregnancy and continue it during early pregnancy.

### **Early diagnosis and treatment**

In some genetic conditions, early diagnosis, sometimes even before the symptoms appear, can lead to specific treatment. For example, all newborn babies in Australasia are screened for phenylketonuria (PKU) by a simple blood test. Diagnosis and treatment within the first month of life are crucial to avoid intellectual disability. Also, some cancers that have a genetic component, e.g. breast cancer, bowel cancer and melanoma, can be detected early enough to enable treatment to take place.

Checking the family health history can be useful to determine if a person or another blood relative is at risk of developing a genetic condition or passing it on to their children.

### **Genetic counselling**

Genetic counselling is available to families and individuals that have concerns about a condition in their family which may have a genetic basis. A team of health professionals that may include clinical geneticists, genetic counsellors and social workers, work together to

provide information and supportive counselling so that families may be better able to understand, and adjust to, the diagnosis of a genetic condition.

Genetic testing, if it is available and appropriate, can also be organised with informed consent. Genetics Services are available in Western Australia and provide genetic counselling to assist informed decision making about genetic testing.

(Information Source: Centre for Genetics Education, NSW. 2008)

## **Support groups**

Support groups provide affected individuals and families with information about the condition and community resources, as well as an understanding and empathic ear.

## **Some points to remember when contacting a group**

Many of the telephone numbers provided in this directory are home numbers, so please be considerate of the hours at which you call. Many support groups do not receive funding and rely on their group members for income. Offering to pay for postage, photocopying and/or materials provided will be appreciated by many groups.

# Acoustic Neuroma Association of WA

Mobile: 0407 427 246

Email: [gail.mayberry9@gmail.com](mailto:gail.mayberry9@gmail.com)

Website: [www.anaa.org.au](http://www.anaa.org.au)

Facebook: <https://www.facebook.com/groups/740322402648020/>

## Acoustic neuroma

Acoustic neuroma is a genetic condition resulting in benign tissue growth in the ear canal. It is characterised by the loss of hearing on one side and is often accompanied by ear ringing and balance disturbance. Most acoustic neuromas occur suddenly without any hereditary component.

## Synonyms of acoustic neuroma

Acoustic neurilemoma

Bilateral acoustic neuroma

Cerebellopontine angle tumour

Perineural fibroblastoma

Neurinoma of the acoustic nerve

Neurofibroma of the acoustic nerve

Schwannoma of the acoustic nerve

Vestibular schwannoma

## Objectives of the group

- To provide support to those who have been diagnosed with acoustic neuroma or other tumours affecting the cranial nerves
- To gather and provide information to patients and their families
- To increase awareness of acoustic neuroma
- To support ongoing research into the cause of acoustic neuroma and its treatment.

## Activities and services

- Provide opportunities for individuals with acoustic neuromas to communicate with others
- Respond to individual member's non-medical needs
- Provide information to the public about symptoms which may indicate a tumour
- Meetings are held three times a year at The Niche, 11 Aberdare Road Nedlands (next to Sir Charles Gardiner Hospital).

## Publications/resources

The parent association in Melbourne produces a quarterly newsletter. Booklets and information sheets are available to members.

# **Albinism Fellowship of Australia**

Head Office:  
PO Box 20729  
WORLD SQUARE NSW 2002

Phone: 1300 22 16 19

WA Contact: Marie  
Email: [albinismwa@albinismaustralia.org](mailto:albinismwa@albinismaustralia.org)

Website: [www.albinismaustralia.org](http://www.albinismaustralia.org)  
Facebook: [www.facebook.com/groups/albinismaustralia](http://www.facebook.com/groups/albinismaustralia)

## **Albinism**

Albinism is a rare genetic condition affecting about 1000 Australians (1 in 17,000). Albinism is inherited via a recessive gene that is passed from both the mother and the father of a person affected with the condition. It causes the body to be unable, or have limited ability, to produce melanin which is responsible for the colouring in skin, hair and eyes. Albinism can affect the eyes, skin and hair (oculocutaneous albinism) or only the eyes (ocular albinism). Most people with albinism have some degree of vision impairment, many will be considered 'legally blind'. The vision impairment is caused by the reduced ability or complete inability to produce melanin which is essential in the development and function of the eye. Melanin is essential for the development of the optic nerve, the retina, the fovea (part of the retina that is responsible for seeing fine detail) and is the main component that makes the eye colour in the iris.

## **Objectives of the group**

The Albinism Fellowship of Australia (AFA) is a national, non-profit organisation established in 2005. It is run by volunteers who have albinism or have been personally touched by albinism in some way. The AFA's key purpose is to provide support, education and fellowship to those with albinism, parents of children with albinism as well as their families and friends. While the albinism community in Australia is small, the AFA provides a united voice to encourage productive developments and support within business, government and media.

## **Activities and services**

- Biennial conference
- State based gatherings
- Parent pack for parents of children with albinism.

## **Publications/resources**

- Quarterly newsletter
- For additional resources, please visit their website [www.albinismaustralia.org](http://www.albinismaustralia.org)

# Alpha-1 Association of Australia

GPO Box 2100  
BRISBANE QLD 4001

Phone: 0410 108 104

Email: [emailus@alpha1.org.au](mailto:emailus@alpha1.org.au)

Website: [www.alpha1.org.au](http://www.alpha1.org.au)

Facebook: [www.facebook.com/Alpha1Australia](http://www.facebook.com/Alpha1Australia)

## Alpha-1 antitrypsin deficiency

Alpha-1 antitrypsin deficiency (Alpha-1) is a condition that is passed from parents to their children through genes. This condition may result in serious lung and/or liver disease at various ages in life. Alpha-1 antitrypsin is a protein that is produced mostly in the liver. Its primary function is to protect the lungs from neutrophil elastase. Neutrophil elastase is an enzyme that normally digests damaged or aging cells and bacteria in lung tissue to promote healing. If neutrophil elastase is left unchecked, it will also attack healthy lung tissue.

## Synonyms of alpha 1 antitrypsin deficiency

Alpha-1  
AATD  
A1AD

## Objectives of the group

The Alpha-1 Association of Australia exists to offer support and information to those affected by or interested in the genetically inherited condition Alpha-1 antitrypsin deficiency.

## Activities and services

- Patient communication and support
- Activism in the interest of promoting research and interest in Alpha-1 antitrypsin deficiency as it relates to Australians affected by or interested in the condition □  
Centralised information hub for Alpha-1 antitrypsin deficiency in Australia.

## Publications/resources

Booklet and posters are available on the website, together with links to discussion.

# **Androgen Insensitivity Syndrome Support Group Australia Inc**

PO Box 103  
COORPAROO QLD 4151

Phone: 0405 143 086

Email: [aissgaustralia@gmail.com](mailto:aissgaustralia@gmail.com)

Website: [www.aissga.org.au](http://www.aissga.org.au)

## **Androgen insensitivity syndrome**

Androgen Insensitivity Syndrome (AIS - formerly known as Testicular Feminising Syndrome) is a genetic condition whereby, due to a variation in the development of the reproductive system, there is a complete (CAIS) or partial (PAIS) inability to utilise testosterone. People with AIS are born with testes and 46XY (male) chromosomes.

## **Synonyms of androgen insensitivity syndrome**

Testicular feminising syndrome  
Intersex/DSD (Disorder of sex development)

## **Objectives of the group**

The AISSGA provides peer support, information and advocacy for intersex people and their families.

## **Activities and services**

- Peer support (telephone/email support at prearranged times)
- Advocacy
- Medical and media liaison
- Government department liaison
- Meetings
- The service is run by volunteers.

## **Publications/resources**

Brochures available via the website.

# Angelman Syndrome Association of WA

PO Box 7184  
APPLECROSS NORTH WA 6153

Contact Person: Leticia Grant, President  
Phone: 0408 445 975

Email: [president@angelmanwa.com.au](mailto:president@angelmanwa.com.au)  
Websites: [www.angelmanwa.com.au](http://www.angelmanwa.com.au) [www.angelmansyndrome.org](http://www.angelmansyndrome.org)

## Angelman syndrome

Angelman syndrome (AS) is a rare neuro-genetic condition caused by a deletion, or chromosome mutation, on chromosome 15. It is characterised by severe intellectual disability, absent speech, sleep disturbance, an unstable jerky gait, seizures and an unusually happy demeanour. The use of the term "Angel/Angels" is loosely applied to any person diagnosed with Angelman syndrome.

## Synonyms of Angelman syndrome

AS  
Happy puppet syndrome (obsolete)

## Objectives of the group

- To increase understanding of AS in the community
- To provide information to students, carers and allied health professionals about AS
- To support, inform, educate, foster research and advocate for families affected by AS.

## Publications/resources

A resource library on Angelman syndrome has been established, encompassing medical journals, family resource books and DVDs. Please contact us for further information.

## Arthritis & Osteoporosis WA

17 Lemnos Street  
SHENTON PARK WA 6008

PO Box 34  
WEMBLEY WA 6913

Phone: (08) 9388 2199  
Fax: (08) 9388 4488  
Country callers: 1800 011 041

Email: [general@arthritiswa.org.au](mailto:general@arthritiswa.org.au)  
Website: [www.arthritiswa.org.au](http://www.arthritiswa.org.au)  
Facebook: <https://www.facebook.com/ArthritisWA>

### Arthritis

There are more than 100 different types of arthritis, some of which have a genetic component. The most common forms of arthritis are osteoarthritis, rheumatoid arthritis, gout and ankylosing Spondylitis. Other conditions include, juvenile idiopathic arthritis (JIA), scleroderma, fibromyalgia, Ross River and Barmah Forrest virus, and osteoporosis.

### Synonyms of arthritis

Degenerative arthritis	Autoimmune disorder
Degenerative joint disease	Bacterial/viral arthritis
Atrophic arthritis	Septic arthritis
Rheumatism	Juvenile idiopathic arthritis
Autoimmune disease	

### Objective of the association

To reduce the incidence and disabling effects of arthritis, osteoporosis and related conditions.

### Activities and services

- Telephone Advisory Service: Information, services and support for individuals with arthritis, osteoporosis and related musculoskeletal conditions
- Education: Seminars and workshops for both the general public and health professionals
- Education and Self-Management Programs: Designed to equip people with the skills, knowledge and confidence to better manage and cope with their disease Programs include Osteoarthritis of Knee Program, Inflammatory Arthritis, Ankylosing Spondylitis and Challenge Your Arthritis
- Exercise Classes: Tai chi for Arthritis, Nordic Pole Walking, Pilates, Gentle Yoga, Osteoporosis exercises
- Support groups
- Hydrotherapy facilities
- Specialist Bookshop, Arthritis Today Magazine & E-News.

### Publications/resources

The Foundation has a range of information sheets and brochures available through their telephone advisory line (numbers above) or online [www.arthritiswa.org.au](http://www.arthritiswa.org.au)



# **Arthrogryposis Group (TAAG) Inc, The Australian**

56 Stevens Close  
WANG WAUK NSW 2423

Phone: 0417 803 573

Email: [berriga@bigpond.com](mailto:berriga@bigpond.com)

Website: [www.taag.org.au](http://www.taag.org.au)

## **Arthrogryposis multiplex congenita**

Arthrogryposis multiplex congenita (AMC) is a condition characterised by the presence of multiple joint contractures (contracture refers to a limited range of motion). AMC may exist in isolation or be part of a genetic condition and varies from mild to severe.

## **Synonyms of Arthrogryposis multiplex congenita**

Congenital multiple arthrogryposis  
Fibrous ankylosis of multiple joints  
Multiple congenital contractures  
Distal arthrogryposis  
Amyoplasia  
Whistling face syndrome

## **Disorder subdivisions**

Amyoplasia congenita  
Guerin-Stern syndrome  
Myopathic arthrogryposis multiplex congenita  
Neurogenic arthrogryposis multiplex congenita

## **Objectives of the group**

- Provide information and contact to assist affected individuals and their families
- Receive, publicise and disseminate information on arthrogryposis
- Liaise with support groups in other countries.

## **Activities and services**

Provide access to library with videos, books and pamphlets.

# **AusDoCC Inc (Australian Disorders of the Corpus Callosum)**

PO Box 533  
ALTONA VIC 3018

Email: [info@ausdoicc.org.au](mailto:info@ausdoicc.org.au)  
Website: [www.ausdoicc.org.au](http://www.ausdoicc.org.au)  
Facebook: [www.facebook.com/AusDoccInc](http://www.facebook.com/AusDoccInc)

## **Disorder of the corpus callosum**

AusDoCC is the key support group for Australian and New Zealand families affected by a congenital disorder of the corpus callosum. This means that they are born missing some or all of their corpus callosum, the major connecting structure between the two hemispheres of the brain. The impacts are far reaching and complex and include associated conditions such as autism, epilepsy, cerebral palsy, developmental delay, psychosocial dysfunction, cognitive impairment, anxiety and more.

Families who are affected by a disorder of the corpus callosum (DCC) are historically isolated, unsupported and frustrated. We are bringing together the world's leading scientists and health and education practitioners, with families from Australia and New Zealand, for a conference in Melbourne in May 2017. We welcome the attendance of your members at a cutting-edge opportunity to hear the best in the world, so that families affected by a DCC can be supported with effective management strategies. It is a disorder that affects 1:4000 live births.

## **Objectives of the group**

- Vision: Supporting individuals, families and caregivers affected by a disorder of the corpus callosum.
- Mission: Advocating, uniting, supporting.

## **Activities and services**

- Interactive, private Facebook group- Australian disorders of the corpus callosum AusDoicc
- Private Facebook for adults with DCC- Australian ACC adults
- 2 yearly conference with local and international presenters and professionals
- Services for families: State get togethers, research program.

## **ausEE Inc**

PO Box 9303  
PACIFIC PARADISE QLD 4564

Email: [sarah.gray@ausee.org](mailto:sarah.gray@ausee.org)  
Website: [www.ausee.org](http://www.ausee.org)  
Facebook: [www.facebook.com/ausEEInc/](http://www.facebook.com/ausEEInc/)

## **Eosinophilic Gastrointestinal Disorder (EGID)**

Eosinophilic gastrointestinal disorders (EGID) occur when eosinophils (pronounced ee-oh-sin-oh-fills), a type of white blood cell, are found in above-normal amounts within the gastrointestinal tract.

Eosinophils are important in your body's defence against parasitic infections (e.g. worms). However, they are also involved in allergy. In some individuals, eosinophils accumulate in the gut in response to food and/or airborne allergens and can cause inflammation and tissue damage.

## **Eosinophilic oesophagitis (EoE)**

Eosinophilic oesophagitis (EoE) is the most common type of Eosinophilic Gastrointestinal Disorder (EGID). The cause of EoE in some individuals appears to be due to an allergy to food(s) and/or aero-allergens.

The current estimated prevalence of EoE is 1–4 cases per 10,000 individuals and rising. People with EoE commonly have other allergic diseases such as asthma or eczema. EoE affects people of all ages, gender and ethnic backgrounds. In certain families, there may be an inherited (genetic) tendency. Males are more commonly affected than females.

## **Objectives of the group**

- To provide support and information to individuals and families affected by eosinophilic gastrointestinal disorders.
- To improve the quality of life for people with eosinophilic gastrointestinal disorders.
- To create greater public awareness and understanding of eosinophilic gastrointestinal disorders, their impact and appropriate management options.
- To help people recognise the symptoms of eosinophilic gastrointestinal disorders.
- To be a credible source of information to physicians and patients.
- To campaign and raise funds to enable further research to be conducted in Australia for eosinophilic gastrointestinal disorders.

## **Activities and services**

- Donations of hand knitted ausEE dolls to gastroenterology clinics in hospitals to be given to kids undergoing endoscopies or surgery for EGIS/EoE.

## **Publication and resources**

- Subscription to monthly eNewsletters via website
- Goal is to have 'Eosinophilic Gastrointestinal Disorders: What you should know' brochure available in all gastroenterology clinics and hospitals in Australia.
- Three published children's picture books about living with an eosinophilic disorder. The first 'Meet Arabella' follows the diagnosis journey and the second 'Being Henry' is about living with EoE. The third book 'Olivia and Kate' is about the siblings of Arabella and Henry.



# Australia Alopecia Areata Foundation

PO Box 5029  
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## Alopecia areata

Alopecia areata (AA) is hair loss from the scalp, eyebrows, eyelashes and the body. The condition occurs equally in males and females. It most commonly occurs in early childhood and late teens, but can occur at any age.

## Synonyms of alopecia areata

Hair loss

## Objectives of the group

- Support research to find a cure or acceptable treatment for AA
- Provide emotional support to those living with AA and their families
- Inform the public and create awareness of AA

## Activities and services

- Attend schools
- Run awareness programs
- Wigs for kids program
- Donate hair campaign
- Support group in each state
- Fund research
- Youth ambassadors
- Discussion forums
- Advocate with dermatologists.

## Publications/resources

Brochures and material are found on the website. Quarterly newsletter sent to those registered.

# **Australian Rare Chromosome Awareness Network (ARCAN)**

PO Box 809  
STANHOPE GARDENS NSW 2768

Phone: 0402 669 075

Email: [pjonas@arcan.org.au](mailto:pjonas@arcan.org.au)

Website: [www.arcan.org.au](http://www.arcan.org.au)

Facebook: <https://www.facebook.com/australianrarechromoawarenessgroup/>

## **Rare chromosomes**

The Network consists of families who are all affected by rare chromosome conditions. It hosts local events for families to meet one another and connect with those who understand. They aim to raise awareness for rare chromosome conditions and help others understand what children go through.

## **Objectives of the group**

ARCAN is a group of parents, who aim to help other parents and families. Network organisers have all walked the rare chromosome path and are at different stages in our journey. Through their experience and support they want to help other families who are also on this rare chromosome path. It can be so lonely when you have a rare chromosome diagnosis and a doctor tells you that your child is only 1 of 5 children in the world with that chromosome anomaly.

## **Activities and services**

- Local events- networking
- Emotional support.

## **Publications/resources**

- Leaflets and flyers
- Information packs to be created.

# **Batten Disease Support and Research Association (Australian Chapter)**

9 Norton Avenue  
KILLARNEY VALE NSW 2261

Phone: (02) 4334 5785

Email: [partner@battens.org.au](mailto:partner@battens.org.au)

Website: [www.battens.org.au](http://www.battens.org.au)

Facebook: [www.facebook.com/AustralianBDSRA](http://www.facebook.com/AustralianBDSRA)

## **Batten disease**

Batten disease or neuronal ceroid lipofuscinoses (NCL) is an inherited condition of the nervous system that usually manifests itself in childhood. Early symptoms of Batten disease usually appear in childhood when parents or doctors may notice a child begin to develop vision problems or seizures. In some cases, the early signs are subtle, taking the form of personality and behaviour changes, delayed learning, and clumsiness or stumbling.

Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Children become totally disabled and eventually die. Batten disease is not contagious nor, at this time, preventable. To date it has always been fatal.

## **Synonyms of Batten disease**

Spielmeyer-Sjogren disease

Vogt-Spielmeyer disease

Neuronal ceroid lipofuscinosis, juvenile

## **Objectives of the group**

Provides support to family, friends and carers of children with Batten disease. Also supports research into Batten disease.

## **Activities and services**

- Phone support
- Email support
- Information packs
- Newsletters
- Family meetings
- Lending library of resources.

## **Publications/resources**

Extensive publications available books and DVDs specific to Batten disease – view the list here [www.battens.org.au/publication-library-list](http://www.battens.org.au/publication-library-list)

# **Bertolotti's Syndrome Support**

Phone: 0424 327 231

Email: [aidanofl@hotmail.com](mailto:aidanofl@hotmail.com)

Facebook: [www.facebook.com/groups/bertolottisydney](http://www.facebook.com/groups/bertolottisydney)

## **Bertolotti's syndrome**

Bertolotti's syndrome is a rare disease relating to an enlargement of the transverse process of a vertebra leading to chronic back pain. This is a congenital disease and onset of the pain is usually delayed until the 30s. It can also have effects on the sacroiliac joint leading to misdiagnosis of back pain.

## **Objectives of the group**

There is a lack of information on Bertolotti's, both through internet searches and from health professionals. This group seeks to raise awareness and provide information for those living with this disease. The support group is run by an individual who has been through the diagnosis process. The aim is to make life easier for those living with this syndrome by providing a support group.

## **Activities and services**

- Facebook group
- Support discussions
- Daily messages
- Information on the disease
- Promotion of 'talking'
- Monthly meetings/Skype meetings
- Photo upload promotion
- Sharing stories and experiences
- Exercise promotion (warm water courses).



## **Bowel Group for Kids (BGK)**

PO Box 40  
OAKDALE NSW 2570

Email: [enquiries@bgk.org.au](mailto:enquiries@bgk.org.au)  
Website: [www.bgk.org.au](http://www.bgk.org.au)  
Facebook: <https://www.facebook.com/pages/Bowel-Group-for-Kids-Australia/220001938162646>

### **Bowel conditions**

Hirschsprung's disease (HD) and Anorectal Malformation Hirschsprung's disease (HSCR) and Anorectal Malformation (ARM) are rare congenital (at birth) conditions that affect one in every 5000 live births. HD is a disorder of the abdomen that occurs when part or all of the large intestine or parts of the gastrointestinal tract have no ganglion cells and therefore cannot function. An imperforate anus is a defect that occurs during the fifth to seventh weeks of foetal development. With these defects, the anus (opening at the end of the large intestine through which stool passes) and the rectum (area of the large intestine just above the anus) do not develop properly.

### **Objectives of the group**

BGK works to raise awareness of **these** conditions. They also aim to provide education, information and support to those with an interest in babies born with congenital bowel disorders, parents, carers and health professionals.

### **Activities and services**

- Online access to education material
- Access to private Facebook site
- National conferences
- Educational pamphlets for schools
- Newsletters
- Phone and in hospital support as required.

### **Publications/resources**

- Newsletters
- Pamphlets
- Articles
- Living with Hirschsprung's disease handbook.

# **Cardio Facio-Cutaneous (CFC) International**

Phone: (08) 9582 2913  
Email: pn543931@bigpond.net.au

Website: [www.cfcsyndrome.org](http://www.cfcsyndrome.org)  
Facebook: [www.facebook.com/groups/cfcangelswithattitude/](http://www.facebook.com/groups/cfcangelswithattitude/)

## **Cardio facio-cutaneous syndrome**

CFC syndrome is a rare genetic condition that typically affects the heart (cardio-), facial features (facio) and skin (cutaneous). It is seen with equal frequency in males and females and across all ethnic groups.

Children with CFC syndrome may have certain features that suggest the diagnosis, such as; relatively large head size, down-slanting eyes, sparse eyebrows, curly hair, areas of thickened or scaly skin, and small stature. Most will also have a heart defect.

Four different genes have been found to be associated with CFC syndrome (BRAF, MEK1, MEK2 and KRAS); however, most individuals with CFC syndrome have a new sporadic mutation in the BRAF gene.

Molecular genetic (DNA) testing for mutations in all of these genes is clinically available. There are only 3 known CFC syndrome cases in Western Australia.

## **Synonyms of cardio facio-cutaneous syndrome**

Cardio-facial-cutaneous syndrome  
CFC syndrome  
Facio-cardio-cutaneous syndrome

## **Objectives of the group**

We offer support for parents, carers, medical professionals etc. who are living with a person with CFC syndrome linking them with sites and support groups. We also can now offer parents of Costello syndrome and Noonan syndrome links to supportive networks, phone numbers or contact as these syndromes have been found to be linked with CFC syndrome.

## **Activities and services**

- Phone calls
- Personal support
- Medical updates
- Current conference for CFC updates
- Website links to services available
- Parent support
- Facebook support
- Online new strategies for living with a CFC child or adult.

# CDH (Congenital Diaphragmatic Hernia) Australia

PO Box 1034  
NORTH GEELONG, VIC 3215

Support Services Referrals: 1800 149 562  
Email: [information@cdh.org.au](mailto:information@cdh.org.au)  
Website: [www.cdh.org.au](http://www.cdh.org.au)  
Facebook: <https://www.facebook.com/CDHAustralia/>

## Congenital Diaphragmatic Hernia (CDH)

CDH is a defect in the diaphragm, which results in abdominal organs being displaced into the chest. These tummy organs take up room that the lungs should have to grow, meaning that CDH babies are born with severely underdeveloped lungs.

CDH occurs in around 1:2500 births. To put it into context, that's 2-3 families being told each week that their child has CDH, or 125 families every year. Half of these children will pass away in their first year.

Every CDH child faces their battle in NICU; of the children who survive, many face a raft of ongoing health complications. These are varied, and range in severity. Examples are: ongoing gastroesophageal reflux; failure to thrive & growth problems; scoliosis & pectus excavatum; bowel obstructions & gastrointestinal disturbances; developmental disorders. Our children often require ongoing medical care, including home oxygen or tracheostomies, PEG feeding, nutritional supplements, ongoing medication.

A generation ago, 90% of CDH babies passed away; the survivors coming through now are the first children with more complex CDH presentations to survive; therefore, management of their ongoing health can be a case of "trial and error". Adding to the challenges, CDH is not well known across all sectors of the medical community, and being considered a "rare" defect, families often face delays in receiving an accurate diagnosis and challenges in accessing appropriate medical care.

## What Does CDH Australia Do?

CDH Australia is a grassroots national health-promotion charity which supports the families of children diagnosed with the birth defect congenital diaphragmatic hernia (CDH). *One day we hope to be redundant; we see a future where CDH does not exist. Until then, we aim to empower families to survive and thrive, and live their best lives.*

CDH Australia was established in 1999; we have supported over a thousand families since our inception. In addition to support, we seek to establish relationships with and between medical professionals, and to support CDH research. Underpinning this is the need to raise awareness of the condition and of our organisation. We believe greater community and political awareness will provide us a firmer footing from which to advocate for our families.

CDH Australia has contact with around 75% of families who are diagnosed with CDH. The majority of these are located in Australia, although we also have a small community in New Zealand.

## Objectives of the group

CDH Australia is a national not-for-profit group to:

- Provide support to families effected by CDH
- Increase awareness of CDH
- Help fund research into the causes of, and treatments for CDH.

### **Activities and services**

- Personal & group support
- Information booklets
- Parent to parent matching (helping families who have experienced similar
- Annual well-being and awareness activities
- Website packed with facts and support.

### **Publications/resources**

- “Expecting A CDH Baby” booklet
- Video: What is CDH?
- CDH Awareness videos
- Fact Sheets

# Charcot-Marie-Tooth Association Australia – WA Division

## WA Coordinators

Phone: Chris - 0407 214 250  
Email: Chris\_murphy@iinet.com.au

Phone: Angela - 6394 1116  
Email: angelacorfe@hotmail.com

## National Office

Phone: (02) 9767 5105  
Fax: (02) 9767 5167

Email: [cmtaa2@cmt.org.au](mailto:cmtaa2@cmt.org.au)  
Website: [www.cmt.org.au](http://www.cmt.org.au)  
Facebook: <https://www.facebook.com/CMTAA-604686846216785/>

## Charcot-Marie-Tooth

CMT or Charcot-Marie-Tooth disease (named after the 3 medical professionals who first identified CMT), is also known as Hereditary Motor and Sensory Neuropathy (HMSN). It is a common but frequently undiagnosed condition. CMT is not a contagious disease but is an inherited neurological disease and in some families has a 50% chance of being passed onto other generations. Research has indicated that as many as 1 in 2,500 people in Australia could have CMT. CMT is not life threatening although people with CMT have it for life. Motor and Sensory Neuropathy (HMSN). It is a common but frequently undiagnosed condition. CMT is not a contagious disease but is an inherited neurological disease and in some families has a 50% chance of being passed onto other generations. Research has indicated that as many as 1 in 2,500 people in Australia could have CMT. CMT is not life threatening although people with CMT have it for life.

## Synonyms of Charcot-Marie-Tooth disease

- Hereditary motor and sensory neuropathy
- Peroneal muscular atrophy

## Objectives of the group

- Provide support through contact with people with CMT
- Distribution of information related to the welfare of people with CMT
- Promote the development of facilities and services for people with CMT
- Actively support CMT research.

## Activities and services

- Conduct information seminars
- Provide an informative website
- Send regular newsletters to members
- Provide additional information regarding CMT.

## Publications/resources

- Newsletter
- Videos of seminars.

# **CHARGE Syndrome Association of Australasia**

WA Coordinator: Sharon Barrey Grassick

Phone: 0410 543 014

Email: sbgrassick@gmail.com

Website: www.chargesyndrome.org.au

## **CHARGE syndrome**

CHARGE is an acronym for:

- C Coloboma: incomplete development of the eye
- H Heart anomalies
- A Atresia of the choanae: a blockage of the nasal passage
- R Restriction of growth and/or development
- G Genitourinary system anomalies
- E Ear anomalies including hearing loss.

## **Synonyms of CHARGE syndrome**

It is also known as the four C's: Coloboma

Characteristic Ear abnormalities

Choanal Atresia

Cranial nerve abnormalities

## **Objectives of the group**

- To provide support to families via networking with others affected by this syndrome
- To raise community awareness
- Provide respite to families.

## **Activities and services**

- Conferences are held every 2 years
- A newsletter is produced quarterly
- Online forum where you may to ask a question or seek information about issues regarding CHARGE Syndrome.

## **Publications/resources**

An information package is available to families of children newly diagnosed with CHARGE on request. Parent and professional representatives are available to discuss CHARGE with any interested persons.

# **Chiari and Syringomyelia Australia**

Incorporating: Tethered Cord, Ehlers Danlos and Craniocervical Instability

Contact: Kirstin Maltby  
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Email: [chiariaustralia@gmail.com](mailto:chiariaustralia@gmail.com)  
Website: [www.chiariaustralia.com.au](http://www.chiariaustralia.com.au)  
Facebook: [www.facebook.com/groups/ChiariSyringomyeliaAustralia/](http://www.facebook.com/groups/ChiariSyringomyeliaAustralia/)

## **Chiari**

Arnold Chiari (Chiari) is a congenital malformation involving the cerebellum, occurring when the section of the skull containing the cerebellum is too small or deformed. This causes the cerebellar tonsils in Chiari I and additionally the brainstem in Chiari II to herniate outside of the skull into the spinal canal. It can also put pressure on the brain tissue that is herniated and block the flow of the cerebrospinal fluid (CSF). Chiari III and Chiari IV are usually fatal in-utero or shortly after birth due to brain developing outside of the skull (Chiari III) or the hindbrain failing to develop (Chiari IV). Due to the blockage of CSF and pressure on parts of the brain this can result in neurological signs and symptoms and possibly secondary conditions like intracranial hypertension, hydrocephalus and syringomyelia.

## **Syringomyelia**

Syringomyelia occurs when a blockage forces the cerebrospinal fluid (CFS) to enter the interior of the spinal cord to create a cyst called a Syrinx. This cyst expands out with time crushing the nerves causing neurological symptoms and pain. There are two major forms of syringomyelia including communicating syringomyelia (as a result of Chiari Malformation) or non-communicating as a complication of trauma, meningitis, haemorrhage, a tumour or arachnoiditis (post-traumatic syringomyelia).

## **Objectives of the group**

- To bring together people with Chiari and Syringomyelia
- Have an area with Australian specific information and support for these conditions
- To offer support and information to those diagnosed and their loved ones\
- To raise awareness and support research
- To become a not-for-profit charity.

## **Activities and services**

Support group (online), support via email, information, awareness products, and literature packets.

# Children's Tumour Foundation of Australia (NF Australia)

Suite 2  
451 Lyons Road West  
FIVE DOCK NSW 2046

PO Box 454  
FIVE DOCK NSW 2046

Phone: (02) 9713 6111  
Fax: (02) 9712 8524  
Email: [info@nfaa.org.au](mailto:info@nfaa.org.au)  
Website: [www.ctf.org.au](http://www.ctf.org.au)  
Facebook: [www.facebook.com/CTFAus](http://www.facebook.com/CTFAus)

## Neurofibromatosis

Neurofibromatosis, or NF, is the term for three distinct genetic conditions - NF1, NF2, and Schwannomatosis - that cause tumours to grow on nerves throughout the body. Occurring in 1 in 2,500 births, NF can lead to blindness, deafness, bone deformities, learning disorders, chronic pain and more. There is no cure.

## Condition Synonyms

Neurofibromatosis Type 1 (NF1)  
Schwannomatosis

Neurofibromatosis Type 2 (NF2)

## Objectives of the group

CTF Australia is the peak national body dedicated to raising funds, awareness and support for NF and NF sufferers. The organisation:

- Provides support to adults, adolescents, children and their families affected by NF
- Provides funding for vital support services and peer-reviewed medical research conducted in Australia to expedite the rate of discovery and the availability of new treatment options and ultimately a cure for NF
- Develops and provides key information resources about NF to patients, their families and the medical community to assist with diagnosis and managing life with NF.

## Activities and services

- Annual NF family camps
- Access to dedicated Support Coordinators in Sydney and Melbourne (with a view to expand the National Support Services Network by placing qualified support coordinators in each state based at major hospitals)
- Access to NF community networking, including online forum and local NF community support groups and contacts
- Information resources available on request.

## Publications/resources

- Information sheets including on: NF1, NF2, optic pathway Gliomas, with more currently in development.
- Translated information about NF1, the most common form of NF (Mandarin, Vietnamese and Arabic)
- Online fundraising portals- registered charity to make fundraising simple and available Australia-wide.



# Chromosome 18 Registry and Research Society Australia

Australasian Coordinator: Marlene  
Phone: (02) 9580 5707  
Email: admin@chromosome18.org.au  
Website: www.chromosome18.org

WA Contact: Veronika  
Phone: (08) 9409 9854

## Chromosome 18

The main conditions are 18q deletion, 18p deletion, trisomy18, tetrasomy 18p, Ring 18 and Pitt-Hopkins Syndrome (with a mutated or deleted TCF4 gene). There are also many unique rearrangements that include translocations, mosaicism, duplications and inversions. Each of the conditions of chromosome 18 has a wide variety of characteristics and severity.

Please refer to our website [www.chromosome18.org](http://www.chromosome18.org) for more information.

## Synonyms of chromosome 18 disorders

18p deletion syndrome	Chromosome 18 Long arm deletion syndrome
18p syndrome	Del (18q) syndrome
Del (18p) syndrome	Monosomy 18q syndrome
Monosomy 18p syndrome	Ring 18
Short arm 18 deletion syndrome	Ring chromosome 18
18q deletion syndrome	Tetrasomy short arm of chromosome 18
18q syndrome	

## Disorder subdivisions

Chromosome 18, monosomy 18p  
Chromosome 18q syndrome  
Chromosome 18 ring  
Chromosome 18 tetrasomy 18p

## Objectives of the group

The Chromosome 18 Registry & Research Society is a not-for-profit organisation. The support group is a contact point for families and individuals that are affected by a chromosome 18 abnormality. We also support research into chromosome 18 abnormalities. Our mission is to help individuals with chromosome 18 abnormalities overcome obstacles to lead happy, healthy and productive lives.

## Activities and services

- Conferences and meetings
- Worldwide parent support network, Facebook – USA and Australasia
- List services for the individual syndrome groups
- Education and public advocacy
- Research through the Chromosome 18 Research Centre, Texas.

## Publications/resources

See [www.chromosome18.org](http://www.chromosome18.org) for publications and resources.

# **CJD(Creutzfeldt-Jakob disease) Support Group Network**

13 Araluen Place  
GLENHAVEN NSW 2156

Phone: 1800 052 466

Email: [contactus@cjdsupport.org.au](mailto:contactus@cjdsupport.org.au)

Website: [www.cjdsupport.org.au](http://www.cjdsupport.org.au)

Facebook: [www.facebook.com/groups/879976008687331](https://www.facebook.com/groups/879976008687331)

## **Creutzfeldt-Jakob disease**

CJD is a rare, fatal and degenerative brain disease of the central nervous system in humans. CJD is one of a group of diseases known as Transmissible Spongiform Encephalopathies (TSE) or prion disease.

## **CJD subdivisions**

Sporadic CJD

Genetic CJD

Acquired forms of prion disease

## **Objectives of the group**

The CJD Support Group Network (CJDSGN) offers support to all Australians affected by Creutzfeldt-Jakob disease (CJD) or other prion diseases.

The CJDSGN provides information and support to families who are caring for, or coming to terms with the loss of a loved one to CJD. They also assist people who are 'at increased risk of CJD' by promoting an environment in the health care setting where patients feel comfortable and confident of receiving equity of care when disclosing they're at risk of CJD status.

## **Activities and services**

- Provides resources to individuals, families and health care professionals to promote knowledge and awareness of CJD
- Provide a national education program free of charge and is committed to an advocacy role on behalf of individuals and families affected by CJD
- Networks with health care professionals, links family members together for mutual support and assists families keen to raise much needed funds for research into CJD in Australia.

## **Publications/resources**

- Information package for CJD families
- Information package for health care professionals'
- DVD- Understanding CJD DVD- What is CJD?
- Brochures- What is CJD?
- Handbook for Health Care Professionals- Patient Care
- Inherited prion disease fact sheet- Frequently asked questions.

# CleftPALS WA

C/- Variety WA  
PO Box 669  
VICTORIA PARK WA 6979

Phone: (08) 9418 3842 or (08) 9407 8441

Email: [membership@cleftpalswa.org.au](mailto:membership@cleftpalswa.org.au)

Website: [www.cleftpalswa.org.au](http://www.cleftpalswa.org.au)

Facebook: [www.facebook.com/groups/cleftpalswa/](http://www.facebook.com/groups/cleftpalswa/)

## Cleft

Early in pregnancy at about 6-8 weeks gestation the usual fusing, or joining up of tissues of the lip and/or palate does not occur, leaving an opening or "cleft".

A cleft sometimes occurs in the lip or the palate, which can be either the soft or hard palate or both, or the lip and palate together. It can occur on one side of the lip or palate, 'uni-lateral cleft', or on both sides, a 'bi-lateral cleft'.

A cleft lip may sometimes be called a hare lip. This term dates back to medieval times when it was believed a mother gave birth to a baby with a cleft lip because a hare had jumped across her path. 'Cleft lip' is a more meaningful term to describe the condition today.

## Synonyms

Cleft palate

Cleft lip

Cleft lip and palate

Hare lip

## Objectives of the group

- Provide information and support to families, individuals and professionals dealing with a cleft condition in Australia and worldwide
- Supply of feeding and other equipment

## Publications/resources

- Quarterly newsletter for both members and professionals containing articles by specialists, from other parents sharing experiences and ideas and information regarding morning teas and other events.
- Facebook group- CleftPALS WA

# Coeliac Western Australia

Cockburn Integrated Health  
Wentworth Parade, SUCCESS WA 6954

Phone: (08) 9494 3734

Email: [wa@coeliac.org.au](mailto:wa@coeliac.org.au)

Website: <https://wa.coeliac.org.au>

Facebook: <https://www.facebook.com/coeliacwa/>

## Coeliac disease

In people with coeliac disease the immune system reacts abnormally to gluten (a protein found in wheat, rye, barley and oats), causing small bowel damage. The tiny, finger-like projections which line the bowel (villi) become inflamed and flattened.

The surface area of the bowel available for nutrient absorption is markedly reduced which can lead to various gastrointestinal and malabsorptive symptoms. A number of serious health consequences can result if the condition is not diagnosed and treated properly.

You must be born with the genetic predisposition to develop coeliac disease. The most important genes associated with susceptibility to coeliac disease are HLA DQ2 and HLA DQ8. Either one or both of these genes are present in virtually every person with coeliac disease.

Only 1 in 30 people (approximately) with one or both genes will get coeliac disease. Environmental factors play an important role in triggering coeliac disease in infancy, childhood or later in life. Some of the symptoms of coeliac disease include:

- Fatigue, weakness and lethargy
- Anaemia
- Diarrhoea
- Constipation
- Cramping and bloating
- Nausea and vomiting
- Flatulence and abdominal distention.

## Objectives of the group

Operating since 1979, Coeliac Western Australia's purpose is to provide information, support and to advocate on behalf of people with, or affected by:

- Dermatitis herpetiformis
- Coeliac disease
- Medically requiring a gluten free diet.

## Activities and services

- Support members by telephone help line and email, workshops, newsletters and social media.
- Produce, scrutinise and disseminate current and accurate information.
- Promote coeliac awareness week (13-20 March annually).

- Coordinate an annual Expo.
- Provide dietetic consultations with Accredited Practicing Dietician, who is an expert in coeliac disease.

### **Publications/resources**

Resources available for purchase are listed on our website.

# ConnecTeD Foundation

PO Box 283  
WESTMEAD NSW 2145  
Phone: 1300 785 163

Email: [info@connectedfoundation.org.au](mailto:info@connectedfoundation.org.au)  
Website: [www.connectedfoundation.org.au](http://www.connectedfoundation.org.au)  
Facebook: [www.facebook.com/connectedfoundation](http://www.facebook.com/connectedfoundation)

## Connective tissue dysplasias

Connective tissue dysplasia encompasses a wide range of disorders. These disorders are caused by defects in the connective tissues such as bone, ligaments, tendons and skin.

Children with these disorders may have:

- Too much joint movement (hypermobility)
- Not enough joint movement (joint contractures - hypo mobility)
- Excessive laxity (looseness) of skin or fragile bones, skin, ligaments etc.
- Short stature
- Excessively tall stature.

The disorders can be variable because many affected people have a mixture of these different symptoms. Most connective tissue dysplasia's follow common patterns of inheritance. These patterns can help genetic counsellors to provide families with information about inheritance in their family.

## Objectives of the group

Supporting all connective tissue disorders including Ehlers-Danlos Syndrome, Loey's-Dietz Syndrome, Marfan's Syndrome, Mucopolysaccharidoses, Osteochondrodysplasias, Osteogenesis Imperfecta, Bone dysplasias/Chondrodysplasias, Arthrogyrosis and related disorders.

Objectives of the group are to provide support, education and awareness for connective tissue disorders

To fundraise for research and to support centres of excellence in Australia concerning connective tissue dysplasias.

## Activities and services

- Support networks
- Education for professionals
- Education for non-professionals, families and carers
- Adolescent and transition workshops.

## Publications/resources

All available on the website: [www.connectedfoundation.org.au](http://www.connectedfoundation.org.au)

# **Cornelia de Lange Syndrome Support Group**

Contact the World Federation of CdLS Support Groups

Name: Jenny  
Phone: 0407 030 269

## **Cornelia de Lange**

Cornelia de Lange is a genetic syndrome characterised by distinctive elf like facial features in association with physical, intellectual and mental retardation.

## **Synonyms of Cornelia de Lange**

Amsterdam syndrome  
Brachmann-de Lange syndrome  
de Lange syndrome

## **Objectives of the group**

- To support the families of children with Cornelia de Lange
- To support research to identify the genetic cause of Cornelia de Lange
- To promote community awareness of Cornelia de Lange.

## **Activities and services**

- Annual national general meeting
- Annual picnic in Sydney in January
- Quarterly conference hook-ups
- E-mail service
- Medical question and information service through links in website addresses as follows: [www.cdlsusa.org/national\\_groups](http://www.cdlsusa.org/national_groups)

## **Publications/resources**

The Group has a variety of brochures available to the community and health professionals. They also provide a quarterly newsletter KIT (Keeping In Touch) and an international bulletin.

# **Cri du Chat Support Group of Australia**

WA Parent Contact: Nadine

Phone: 0434 744 545

National: (03) 9775 9962

Email: [info@criduchat.asn.au](mailto:info@criduchat.asn.au)

Website: [www.criduchat.asn.au](http://www.criduchat.asn.au)

Facebook: [www.facebook.com/CriDuChatSupportGroupOfAustraliaInc?fref=ts](https://www.facebook.com/CriDuChatSupportGroupOfAustraliaInc?fref=ts)

## **Cri du chat syndrome**

Cri du chat syndrome results from the loss or deletion of a significant portion of the genetic material from the short arm of one of the pair of number five chromosomes. It is a relatively rare genetic condition with an estimated incidence of between around 1 in 25,000 to 1 in 50,000 births. There are more children being diagnosed now that genetic testing is carried out more frequently and is more accurate. The incidence appears to be the same in most countries, ethnic groups and regions. To date, there is no single environmental factor implicated in the incidence of this syndrome.

The most distinctive characteristic, and the one for which the syndrome was originally named in 1963 by geneticist Jerome Lejeune, is the distinctive high pitched, monotone, cat-like cry. "Cri du chat" is French for "cat's cry". The cry is thought to be the result of structural abnormality and low muscle tone. Although the voice will naturally lower as the child grows, the characteristic high pitch often persists into adulthood. In addition to the cry, there are a number of distinguishing characteristics present in infancy which aid in recognition of the syndrome. Not every child will have every feature. Those only mildly affected may have very few or they may be less obvious. More information about the syndrome is available at [www.criduchat.asn.au](http://www.criduchat.asn.au).

## **Synonyms of Cri du chat (CDC)**

5p minus (5p-)

Lejeune's syndrome

## **Objectives of the group**

The aim of this group is to provide information, friendship and support to those who have a family member or friend with Cri du chat syndrome or other anomaly of chromosome five. Membership is open to anyone with an interest in the syndrome, however our specific focus is on supporting families from Australia, New Zealand and the wider southern hemisphere.

## **Publications/resources**

- Website with related links/publications
- Facebook Page.



# **Cushing's Disease Support**

Contact: Sylvia  
Phone: (08) 9402 4394  
Email: sylvia17@westnet.com.au

## **Cushing's disease**

Cushing's disease (hypercortisolism or hyperadrenocorticism) is a hormonal disorder caused by prolonged exposure of the body's tissues to the hormone cortisol.

Many people suffer the symptoms of Cushing's Disease (CD) because they take steroids for asthma, , lupus and other inflammatory diseases or for immunosuppression after transplantation. Others develop CD because of over production of cortisol by the body due to a tumour on the pituitary or adrenal glands or elsewhere on the body.

Symptoms of this condition are weight gain, reddening of the face and neck, excessive body and facial hair, raised blood pressure, raised blood sugar levels, the loss of mineral from the bone (causing osteoporosis, brittle bones) and severe fatigue.

Other symptoms include excessive sweating and sometimes emotional and mental changes. This condition is treatable and individuals can return to good health with treatment.

## **Synonyms of Cushing's disease**

Cushing's syndrome  
Adrenal cortex adenoma  
Adrenal hyperfunction resulting from adrenocorticotrophic hormone (ACTH) excess  
Ectopic ACTH syndrome

## **Objectives of the group**

- To provide emotional and psychological support for individuals with CD
- To provide support on a one to one basis
- To ease suffering due to CD
- To provide education from firsthand experience.

## **Activities and services**

The group operates via a telephone and email support network. It provides an opportunity for newly diagnosed individuals to make contact with other people who have experienced CD. It provides a point of contact for people affected by post-Cushing's syndrome.

## **Other support**

The Australian Pituitary Foundation is a national peer-support charity for people with pituitary conditions (including CD), their families and those who care for them.

Through social interaction, networking, education and hands on workshops we provide practical support and information in addition to promoting pituitary awareness throughout Australia.

Phone: 1300 331 807

Email: [support@pituitary.asn.au](mailto:support@pituitary.asn.au)

Website: [www.pituitary.asn.au](http://www.pituitary.asn.au)

# Cystic Fibrosis

Cystic Fibrosis WA  
The Niche – Suite C  
11 Aberdare Road  
NEDLANDS WA 6009

PO Box 959  
NEDLANDS WA 6909

Phone: (08) 6457 7333

Fax: (08) 6457 7344

Email: [info@cfwa.org.au](mailto:info@cfwa.org.au)

Website: [www.cysticfibrosis.org.au/wa](http://www.cysticfibrosis.org.au/wa)

Facebook: [www.facebook.com/CysticFibrosisWA](http://www.facebook.com/CysticFibrosisWA)

## Cystic Fibrosis

Cystic Fibrosis (CF) is an inherited recessive genetic condition, which affects the lungs, digestive system and the sweat glands. In CF the body produces thick, sticky mucus, which prevents digestive enzymes reaching the food in the digestive system. Mucus collects in the lungs, clogging the airway and trapping bacteria which can cause infection. It is the most common recessive genetic condition affecting Australians.

## Objectives of the group

- To contribute to the social, physical and emotional wellbeing of those individuals and families affected by CF
- To assist in the promotion of research which will improve the lives of those with CF or find a cure for CF.

## Activities and services

Cystic Fibrosis WA (CFWA) offers support through a comprehensive home care program that includes:

- Home Support Service: regular visits by a CFWA home care worker to assist with airway clearance and respite
- Transplant support
- Education and information
- Recreation programs
- Counselling
- Coffee mornings for carers, families and individuals with CF
- CFWA is also involved with advocacy and lobbying for both individuals and the wider CF community
- Equipment is available for loan to members.

## Publications/resources

- CFWA provides books, videos, brochures and booklets for the community and health professionals. A listing of printed material (pamphlets and brochures) is available on the website; library publications are listed on the online library at [www.cfwalibrary.org.au](http://www.cfwalibrary.org.au). Library publications are available for loan free of charge to members
- CFWA publishes a quarterly newsletter, RED Magazine, as well as a fortnightly e-mail, RED E-news.

# **Cystinosis Support Group, Australian**

Contact: Sue

Phone: (08) 9531 2135 or 0407 196 685

Email: [cystinosis.australia@bigpond.com](mailto:cystinosis.australia@bigpond.com)

Website: [www.cystinosis.com.au](http://www.cystinosis.com.au)

## **Cystinosis**

Cystinosis is an inherited (autosomal recessive) metabolic disease characterised by an abnormal accumulation of amino acid cystine in various organs of the body such as the kidney, eyes, muscles, pancreas and brain. Different organs are affected at different stages.

## **Synonyms of Cystinosis**

Intermediate cystinosis

Nephropathic cystinosis

Non-nephropathic cystinosis

## **Objectives of the group**

To provide support, information and contact with families in Australia and New Zealand that are affected by cystinosis.

## **Activities and services**

- Provide support to newly diagnosed families and (with permission) put them in touch with others in a similar situation within their state
- To assist families in contacting doctors in other states that know about cystinosis if they should travel and need medical assistance
- Hold conferences and gatherings where families and doctors can discuss cystinosis
- Raise funds to support families affected by cystinosis.

## **Publications/resources**

The group can provide information and resources to assist families affected by cystinosis.

# **Deafblind West Australians (Formerly Usher Syndrome Support Group of WA)**

11 Kitchener Ave  
BURSWOOD WA 6100

Phone: (08) 9473 5462

Email: [Karen.wickham@senses.org.au](mailto:Karen.wickham@senses.org.au)

Website: [www.dbwa.org.au](http://www.dbwa.org.au)

## **Usher syndrome**

Usher syndrome is a genetic condition which causes hearing loss from birth and progressive loss of sight due to retinitis pigmentosa (RP), which causes degeneration of the retina. Often the first symptom of RP is night blindness, followed by narrowing side vision leading to what is called "tunnel vision". The condition follows autosomal recessive inheritance.

## **Objectives of the group**

The Usher Syndrome Support Group was established by Senses Australia in 2007 to provide support for people who have Usher syndrome, their families, carers and friends. The group meets regularly to provide information, education and recreation opportunities and to establish fun supportive networks for people with Usher syndrome.

## **Activities and services**

Group activities include:

- Water skiing
- Canoeing
- Bushwalking/rock climbing
- Educational workshops
- Group therapy
- Archery
- Dinners/social networking.

# **DEBRA Australia (Dystrophic Epidermolysis Bullosa Research Association)**

PO Box 226  
PITTSWORTH QLD 4356

Phone: (07) 4693 7003

Email: [secretary@debra.org.au](mailto:secretary@debra.org.au)  
Website: [www.debra.org.au](http://www.debra.org.au)  
Facebook: [www.facebook.com/debra.aust](http://www.facebook.com/debra.aust)

## **Epidermolysis bullosa (EB)**

Rare genetic skin blistering condition affecting 1,000 people in Australia. Blisters can occur internally and externally leading to a lifetime of painful dressing regimes and secondary complications.

## **Synonyms of epidermolysis bullosa**

Epidermdysis bullosa (EB)

## **Objectives of the group**

DEBRA: Dystrophic Epidermolysis Bullosa Research Association.

- Support and educate families living with EB
- Provide advocacy
- Educate health professionals
- Support EB research to ultimately one day find a cure
- Working collaboratively with other DEBRA organisations on an international basis to improve care and quality of life outcomes for families living with EB
- Create awareness and fundraise to carry out programs in place.

## **Activities and services**

- Family support program
- EB nurse program
- Biennial EB camp
- Annual health professionals EB conference
- Advocacy

## **Publications/resources**

What is EB brochure available online [www.debra.org.au](http://www.debra.org.au)

## **Dementia Australia**

155 Oak Street  
PARKVILLE VIC 3052

Phone: 1800 100 500

Email: [helpline.nat@dementia.org.au](mailto:helpline.nat@dementia.org.au)

Website: <https://www.dementia.org.au/>

Facebook: <https://www.facebook.com/DementiaAustralia/>

### **Dementia**

Dementia Australia is the national peak body and charity for people of all ages, living with all forms of dementia, their families and carers. It provides advocacy, support services, education and information to all Australians impacted by dementia.

Dementia Australia represents more than 436,000 Australians living with dementia and the almost 1.5 million Australians involved in their care.

### **Synonyms of Dementia**

Pre-senile dementia

### **Activities and services**

- Dementia Australia offers a range of support services for people living with dementia and their families and carers and professional education options for people working with people affected by dementia.

#### *Dementia-friendly communities*

A dementia-friendly community is a place where people living with dementia are supported to live a high quality of life with meaning, purpose and value.

Dementia Australia has developed resources to assist all Australians in understanding what they can do to contribute to a dementia-friendly nation.

For more information visit: [www.dementiafriendly.org.au](http://www.dementiafriendly.org.au).

## **Diabetes WA (DWA)**

Level 3, 322 Hay Street  
SUBIACO WA 6008

172 Campbell St  
BELMONT WA 6104

Diabetes Information Advice Line: 1300 001 880

Phone: (08) 9325 7699

Fax: (08) 9221 1183

Website: [www.diabeteswa.com.au](http://www.diabeteswa.com.au)

Facebook: [www.facebook.com/diabeteswa](http://www.facebook.com/diabeteswa)

### **Diabetes mellitus**

Diabetes is a disorder in which the body cannot make proper use of carbohydrate in food because it does not make enough effective insulin. There are three major types; Type 1, Type 2 and gestational diabetes and information about these and their management can be found on the website listed above.

### **Objectives of the group**

- To support people living with, at risk and affected by diabetes
- To create greater awareness of diabetes in the WA community
- To help prevent or delay the onset of type 2 diabetes through education.

### **Activities and services**

- Providing diabetes-related products to assist people in managing their diabetes
- Support and education programs for people living with diabetes
- Advocating on behalf of all people living with diabetes and their families
- Membership for people living with and at risk of diabetes
- A telephone counselling and information service staffed by credentialed diabetes educators (individual appointments may be made)
- Links people with diabetes to a range of support services and networks.

### **Publications/resources**

A range of free resources available to order at  
[www.diabeteswa.com.au/Living\\_With\\_DiabetesOnline\\_Resources](http://www.diabeteswa.com.au/Living_With_DiabetesOnline_Resources).

Diabetes WA also publish e-Newsletters. To sign up visit  
[www.diabeteswa.com.au/healthjourney](http://www.diabeteswa.com.au/healthjourney).

The quarterly magazine for all people with diabetes, 'Diabetes Matters', is also published. To receive the member's magazine call 1300 136 588.



# Down Syndrome WA

Suite 3  
2 Canning Hwy  
SOUTH PERTH

PO Box 338  
BENTLEY WA 6982

Phone: (08) 9368 4002, 1800 623 544

Fax: (08) 9368 4006

Email: [admin@downsyndromewa.org.au](mailto:admin@downsyndromewa.org.au)

Website: [www.downsyndromewa.org.au](http://www.downsyndromewa.org.au)

## Down syndrome

Down syndrome is a genetic condition that results when a person has an extra chromosome 21 in all of their cells. It occurs worldwide approximately once in every 800 births.

People with Down syndrome have some characteristic physical features, some health and developmental challenges, as well as some level of intellectual disability. Most of the young people growing up with Down syndrome today will live ordinary lives with support in the community. They have the same needs and aspirations in life as anyone else.

## Synonyms of Down syndrome

Trisomy 21, Mosaic 21, Translocation 21

## Objectives of the group

- To provide support to parents, advocate for and enrich the lives of people with Down syndrome and to educate the community
- To provide support and information to individuals with Down syndrome and their families
- To help raise awareness of Down syndrome in the community
- To support the community in developing the potential of individuals with Down syndrome to lead a valued life.

## Activities and services

- One to one parent support
- Hospital or home visits
- Peer to peer support including Facebook
- New parent morning teas
- Information seminars for families
- Developmental playgroup
- Skills development and social club program for young adults with Down syndrome
- Education consultancy, trouble shooting, strategies for teachers
- Talks to schools, hospitals, community groups
- Professional development and training for teachers, support workers and child care staff
- Awareness and fundraising events, such as; World Down Syndrome Day – March, Butterfly Ball – June, and; Step UP! And Walk – October.

## Publications/resources

Down Syndrome WA provides a monthly e-news, a resource library for families, as well as a comprehensive website. A New Parent Resource is available to families as well as a National Journal. Down Syndrome WA (along with other State and Territory Associations) is a member of Down Syndrome Australia.

# Dyslexia SPELD Foundation WA Inc

PO Box 409  
SOUTH PERTH WA 6951

Phone: (08) 9217 2500 Fax: (08) 9217 2599

Email: [support@dsf.net.au](mailto:support@dsf.net.au)  
Website: <https://dsf.net.au/>  
Facebook: [www.facebook.com/dyslexiaspeld](http://www.facebook.com/dyslexiaspeld)

## Specific learning disabilities

A specific learning disability can be defined as a significant delay in one or more areas of learning occurring in an individual of average to above average intelligence with intact hearing, vision and emotional status. The genetic component of specific learning disabilities is still being investigated.

**Dyslexia** is commonly associated with difficulties with phonological awareness and processing. This refers to the ability to hear and manipulate the separate sounds within words (phonemes). Spelling, comprehension, reading accuracy, reading rate, word identification and phonological coding are all affected. There is a strong hereditary component in dyslexia - that is, it runs in families.

## Synonyms of dyslexia

Congenital word blindness	Primary reading disability
Developmental reading disorder	Specific reading disability

## Objectives of the group

The Dyslexia SPELD Foundation provides a range of services and family support throughout Western Australia to enable children and adults with specific learning difficulties to realise their potential.

## Activities and services

- Meetings, information evenings, seminars and workshops for professionals and parents
- Psychological assessments and counselling, tutoring and teacher in-service courses
- Individual consultations are available with a computer specialist for those seeking advice about software
- An information meeting is held on the 1st Wednesday of every month.

## Publications/resources

The Foundation provides information by phone and through open monthly meetings for the general community. Library resources are available for members. Health professionals can obtain information by phone. Other resources include: A bulletin published for members three times a year. Resource material for tutors, classroom teachers and parents to assist with the support and education of students with specific learning difficulties.

## **Ectodermal Dysplasia Support Group (ozED)**

Phone: (03) 9755 5626

WA Contact: Leanne Lewis  
Phone: (08) 9454 4967

Email: [info@ozed.org.au](mailto:info@ozed.org.au)  
Website: [www.ozed.org.au](http://www.ozed.org.au)  
Facebook: <https://www.facebook.com/groups/34955153863/>

### **Ectodermal dysplasia**

Ectodermal dysplasia (ED) has over 150 different forms. The effects of ED include abnormalities to the hair, teeth, and nails, sweating ability, skin disorders such as dermatitis and other ear, nose and throat issues. Also known as anhidrotic ectodermal dysplasia.

### **Disorder subdivisions**

There are numerous subdivisions of ED and information about these can be found by visiting the links on the website above.

### **Objectives of the group**

- Provide support and referral advice to all persons and families that are diagnosed with ED
- Provide advice and information to health care professionals to assist in provision of quality care to people and families affected by ED
- Recognise all of the ED forms as equal; no categorising or prioritising of any forms
- Conduct its business, including fundraising, political lobbying and international liaisons in the highest professional manner.

### **Activities and services**

- Support, referral and advice to all persons and families that are diagnosed with ED
- Information to health care professionals to assist in the provision of quality care to people and families affected by ED
- Annual conference and family days are organised for each state.

### **Publications/resources**

Education packages for health and other community professionals that interact with ED people and their families are available.

# Epilepsy Association of WA Inc

The Niche, Suite B  
11 Aberdare Road  
NEDLANDS WA 6009

Helpline: 1300 852 853  
Phone: (08) 6457 7699

Email: [eo@epilepsywa.asn.au](mailto:eo@epilepsywa.asn.au)  
Website: [www.epilepsywa.asn.au](http://www.epilepsywa.asn.au)  
Facebook: <http://www.facebook.com/epilepsywa>

## Epilepsy

Epilepsy is a sudden disturbance of function of the brain lasting for a few seconds or minutes, and then returning to normal. Epilepsy can occur at any age and may be as the result of many factors, including head injuries, brain infection and stroke. There are over 40 types of seizures with two basic categories, generalised-affecting the whole brain, and partial-originating from a specific focal point in the brain. Some types of epilepsy may be inherited.

## Synonyms of epilepsy

Convulsions

Seizures

## Disorder subdivisions

Tonic clonic seizures  
Simple partial seizures  
Complex partial seizures

Absence seizures  
Pseudo seizures  
Myoclonic seizures

## Objectives of the group

We are committed to enhancing the quality of life of people having seizures and to improving community attitudes toward seizure disorders. We aim to educate and support individuals and families with epilepsy in addition to their friends and the community as well as assisting individuals with epilepsy to integrate successfully into the community thus allowing them to enjoy a full and productive lifestyle.

## Activities and services

- Awareness and education programs
- Seminars and events
- Advocacy
- Support groups.

## Publications/resources

The Association has a resource library of information brochures, books, audiotapes, DVDs and videos on a range of related topics.

# Even Keel Bi-Polar Disorder Support Assoc. Inc

165 Wright St  
KEWDALE WA 6105

Phone: (08) 9477 0460  
Email: office@evenkeel.org.au  
Website: www.evenkeel.org.au

## Bipolar disorder

Bipolar, also known as manic depression is a brain disorder that involves unusual shifts in mood, energy and activity levels. At its most severe, the affected person's ability to carry out regular tasks is impaired.

The symptoms can range from a state called mania, in which there can be an overly joyful or overexcited state to an extremely hopeless and sad state, called a depressive episode.

Bipolar disorder is often diagnosed in a person's late teens or early twenties and affects 2 out of 100 people in the population.

Bipolar disorder is currently thought to be a life long illness. There are many treatments available for a person with bipolar disorder and many can live a satisfying and productive life.

## Synonyms of bipolar disorder

Manic depression  
Manic depressive illness

Manic-depressive psychosis  
Affective disorder  
Major affective disorder

## Disorder subdivisions

Bipolar I  
Bipolar II

Bipolar III (cyclothymia)  
Bipolar not otherwise specified

## Objectives of the group

- To reduce the stigma associated with mental illness
- To empower individuals who have bipolar disorder and other mental illnesses
- To provide information and community education about bipolar disorder and other mental illnesses
- To provide friendship and support to individuals experiencing mental illness and their friends, family and carers.

## Activities and services

Support groups are held for people with bipolar disorder and other mental illnesses. Meetings are held weekly in Subiaco and monthly in Midland, Riverton, Perth City, Rockingham, Yokine, Cannington, Fremantle and Mandurah. With Joondalup meetings held twice a month.

## Publications/resources

We have an extensive library and resource centre, with borrowing facilities available for members and a newsletter sent out quarterly.

## Fabry Australia

PO Box 106  
MENTONE VIC 3194

Contact: Megan Fookes (Managing Director)  
Phone: 0418 452 940  
Email: [director@fabry.com.au](mailto:director@fabry.com.au)

Contact: Anne Hunter (National Office Secretary)  
Phone: 0422 201 013  
Email: [secretary@fabry.com.au](mailto:secretary@fabry.com.au)

Website: [www.fabry.com.au](http://www.fabry.com.au)  
Facebook: [www.facebook.com/fabrysupportgroupaustralia](http://www.facebook.com/fabrysupportgroupaustralia)

### Fabry disease

Fabry Disease is a rare genetic disease caused by a deficiency of an enzyme essential for the breakdown of waste products in the cells of the body. In individuals living with Fabry Disease, these waste products build up in their cells and this causes the symptoms of Fabry Disease which can include pain, impaired sweating, skin angiokeratomas, kidneys, brain, heart, and nervous system and is sometimes referred to as a storage disorder. The condition can lead to more serious problems such as heart failure, an enlarged heart, increased risk of stroke or heart attack and kidney failure.

Fabry Australia is a voluntary support group, founded in 1994. It represents over 200 adults and children (both male and female) affected by Fabry Disease, along with their families and health professionals. Fabry Australia is a registered charity supported by voluntary donations, educational grants, fundraising and is managed by the members themselves. Mission: Uniting the Australian Fabry community, by working together to improve the lives of those affected by Fabry Disease.

### Objectives of the group

Fabry Australia is committed to the following aims / desired outcomes:

- Improve contacts, information and support among people affected by Fabry disease, and their families, within Australia.
- Bring about more public awareness of Fabry Disease.
- Improve the medical services to Fabry patients in Australia.
- Promote and support research into Fabry Disease.
- Share information on Fabry Disease, ongoing management and care as well as available treatments /therapies, to Fabry patients, disease.
- Co-operate and collaborate with other Fabry related groups and individuals interested in rare diseases to promote common their supporting carers and related Fabry stakeholders.
- Build links with families, clinicians, researchers and support groups, to strengthen and support local knowledge about Fabry disease.
- Co-operate and collaborate with other Fabry related groups and individuals interested in rare diseases to promote common interests.

- Help raise money in order to support with achieving these aims.

### **Activities and services**

Fabry Australia offer a range of services and implement projects in an effort to further support its membership.

- Fabry Australia website
- Fabry Australia Social Media
- Australian Fabry Expert meeting and Conferences
- State Fabry Patient Meetings
- Fabry Disease Advocacy
- Educating families and doctors
- Funding Fabry Research
- Fabry Educational Materials
- Visiting International Fabry Experts
- Fundraising activities
- Fabry Awareness Month Campaign
- Fabry Australia Membership Family Retreats / Camps

## Face Up

Email: [info@faceup.org.au](mailto:info@faceup.org.au)  
Website: <http://www.faceup.org.au/>  
Facebook: <https://www.facebook.com/faceupinc>

FaceUp is the first charity of its kind in Western Australia.

It was established in July 2016 and is dedicated to supporting and empowering people with visible facial differences and their families.

FaceUp aims to increase community awareness and work to influence attitudes toward people with visible facial differences.

### Objectives of the group

- Support and help people of all ages with facial disfigurements including but not limited to; birth defects, and their families
- Assist in the prevention and reduction of psychological and potentially physical harm to people with visible facial differences
- Support people with visible facial differences to learn to love themselves for who they are
- To bring empathy, understanding and acceptance to our community
- Support people with visible facial differences to make friends and to be confident enough in public to be involved in regular activities like sports and other recreational activities

### Activities and services

- Coaching and training
- Empowerment and advocacy
- Education and influencing
- Activities and networking forums.



# **Familial Hypercholesterolaemia Support Group of WA**

Contact: Annette  
Phone: 0408 276 780

Email: fhfamilysupportgroup@hotmail.com.au  
Website: www.fhfamilysupportgroup.websyte.com.au

## **Familial hypercholesterolaemia**

Familial hypercholesterolaemia (FH) is an inherited disorder of cholesterol metabolism leading to premature coronary heart disease. 1 in 300 West Australians have FH but 80% of cases are not diagnosed. The early diagnosis and treatment of FH can delay or prevent the onset of coronary heart disease.

## **Synonyms of familial hypercholesterolaemia**

Hyperlipoproteinemia, Type IIA  
LDL receptor disorder

## **Disorder subdivisions**

Heterozygous familial hypercholesterolaemia  
Homozygous familial hypercholesterolaemia

## **Objectives of the group**

The aim of establishing an FH Support Group is to improve information, communication and support services for families with FH in Western Australia and help prevent premature deaths caused by high cholesterol and cardiovascular disease.

The FH support group encourage the involvement of medical, nursing and allied health professionals, families, carers, supporters and advocates, in partnership with patients to help support those West Australians at risk of inherited high cholesterol and cardiovascular disease.

## **Activities and services**

- Meetings every 2 months and information seminars
- Providing information, advice and support
- Supporting health professional awareness
- Providing a network for individuals and their families with inherited high cholesterol
- Promoting healthier lifestyle options.

## **Publications/resources**

The group can provide information on FH.

# **Fragile X Association of Australia Inc**

Suite 204 20 Dale Street  
BROOKVALE NSW 2100

Phone: (02) 9907 2366

Free call: 1300 394 636

Email: [support@fragilex.org.au](mailto:support@fragilex.org.au)

Website: [www.fragilex.org.au](http://www.fragilex.org.au)

Facebook: [www.facebook.com/fxAus](http://www.facebook.com/fxAus)

## **Fragile X syndrome**

Fragile X syndrome (FXS) is a genetic condition causing intellectual disability, behavioural and learning challenges and notable physical characteristics. It is also the most common single gene cause of autism worldwide. It appears in people of all ethnic, racial and socio economic backgrounds. Though FXS occurs in both genders, males are generally affected with greater severity. Every week in Australia one child is born who is fully affected and 20 children are born who are carriers. It is estimated that 5 per cent of people with a diagnosis of an autism spectrum disorder also have Fragile X. Although there is currently no cure, early intervention by health and educational professionals can assist people living with Fragile X to reach their full potential.

## **Conditions related to Fragile X syndrome**

Fragile X-Associated Tremor Ataxia

Fragile X-Associated Primary Ovarian Insufficiency

## **Objectives of the group**

Fragile X Association is a national member-based non-profit organisation and is a registered charity. Their objectives are to provide services and support to individuals affected by Fragile X-associated disorders, and to raise awareness of these conditions.

## **Activities and services**

- Helpline 5 days per week
- Information - website
- Family Support counselling (by phone/email)
- Workshops and seminars
- Putting Fragile X families in touch with each other
- Awareness raising.

## **Publications/resources**

- Website
- YouTube channel
- Articles

## **Friedreich Ataxia Network (FAN)**

12 Lowrie Court  
CLEVELAND QLD 4163

President: Terry O'Hanlon  
Phone: 0414 632 986

Email: [info@fan.asn.au](mailto:info@fan.asn.au)  
Website: [www.fan.asn.au](http://www.fan.asn.au)  
Facebook: [www.facebook.com/groups/fanqld/](https://www.facebook.com/groups/fanqld/)

### **Friedreich ataxia**

Friedreich ataxia is a neurological disorder, a progressive condition, with symptoms starting with poor balance and coordination and more severely including scoliosis, diabetes, dysarthria & dysphagia, blindness, neuropathy of lower limbs and heart problems including cardiomyopathy.

### **Objectives of the group**

FAN is a Queensland based network that provides support for 'FAers' - those with the condition, carers, family and friends. Research is ongoing to find treatments and ultimately a cure for Friedreich ataxia FAers in the meantime.

### **Activities and services**

- We host get-togethers where anyone impacted by FA can meet others to share experiences, exchange thoughts and advice
- We advocate for and monitor the FA clinic run in Brisbane by QLD Health
- We maintain a database of QLD FAers to provide an accessible patient group for a clinical trial if such opportunity arises
- We support FA education to our members and health professionals caring for them
- We provide limited direct funding for equipment that will enhance the life of someone with FA.

### **Publications/resources**

- We maintain a website ([www.fan.asn.au](http://www.fan.asn.au)) with current knowledge of FA, progress in research to find treatments & a cure, information about help available from state & federal governments to FAers and carers
- We have a group on Facebook (search: Friedreich Ataxia - FAers, family, carers and friends can pose questions and receive answers from others who share and understand their situation.

## **GAPPS Support**

Please contact Genetic and Rare Disease Network (GaRDN)

Phone: 1300 770 995  
Email: [hello@gardn.org.au](mailto:hello@gardn.org.au)

### **GAPPS**

Gastric adenocarcinoma and proximal polyposis of the stomach (GAPPS) is an inherited disorder.

At this stage it is only known to affect members of a large Australian family, all descended from Matthias Powell.

It is characterised by a growth of gastric polyps which have a risk of developing into stomach cancer. Many people have gastric polyps but do not have GAPPS.

There is no genetic test, so diagnosis can only be obtained by having an endoscopy.

Many family members, in consultation with their doctors, have elected to have a total gastrectomy to remove the risk of gastric cancer.

### **Objectives of the group**

- To provide support to individuals and their families affected by GAPPS
- To facilitate information sharing between families
- To promote partnership between families and health professionals.

### **Activities and services**

- Facilitate meetings/get-togethers with GAPPS family members □ General support and information
- Person to person support when required.

# **Gelastic Seizure Support Hub**

PO Box 88  
TOOWOOMBA QLD 4350

Contact: George W Helon  
Phone: (07) 4635 9588  
Mobile: 0419 768 792

Email: [ghelon@yahoo.com.au](mailto:ghelon@yahoo.com.au)  
Facebook: [www.facebook.com/Gelastic.Seizures](http://www.facebook.com/Gelastic.Seizures)

## **Gelastic seizure**

Gelastic: of or pertaining to laughter; from the Greek 'Gelastikos' i.e. 'able to laugh'. A rare syndrome related type of neurophysical event, a gelastic seizure could best be described as a 'paroxysm' - a sudden and involuntary outburst of emotion, action or activity.

Usually associated with the presence of a hypothalamic hamartoma (HH) - a benign, non-cancerous, non-malignant and usually not life-threatening tumour.

## **Synonyms of gelastic seizure**

Gelastic paroxysm  
Hypothalamic hamartoma (HH)  
Pallister-Hall syndrome (PHS)  
Laughing seizures

## **Objectives of the group**

- To educate and inform the public
- To facilitate contact and support for gelastic seizure patients, their families, carers, medical providers, allied health professionals and interested researchers worldwide.

## **Activities and services**

Worldwide network support group offering:

- Phone support by appointment only
- Email and online support 24/7
- Public advocacy and peer support
- Guest speakers by negotiation
- Library services
- Fact sheets.

## **Publications/resources**

- Web presence at [www.facebook.com/Gelastic.Seizures](http://www.facebook.com/Gelastic.Seizures)
- Information article: Anatomy of a Gelastic (or Laughing) Seizure by George W.Helon available at [www.facebook.com/Gelastic.Seizures/notes](http://www.facebook.com/Gelastic.Seizures/notes)
- See also Pallister-Hall Syndrome (PHS) Support Hub.

# **HAE Australasia (Hereditary Angioedema)**

WA Representative - [Fiona@haeaustralasia.org.au](mailto:Fiona@haeaustralasia.org.au)

PO Box 285  
Webb St  
NARRE WARREN VIC 3805

Email: [info@haeaustralasia.org.au](mailto:info@haeaustralasia.org.au)  
Website: [www.haeaustralasia.org.au](http://www.haeaustralasia.org.au)  
Facebook: [www.facebook.com/HAEAustralasia](http://www.facebook.com/HAEAustralasia)

## **Hereditary angioedema**

Hereditary angioedema (HAE) results from a deficiency or dysfunction of the protein C1 inhibitor. Symptoms include oedema (swelling) of the limbs, trunk, face, genitals, gastrointestinal tract, tongue, inside the mouth or laryngeal swelling.

## **Synonyms of hereditary angioedema**

HAE	C1-inhibitor deficiency
C1-esterase inhibitor deficiency	

## **Hereditary angioedema subdivisions**

Hereditary angioedema Type I	Hereditary angioedema Type III
Hereditary angioedema Type II	

## **Objectives of the group**

- Enhance the support and quality of life for those living with HAE
- Provide broad based education to patients and carers
- Provide broad based education to medical professionals to appropriately diagnose and treat HAE patients
- Look at ways to make treatments more accessible
- Assist with HAE research
- Increase community awareness of HAE.

## **Activities and services**

- Bi Annual patient and carers conference
- Local 'HAE Meet Ups'
- Local HAE Healthy Minds Workshops
- Facebook support group
- Twitter
- Telephone support
- Advocacy
- Fundraising and awareness activities.

## **Publications/resources**

- Education/information packs
- Regular newsletters
- Flyers.

# Haemochromatosis Australia

PO Box 6185  
MERIDAN PLAINS QLD 4551

Phone: 1300 019 028

Email: [admin@ha.org.au](mailto:admin@ha.org.au)

Website: [www.ha.org.au](http://www.ha.org.au)

Facebook: [www.facebook.com/haemochromatosis](http://www.facebook.com/haemochromatosis)

## Haemochromatosis

Hereditary haemochromatosis is an inherited iron overload disorder. Early symptoms include fatigue, weakness, joint pain and sexual dysfunction. If untreated more serious symptoms may develop including liver cirrhosis and cancer, cardiomyopathy, osteoarthritis and diabetes.

## Objectives of the group

Haemochromatosis Australia is the support, advocacy and health promotion group for people with haemochromatosis and their families.

- Support people with haemochromatosis and improve early detection and treatment
- Raise community awareness
- Improve professional awareness
- Foster and support research
- Improve public health policy
- Good governance of administration

## Activities and services

- Haemochromatosis awareness week
- Info line: 1300 019 028
- Public information meetings
- Membership

## Publications/resources

- Videos- DVD and YouTube: [www.youtube.com/haemochromatosisAust](http://www.youtube.com/haemochromatosisAust)
- Print publications  
Newsletters

# Haemophilia Foundation WA Inc

City West Lotteries House  
2 Delhi Street  
WEST PERTH WA 6005

Phone: (08) 9420 7294  
Email: office@hfwa.org  
Website: www.hfwa.org  
Facebook: www.facebook.com/HaemophiliaFoundationAustralia

## Haemophilia

Haemophilia is a genetic condition transmitted on the X chromosome and occurs almost exclusively in boys. The deficiency in clotting factor produces a wide range of bleeding episodes which are mostly internal and are usually into the joints or muscles. These may occur spontaneously or as the result of injury. The bleeding is stopped by the infusion of the appropriate clotting factor by intravenous injection.

**Von Willebrand disease** is another bleeding condition where there is a combined deficiency of factor VIII and a platelet abnormality. Von Willebrand disease is not carried on a sex linked (X) chromosome therefore affecting males and females equally. There are a number of synonyms of haemophilia, including; antihæmophilic (US spelling antihemophilic) factor deficiency, antihæmophilic globulin deficiency, and Christmas disease.

The subdivisions are: haemophilia A (factor VIII) deficiency), haemophilia B (factor IX) deficiency and haemophilia C (factor XI deficiency). There are also a number of Von Willebrand disease subdivisions that can be found on the website.

## Objectives of the group

- To provide support, peer support/mentoring information and advocacy to individuals and their families and carers with haemophilia or Von Willebrand disease
- To promote a better lifestyle for individuals with haemophilia
- To increase public awareness of this condition.

## Activities and services

- Seminars and conferences
- Child and family camps and outings
- Counselling service
- Limited financial support for equipment for members
- Monthly committee meeting with additional meetings as required and an annual general meeting.
- Treatment updates and information sessions.

## Publications/resources

The foundation provides members with bi-monthly state and national newsletters. Restricted library and internet use, as well as brochures and other literature covering a wide range of (bleeding condition) issues are available to the community.



## **Heart Kids WA Inc.**

PO Box 1554  
WEST PERTH WA 6872

Phone: (08) 9340 7996

Email: [office@heartkids.org.au](mailto:office@heartkids.org.au)

Website: [www.heartkidswa.org.au](http://www.heartkidswa.org.au)

Facebook: [www.facebook.com/HeartKidsWA](http://www.facebook.com/HeartKidsWA)

### **Heart defects**

Congenital heart defects result from abnormalities in the foetal development of the heart and major vessels during pregnancy. It may be caused by maternal illness, environmental exposure or as part of a genetic condition. Some children may not develop heart problems until later in childhood (acquired).

### **Objectives of the group**

To provide support for children and their families with congenital or acquired heart disease and their families.

### **Activities and services**

The group supports families and children in various ways, including:

- Visiting families in hospital
- Coffee and chats once a month
- Information evenings
- Support events
- Fundraising events
- Family camp and teen camp
- Community awareness, through displays at community events and medical centres.

## **HHT Alliance (Hereditary Hemorrhagic Telangiectasia)**

C/o Genetic Medicine  
The Royal Melbourne Hospital Level 2 Centre, Grattan Street  
PARKVILLE VIC 3050

Phone: (03) 9342 7151  
Fax: (03) 9342 4267

Email: [hht@mh.org.au](mailto:hht@mh.org.au)  
Website: [www.hht.org.au](http://www.hht.org.au)

### **Hereditary Hemorrhagic Telangiectasia**

Hereditary Hemorrhagic Telangiectasia is an autosomal dominant condition that affects approximately 1 in 3,000 to 1 in 10,000 people. It is a rare condition also known as Osler Weber Rendu syndrome.

A person living with HHT has a tendency to form blood vessels that lack normal capillaries between an artery and vein. This means that arterial blood under high pressure flows directly into a vein without first having to squeeze through very small capillaries. This place where an artery is connected directly to a vein tends to be a fragile site that can rupture and bleed.

### **Objectives of the group**

The HHT Alliance stands for:

H: Hope - Hope for a future cure

H: Health - To understand the Health of those diagnosed with HHT

T: Treatment - To promote research into new treatments and management of HHT

### **Activities and services**

As HHT is a rare genetic condition little research has been conducted into the impact HHT has on patients diagnosed with the condition.

Through the HHT Alliance, we are inviting people and their proxies from all over Australia to submit information about their HHT diagnosis so that researchers can develop new insights into this genetic condition.

### **Publications/resources**

Further information about HHT is also available from HHT Foundation International Inc. at: <http://hht.org/about-hht>

## **Homocystinuria (HCU) Network Australia**

PO Box 7484  
BAULKHAM HILLS NSW 2154

Phone: 0411 488 476

Email: [info@hcunetworkaustralia.org.au](mailto:info@hcunetworkaustralia.org.au)

Website: [www.hcunetworkaustralia.org.au](http://www.hcunetworkaustralia.org.au)

Facebook: <https://www.facebook.com/HCUNetworkAustralia/>

### **Homocystinuria (HCU)**

Homocystinuria (HCU) is a rare genetic disease. Untreated it is progressive and affected individuals suffer multi-systemic disorder of the central nervous system (CNS), ocular, skeletal, and cardiovascular system. Symptoms are highly variable but include intellectual disability, developmental delay and learning difficulties, psychiatric disturbances, behavioural problems, dislocation of the lens of the eye resulting in blindness if untreated, seizures, osteoporosis and skeletal abnormalities, premature heart attack and stroke. As a whole, the complications of the disease lead to a reduced quality of life and shorter life expectancy.

Whilst no cure has been discovered for homocystinuria, nearly one half of individuals affected benefit from Vitamin B6 supplementation. For the remainder, the current mainstay of treatment is a strict low protein diet coupled with supplementation with B6, B12 and folate, together with a 'medical formula'. This treatment regime aides in the restoration of metabolic balance in the affected individual.

### **Objectives of the group**

HCU Network Australia is a health promotion charity established in 2014 with the vision to be a driving force in the journey to a cure, improving quality of life along the way. Our aim is to improve health outcomes of those affected by HCU by supporting patients and their families, raising awareness and supporting and encouraging research into diagnosis, treatments and ultimately a cure.

## **Huntington's WA (Inc)**

The Niche  
Centre for Neurological Support  
Suite B, 11 Aberdare Road  
NEDLANDS WA 6009

Phone: (08) 6457 7599

Fax: (08) 6457 7597

Email: [admin@huntingtonswa.org.au](mailto:admin@huntingtonswa.org.au)

Website: [www.huntingtonswa.org.au](http://www.huntingtonswa.org.au)

Facebook: [www.facebook.com/huntingtonswesternaustralia](http://www.facebook.com/huntingtonswesternaustralia)

### **Huntington's disease**

Huntington's disease (HD) is an inherited 'neurodegenerative disease' which means that it causes the progressive death of nerve cells in the brain. HD is a complicated disease that affects the body, mind and emotions. There are symptoms of HD that are easy to see, like chorea (involuntary movements,) and there are those that are less visible, such as forgetfulness, impulsiveness or depression.

Symptoms of HD may vary widely from person to person, even within the same family. Symptoms appear gradually and with support, people with HD can maintain their independence for years. HD affects all aspects of a person's life and treating it requires a broad-based approach. Referrals to professionals who understand HD can be provided by Huntington's WA.

### **Objectives of the group**

Huntington's WA (HWA) was specifically formed to enable the best possible quality of life for people with HD, their families and carers, through advocacy, support and education.

### **Activities and services**

- Support Groups: HWA facilitates groups for members of the Huntington's community including teens, gene positive people, early symptomatic people and carers. Each year HWA organises retreats for carers and 'getaways' for all the other groups.
- Regional Support: HWA visits a number of regional communities (by request) and holds quarterly support meetings in Bunbury
- Counselling and home visits: specialised HD advisers are available to visit or provide counselling and one on one support to members of the community impacted by HD.
- Education: HWA promotes awareness and understanding of HD through education forums for both community members and health professionals.
- Advocacy and referral: HWA provides referral to appropriate agencies.

### **Publications/resources**

A quarterly newsletter is available to provide local, national and international Huntington's research updates, care news and a guide to local community events. The Association also has a library of books, articles and information booklets available on request. Detailed information is also available on the HWA website [www.huntingtonswa.org.au](http://www.huntingtonswa.org.au)

# Hypersomnolence Australia

249 Nottingham Road  
ALGESTER QLD 4115

Phone: 0405 917 736  
Email: [hypersomnolenceaustralia@gmail.com](mailto:hypersomnolenceaustralia@gmail.com)

Website: [www.hypersomnolenceaustralia.com](http://www.hypersomnolenceaustralia.com)  
Facebook: [www.facebook.com/hypersomnolenceaustralia](http://www.facebook.com/hypersomnolenceaustralia)

## Idiopathic hypersomnia (IH)

Idiopathic Hypersomnia (IH), sometimes referred to as Idiopathic Hypersomnolence, is a neurological sleep/wake disorder characterised by excessive sleep and chronic daytime sleepiness. It is a debilitating condition often profoundly affecting work, education, and relationships.

Most people can feel tired, fatigued and at times, excessively sleepy, particularly when they do not get enough sleep. However what sets people with IH apart is that they experience extreme sleepiness despite getting adequate or typically more than adequate hours of sleep. Their sleep may be deep and uninterrupted, but it is not refreshing. Despite extraordinary amounts of good quality sleep people with IH are in an almost constant state of sleepiness. Many people with IH also experience extreme and prolonged difficulty awakening from sleep, accompanied by confusion, disorientation, irritability and poor coordination with an uncontrollable desire to go back to sleep. It can also include automatic behaviour ie: performing tasks without consciously knowing it and not remembering you have done them eg: turning off alarm clocks or answering your phone. This is known as “sleep drunkenness”. IH is a lifelong disorder with only rare spontaneous remissions reported. Symptoms typically begin in adolescence or young adulthood.

The cause of Idiopathic Hypersomnia is unknown.

## Objectives of the group

Our primary purpose is to support the medical community work towards a better understanding of Idiopathic Hypersomnia, effective treatment options, and identifying biomarkers that will lead to more appropriate diagnostic tools and ultimately a cure for Idiopathic Hypersomnia. We are committed to being a strong advocacy, raising awareness and educating others about Idiopathic Hypersomnia.

Our goal is to not just change the process to diagnosis but also the level of care and services available to patients post diagnosis.

## Activities and services

- Maintain a registry of people diagnosed with Idiopathic Hypersomnia that will be used with permission for aiding research and education
- Help to improve the level of education with regards to Idiopathic Hypersomnia within the medical community
- Provide support for clinical trials and research and to provide a link between researchers and people with Idiopathic Hypersomnia

- Increase public awareness and education of Idiopathic Hypersomnia
- Advocate for the interests of people with Idiopathic Hypersomnia
- Provide support and information to patients and their family and friends
- Provide information, referrals and advocate for better management of sleep and general health
- Host and promote the annual International Idiopathic Hypersomnia Awareness Week

### **Publications/resources**

Brochure/Factsheet <https://goo.gl/JvTgcm>

Information about our patient registry

<http://www.hypersomnolenceaustralia.org.au/patientregistry>

# **Immune Deficiencies Foundation Australia (IDFA)**

PO Box 969  
PENRITH NSW 2751

Phone: 0409 945 114

Email: [info@idfa.org.au](mailto:info@idfa.org.au)

Website: [www.idfa.org.au](http://www.idfa.org.au)

Facebook: [www.facebook.com/ImmuneDeficiencyFoundationAustralia](http://www.facebook.com/ImmuneDeficiencyFoundationAustralia)

## **Primary immune deficiencies**

An immune deficiency is a disorder where the immune response (the body's defence system) is reduced or absent.

Primary immune deficiencies (PIDs) are caused by defects in the genes that control the immune system, so people with PID are born missing some or all of the parts of the immune system.

The World Health Organisation (WHO) recognises more than 240 primary immune deficiency diseases. PIDs can be mild, severe and in some cases fatal. IDFA is committed to providing education, resources, social and emotional support, linking members via teleconferencing and events, and improving patient care and quality of life for those affected by primary immune deficiencies.

## **Objectives of the group**

- Vision: A future where primary immune deficiencies (PIDs) are diagnosed early, communities are aware of the signs and symptoms of PIDs, and those affected by PIDs feel supported
- Goals: awareness, membership, advocacy and communication
- Health promotion charity raising awareness of PIDs and increasing quality of life for those with PIDs.

## **Activities and services**

- National conference
- Closed Facebook group
- YAMs (young adult members)
- Education, support and events for PID community.

## **Publications and resources**

PID Resource Pack (all available online at [www.idfa.org.au](http://www.idfa.org.au)).

# **Kawasaki Disease Foundation Australia**

PO Box 585  
BRENTFORD SQUARE VIC 3131

Phone: (03) 9894 1257  
Email: [info@kdfoundation.org.au](mailto:info@kdfoundation.org.au)

Website: [www.kdfoundation.org.au](http://www.kdfoundation.org.au)  
Facebook: [www.facebook.com/pages/Kawasaki-Disease-Foundation-Australia/215578498590959](https://www.facebook.com/pages/Kawasaki-Disease-Foundation-Australia/215578498590959)

## **Kawasaki disease**

Kawasaki disease is an acute illness mainly affecting infants and preschool children, but it may occur at any age.

It is characterised by prolonged fever (lasting for at least 4-5 days), rash, red eyes, swollen and red hands and feet, red lips and tongue and enlarged lymph nodes (gland) in the neck.

Not all features occur in every child and they may appear over a number of days. There is no diagnostic test and the diagnosis is made on these clinical features, nonspecific blood tests, which often show inflammation, and exclusion of serious infections and other diagnoses.

Kawasaki disease results from an inflammation of the arteries (vasculitis) and importantly may damage the coronary arteries that supply blood to the heart. The cause is unknown. It is thought to result from one or more infectious triggers in genetically susceptible children.

There are about 200 Kawasaki disease cases diagnosed each year in Australia. It is more common in Asians (especially North East Asians), and in boys.

## **Objectives of the group**

- To provide education, information and support to families affected by Kawasaki disease
- To provide education, information and assistance to professionals who provide services to families affected by Kawasaki disease
- To provide community awareness about Kawasaki disease in order to assist with early diagnosis and treatment
- To maintain links with organisations concerned with Kawasaki disease in other countries
- To assist with research into Kawasaki disease.

## **Activities and services**

- One to one parents support
- Networking between (members) families
- Informative website: [www.kdfoundation.org.au](http://www.kdfoundation.org.au)
- Information sessions for families.



## **Publications and resources**

The foundation has available on its website resources for parents as well as other Kawasaki disease groups overseas. We also provide e-news updates and have available a printed awareness brochure.

## **Klinefelter's Support Group WA**

199/831 Mandurah Rd  
BALDIVIS WA 6171

WA Contact: Kerry Wood  
Phone: (08) 9523 2474  
Email: wood.kerry7@bigpond.com

Website: [www.axys.org.au](http://www.axys.org.au)

This website is the Australian X and Y Spectrum Support and a Facebook page is available if you are accepted by messaging.

### **Klinefelter's syndrome**

Klinefelter's syndrome is a genetic condition affecting males in which there are three sex chromosomes (XXY) instead of two (XY). Individuals with this condition are considered male, however the condition may be characterised by the enlargement of the breasts, the absence of facial and body hair, the failure of normal sperm production, and small testes.

### **Objectives of the group**

One of the important aspects of the Klinefelter's support group is to provide support, information and resource materials about Klinefelter's syndrome to affected individuals and families in Western Australia to help men and boys with this syndrome adjust to life.

The Klinefelter's support group also endeavours to provide health professionals and the community with information about our support group and Klinefelter's syndrome.

### **Activities and services**

Information is provided by the group upon request.

# **Learning and Attentional Disorders Society of WA (LADS)**

The Niche, Suite B  
11 Aberdare Rd (cnr Hospital Ave)  
NEDLANDS WA 6009

Phone: (08) 6457 7544

Email: admin@lads.org.au  
Website: ladswa.nationbuilder.com

## **Learning and attention disorders**

Learning and attention disorders are characterised by the inability to acquire or retain information or skills as a result of a deficit in attention, memory or reasoning. Individuals with Attention Deficit Hyperactivity Disorder, may daydream, not seem to listen, find it hard to start or stay on task, procrastinate, be forgetful and disorganised, and may be easily frustrated and quick to anger. A number of genetic conditions are characterised by learning and attention disorders.

## **Synonyms of attention deficit hyperactivity disorder**

Attention deficit disorder (ADD)  
Attention deficit hyperactivity disorder (ADHD)  
Hyperkinetic disorder

## **Attention deficit hyperactivity disorder**

ADHD, predominantly inattentive type  
ADHD, predominantly hyperactive impulsive type  
ADD, combined type

## **Objectives of the group**

- To increase awareness and acceptance of ADHD
- To improve the quality of life for individuals with this condition
- To provide up to date information, support and referral advice to adolescents, adults and parents of children with learning difficulties and attention disorders.

## **Activities and services**

- Talks, seminars, courses and workshops
- Support is provided through informal meetings and a drop in centre
- Meetings are held monthly
- A well-stocked library.

## **Publications/resources**

The Society publishes a newsletter and has a resource library with an extensive range of books, journal articles, videos and audiotapes available to members for loan. Information kits are available to members and non-members.

# **Leukodystrophy Support Group, Australian**

Nerve Centre Building  
54 Railway Road  
BLACKBURN VIC 3130

Phone: 1800 141 400  
Email: [info@leuko.org.au](mailto:info@leuko.org.au)

Family Advocate: Anne Patricia 0418 755 994

Website: [www.leuko.org.au](http://www.leuko.org.au)  
Facebook: [www.facebook.com/leukodystrophyaustralian](http://www.facebook.com/leukodystrophyaustralian)

WA committee member available for support. Please contact GaRDN for contact details.

## **Leukodystrophy**

Leukodystrophy refers to a group of over 40 identified (plus more unidentified) conditions (mostly serious) that affect the myelin, or white matter, of the brain and spinal cord, which in turn, impacts the ability of the nervous system to send its signals throughout the body.

## **Leukodystrophy subdivisions**

There are many different types of leukodystrophy. An extensive list can be found on <http://alds.org.au/about-leukodystrophy/different-types-of-leukodystrophy>.

## **Objectives of the group**

- To provide assistance and information to those affected by leukodystrophy and support research into leukodystrophy
- Provide professional counselling, encouragement and support to those affected
- Act as a source of information on the range of leukodystrophies
- Raise funds and provide financial support for families affected
- Increase public awareness of leukodystrophy.

## **Activities and services**

- Website
- Quarterly newsletter
- Social gatherings
- ALDS Family Information Kit
- Fundraising activities
- Providing information and advocacy
- Peer support
- Provide professional liaison with local service (with consent)
- Provide emotional and practical support to those affected.

# **Liver Foundation of Western Australia**

Suite 17, Gallery Suites 185 High Street  
FREMANTLE WA 6160

PO Box 327  
Nedlands WA 6009

Phone: (08) 9336 3178  
Fax: (08) 9433 5880

Email: [contact@liverfoundation.org.au](mailto:contact@liverfoundation.org.au)  
Website: [www.liverfoundation.org.au](http://www.liverfoundation.org.au)  
Facebook: [www.facebook.com/liverfoundationwa](http://www.facebook.com/liverfoundationwa)

## **Liver disease**

Liver diseases, which includes but is not limited to alcoholic liver disease, hepatitis A, B, C, D and E, autoimmune liver disease, non-alcoholic fatty liver disease and genetic liver disease.

## **Objectives of the group**

The Liver Foundation of WA is established to help, support and give information to the medical fraternity and to the general public about diagnosis, prevention and treatment of liver related diseases. We also promote awareness about liver health by supporting research and educational activities.

## **Activities and services**

- Liver clinics with alcohol education in the Pilbara
- Crazy Sock program in schools in WA
- Sporting activities sponsored by Liver Foundation of WA
- Medical research.

## **Publications/resources**

- Resources available on website [www.liverfoundation.org.au](http://www.liverfoundation.org.au)
- Book: Little Liver Stories
- Short film: 20 Years of Liver Transplant in WA.

## **LQTS Support Group of WA Inc**

53 Boston Way  
BOORAGOON WA 6154

Phone: (08) 9364 4109

Email: [tiko@iinet.net.au](mailto:tiko@iinet.net.au)

Website: [www.sads.org.au](http://www.sads.org.au)

Facebook: <https://www.facebook.com/SADSAustralia>

### **Long QT syndrome**

Long QT syndrome (LQTS) is an abnormality of the heart electrical system. The mechanical function of the heart is normal however the electrical problem is due to defects in heart muscle cell structures called ion channels. These electrical defects pre-dispose affected persons to a very fast heart rhythm (arrhythmia) which leads to sudden loss of consciousness (syncope) and may cause sudden cardiac death. The QT refers to an interval measured on the electrocardiogram (ECG). This syndrome can be inherited or acquired.

Unfortunately, many times the cause of the syncope is overlooked and the events are called simple fainting spells or seizures. Most often, these events occur during physical exertion or emotional stress. In some they occur during sleep. Fortunately, most deaths are preventable if the condition is recognised and treated.

### **Disorder subdivisions**

Romano-Ward type

Jervell and Lange-Neilson type

Acquired long QT syndrome

### **Objectives of the group**

To raise awareness, provide information and support the families of children and young adults who are genetically predisposed to sudden death due to heart rhythm abnormalities.

### **Activities and services**

The group provides information and educational resources to the lay public and to health care providers in order to promote early diagnosis and treatment.

### **Publications/resources**

Online materials are available for information and public distribution from the website.

## **Lupus Group of WA Inc, The**

Room N 1.03, Level 1  
Colonial House  
Royal Perth Hospital  
Murray St  
PERTH WA 6000

c/o GPO Box X2213  
PERTH WA 6847

Phone: (08) 9224 3144 (24 hour message bank)

Email: [admin@lupuswa.com.au](mailto:admin@lupuswa.com.au)

Website: [www.lupuswa.com.au/content/page/contact.html](http://www.lupuswa.com.au/content/page/contact.html)

### **Lupus**

Lupus is a widespread and chronic (lifelong) autoimmune disease that, for unknown reasons, causes the immune system to attack the body's own tissues and organs, including the joints, kidneys, heart, lungs, brain, blood, or skin. The symptoms of lupus often mimic other less serious illnesses and can range from mild to life-threatening. Lupus can go into periods where symptoms are not present, called remission and although lupus can affect any part of the body, most people experience symptoms in only a few organs.

### **Synonyms of lupus**

Disseminated lupus erythematosus

Lupus erythematosus

### **Disorder subdivisions**

Discoid lupus erythematosus (DLE)  
Drug-induced lupus erythematosus

Systemic lupus erythematosus

### **Objectives of the group**

To act as the principle resource for lupus patients, their families, doctors and the community to provide educational awareness, support and facilitation of research activities.

### **Activities and services**

The Group provides support services that are reviewed by the Board. Some of the services include community awareness via:

- Seminars, workshops, community expo's and educational displays
- Phone-line help: Assistance with 'living with lupus', provision of mutual support to share information, personal experiences and coping strategies
- Advocacy, contribute to relevant policy
- Fundraising.

### **Publications/resources**

Fact sheets on a wide range of issues relating to lupus. Brochures and a newsletter are available to members.

# **Lyme Disease Association of Australia**

PO Box 137  
STOCKTON NSW 2295

Email: [info@lymedisease.org.au](mailto:info@lymedisease.org.au)  
Website: [www.lymedisease.org.au](http://www.lymedisease.org.au)  
Facebook: [www.facebook.com/LymeDiseaseAustralia](http://www.facebook.com/LymeDiseaseAustralia)

## **Lyme disease**

Lyme disease is an infection caused by a bacteria (spirochete) that infects humans from the bite of ticks which are infected with the bacteria. In fact, ticks can spread multiple diseases, these are known as “co-infections”. Lyme disease is called “The Great Imitator” because it can imitate many other diseases such as MS, Parkinson’s disease, motor neurone disease (ALS), chronic fatigue syndrome, fibromyalgia, Guillain-Barre syndrome, juvenile rheumatoid arthritis, Cushing’s disease, , Lupus, Alzheimer’s disease etc.

Lyme disease can affect any organ in the body including muscles & joints, the heart, gastro-intestinal system & neurological system (including the brain).

## **Synonyms of Lyme disease**

Borreliosis  
Multi system infectious disease syndrome (MSIDS)  
Lyme and co-infections

## **Objectives of the group**

Awareness, prevention, research, recognition, advocacy and support of Lyme disease patients.

## **Activities and services**

- Doctor referral
- Research
- Awareness and fundraising material distribution
- Government liaison
- Lobbying
- Media liaison
- Awareness campaign leadership.

## **Publications/resources**

All are available online at [www.lymedisease.org.au](http://www.lymedisease.org.au)



# **Machado Joseph Disease (MJD) Foundation Ltd**

PO Box 414  
ALYANGULA NT 855

Phone: 1300 584 122  
Email: [nadia.lindop@mjd.org.au](mailto:nadia.lindop@mjd.org.au)

Website: [www.mjd.org.au](http://www.mjd.org.au)  
Facebook: [www.facebook.com/MJDfref=ts](https://www.facebook.com/MJDfref=ts)

## **Machado Joseph disease**

The MJD Foundation seeks to improve the quality of life of Indigenous Australians and their families living with MJD. MJD is a hereditary neuro-degenerative condition. It is in a 'family' of neuro-degenerative disease that includes Huntington's disease.. There is no known cure for MJD. Progression to dependence occurs over 5-10 years; most people are wheelchair bound and are fully dependent for all activities of daily living within 10-15 years.

## **Objectives of the group**

- To provide improved services by working with and supplementing those services provided by the government
- Contributing to local and international research, to gain an understanding of how this research can benefit Indigenous Australians living with MJD
- Implementation of practical solutions to help individuals understand and manage their symptoms
- Provide a greater level of community infrastructure and transportation options to support the needs of individuals and their families living with MJD
- Facilitating comprehensive genetic education programs
- Advocating on behalf of individuals and families to all levels of the community and government
- Ensuring flow on benefits of individuals and families to the wider international MJD community through research and advocacy.

## **Activities and services**

- Research-funding for research projects
- Education for families, health care professionals and other service providers
- Equipment purchase to improve quality of life for those living with MJD
- Advocacy for our clients
- Improved services for our clients and their carers.

# **Mental Illness Fellowship Western Australia Inc**

Level 3  
9 The Avenue  
PO BOX 1947  
MIDLAND DC WA 6936

Phone: 1800 985 944  
Lorikeet Centre: (08) 9237 8951

Email: [info@mifwa.org.au](mailto:info@mifwa.org.au)  
Website: [www.mifwa.org.au](http://www.mifwa.org.au)

## **Schizophrenia**

Schizophrenia is a condition characterised by disturbances in a person's thoughts, perceptions, emotions and behaviour. It affects approximately one in every 100 people worldwide and first onset commonly occurs in adolescence or early adulthood. Schizophrenia is not a single illness. It is in fact a cluster of illnesses, which have overlapping signs and symptoms. It is therefore important to acknowledge the unique experience of each person living with schizophrenia.

## **Synonyms of schizophrenia**

Schizophrenic disorder  
Schizophrenic psychosis  
Dementiapræcox

## **Objectives of the group**

- To provide fellowship, a place where people affected by mental illness can meet and find understanding and support
- To promote an understanding of issues relating to mental illness in the community; and to contribute to innovation and systemic reform in mental health service delivery.

## **Activities and services**

- Workshops and training
- Self-Care for carers courses
- Various activities during Schizophrenia Awareness Week, Mental Health Week and Carers Week
- Accredited rehabilitation service for people with a history of mental illness: contact Lorikeet Centre
- Support groups for carers- call for details.

## **Publications/resources**

The Fellowship has a resource library with books, videos, audiotapes and reports, which are available for loan for members (a small fee applies to non-members). A quarterly newsletter is published for members as well as regular mail outs.

# **Mitochondrial Disease Foundation, Australian (AMDF)**

Unit 3  
21 Mary St  
Surry Hills NSW 2010

Phone: (02) 8033 4113  
Helpline: 1300 977 180

Email: [info@amdf.org.au](mailto:info@amdf.org.au)  
Website: [www.amdf.org.au](http://www.amdf.org.au)  
Facebook: [www.facebook.com/australianmitochondrialdiseasefoundation](http://www.facebook.com/australianmitochondrialdiseasefoundation)

## **Mitochondrial disease**

Mitochondrial disease is a debilitating genetic disorder that robs the body's cells of energy, causing multiple organ dysfunction or failure and potentially death. There are currently few treatments available and no cure.

## **Subdivisions of mitochondrial disease**

There are a significant number of subdivisions of mitochondrial disease and information about these can be found by contacting AMDF.

## **Objectives of the group**

The AMDF is committed to funding research into mitochondrial disease through differing fundraising activities. As well as this, the AMDF aims to support sufferers of the disease, along with their families. Lastly, we aim to educate the general public and the medical profession about mitochondrial disease.

## **Activities and services**

AMDF holds regular information sessions in different states, in which patients can hear the latest research, as well as network with other patients. AMDF also host a helpline run by a volunteer who is a former GP. The helpline is available if patients have questions regarding the disease, diagnosis, symptoms etc.

## **Publications/resources**

AMDF has published a booklet for medical practitioners. We often provide these to patients who have difficulty explaining the disease to their doctors.

## **Motor Neurone Disease Association WA (MNDAWA)**

The Niche Suite B  
11 Aberdare Rd  
NEDLANDS WA 6009

Phone: 08 6457 7355  
Fax: 08 6457 7332

Email: [admin@mndawa.asn.au](mailto:admin@mndawa.asn.au)  
Website: [www.mndawa.asn.au](http://www.mndawa.asn.au)  
Facebook: [www.facebook.com/MNDAWesternAustralia](http://www.facebook.com/MNDAWesternAustralia)

### **Motor neurone disease**

Motor Neurone Disease (MND) is a progressive neurological condition that attacks the motor neurones (nerves). It is the name given to a group of diseases in which the nerve cells (neurones) controlling the muscles that enable us to move, speak, swallow and breathe undergo degeneration and die. The cause of MND is unknown and not all symptoms of the disease necessarily happen to everyone with the patterns of weakness varying from person to person. MND is diagnosed in people of all ages, men and women. Currently there is no cure, however symptoms can be managed to help the person achieve the best possible quality of life.

### **Synonyms of motor neurone disease**

MND  
ALS  
Lou Gehrig's disease

### **Objectives of the group**

- To provide support, information and equipment to people living with MND and their families
- To provide education to health service providers
- To advocate for people living with MND
- To raise public awareness about MND
- To contribute to MND research.

### **Activities and services**

- Case co-ordination
- Emotional support
- Information and education
- Equipment service
- Advocacy.

### **Publications/resources**

- MNDAWA 'Care Advisory Service' leaflet
- Motor Neurone Disease Association WA leaflet
- 'mnd matters' newsletter, 2 monthly
- [mndawa.asn.au](http://mndawa.asn.au)
- Fact sheets.

# Mucopolysaccharide and Related Diseases Society

Operations Manager: Vanessa Ede-Scott

Phone: 1800 045 515

Email: [info@mpssociety.org.au](mailto:info@mpssociety.org.au)

Website: [www.mpssociety.org.au](http://www.mpssociety.org.au)

## Mucopolysaccharide and related diseases

The Mucopolysaccharidoses (MPSs) are rare genetic disorders in children and adults. They involve an abnormal storage of mucopolysaccharides, caused by the absence of a specific enzyme. Without the enzyme, the breakdown process of mucopolysaccharides is incomplete. Partially broken down mucopolysaccharides accumulate in the body's cells causing progressive damage. The storage process can affect appearance, development and the function of various organs of the body. Each MPS disease is caused by the deficiency of a specific enzyme...

### Synonyms of mucopolysaccharide disease

Mucopolysaccharidoses

### Disorder subdivisions

- Table of Diseases
- MPS I - Hurler, Hurler-Scheie and Scheie syndromes
- MPS II - Hunter syndrome
- MPS III - Sanfilippo syndrome
- MPS IV - A and B Morquio syndrome
- MPS VI - Maroteaux-Lamy syndrome
- MPS IX Hyaluronidase deficiency
- MPS VII Sly syndrome
- ML II & ML III Mucopolidoses

### Objectives of the group

- To provide support and information to families and other interested people
- To promote a partnership between families and professionals
- To promote a community awareness of the mucopolysaccharide conditions and its impact on families and carers.

### Activities and services

- act as a support group for our members through
  - MPS Awareness Day and Family days
  - The publication of newsletters,
  - the holding of biennial National Conferences and
- promote a partnership between families and professionals, both locally and internationally;
- promote community awareness of MPS and its impact on families and carers.

### Publications/resources

The Society has various booklets and information sheets available that cover the different syndromes as well as helpful ideas.

## **Multiple Sclerosis Society of WA Inc (MSWA)**

Locked Bag 2  
BENTLEY DC WA 6983

Phone: (08) 9365 4888

Fax: (08) 9451 4453

Email: [enquiries@mswa.org.au](mailto:enquiries@mswa.org.au)

Website: [www.mswa.org.au](http://www.mswa.org.au)

### **Multiple sclerosis**

Multiple sclerosis (MS) is a chronic, neurodegenerative condition that affects the central nervous system (CNS). The body's own immune system appears to attack the protective sheath of fatty protein, called myelin, which surrounds the nerves in the brain, spinal cord and the optic nerve. An attack results in inflammation and development of one or more lesions, resulting in scarring or sclerotic plaques, forming on the nerves. These lesions interfere with the nerve messages sent back and forth through the brain and spinal cord. We do know the condition is not contagious, and while MS is not directly inherited, genetics does play an important role in who gets the disease.

### **Objectives of the group**

The Multiple Sclerosis Society WA supports people with MS (not considered a rare disease) but also provides support and services to people with other genetic and/or rare neurological conditions including MND, Huntington's, Cerebellar Spino Atrophy etc.

We are a large service provider with health and disability programs in the metro and some regional areas.

Our objectives are to provide quality and timely information, support and services, to enhance their quality of life and maximise independence.

We also provide high support accommodation and residential respite options.

### **Activities and services**

- Nursing and allied health services
- In home care and supports
- Centre based programs eg recreational and social activities
- Residential respite options
- High support accommodation options
- Education programs.

### **Publications/resources**

- Numerous fact sheets, information resources, research updates
- Quarterly magazine
- Monthly E-newsletter.

# Muscular Dystrophy WA

Suite B The Niche  
11 Aberdare Rd  
NEDLANDS WA 6009

PO Box 680  
NEDLANDS WA 6909

Phone: (08) 9380 3400  
Email: [info@mdwa.org.au](mailto:info@mdwa.org.au)  
Website: [www.mdwa.org.au](http://www.mdwa.org.au)

## Muscular dystrophy

Muscular dystrophy is a neuromuscular, genetic disorder which results in the progressive deterioration of muscle strength and function.

There are many other types of neuromuscular conditions, affecting people of all ages. While most forms of muscular dystrophy occur in babies or children, some others appear in late adolescence or adulthood. The incidence of persons with a neuromuscular condition is estimated to be 100 per 100,000 head of the population. It is estimated that there are more than 20,000 people in Australia who have some form of neuromuscular condition.

Researchers and doctors are learning more about the causes of muscular dystrophy and various treatments are being trialled to stop the progression of the disease. Nonetheless, through early diagnosis, proper therapy and support an increased quality of life is possible.

### Objectives of the group

At Muscular Dystrophy WA our mission is to enrich the lives of Western Australians living with muscular dystrophy and neuromuscular conditions.

We start with the people.

**The condition does not define a person.** It's their ambitions and personalities that make them unique.

We believe in practical personal support today. Not just hope for a cure in the future.

We work with hundreds of families to provide personalised support, advice and services. By connecting families, communities and care, people can make their own decisions about their life and well-being.

Most importantly, we understand everyone wants to live a life they love and to reach their full potential.

### Activities and services

Muscular Dystrophy WA provides a range of support services for people living with a neuromuscular condition and those that support them.

- Advocacy – we provide information, linkages, representation and guidance on a range of issues including National Disability Insurance Scheme, service provision and funding.
- CoughAssist machines – currently we provide over 60 families with long-term loans of these potentially life-saving machines
- Counselling – free individualised counselling is available to people with neuromuscular conditions and their families.
- Duke of Edinburgh International Award – we deliver this self-development program to young adults with neuromuscular conditions.
- Education and Information – a range of education and information options are available - MDWA Education sessions, Community Group education, Fact sheets, Newsletters
- Hales Mates – in conjunction with Hale School, we provide a peer mentoring program for school-aged children with a neuromuscular condition.
- Research – we continue to provide funding to support vital research into innovative therapies for neuromuscular conditions.
- Social Support Program
  - School Aged Children
  - Young Adults Social Group
  - Adults with a neuromuscular condition
  - Carers

## **Myasthenic Association (The Australian, in NSW Inc)**

17 Aldridge Avenue  
CORRIMAL EAST NSW 2518

Contact Name: Glenda  
Phone: (02) 4283 2815

Email: [info@myasthenia.org.au](mailto:info@myasthenia.org.au)  
Website: [www.myasthenia.org.au](http://www.myasthenia.org.au)

### **Myasthenia gravis (MG)**

Myasthenia gravis is a chronic autoimmune disease characterised by varying degrees of weakness of the skeletal (voluntary) muscles of the body. The name 'myasthenia gravis', which is Latin and Greek in origin, literally means "grave muscle weakness". It is a chronic autoimmune disease characterised by varying degrees of weakness of the skeletal (voluntary) muscles of the body. The name 'myasthenia gravis', which is Latin and Greek in origin, literally means "grave muscle weakness".

With current therapies, however, most cases of myasthenia gravis are not as 'grave' as the name implies. In fact, for the majority of individuals with myasthenia gravis, life expectancy is not lessened by the disorder.

### **Objectives of the group**

- To provide support and information to myasthenics, their families, supporters/carers.
- To bring informed knowledge of myasthenia gravis and their needs and problems of myasthenics to the notices of the general public and the medical profession.
- To encourage research.

### **Activities and services**

We hold quarterly meetings as well as support by phone or email.

### **Publications/resources**

- Quarterly newsletter for members or interested parties
- Pamphlet on 'Drugs that may aggravate Myasthenia Gravis'.



# **Noonan Syndrome Awareness Association**

Email: [brettzani@noonansyndrome.com.au](mailto:brettzani@noonansyndrome.com.au)  
Website: <https://noonansyndrome.com.au/>  
Facebook: <https://www.facebook.com/NoonanSyndromeAwarenessAssociation/>

## **Noonan Syndrome**

Noonan Syndrome is a genetic condition that affects many areas of the body and occurs in as many as 1 in 1000 individuals.

## **Objectives of the group**

- Increase awareness of Noonan syndrome in the community and health professionals
- Reduce Noonan syndrome misdiagnosis rates
- Facilitate access of services and improve outcomes for people with Noonan syndrome

## **Activities and services**

- Seminars and PD for health professionals
- Source of resources for individuals with Noonan syndrome, teachers/educators, carers and health professionals
- Support and liaise with people with Noonan syndrome

## **Publications/resources**

Are listed on the NSAA website.

# **Osteogenesis Imperfecta Society of Australia**

Contact name: Fern  
Phone: 0409 681 869

Email: [wa@oiaustralia.org.au](mailto:wa@oiaustralia.org.au)  
Website: [www.oiaustralia.org.au](http://www.oiaustralia.org.au)  
Facebook: [www.facebook.com/groups/77895455278/](http://www.facebook.com/groups/77895455278/)

## **Osteogenesis imperfecta (OI)**

OI is a genetic disorder of collagen, a protein which forms the framework for the bone structure. In OI collagen may be of poor quality or there may not be enough to support the mineral structure of the bones. This makes bones weak and fragile and likely to fracture at any given time without trauma.

## **Synonyms of osteogenesis imperfecta**

Brittle bones

## **Objectives of the group**

- To offer information and support to individuals and families affected by OI
- To encourage and support research
- To provide public and professional education and create awareness.

## **Activities and services**

- National conference every 2 years
- Wishbone day celebration
- Meetings and social get togethers during the year
- Support for newly diagnosed families.

## **Publications/resources**

- OI Society of Australia website [www.oiaustralia.org.au](http://www.oiaustralia.org.au)
- OI Society Private Facebook Groups – for adults and families  
[www.facebook.com/groups/77895455278/](http://www.facebook.com/groups/77895455278/)

## **Our Faces Support**

27 Schubert Street  
STRATHPINE QLD 4500

Phone: (07) 3881 3037

Email: [jenny.woolsey@hotmail.com](mailto:jenny.woolsey@hotmail.com)

Website: <https://www.ourfacesaustralia.org/>

Facebook: <https://www.facebook.com/groups/1847184975533482/>

## **Our Faces Support**

Our Faces Support is a new Australia-wide support group for individuals, family and friends who are affected by having a facial difference. The facial difference can be from birth or acquired as a result of an accident or illness.

### **Facial differences**

Craniofacial syndromes/conditions

Burns

Scars

Birthmarks

Cleft lip

### **Objectives of the group**

- To provide support and information to families and other interested people
- To promote awareness in the community about the impact of having a facial difference.

### **Activities and services**

- Phone, email and Facebook group support are offered
- As the group grows state get-togethers will be held.

### **Publications/resources**

- A three monthly newsletter
- Website with resources and links.

# **Pallister-Hall Syndrome (PHS) Support Hub**

PO Box 88  
TOOWOOMBA QLD 4350

Contact: George W Helon  
Phone: (07) 4635 9588  
Mobile: 0419 768 792

Email: [ghelon@yahoo.com.au](mailto:ghelon@yahoo.com.au)  
Facebook: [www.facebook.com/Pallister.Hall.Syndrome](http://www.facebook.com/Pallister.Hall.Syndrome)

## **Pallister-Hall syndrome**

Pallister-Hall syndrome (PHS) is an extremely rare genetic disorder that may be apparent at birth. It is a multiple anomaly congenital syndrome not a disease, and it affects all races of people. PHS follows a pattern of genetic inheritance in families called autosomal dominant inheritance.

Primary diagnostic features include: the presence of hypothalamic hamartoma (HH), bifid epiglottis, gelastic seizures, syndactyly, polydactyly, nail dysplasia and cranio-facial abnormalities.

Quite contrary to much earlier medical literature and resources, PHS is no longer considered a disorder that will result in early death; in fact, patients - given the correct medical intervention- will more than likely live a long and fruitful life.

## **Objectives of the group**

- To educate and inform the public
- To facilitate contact and support for PHS patients, their families, carers, medical providers, allied health professionals and interested researchers worldwide.

## **Activities and services**

Worldwide network support group offering:

- Phone support by appointment only
- Email and online support 24/7
- Public advocacy and peer support
- Guest speakers by negotiation
- Library services
- Fact sheets.

## **Publications/resources**

- Web presence at [www.facebook.com/Pallister.Hall.Syndrome](http://www.facebook.com/Pallister.Hall.Syndrome).
- Patient resource guide development in progress.
- See also Gelastic Seizure Support Hub.

# **Pallister-Killian Syndrome Foundation of Australia**

PO Box 4307  
MYAREE WA 6960

Mobile: 0401 044 814

Email: [nic.acquarola@pks.org.au](mailto:nic.acquarola@pks.org.au)  
Website: [www.pks.org.au](http://www.pks.org.au)

## **Pallister-Killian syndrome**

Pallister-Killian syndrome (PKS) is a rare chromosomal disorder in which there are 2 extra copies of 12p. Features and severity varies, with the main features including: hypotonia, developmental delay, hearing and vision impairment, mild to profound intellectual impairment, seizures, apnoea, congenital heart defects and gastro-oesophageal reflux. The number of affected cells does not correlate to disease severity.

PKS is not related to environmental, dietary or lifestyle factors. It is also not related to the age of the parents, no recorded incidence of familial inheritance or subsequent pregnancies with PKS. Approximately 20 known individuals live with this condition in Australia and New Zealand.

## **Synonyms of Pallister-Killian syndrome**

Mosaic tetrasomy 12p  
Teshler-Nicola  
Isochromasome 12p  
Pallister mosaic aneuploidy syndrome

## **Objectives of the group**

- Maximise the quality of life for individuals diagnosed with PKS
- Facilitating physical, emotion and educational support through the provision of equipment and services
- Facilitate the provision and enhancement of support to parents or guardians
- Assisting and liaising with medical institutions with their service provision
- Promote medical and scientific research in PKS
- Operate as a not for profit, benevolent and charitable entity.

## **Activities and services**

- Bi-annual nation general meeting/conferences
- Support group discussion through social media (closed group)
- Provision of an informative website
- Parent links/networking.

## **Publications/resources**

- Brochures and newsletters
- Webpage.

## **Parenteral Nutrition Down Under (PNDU)**

128 Rainbow Street  
RANDWICK NSW 2031

Email: contactpndu@gmail.com  
Website: www.pndu.org  
Facebook: email PNDU for an invitation to join the private Facebook group

### **Intestinal failure (IF)**

Intestinal failure (IF) is a reduction in gut function to below the minimum necessary for absorption of nutrients, water, vitamins and electrolytes. This means it becomes impossible to eat or absorb goodness from food of any kind- whether it is normal food or special formulas, modified diets or tube feeds - so the patient needs parenteral nutrition (intravenous feeding) to live. Parenteral nutrition therapy can be done at home (Home Parenteral Nutrition or HPN).

### **Disorder Subdivisions**

Total parenteral nutrition or home parenteral nutrition  
Intravenous nutrition

### **Objectives of the Group**

PNDU is a support group run specifically by and for Australian and New Zealand HPN consumers and their families/carers. PNDU aims to support, research and inform consumers, carers and providers of parenteral nutrition for IF. PNDU brings HPN consumers and carers together to provide comfort, strength and support to each other, as well as work with the medical profession and industry for the benefit of all HPN consumers in Australia and New Zealand.

### **Activities and services**

- Private email forum
- Letter-writing companion for Australian and New Zealand HPN consumers/carers
- Social gatherings
- HPN workshops
- Awareness raising (including annual HPN Awareness Week)
- Member surveys
- Representation at medical conferences
- Involved in international collaboration of HPN patient organisations and to various bodies about HPN issues.

### **Publications/resources**

- Quarterly e-newsletter
- Travel information booklet
- Restaurant card
- "Going Home on HPN" hospital leaflet.

## Parkinson's Western Australia

The Niche  
11 Aberdare Road  
NEDLANDS WA 6009

Phone: (08) 6457 7373  
Email: info@parkinsonswa.org.au  
Website: www.parkinsonswa.org.au  
Facebook: www.facebook.com/ParkinsonsWA

### Parkinson's disease

Parkinson's is a progressive neurological condition characterised by motor and non-motor symptoms.

#### Disorder subdivisions

Neurological disorders	Dyskinesia
Aged care	Progressive supranuclear palsy
Multiple system atrophy	Cortico basal degeneration
Lewy body dementia	

#### Objectives of the group

Parkinson's WA is a not-for-profit organisation providing a specialist nursing service, support networks, education and training and research. Parkinson's WA are working to encourage, develop and implement programs for the cure, treatments and care of people with Parkinson's.

#### Activities and services

Parkinson's WA provides a range of services to people with Parkinson's, their families and carers, including:

- Information and referral to Parkinson's Nurse Specialists – visits in the home and Aged care facilities.
- Upskilling allied health professionals in best practice management of Parkinson's
- Clinical services
- Seminars
- Support groups
- Support therapies e.g. Dancing, Singing, Yoga and Tai Chi
- Research.

#### Publications/resources

Information sheets for people with Parkinson's and health professionals

- Telephone support
- Nurse consultations and home visits
- DVD of seminars
- GP on line course
- Specialised Health Professional manuals e.g. Guidelines for Speech Pathologists and Guidelines for Physiotherapy practice in caring for people with Parkinson's.

## **Pompe Association, Australian**

Phone: (02) 9629 2842

Email: [president@australianpompe.com](mailto:president@australianpompe.com)

Website: [www.australianpompe.com](http://www.australianpompe.com)

### **Pompe disease**

Pompe disease is an ultra-rare disease and is caused by deficiency or total lack of the enzyme acid alpha glucosidase.

This enzyme is responsible for breaking down excess glycogen which normally accumulates in the muscles.

As the Pompe patient is deficient in this enzyme, the excess glycogen is not broken down and continues to accumulate in the muscle-cells. This results in progressive muscle damage and severe muscle weakness, so that normal muscle function is increasingly impaired.

Respiratory muscles are also involved, severely affecting pulmonary functions. Treatment for Pompe disease is currently only funded by the federal government for patients who meet the treatment guidelines and are diagnosed between 0 and 18 years old.

### **Objectives of the group**

The Australian Pompe Association is a voluntary support group that represents some 34 patients in Australia. Pompe affects males and females equally and there are Pompe patients in every state and territory. The Association is a registered charity supported by member and voluntary donations, education grants, fundraising and it managed by the members themselves. Their objectives are to act as a support network for Pompe patients and families, to press for treatment funding all Pompe patients in Australia and to increase public awareness about Pompe disease.

### **Activities and services**

- Website
- Member information
- National conferences
- Advocacy.



## **Retina Australia (WA)**

PO Box 183  
Osborne Park WA 6917

Phone: (08) 9388 1488

Fax: (08) 9388 1447

Email: [info@rawa.com.au](mailto:info@rawa.com.au)

Website: [www.rawa.com.au](http://www.rawa.com.au)

Facebook: <https://www.facebook.com/RetinaAustralia/>

### **Retinitis pigmentosa**

Degenerative retinal diseases affect millions of people worldwide, causing loss of vision in adults and children alike. The many diseases within this group are caused by any of thousands of known genetic mistakes in approximately 200 genes. These genetic mistakes and therefore the diseases they cause, are inherited. An individual may inherit such a genetic mistake from a parent, and pass it down to their own children, completely unaware of this silent problem until a family member, be it adult or child, is diagnosed with a sight stealing disease. The roller coaster begins.

Based on current statistics, the most likely diagnosis would be

- 1) Retinitis Pigmentosa<sup>[98]</sup>
- 2) Macular Dystrophy (MD) ), which affects central vision critical in daily life, or
- 3) the age related form of MD, Macular Degeneration (AMD) which requires both a genetic predisposition and an environmental cue before disease onset later in adult life.'

### **Synonyms of retinitis pigmentosa**

Progressive pigmentary retinopathy  
Rod-cone dystrophy

### **Objectives of the group**

- To raise funds for research that will enable prevention, treatment and cures for inherent retinal diseases.
- To raise awareness of RP and associated dystrophies
- To offer support for newly diagnosed individuals
- To ascertain the cause and means to cure or arrest all forms of hereditary retinal dystrophies
- To seek by all practical means ways to assist those affected and their families
- To gather and disseminate information on RP and all forms of hereditary retinal dystrophies
- To affiliate with Retina Australia and Retina International and to cooperate with all blind welfare and low vision agencies.

### **Activities and services**

- Research funding
- Information
- Fundraising.

## **Publications/resources**

The Foundation provides a newsletter and information booklets to the community. A medical library has brochures and booklets for health professionals.

# **Rett Syndrome Association of WA**

Phone: 0431 200 205  
Email: sanaafay@hotmail.com

## **Rett syndrome**

Rett syndrome is a neurological disorder occurring primarily in girls, in which individuals exhibit reduced muscle tone, autistic-like behaviour, stereotyped hand movements consisting mainly of wringing and waving, loss of purposeful use of the hands, diminished ability to express feelings, avoidance of eye contact, a lag in brain and head growth, gait abnormalities and seizures.

## **Synonyms of Rett syndrome**

Autism  
Ataxia and loss of purposeful hand use  
RS  
RTS

## **Objectives of the group**

- To collect and disseminate accurate and objective information regarding the cause, identification, treatment, prevention and cure of Rett syndrome
- To promote the general welfare of individuals with Rett syndrome
- To support families in coping with the condition
- To develop understanding and awareness of Rett syndrome
- To further the advancement of study, research, therapy and care.

## **Activities and services**

- General support and information
- Parent to parent support when required
- Contribute to the Australian Rett syndrome study
- Meetings throughout the year and an end of year Christmas picnic.

## **Publications/resources**

The Association provides an Australian quarterly newsletter and other overseas newsletters, a parent contact list, pamphlets, journal articles, books and videos.

# **Sanfilippo Children's Foundation (SCF)**

PO Box 475  
FRESHWATER NSW 2096

Phone: 1800 664 878

Email: [info@sanfilippo.org.au](mailto:info@sanfilippo.org.au)

Website: [www.sanfilippo.org.au](http://www.sanfilippo.org.au)

Facebook: [www.facebook.com/Sanfilippochildrensfoundation](http://www.facebook.com/Sanfilippochildrensfoundation)

## **Sanfilippo syndrome**

Sanfilippo syndrome (AKA MPSIII) is a metabolic disorder resulting in severe central nervous system (CNS) damage. The lack of enzyme prevents the body from going through its natural recycling process, causing cellular malfunction.

## **Subdivisions of Sanfilippo syndrome**

MPS III type A, B, C, D

## **Objectives of the group**

The mission of the Sanfilippo Children's Foundation is to drive research, enable access to effective treatments and improve the quality of life for Australian children affected by the fatal childhood disease, Sanfilippo syndrome.

This will be achieved by:

- Enabling participation in clinical trials or special access to emerging treatments until such time as an effective treatment is developed and available on the market
- Improving the diagnosis path- more accurate and earlier diagnosis to enable appropriate treatment
- Raising awareness of the disease among the community, including the medical profession
- Providing diagnosed families with clear, up-to-date information on the disease and the latest medical advances
- Fundraising to achieve those aims.

## **Senses Australia**

PO Box 143  
BURSWOOD WA 6100

Phone: (08) 9473 5400

Fax: (08) 9473 5499

Email: [admingroup@senses.org.au](mailto:admingroup@senses.org.au)

Website: [senses.org.au](http://senses.org.au)

### **Senses Australia**

Senses Australia is a not-for-profit organisation assisting individuals of all ages with disability to meet their goals and aspirations through the provision of contemporary and responsive services.

Senses Australia has a specialist Deafblind Service which supports those who have a combined loss of hearing and vision which significantly affects communication, socialisation, mobility and daily living. Deafblind individuals are affected to varying degrees with most having some level of hearing or vision. The conditions of deafness or blindness may be acquired or present at birth. The main causes are congenital rubella syndrome, Usher syndrome, CHARGE syndrome and ageing.

### **Objectives of the group**

To assist individuals of all ages with disability who meet Senses Australia's eligibility criteria, to meet their goals and aspirations through the provision of contemporary and responsive services.

### **Activities and services**

The Senses Australia provides a range of services for preschool and school age children, teenagers and adults. Senses Australia's services are for the person with disability, their family, other professionals and service providers.

- Accommodation a high quality service supporting people in the living option of their choice.
- Individual funding a person-centred service which encourages self direction and access to a range of meaningful activities and involvement in community life while developing skills, enjoyment and satisfaction.
- Therapy including physiotherapy, occupational therapy, speech pathology, psychology and social work are offered, which contribute to the individual's wellbeing, independence, positive relationships and development of abilities and participation in community life.
- Respite flexible support for the family in their caring role – in or out of home – and a positive experience for the individual.
- Deafblind service a high quality information, resources and training service for individuals with congenital and acquired deafblindness.
- Support Groups run throughout the year to meet the needs of our clients as well as their families and carers.

### **Publications/resources**

Senses Australia produces a newsletter 'In Touch' and can be viewed or downloaded from the website: [www.senses.org.au/resources-products/in-touch](http://www.senses.org.au/resources-products/in-touch)

## **Short Statured People's Association WA Branch**

Phone: (08) 9295 0636

Email: [eleanorallansnook@gmail.com](mailto:eleanorallansnook@gmail.com)

Website: [www.sspa.org.au](http://www.sspa.org.au)

Facebook: <https://www.facebook.com/sspanational/>

### **Short stature**

Short stature may occur as one of many genetic conditions causing the abnormal growth or development of the skeleton. Short stature is identified as height less than 1.45 metres tall at maturity and often with disproportionate limbs and skull. There are many different forms of short stature, with different modes of inheritance and different rates of occurrence.

### **Objectives of the group**

To support individuals of short stature and their families and parents who have short statured babies. Also to assist the assimilation of short statured people into society with the goal of equality of educational and social status and of employment opportunities.

### **Activities and services**

- Social meetings approximately 3-4 times per year
- Picnics and other recreational activities for children
- Advice regarding pre-school and school choices
- Support through mutual discussion and contact with other parents
- Community education through seminars and videos on short stature
- Journal informing members of the work being carried out by the national group and state sub-groups
- An annual national convention
- Promotion of personal development and self-esteem through contact with others of short stature and of average height
- Advice and assistance on career and TAFE opportunities
- Promotion of research into causes and treatment through National Medical & Scientific Advisory Board.

### **Publications/resources**

The Association has a range of publications and guides available upon request.

## **Red Nose WA (Formerly Sids and Kids WA)**

33 Sixth Avenue  
KENSINGTON WA 6151

Phone: (08) 6365 7164  
Country Free call: 1800 199 466  
24 hour telephone support in WA: 1800 686 780

Email: [gerrywalsh@rednose.com.au](mailto:gerrywalsh@rednose.com.au)  
Website: <https://rednose.com.au/>  
Facebook: [www.facebook.com/rednoseaus](http://www.facebook.com/rednoseaus)

### **Objectives of the group**

SIDS and Kids Western Australia is a not-for-profit organisation dedicated to saving babies' lives from pregnancy through childhood and to providing loss and grief support to those affected by the death of a child from sudden infant death syndrome (SIDS).

- To educate the public and professionals raise funds for scientific and medical research into the cause and prevention of SIDS and stillbirth
- To give support to parents, family and those affected by the death of a child from pregnancy through childhood, regardless of the cause of death (e.g. miscarriage, stillbirth, SIDS, neonatal death, accident, illness)
- To educate the public in the nature, incidence, causes and how to reduce the risk of sudden unexpected death in infancy to save babies lives.

### **Activities and services**

Services are free to anyone affected by the death of a child from conception onwards:

- Support and counselling services
- Education/prevention
- Research.

### **Publications/resources**

SIDS and Kids WA have a variety of booklets and articles relating to different aspects of grief. They also have an on-line catalogue for research articles, which can be found on the National Website [www.sidsandkids.org/research/](http://www.sidsandkids.org/research/). SIDS and Kids WA offer free subscription to their newsletter.

# **Sjögren's Syndrome Association Inc, Australian**

Phone: (02) 6672 4463  
Mobile: 0438 347 444

Email: bevnorton@sjogrens.org.au  
Website: www.sjogrens.org.au  
Facebook: www.facebook.com/SjogrensAustralia  
Twitter: www.twitter.com/SjogrensAust

## **Sjögren's syndrome**

A disorder in which the body's immune system mistakenly attacks its own moisture producing glands. Lymphocytes, a type of white blood cell, infiltrate and destroy these glands causing decreased production of saliva and tears.

The hallmark symptoms of Sjögren's syndrome are dry eyes and dry mouth, but it is a systemic disease, affecting many organs and may cause fatigue. It is one of the most prevalent autoimmune disorders, striking as many as 0.5% of Australians.

## **Synonyms of Sjögren's syndrome**

Sjögren's peripheral neuropathy  
Associated immune diseases

## **Objectives of the group**

- Increase awareness of Sjögren's
- Educate health care professionals about Sjögren's
- Information and referral service for patients, carers and their families.

## **Activities and services**

- Membership drives
- Telephone support
- Support on Skype and Facebook
- Support through our website and health unlocked interactive website (<http://sjogrens-aus.healthunlocked.com>) which has Sjögren's solutions in video modules.

## **Publications/resources**

- Newsletter posted to members
- Publications on website.



# **SOFTWA (Support Organisation for Trisomy and Related Disorders of WA)**

Contact: Mandy  
Phone: (08) 9591 3316  
Mobile: 0422 106 354

Email: [mandycain@bigpond.com](mailto:mandycain@bigpond.com)  
Website: [www.trisomy9.org/support.htm](http://www.trisomy9.org/support.htm)

## **Trisomy and Other Related Disorders**

Trisomy disorders have a range of different characteristics including physical and intellectual disability. There are different Trisomy conditions, including Trisomy 18 ), trisomy 13 (Patau syndrome), trisomy 9 and trisomy 22.

### **Synonyms of trisomy disorders**

Chromosomal triplication	Cat eye syndrome
Trisomy 9 mosaic	Trisomy 22 mosaic
Trisomy 9 mosaicism syndrome	Trisomy 22 mosaicism syndrome
Trisomy 9 (Complete trisomy 9 syndrome) included	Chromosomal triplication

### **Disorder subdivisions**

Trisomy 18  
Trisomy 13  
Trisomy 9  
Trisomy 22

### **Objectives of the group**

To help and provide support to families dealing with a child with a rare chromosomal disorder.

### **Activities and services**

- Provide information and about the condition
- Provide links to other individuals dealing with the same condition
- Organise social events including picnics and other recreational activities.

### **Publications/resources**

The organisation provides information on a range of rare chromosomal conditions to the community and health professionals.

# **Spinocerebellar Ataxia Australia Inc**

PO Box 115  
HOWLONG NSW 2643

Email: mdunn6418@gmail.com  
Website: www.scars.org.au  
Facebook: www.facebook.com/scarsau

## **Spinocerebellar ataxia (SCA)**

'Ataxia' means 'absence of order'. People with ataxia have problems of coordination. This is because parts of the nervous system that normally control coordination and balance are affected. Ataxia is the principal symptom of a group of neurological disorders called the cerebellar ataxias. Most are progressive. Ataxia may also be a symptom of other conditions such as multiple sclerosis or cerebral palsy.

## **Objective of the group**

To promote and support spinocerebellar ataxia in all forms and to lead the quest for knowledge, diagnosis, treatment and prevention of SCA with support from communities worldwide. The group is run entirely by volunteers. There are support groups in Melbourne and Brisbane, as well as in South Australia run by Muscular Dystrophy SA.

## **Activities and services**

- Website [www.scars.org.au](http://www.scars.org.au)
- Membership
- Ataxia Awareness Day annually
- Fundraising activities for research.

## **Publications/resources**

- Brochure
- Quarterly newsletter.

# **Syndromes Without A Name (SWAN) Australia**

PO Box 390  
FAIRFIELD VIC 3078

Phone: 0404 280 441

Email: [info@swanaus.org.au](mailto:info@swanaus.org.au)  
Website: [www.swanaus.org.au](http://www.swanaus.org.au)  
Facebook: [www.facebook.com/SWANAustralia](http://www.facebook.com/SWANAustralia)  
Twitter: [www.twitter.com/swanaus](http://www.twitter.com/swanaus)

## **Objectives of the group**

SWAN provides information and support to families caring for a child with an undiagnosed or rare genetic condition for which there is no other support group.

We aim to increase the awareness and understanding of the impact of the prevalence of undiagnosed genetic conditions within the wider community. We provide information and emotional support to families to assist them with establishing enduring mutually supportive relationships. We want to limit the isolation and the negative social, emotional and financial impacts experienced by families.

We advocate for further funding into genetic research, so that more testing is available, turnaround time on testing is decreased and more accurate results are achieved. We strive to obtain better resources and pathways for our children without a diagnosis in the future.

## **Activities and services**

- Workshops
- Seminars
- Social events
- Undiagnosed Children's Awareness Day
- Networking events.

## **Publications/resources**

- Brochure
- A4 Information Sheet.

# Tarlov Cyst Disease Society of Australia

28 Buena Vista Rd  
WOODFORD NSW 2778

Email: tarlovcystsocietyaustralia@gmail.com  
Facebook: Facebook public page:  
<https://www.facebook.com/tarlovcystsocietyaustraliaandnewzealand/>  
Facebook support page:  
<https://www.facebook.com/groups/tarlovcystaustralia/>

## Objectives of the group

The Tarlov Cyst Society, Australia is a support group for Tarlov Cyst Disease patients within Australia and a sister group to Tarlov Cyst Disease Society, Canada. We exist for the sharing of information and support with one another. Between our members we share a wealth of information and are more than an online community, we are a family.

We are of all ages, backgrounds, and social classes and have come together to help and support each other. We are not specifically a religious or ethnic group, meaning all are welcome here.

We aim to promote awareness of the rare genetic disease Tarlov Cyst Disease.

Tarlov cysts are also known as perineural/perineurial, or sacral nerve root cysts. They are dilations of the nerve root sheaths and are abnormal sacs filled with cerebrospinal fluid that can cause a progressively painful radiculopathy (nerve pain). Currently, as far as we are aware, there are no medical specialists within Australia who have success in the treatment of the disease. Patients are reliant on nerve pain medications to cope with the varying degrees of pain, some also require opiates to manage pain levels under medical supervision.

## Activities and services

- Email and online support
- Fact sheets
- Peer group
- Personal advocacy

## **Thalassaemia Australia Inc**

Phone: (03) 9888 2211

Fax: (03) 9338 2150

Email: [info@tasca.org.au](mailto:info@tasca.org.au)

Website: [tascsa.net.au](http://tascsa.net.au)

### **Thalassaemia**

The thalassaemia's are a group of inherited disorders in which the production of normal haemoglobin is partly or completely suppressed because of a defective synthesis of one or more of its component globin chains. There are three types; major, minor and intermedia, each differing in severity. Thalassaemia major results in severe anaemia requiring frequent blood transfusions.

### **Synonyms of Thalassaemia**

Cooley's anaemia

Mediterranean anaemia

### **Objectives of the group**

To provide community education in secondary schools, GPs, CALD groups and other medical bodies. We also have a role in patient support and advocacy.

### **Activities and services**

- Community education
- Fund raising events
- Patient support and advocacy.

### **Publications/resources**

All resources can be found on our website at [www.thalassaemia.org.au](http://www.thalassaemia.org.au)

## **Tourette Syndrome Support Group, Perth**

Contact: Lydia Roberts

Phone: 0437 234 180

Email: [lydyrob@gmail.com](mailto:lydyrob@gmail.com)

Facebook: [www.facebook.com/TouretteSyndromeAssociationOfAustralia#!/TouretteSyndromeAssociationOfAustralia](https://www.facebook.com/TouretteSyndromeAssociationOfAustralia#!/TouretteSyndromeAssociationOfAustralia)

### **Tourette syndrome**

Tourette syndrome is a genetic condition affecting neurological movement beginning between 2 and 16 years of age. It is characterised by rapidly repetitive multiple movements (tics) and involuntary vocalisations.

Approximately 10% of individuals with Tourette syndrome have a family history of the condition.

### **Synonyms of Tourette syndrome**

Chronic motor tic

Maladie de tics

Chronic multiple tics

Paulitis

Gilles de la Tourette's disease

Tics

Gilles de la Tourette's syndrome

Tourette's disorder

GTS

TS

Habit spasms

### **Objectives of the group**

- To support individuals with Tourette syndrome and their family and carers
- To increase public and professional awareness of this syndrome.

### **Activities and services**

- Telephone support service run by volunteers, contact number: (02) 9382 3726.
- Facebook forum which aims to help break down isolation many families and individuals with Tourette's feel and provides an opportunity to share experiences and advice. This is a social group designed for TSAA members & their friends & family.

### **Publications/Resources**

The organisation provides education packs to members, the public and health professionals at low cost. Please contact them for more details.

# **Tuberous Sclerosis Australia**

WA Contact – Nicole Stone

18 Central Rd  
BEVERLY HILLS NSW 2209

Phone: 1300 733 435 (Australia only)  
Email: [info@tsa.org.au](mailto:info@tsa.org.au)  
Website: [www.tsa.org.au](http://www.tsa.org.au)  
Facebook: [www.facebook.com/AuTSC](http://www.facebook.com/AuTSC)

## **Tuberous sclerosis complex**

Tuberous sclerosisComplex (TSC or TS) is a complex genetic disorder which affects more than 2500 individuals in Australia and thousands more carers, families and friends who live with the impact of the disease.Complex (TSC or TS) is a complex genetic disorder which affects more than 2500 individuals in Australia and thousands more carers, families and friends who live with the impact of the disease.

TSC tumours can grow in any organ of the body, commonly affecting the brain, heart, kidneys, skin and lungs. TSC can cause epilepsy, developmental delay and autism. Recently approved medicines for TSC are offering new treatment options for people with TSC.

## **Synonyms of tuberous sclerosis complex**

TSC  
TS  
Tuberous sclerosis

## **Objectives of the group**

TSC works to empower TSC affected families through information and support.  
We:

- Provide access to current information and resources
- Help TSC families build relationships and support networks
- Actively encourage best practice management and care for TSC affected people in Australia.

## **Activities and services**

- Education and social events
- Phone, email support and information
- Advocacy for improved health and other services
- Funding TSC research in Australia.

## **Publications/resources**

- Website ([www.tsa.org.au](http://www.tsa.org.au)) including TSC information pages, a resource directory and personal stories
- Monthly email newsletter

- Reach Out printed magazine.



# Turner Syndrome Association of Australia Ltd (WA)

Phone: (07) 3298 6635 or 0419 655 974

Email: [info@turnersyndrome.org.au](mailto:info@turnersyndrome.org.au)

Website: [www.turnersyndrome.org.au](http://www.turnersyndrome.org.au)

## Turner syndrome

Turner syndrome (TS) results from a chromosomal abnormality in which a female infant is born with only one X chromosome instead of the usual two, or is missing part of one X chromosome. In most cases, untreated females with this disorder are typically short in stature with an average final adult height of 140 centimetres and may have a variety of associated physical features and medical problems.

Because females with TS don't have proper ovarian development, they usually don't develop all of the secondary sexual characteristics expected during adolescence and are infertile as adults. Other health problems that may occur with TS include kidney and heart abnormalities, high blood pressure, obesity, diabetes mellitus, cataracts, thyroid problems, arthritis, as well as some learning difficulties (despite normal intelligence) and girls with TS are prone to hearing problems.

## Synonyms of Turner syndrome

45 X syndrome	Ovarian dwarfism, Turner type
Bonnevie-Ulrich syndrome	Ovary aplasia, Turner type
Chromosome X, monosomy X	Pterygolympangiectasia
Gonadal dysgenesis (45, X)	Schereshevskii-Turner syndrome
Monosomy X	Turner-Varny syndrome
Morgagni-Turner-Albright syndrome	XO syndrome

## Objectives of the group

- To assist individuals and families to have a healthier and better adjusted life
- To educate and to help people gain a greater knowledge of TS.

## Activities and services

- Education conferences
- Support group meetings.

## Publications/resources

Resources can be found on the website.

## **VCFS 22q11 Foundation**

Email: secretary@22q.org.au

Website: www.22q.org.au

### **Velocardiofacial syndrome (VCFS/ 22q11)**

Velocardiofacial syndrome is a genetic syndrome (a syndrome means a pattern of features occurring together).

VCFS/deletion 22q11 is the most common microdeletion syndrome,, the most common syndrome associated with cleft palates and the second most common syndrome associated with congenital heart disease.

VCFS is a multiple anomaly syndrome caused by a sub microscopic deletion of genetic material from the long arm of chromosome 22. In fact, over 180 disorders might occur in VCFS and they cover nearly every organ system in the body with broad reaching effects on development and behaviour, including speech, language, personality, mood, learning, attention, and temperament.

### **Objectives of the group**

The VCFS 22q11 Foundation supports people affected by 22q. We offer annual conferences, an informative website and regional contacts.

### **Activities and services**

- Social events
- Awareness days/weeks
- Support via social media
- Conferences.

### **Publications/resources**

All resources are available online [www.vcfs22q.org.au](http://www.vcfs22q.org.au)

## VHL (von Hippel-Lindau) Alliance

49 Adams Rd  
MARIGINIUP WA 6078

WA contact: Michael  
Phone: 0419 949 183

Email: michael@west-coast.com.au  
Website: www.vhl.org

### von Hippel-Lindau (VHL) syndrome

von Hippel-Lindau syndrome is a genetic condition involving the abnormal growth of blood vessels in some parts of the body which are particularly rich in blood vessels. It is caused by a flaw in one gene, the VHL gene, on the short arm of chromosome 3, which regulates cell growth. Tumours will develop in one or more parts of the body.

Many of these tumours involve the abnormal growth of blood vessels in different organs of the body. While blood vessels normally branch out like trees, in people with VHL little knots of blood capillaries sometimes occur in the brain, spinal cord or retina. These are called angiomas or hemangioblastomas.

Tumours in other parts of the body may themselves cause problems or problems may develop around them. VHL is different in every patient. Even in the same family, people may show only one or several features of VHL. Since it is impossible to predict exactly which one or more manifestations of VHL each person will have, it is important to continue to check for all the possibilities throughout a person's lifetime.

### Disorder subdivisions

Kidney	Inner ear
Brain and spinal cord (CNS)	Pheochromocytoma/paraganglioma
Retina	Reproductive organs
Pancreas	

### Objectives of the group

- To increase awareness of VHL within the community, government and medical profession
- To ensure that people with VHL, their carer's, families and support networks have access to the resources required to maximise their lifestyle
- To support research into VHL.

### Publications/resources

VHL Handbook: What you need to know about VHL- A reference handbook for people with von Hippel-Lindau, their families, and support personnel, published by the VHL Alliance.

# **Williams Syndrome Association of Western Australia (WSAWA Inc)**

26 Lapwing Rd  
DALYELLUP WA 6230

Phone: 0417 946 007 or 0408 913 348

Email: [wsawa@wsawa.org](mailto:wsawa@wsawa.org)

Website: [www.wsawa.org](http://www.wsawa.org)

Facebook: [www.facebook.com/WilliamsSyndromeAssociationOfWesternAustralia](http://www.facebook.com/WilliamsSyndromeAssociationOfWesternAustralia)

## **Williams syndrome**

Williams syndrome (WS) is a rare genetic condition that affects 1 in 50,000 people worldwide. WS is characterised by medical problems including cardiovascular disease, developmental delays and learning disabilities. These occur besides striking verbal abilities, highly social personalities and an affinity for music with no concept of 'stranger danger'.

## **Synonyms of Williams syndrome**

WS

Williams-Beuren syndrome (WBS)

## **Objectives of the group**

- Devoted to improving the lives of individuals with WS and their families
- Strives to locate individuals with the syndrome and their families.

## **Activities and services**

- Disseminates timely and accurate medical, education and assistance group information (including relevant governmental agencies)
- At least 2 social gatherings a year and an AGM with free entertainment, with one being the fully catered Christmas party with presents for the kids
- Assist members by paying for the second parent to fly interstate for medical needs
- Family weekends away once every 2 years, subsidised by the association
- A medical conference at PMH in 2 to 3 years for the members and other interested parties that will have medical, educational and disabilities support and health experts as guest speakers, paid by the association.

## **Publications/resources**

- Website ([www.wsawa.org](http://www.wsawa.org)) has links to other Australian WS websites, overseas websites, information and blogs on WS plus local WA website links to support services and information
- Library for members (being established) with texts, stories and journals on WS.

# **XYY Syndrome Association of Australia Inc.**

PO Box 3286  
MANDURAH EAST WA 6210

Phone: 0427 064 157

Email: [xyyaustralia@gmail.com](mailto:xyyaustralia@gmail.com)

Website: [xyyaustralia.org](http://xyyaustralia.org)

Facebook: [www.facebook.com/XYY-Syndrome-Association-of-Australia-Inc-198366960788495/?ref=br\\_rs](https://www.facebook.com/XYY-Syndrome-Association-of-Australia-Inc-198366960788495/?ref=br_rs)

## **XYY Syndrome**

XYY Syndrome is caused by an extra Y Chromosome being present in the cells of males with XYY Syndrome. Most males with this condition are taller than average, although they generally have normal levels of the male sex hormone testosterone. Their sexual development and ability to father children is not known to be affected by the extra Y Chromosome.

Males with XYY Syndrome often have an increased risk of behavioural, social, and emotional difficulties compared with their unaffected peers. These problems include attention deficit hyperactivity disorder (ADHD); depression; anxiety; and Autism Spectrum Disorder, which is a group of developmental conditions that affect communication and social interaction. XYY Syndrome is associated with an increased risk of learning difficulties and delayed development of speech and language skills.

Other possible signs and symptoms include delayed development of motor skills (such as sitting and walking), weak muscle tone (Hypotonia), hand tremors or other involuntary movements (motor tics), seizures, asthma, flat feet (pes planus), little fingers that curve inwards clinodactyly and abnormal side-to-side curvature of the spine (scoliosis). These characteristics vary widely among affected boys and men.

Reference: <https://www.ncbi.nlm.nih.gov/gtr/conditions/C3266843/>

Healthline: <https://www.healthline.com/health/xyy-syndrome#treatment>

## **Objectives of the group**

Our focus is to help the community have a positive understanding of XYY Syndrome and give support to those who need it.

- Creating awareness of XYY Syndrome by obtaining and providing the latest research for XYY families and the community
- Networking with government services, medical specialists, general practitioners and educators to create a better understanding of XYY Syndrome

- Rare Syndrome recognition for XYY Syndrome within State and Federal Government and educational systems

### **Activities and services**

- Facebook support association page: [https://www.facebook.com/XYY-Syndrome-Association-of-Australia-Inc-198366960788495/?ref=br\\_rs](https://www.facebook.com/XYY-Syndrome-Association-of-Australia-Inc-198366960788495/?ref=br_rs)
- Support page: <https://www.facebook.com/groups/475724605966667/>
- Information days
- Networking to create better understanding of XYY Syndrome
- Research projects

### **Publications/resources**

- Library for members (being established) with texts, stories and journals on website <https://xyyaustralia.org/>
- Brochures, awareness cards and the creation of information packs

## **Support Organisations**

### **Activ Foundation**

327 Cambridge St  
WEMBLEY WA 6913

PO Box 446  
WEMBLEY WA 6913

Phone: 1800 622 351 or (08) 9387 0555  
Fax: (08) 9387 0474

Email: [records@activ.asn.au](mailto:records@activ.asn.au)  
Website: [www.activ.asn.au](http://www.activ.asn.au)  
Facebook: <https://www.facebook.com/ActivFoundation>

### **Objectives of the group**

Activ is a family based, leading community benefit organisation and has been providing services and support to people living with disability since 1951. Activ's purpose is to provide a range of services and support that enables people with disability and their families to pursue a better life.

### **Activities and services**

- Supported accommodation options, including individual, shared and group home settings, to children and adults living with intellectual and/or physical disabilities
- Assistance for individuals living within their own homes and respite (out of home and in home) for both adults and children
- Individualised services (self-managed, shared management and/or coordination)
- Support to individuals through Home and Community Care (HACC) programs

- Supported employment options to people with disabilities in Activ Business Services
- Training including nationally recognised courses and qualifications
- Alternatives to Employment (ATE) and Post School Opportunities (PSO) programs
- Transport services to workplaces and other venues
- The opportunity for people with disability to experience holiday, leisure and adventure activities locally, interstate and overseas.

### **Publications/resources**

Activ's Library is the largest library in the Southern hemisphere that focuses on the single topic of disability. There is a vast collection of books, journals, training materials and resources suitable for parents, people with disability, carers, teachers and professional staff. The librarians can be contacted on (08) 9387 0458 or email: [library@activ.asn.au](mailto:library@activ.asn.au)

### **Related websites**

[www.parentportal.activ.asn.au](http://www.parentportal.activ.asn.au)

[www.citytosurf.activ.asn.au](http://www.citytosurf.activ.asn.au)

# Association for the Wellbeing of Children in Healthcare

Gladesville Hospital Bldg 40B  
Cnr Victoria & Punt Roads  
GLADESVILLE NSW 2111

Phone: (02) 9817 2349  
Parent line: 1800 244 396

Email: [awch@awch.com.au](mailto:awch@awch.com.au)  
Website: [www.awch.org.au](http://www.awch.org.au)  
Facebook: <https://www.facebook.com/awchkids/>

Non-profit organising promoting and raising awareness of the emotional and social needs of children and young people in healthcare.

## Objectives of the Group

Parents, professionals and community members who work together to ensure the emotional and social needs of children, adolescents and their families are recognised and met within hospitals and the health care system in Australia.

## Activities and services

- Support group information for chronic illness and rare diseases
- Hospital Ward Grandparent Scheme
- Hospital Familiarisation Program (AWCH WA).

## Publications/resources

- Books and DVDs available on loan from library
- Information brochures available about our organisation and services
- Hospital preparation and hospitalisation resources.



## **Asthma Foundation of WA**

36 Ord Street  
WEST PERTH WA 6005

PO Box 864  
WEST PERTH WA 6872

Phone: (08) 9289 3600  
Fax: (08) 9289 3601

Email: [ask@asthmawa.org.au](mailto:ask@asthmawa.org.au)  
Website: [www.asthmawa.org.au](http://www.asthmawa.org.au)

### **Asthma**

Asthma is a condition in which the airways in the lungs become narrow causing wheezing, coughing or difficulty in breathing. Asthma tends to run in families and generally the stronger the family history of asthma the more likely it is that the child will develop it.

### **Objectives of the group**

- To raise awareness of asthma in the community
- To raise funds for asthma research
- To provide educational and support services to individuals with asthma, their families, carers and health professionals.

### **Activities and services**

The Asthma Foundation of WA has a wide range of activities and services for the community and health professionals including:

- Free Community education sessions
- Free asthma training for school and preschool staff
- National Asthma Week is held in September each year for the community and health professionals
- 1800 ASTHMA free call telephone advice for people with asthma and their carers
- Asthma therapy equipment such as nebulisers and spacer devices are available for sale
- Telehealth education available for rural and remote community members on asthma and COPD
- Monthly telehealth professional development seminars for rural and remote health professionals
- The Coach Program – a confidential, free health coaching service delivered by regular scheduled phone calls by Asthma Foundation coaches to help people with asthma improve their general health and asthma control
- Manage My Asthma Kids Club – free membership
- Full lung function clinic – bulk billed

### **Publications/resources**

Asthma Foundation WA provides brochures, booklets and pamphlets to the community and health professionals.

# Be Inspired Foundation

P.O. Box 1811  
Subiaco, WA, 6904

Phone: 1300 799 915

Email: [info@beinspiredfoundation.com.au](mailto:info@beinspiredfoundation.com.au)

Website: [www.beinspiredfoundation.com.au/](http://www.beinspiredfoundation.com.au/)

Facebook: [www.facebook.com/beinspiredaustralia/](https://www.facebook.com/beinspiredaustralia/)

LinkedIn: [www.linkedin.com/company/be-inspired-foundation/](http://www.linkedin.com/company/be-inspired-foundation/)

Twitter: [twitter.com/BeInspiredFound](https://twitter.com/BeInspiredFound)

YouTube: [www.youtube.com/channel/UCw6QIzke-sdHeZ1pvMrOQTA](https://www.youtube.com/channel/UCw6QIzke-sdHeZ1pvMrOQTA)

Instagram: [instagram.com/beinspiredfoundation/](https://www.instagram.com/beinspiredfoundation/)

The mission of the Be Inspired Foundation (BIF) is to create access to wellness for young, disadvantaged Australians living with chronic disease, cancer or major trauma, by providing funding for evidence based integrated health services including exercise, diet and counselling.

Our slogan is "Building Lifelong Independence"

<https://www.youtube.com/watch?v=IXJxOHJQ7-M>

## Objectives of the group

To help beneficiaries to reach maximal medical improvement and reduce permanent impairment caused by chronic disease, disability, cancer or major trauma by funding integrated, multi-disciplinary treatment plans. We aim to empower beneficiaries with long-term strategies to manage their conditions and to improve their quality of life.

## Activities and services

The active rehabilitation services we fund include exercise rehabilitation, diet advice and counselling.

<https://www.beinspiredfoundation.com.au/services/active-rehabilitation/>

## Publications/resources

ESSA Conference - Poster 1

Be Inspired Foundation: Creating access to wellness for disadvantaged and young Australians living with cancer, major trauma, or chronic disease. (April 2016)

<http://dx.doi.org/10.13140/RG.2.2.13117.36329>

Conference: Exercise and Sport Science Australia (ESSA) - Research To Practice 2016

ESSA Conference - Poster 2

Active Rehabilitation Improves Function and Health of a Morbidly Obese 15 Year Old Boy: A Case Study of Treatment Provided by the Be Inspired Foundation (March 2018)

<http://dx.doi.org/10.13140/RG.2.2.33250.02245>

Conference: Exercise and Sport Science Australia (ESSA) - Research To Practice 2018

## **Cancer Council WA**

Cancer Nurses/Cancer Information and Support Services  
Level 3/420 Bagot Rd  
SUBIACO 6008

Cancer Council WA Phone: 13 11 20 (business hours)  
Email: [questions@cancerwa.asn.au](mailto:questions@cancerwa.asn.au)  
Website: [www.cancerwa.asn.au](http://www.cancerwa.asn.au)

### **Cancer Council**

The Cancer Council WA Cancer Council provides information about cancer related issues and support services for individuals, families and communities affected by cancer. Not only are we supported by health care professionals in updating our information, but we also support them in caring for cancer patients, their families and communities.

### **Activities and services**

- Information on specific cancers
- Information on chemotherapy, surgery, radiotherapy
- Information on new treatments and clinical trials
- Someone confidential and professional to talk to about what you're going through
- Information on prevention, screening and risk factors for cancer
- Information on any of the Cancer Council's support services, including counselling, financial assistance and accommodation for country people accessing health service in the metropolitan area
- Helpline Nurse
- Information about services in your community
- Information on how best to navigate your way around the health care system (i.e. finances, appointments, health care professionals)
- Information on any of the Cancer Council's support services, including counselling, financial assistance and accommodation for country people accessing health service in the metropolitan area
- Cancer Nurse

### **Publications/resources**

The Cancer Council WA provides a number of brochures relating to Cancer Council Services, Cancer Treatments, Coping with Cancer and Prevention. Most publications are available online, or contact the Cancer Council on 13 11 20.

## **Carers Association of WA (Inc)**

182 Lord Street  
PERTH WA 6000

Phone: 1300 CARERS (1300 227 377)  
8.00 am 5.00 pm Mon - Fri  
Carer Counselling Line: 1800 007 332  
Fax: (08) 9228 7488

Email: [info@carerswa.asn.au](mailto:info@carerswa.asn.au)  
Website: [www.carerswa.asn.au](http://www.carerswa.asn.au)  
Facebook: [www.facebook.com/pages/Carers-WA/444158565618199](https://www.facebook.com/pages/Carers-WA/444158565618199)

### **Objectives of the group**

Carers WA is a non-profit, community based organisation and registered charity dedicated to improving the lives of the estimated 310,000 unpaid family carers in WA. Carers WA provide information support and social activities for unpaid family carers who are providing care to a family member or friend with a disability, chronic or mental illness, or who is frail aged.

### **Activities and services**

- Counselling
- Information and advice
- Workshops and social activities
- Young carers program (8-25 year olds).

**Publications/resources** Care Support Kit.

## **Centre for Genetics Education**

RNS Community Health Centre  
Level 5, 2c Herbert St  
ST LEONARDS NSW 2065

Phone: (02) 9462 9599

Email: [contact@genetics.edu.au](mailto:contact@genetics.edu.au)

Website: [www.genetics.edu.au](http://www.genetics.edu.au)

### **Centre for Genetics Education**

The Centre for Genetics Education (CGE) is the education arm of the NSW Clinical Genetics Service with a clear mission to prepare NSW non-genetics trained health professionals with the skills, tools and knowledge to manage the impact of genetic and genomic technologies on their practice. Research and engaging GPs continues to be a major challenge.

### **Objectives of the group**

- Facilitate the integration of genetics and genomics into the knowledge, skills and clinical practice of relevant non-genetics trained health professionals in NSW
- Contribute to relevant policy development at state and national levels and provide leadership in genetics and genomics education.

### **Activities and services**

As part of its core business, the Centre continues to update and build on its established patient resources and programs, respond to education requests in priority areas, participate in research to promote an evidence-base for practice and working closely with its key partners, NSW Clinical Genetic Services and Genetic Alliance Australia (formerly AGSA). The Centre is a RACGP Accredited Activity Provider.

### **Publications/resources**

The website has a wide range of resources and information including 69 fact sheets, patient booklets and pamphlets, decision aids around genetic testing, contact details of and GP online training modules.

Resources include (all available online):

- Prenatal Testing and Counselling- Test to check your baby's health before birth
- Genetics Services and Counselling: Why knowing about your genes is important to your future
- When your unborn baby has a problem, how to manage the weeks ahead
- Information for women considering preventative Mastectomy because of strong family history of breast cancer.

# Children and Young People with Disability Australia (CYDA)

20 Derby St  
COLLINGWOOD VIC 3066

PO Box 172  
CLIFTON HILL VIC 3068

Phone: 1800 222 660  
(03) 9417 1025  
Email: [info@cyda.org.au](mailto:info@cyda.org.au)  
Website: [www.cyda.org.au](http://www.cyda.org.au)  
Facebook: [www.facebook.com/CDISAUS](http://www.facebook.com/CDISAUS)  
Twitter: @CDA39

CYDA is the national representative organisation that represents children and young people with disability, aged 0-25 years. CYDA has a national membership of 5000 with the majority being families. CYDA provides a link between the direct experiences of children and young people with disability and their families to the federal government and other key stakeholders. This link is essential for the creation of a true appreciation of the experiences of and challenges for children and young people with disability and their families.

## Objectives of the group

CYDA's vision is that children and young people with disability living in Australia are afforded every opportunity to thrive, achieve their potential and that their rights and interests as individuals, members of a family and their community are met. CYDA's purpose is to advocate systematically at the national level for the rights and interests of all children and young people with disability living in Australia as individuals, members of a family and their community.

## Activities and services

CYDA undertakes the following to achieve its purpose:

- **Listen and respond** to the voices and experiences of children and young people with disability
- **Advocate** for children and young people with disability for equal opportunities, participation and inclusion in the Australian community
- **Educate** national public policy-makers and the broader community about the experiences of children and young people with disability
- **Inform** children and young people with disability, their families and care givers about their citizenship rights and entitlements
- **Celebrate** the success and achievements of children and young people with disability.

## Publications/resources

- CYDA's national publication, Listen Up!
- CYDA issues papers, available at [www.cdyda.org.au/cyda-issue-papers](http://www.cdyda.org.au/cyda-issue-papers)
- Information about the Royal Commission into Institutional Responses to Child Sexual Abuse, available at [www.cyda.org.au/royal-comission-home](http://www.cyda.org.au/royal-comission-home)
- CYDA's organisational website contains a wide range of resources of relevance to children and young people with disability including updates on current issues and inquiries, links to relevant media and information about policy, legislation and key issues.

# CLAN WA

Level 1, 165 Wright St  
KEWDALE WA 6105

PO Box 1009  
CLOVERDALE WA 6985

Phone: (08) 9477 0400  
Fax: (08) 9477 0444

Email: [admin@clanwa.com.au](mailto:admin@clanwa.com.au)  
Website: [www.clanwa.com.au](http://www.clanwa.com.au)  
Facebook: [www.facebook.com/CLANWA](http://www.facebook.com/CLANWA)

CLAN WA works alongside families in WA to enhance resilience, encourage healthy relationships, effective parenting, support networks and community participation.

## Objectives of the group

Strengthen family life by encouraging healthy relationships, effective parenting, emotional and mental health, support networks and community participation. To provide parenting support and parenting information.

## Activities and services

CLAN WA work across the metropolitan area and in the Peel Rockingham region.

- **Community Workshops-** CLAN WA provides a range of workshops for families encouraging healthy relationships, effective parenting, and community participation
- **Family Support-** The CLAN WA Family Support program is a free and confidential home visiting service offered to families with children aged 0 – 18 years. Trained volunteers visit families in their home for around 2 hours per week to assist parents to find ways to meet their children's needs and manage behaviour
- **Professional Training-** CLAN WA can provide professional training for community organisations, corporate and government agencies
- **Talk It Out-** a program to assist families living in the City of Swan. This is aimed at children 0 - 18 years, who are experiencing emotional difficulties in their lives or are at risk of developing mental health issues.

# **ConnectGroups Support Groups Association WA Inc**

10 Almondbury Road  
BOORAGOON WA 6154

Phone: (08) 9364 6909  
Email: [info@connectgroups.org.au](mailto:info@connectgroups.org.au)  
Website: [www.connectgroups.org.au](http://www.connectgroups.org.au)  
Facebook: [www.facebook.com/pages/ConnectGroups-Support-Groups-Association-WA-Inc/322384177821587](https://www.facebook.com/pages/ConnectGroups-Support-Groups-Association-WA-Inc/322384177821587)

## **Objectives of the group**

ConnectGroups Support Groups Association WA Inc. (Formally W.I.S.H) WA Peak body for Self Help and Support Groups providing a referral pathway to individuals and families; offers assistance to new and emerging Support Groups with start-up and ensures long term viability of its existing Support Groups through sector development and advocacy.

## **Activities and services**

- Referral service to individuals/government and NGOs
- Training and development to existing and emerging groups
- Research
- Advocacy
- Awareness events/information sessions.

## **Publications/resources**

- Havoc research project 'Victims of Crime'
- Governance manual
- PIF (Pay It Forward) Grants
- Online resources how to set up a support group etc.
- Good practice support groups minimum standards.



## **Deaf Society Inc, Western Australian**

Head Office  
Suite 46/ 5 Aberdeen St  
EAST PERTH WA 6004

PO Box 8558  
Perth BC WA 6849

Phone: (08) 9441 2677  
TTY: (08) 9441 2655  
Fax: (08) 9441 2616

Email: [wadeaf@wadeaf.org.au](mailto:wadeaf@wadeaf.org.au)  
Website: [www.wadeaf.org.au](http://www.wadeaf.org.au)  
Facebook: [www.facebook.com/wadeaf](http://www.facebook.com/wadeaf)

### **Deafness**

The terms 'deafness' and 'hard of hearing' refer to partial or total loss of hearing in one or both ears. This may be present from birth e.g. congenital rubella syndrome or may result from injury, disease and/or exposure to loud noise during one's lifetime. Aging is also a cause of deafness.

### **Objective of the group**

The WA Deaf Society exists so that deaf and hard of hearing people experience full citizenship and enjoyment of life.

### **Activities and services**

- Interpreting services
- Employment services
- Information and referral services
- Advocacy services
- Educational services e.g. Auslan classes, Deaf Awareness Training
- LEAP (Language Early Access Program for children 0 – 6 years)
- Seniors activity programs.

### **Publications/resources**

The Society produces a quarterly newsletter, Deaf Magazine, pamphlets relating to the Society's services, an Annual report and resource information.

# Developmental Disability WA

2 Delhi Street  
WEST PERTH WA 6005

Phone: (08) 9420 7203  
Email: [ddwa@ddwa.org.au](mailto:ddwa@ddwa.org.au)  
Website: [www.ddwa.org.au](http://www.ddwa.org.au)  
Facebook: [www.facebook.com/DDCofWAlnc](http://www.facebook.com/DDCofWAlnc)

Developmental Disability WA (DDWA) was established in 1985 and is a trusted source of independent information, advocacy, education and support for people with intellectual and other developmental disability, their families and the people who support them. Membership is free for individuals.

## Objectives of the group

People with developmental disabilities and their families live their lives their way.

### Advocacy

- To support people with developmental disabilities and their families to have a strong voice and seek change where needed.
- To influence government and other decision makers to make positive and lasting change.

### Knowledge

- To build the expectations and capacity of people with developmental disability and their families.
- To inform people and families about their rights, choices and options to equitable services and supports.

### Community

- To support people with developmental disabilities and their families to live their everyday lives.
- To partner with others to develop more connected and inclusive communities.

## Activities and services

### Advocacy

- Member and sector forums and surveys
- Individual Advocacy and Service Negotiation
- General representation on disability sector and other committees
- Issue based representations to government and other decision makers

## Knowledge

- Workshops
- Resource books and tools
- Information: Newsletters, Facebook, Website, YouTube
- Peer Groups
- Discussion Groups/Forums
- Conferences
- Webinars

## Community

- Positive Behaviour Support – Side by Side Program
- Professional Development – Training for Disability, Education, Justice, Child Protection, Local Govt., Mental Health, Emergency Services, Retail, Hospitality, Transport, Communications staff.

# Disability Services Commission

146-160 Colin Street  
WEST PERTH WA 6005

PO Box 441  
West Perth WA 6872

Phone: (08) 9426 9200  
Free call: 1800 998 214

Website: [www.disability.wa.gov.au](http://www.disability.wa.gov.au)

The Disability Services Commission is the State Government agency responsible for advancing opportunities, community participation and quality of life for people with disability.

Established in 1993 under the Disability Services Act 1993, the Commission provides a range of direct services and support and also funds non-government agencies to provide services to people with a disability, their families and carers.

The Commission also partners and collaborates with disability sector organisations, business and government, and other stakeholders to improve participation, inclusion and access for people with disability across the community.

## Objectives of the group

The Commission's vision, called Count Me In, is that all people live in welcoming communities that facilitate citizenship, friendship, mutual support and a fair go for everyone.

The vision has three areas of focus:

- Economic security and well-designed and accessible communities and homes
- Participation and contribution for people with disability in all aspects of life
- The provision of personalised supports and services which are innovative, flexible and person-centred.

## Activities and services

The Commission's roles and responsibilities are vast, including direct service delivery and progressive improvements to social and community inclusion and policy.

The Commission is undertaking a number of strategic initiative to facilitate better outcomes for people with a disability, their families and carers, to improve processes for disability service providers and build capacity across the disability sector and to ensure continuous improvement of the Commission services.

These projects include: WA NDIS My Way- a procurement reform project and the establishment of a disability justice centre.

Key operational programs which underpin the Commission's work include: Local Area Coordinator, Community and Family Living Initiative, Commission therapy services,

Country Resource and Consultancy, Disability Access and Inclusion Plans and the Count Me In strategy.

### **Publications/resources**

Disability Update

Sector e-Bulletin

WA NDIS My Way e-newsletter

YouTube- DisabilityWA

Twitter- DisabilityWA.

## **EDGE Employment Solutions**

38 Hood Street  
SUBIACO WA 6008

Phone: (08) 9286 6000

Email: [edge@edge.org.au](mailto:edge@edge.org.au)

Website: [www.edge.org.au](http://www.edge.org.au)

Facebook: [www.facebook.com/EdgeEmployment](http://www.facebook.com/EdgeEmployment)

EDGE is a disability employment service who assist people with disability or medical conditions to secure employment, and we also provide on-the-job support. We are completely free of charge. We can work with individuals that live within the North metropolitan and central Western metropolitan suburbs.

### **Objectives of the group**

Assist people with disability to gain employment in the community which enhances inclusion in the workplace and provide flow on effects into the personal, social and financial aspects of life.

### **Activities and services**

- Assist with job search interview training
- Resume assistance
- Job search
- Assistance with job support on-the-job training/support
- Educating employers
- Assistance with workplace modifications/equipment.

## **Ethnic Disability Advocacy Centre (EDAC)**

320 Rokeby Road  
SUBIACO WA 6008

Phone: (08) 9388 7455  
Fax: (08) 9388 7433  
Free call: 1800 659 921

Email: [admin@edac.org.au](mailto:admin@edac.org.au)  
Website: [www.edac.org.au](http://www.edac.org.au)

### **Objectives of the group**

To promote advocacy, support, information, referral, networking, lobbying and training for people with disabilities from culturally and linguistically diverse backgrounds (CaLD) and their families and carers.

Operating hours are Monday- Friday 9am-5pm. No fees are payable for service and there is no cost for the use of interpreters. Ethnic people with disabilities, families and carers can access our services.

### **Activities and services**

- EDAC assists people with a disability to resolve problems with government offices such as Department of Social Services, Housing, Education, Health, State Administrative Tribunal and others
- EDAC provides information on disability support services, community groups and government agencies/policies
- We have some translated resources and can arrange an interpreter to ensure you understand your options and rights
- EDAC provides a meeting place and support for the Multicultural Family Support Group and Women with Disabilities
- EDAC is a peak body and strong advocate in raising awareness of issues impacting upon people with disabilities from CaLD backgrounds.

## **Genetic Alliance Australia**

C/- Garvan Institute of Medical Research  
Level 6  
384 Victoria Street  
DARLINGHURST NSW 2010

Phone: (02) 9295 8359

Email: [info@geneticalliance.org.au](mailto:info@geneticalliance.org.au)

Website: [www.geneticalliance.org.au](http://www.geneticalliance.org.au)

Facebook: [www.facebook.com/GeneticAllianceAustralia](http://www.facebook.com/GeneticAllianceAustralia)

This organisation is an umbrella group for rare genetic conditions/diseases that provides peer support and information. They facilitate contact between families/individuals affected by the same or similar condition through their extensive contact register. Other services are genetic seminars, counselling, sibling workshops, annual BRCA 1/2 information day, NSW rural outreach program and networking events such as genetic awareness week.

### **Objectives of the group**

- Provide a contact point for families who are affected by genetic conditions so rare that they do not have their own support group
- Facilitate access to individual support groups for those families with particular genetic disorders
- Provide a forum for the exchange of information between support groups regarding available community services
- Educate the medical and allied health professionals and the community about genetic disorders
- Consult with government bodies, both Federal and State, for appropriate funding for genetic services.

### **Publications/resources**

Quarterly newsletter.



## Genetic Services of Western Australia

Agnes Walsh House  
374 Bagot Rd  
SUBIACO WA 6008

PO Box 134  
SUBIACO WA 6904

Phone: (08) 6458 1525  
Fax: (08) 6458 1678

Email: [gswa@health.wa.gov.au](mailto:gswa@health.wa.gov.au)  
Website: [www.kemh.health.wa.gov.au/services/genetics/](http://www.kemh.health.wa.gov.au/services/genetics/)

### Objectives of the group

Genetic Services of Western Australia (GSWA) provides a range of diagnostic, treatment, counselling and investigative services for individuals and families with suspected or known hereditary (genetic) conditions.

These services are provided through a multidisciplinary team which includes clinical geneticists and genetic counsellors, working closely with DNA and cytogenetic laboratories and other associated disciplines.

### Activities and services

- Early diagnosis and treatment: Early detection of conditions in high risk families. Diagnosis of a predisposition to some cancers before symptoms appear can provide the opportunity for preventative measures
- Screening tests: A range of screening tests can be carried out during pregnancy (prenatal) to detect some genetic conditions
- Newborn babies are screened for selected conditions which are often treatable if detected early
- Genetic counselling to date information, assists with understanding the diagnosis and treatment of conditions, the risks of the condition occurring to other family members and information about what tests are available to clarify these risks and decision making
- Genetic counselling to date information, assists with understanding the diagnosis and treatment of conditions, the risks of the condition occurring to other family members and information about what tests are available to clarify these risks and decision making
- Laboratory services: DNA tests examine an individual's DNA for gene changes. Cytogenetic tests examine an individual's chromosomes for chromosome abnormalities. Biochemical tests involve testing for certain substances in the body (such as proteins) for evidence of hereditary conditions and birth defects
- Familial Cancer Program: This program is run by GSWA and provides comprehensive services to individuals and families with a significant family history of breast, ovarian, bowel and other related familial cancers.

### Publications/resources

Information brochures about the above services are available on request or visit the website for further information.

## Health Consumers' Council (WA) Inc

Unit 6 Wellington Fair  
40 Lord Street  
EAST PERTH WA 6004

GPO Box C134  
PERTH WA 6839

Phone: (08) 9221 3422                      Free call: 1800 620 780  
Email: info@hconc.org.au  
Web: www.hconc.org.au  
Facebook: www.facebook.com/hconcwa

Open 9.00am – 4.30pm Mon – Fri (except public holidays)

### Health Consumers' Council

The Health Consumers' Council of WA (HCC) is an independent voice, advocating for patients in Western Australia. It offers a unique perspective on health policy and service delivery matters.

### Objectives of the group

Our Purpose: To raise awareness of, and advocate for, health consumers' rights in Western Australia.

Our Vision: To be an independent, authoritative and effective voice of and for health consumers in Western Australia.

Our Values: The Health Consumers Council believes that people are entitled to

- Be treated with respect, dignity and understanding
- Be informed about their rights and have those rights protected and enhanced
- Receive safe evidenced based care
- Be informed about their condition and any proposed treatment
- Have equitable access to health services
- Have access to information about themselves held by health professionals and the right to correct anomalies.

### Activities and services

- We operate a health information and referral telephone service
- We advocate for individuals with problems with the health system and inform them of their rights
- We run the Health Issues Group where you can discuss health issues affecting you and action can result
- We organise workshops and information sessions on health issues of interest to the community
- We coordinate the placement of consumer representatives on committees.

### Publications/resources

We produce four "Health Matters" newsletters a year. These contain information on current health concerns, changes to the health system and updates on the work of the Health Consumers' Council. All contributions are welcome. We also have a fortnightly eNewsletter that includes upcoming events, news and health articles. This is also available to non-members.

# HelpingMinds - Mental Health Carers and Friends Association

182 Lord St  
PERTH WA 6000

Counselling information and admin

Phone: (08) 9427 7100

Fax: (08) 9427 7119

Rural Free call: 1800 811 747

Email: [info@helpingminds.org.au](mailto:info@helpingminds.org.au)

Website: [www.helpingminds.org.au](http://www.helpingminds.org.au)

## Mental illness

Mental illness includes a variety of conditions such as depression, bipolar, anxiety, schizophrenia, eating disorders, and obsessive compulsive disorder . Mental illness can affect individuals from all walks of life and research has shown that some individuals have a genetic predisposition to mental illness.

## Objectives of the Group

Our job is to support anyone that is experiencing mental health distress. This includes both the person that is experiencing mental health issues as well as their family and friends who are providing support both emotionally or practically.

## Activities and Services

Our services include:

- Counselling
- Advocacy
- Respite
- Support Groups
- Online Education
- NDIS support
- Peer Support
- Community and School Education Programs
- Workplace Wellness

## Publications/resources

HelpingMinds has resources available on our website and offers online education programs. The HelpingMinds head office also has a library, including videos.

# Immunodeficiency Research Centre

Isfahan University of Medical Sciences  
ISFAHAN, IRAN

Phone: 00989133177356

Email: nrg\_rahmanian@yahoo.com

Website: [www.educationiran.com/en/Isfahan/mui/wrapper/19819/Acquired-Immunodeficiency-Research-Center](http://www.educationiran.com/en/Isfahan/mui/wrapper/19819/Acquired-Immunodeficiency-Research-Center)

The Immunodeficiency Research Centre was established to conduct scientific research projects with innovative valid methods for diagnosis and treatment of immunodeficiency diseases in order to contribute to the improvement of affected patients.

## Objectives of the group

Acquired Immunodeficiency Research Centre has three main goals:

- Raising awareness
- Early diagnosis
- Improving treatment outcome of immunodeficient patients

## Publications/resources

Recent publications from the Immunodeficiency Research Centre include:

- Yazdani, R., et al., Comparison of common monogenic defects in a large predominantly antibody deficiency cohort. *The Journal of Allergy and Clinical Immunology: In Practice*, 2018.
- Sherkat, R., et al., Establishment and Development of the First Biobank of Inflammatory Bowel Disease, Suspected to Primary Immunodeficiency Diseases in Iran. *Advanced biomedical research*, 2018. 7.
- Sheikhabahaei, S., et al., Pregnancy, child bearing and prevention of giving birth to the affected children in patients with primary immunodeficiency disease; a case-series. *BMC pregnancy and childbirth*, 2018. 18(1): p. 299.
- Sadati, Z.A., et al., Comparison of the Percentage of Regulatory T cells and their p-STAT5 Expression in Allergic and Non-Allergic Common Variable Immunodeficiency Patients. *Immunological investigations*, 2018: p. 1-12.
- Nekooie-Marnany, N., et al., Impaired IL-12-and IL-23-Mediated Immunity Due to IL-12R $\beta$ 1 Deficiency in Iranian Patients with Mendelian Susceptibility to Mycobacterial Disease. *Journal of clinical immunology*, 2018. 38(7): p. 787-793.
- Abolhassani, H., et al., Clinical, immunologic, and genetic spectrum of 696 patients with combined immunodeficiency. *Journal of Allergy and Clinical Immunology*, 2018. 141(4): p. 1450-1458.
- Talaei, Z., et al., Recurrent vulvovaginal candidiasis: Could it be related to cell-mediated immunity defect in response to Candida antigen? *International journal of fertility & sterility*, 2017. 11(3): p. 134.
- Sherkat, R., et al., EBV lymphoproliferative-associated disease and primary cardiac T-cell lymphoma in a STK4 deficient patient: a case report. *Medicine*, 2017. 96(48).
- Bahrami, E., et al., Myb-like, SWIRM, and MPN dom

## Independent Living Centre WA

The Niche, Suite A  
11 Aberdare Road  
NEDLANDS WA 6009

Phone: (08) 9381 0600 or free call 1300 885 886  
Email: [general@ilc.com.au](mailto:general@ilc.com.au)  
Website: [www.ilc.com.au](http://www.ilc.com.au)  
Facebook: <https://www.facebook.com/independentlivingcentre/>

The Independent Living Centre WA (ILC) provides information and advice, assessment, training, funding and hire services that enable Western Australians of all ages and abilities to live more independent and fulfilling lives.

### Objectives of the group

To guide people's choices and to access assistive technology and services for independence. Support people's independence and wellbeing through information, advice, assessment, referral, funding, hire and respite services.

### Activities and services

Assistive Equipment Services: provides free information and advices to help people choose and access the most appropriate assistive equipment items.

- ILC Hire: A range of assistive equipment items are available for hire through our specialist hire service.
- ILC Tech: provides information, advice and training on augmentative and alternative communication equipment, computer access and technology to support learning.
- ILC Training: offers a range of professional training and community information sessions delivered by health professionals on a range of topics.
- Occupational Therapy Driver Assessment: this service provides comprehensive assessment and rehabilitation services for people with disabilities or medical conditions.
- Multicultural Aged Care Service: an information service designed to educate, support and partner aged care providers and people from culturally and linguistically diverse communities to meet the diverse needs of older people.
- Respite and Carelink: The ILC Commonwealth respite and Carelink centre can provide short term and emergency respite services to people who provide unpaid support to another person with an ongoing disability or illness.
- Noah's Ark WA: The Noah's Ark Toy Library for children with learning disabilities and additional needs provides resources and information to help families and agencies promote children's development through play.

### Publications/resources

A range of resources are available from our website and include newsletters, information resources to help individuals, carers and organisation with assistive equipment and technology; augmentative and alternative communication resources including tablets and smartphones, computer access and software; research and project reports and useful links to a range of government agencies and community based organisations that provide support, information, advice and resources for individuals with a disability, their families and carers.

## **Intelife**

11 Kirke Street  
BALCATTWA WA 6021

Phone: (08) 6169 1100

Fax: (08) 9240 1846

Email: [centralreception@intelife.org](mailto:centralreception@intelife.org)

Website: [www.intelife.org](http://www.intelife.org)

Facebook: <https://www.facebook.com/intelifegroup/>

At Intelife we recognise each person as an individual and with different needs, and because of this we deliver flexible, holistic and unique programs, achieving the best fit for people with disability to realise their potential. We are committed to providing individually tailored opportunities for our people to participate in employment, social, educational and cultural activities, and have continued to expand on our range of services since our inception in 1991.

### **Objectives of the group**

Vision: Excellence in individualised support services for people with a disability.

Purpose: Helping people with a disability to achieve their potential and participate in the economic, social and cultural life of their community. What we stand for: an inclusive community.

### **Activities and services**

- Lifeskills plus (alternatives to employment)
- Respite service
- Holiday program
- Aboriginal Engagement Program
- Training services
- Australian Disability Enterprise
- Disability Employment Service.

### **Publications/resources**

- Brochures for each program
- Annual reports
- Media releases (online)
- External newsletter (in production).

# **Ishar Multicultural Women's Health Centre**

21 Sudbury Road  
MIRRABOOKA WA 6061

Phone: (08) 9345 5335

Email: [info@ishar.org.au](mailto:info@ishar.org.au)

Website: [www.ishar.org.au](http://www.ishar.org.au)

Ishar Multicultural Women's Health Centre Inc. (Ishar) is an incorporated not for profit organisation which focuses on the delivery of services related to the health and wellbeing of women and their families, particularly those from culturally diverse backgrounds, living in the Perth metropolitan area. Since its inception in 1992 the organisation has been gradually expanding its services in both the demography of clients assisted and in the range of services provided.

## **Objectives of the group**

The mission of Ishar is to provide inclusive, holistic and culturally sensitive services for women and their families, promoting healthy communities.

## **Activities and services**

Ishar is available to provide generic support which takes into account the special cultural needs of women from culturally and linguistically diverse backgrounds. Activities and services offered are listed below, for more information on any of these services please visit their website or contact them directly.

### **Health Services and Health Promotion**

- Women's Health Clinic (Female GP and Nurse)
- Well Women's check
- Dietitian
- Counselling
- Clinical Psychologist
- Pregnancy and Postnatal Support
- Domestic Violence Support program
- Exercise and Fitness Program.

### **Carers and Family Support**

- Carers Support Services
- 40+ Women's Lifestyle Program
- Neighbourhood Mothers Program
- Family Settlement Support Program
- Settlement Grant Program.

## **Kalparrin**

Perth Children's Hospital  
15 Hospital Avenue, Nedlands WA 6009

c/- Child & Adolescent Health  
Locked Bag 2010,  
Nedlands WA 6840

Phone: (08) 6456 0035  
Free call: 1800 066 413

Email: [kalparrinwa@health.wa.gov.au](mailto:kalparrinwa@health.wa.gov.au)  
Website: [www.kalparrin.org.au](http://www.kalparrin.org.au)  
Facebook: [www.facebook.com/groups/WASpecialFamilies/](http://www.facebook.com/groups/WASpecialFamilies/)

Kalparrin is an organisation committed to providing practical and emotional support to:

- Families of children with special needs and disability who require ongoing care and support
- Families of children with special needs who are under the unique stress of awaiting diagnosis or whose diagnosis is unknown
- Families of children with special needs requiring empathy, advocacy, support and empowerment in times of crisis
- Professional and students and staff of other agencies seeking information about a range of disabilities and services and self-help/support groups.

### **Objectives of the group**

To make a positive difference to the lives of families and carers of children with special needs through the provision of practical and emotional support.

### **Activities and services**

- Information, support and referral to range of supporting services
- A drop in centre: with a parent lounge and a toy room for children
- A parent link, linking parents with support groups and other families through a confidential computer database
- A weekend mother's camp twice a year. These weekends are a needed break for many mothers and involve workshops on various topics and pampering
- Family Fun days for families of children with special needs
- Siblings' day events in school holidays.

### **Publications/resources**

The Centre has a range of information including internet resources and a newsletter.



## **Kidney Health Australia**

80 Birdwood Parade  
DALKEITH WA 6009

GPO Box 9993  
MELBOURNE VIC 3001

Phone: 0481 914 629  
Free call Kidney Health Information Service: 1800 454 363

Email: Justine.bell-morris@kidney.org.au  
Website: www.kidney.org.au

### **Kidney disease**

Many conditions are associated with kidney disease including urinary infection, kidney stones, glomerulonephritis, urinary incontinence and renal failure.

Kidney abnormalities can exist in isolation or they can be part of other conditions. Some of these conditions have a genetic basis.

### **Objectives of the group**

Kidney Health Australia, formerly the Australian Kidney Foundation, was officially formed 35 years ago. It is a non-profit organisation with a mission to be the lead organisation promoting kidney and urinary tract health through research, advocacy, education and health service excellence. It achieves this by:

- Implementing awareness and education programs in conjunction with renal specialists, health professionals and educators to reduce the incidence of kidney disease in the community
- Funding cutting edge medical research into cures for kidney and urinary tract disease
- Providing a network of education, care and support for patients, their families and carers
- Promoting organ donation.

### **Activities and services**

The Foundation offers a range of non-medical support services for individuals with kidney disease, their families and members of the general community including:

- Information and services to individuals with genetic kidney disease, their families and their carers
- Individual support programs and services and links to other support networks
- Information seminars throughout the year
- Guest speakers to present a range of issues relating to kidney disease and organ donation.

### **Publications/resources**

Kidney Health Australia publishes a monthly newsletter and has a large number and range of regularly updated publications and resources for individuals with kidney disease, their families, and the community and health professionals.

## **Linkwest**

Suite 7, Oasis Lotteries House  
37 Hampden Road  
NEDLANDS WA 6009

Phone: (08) 9485 8929  
Email: office@linkwest.asn.au

Website: www.linkwest.asn.au  
Facebook: www.facebook.com/linkwest

### **Objectives of the group**

Linkwest is the Peak Body for Neighbourhood and Community Resources Centres in WA. We are a member-based service, with membership from across the state. Many members have self-help groups running out of their premises. We aim to provide our members information, training, resources and advocacy to enable them to develop vibrant, inclusive and connected communities.

### **Activities and services**

We offer our members:

- Training and networking opportunities
- Resources
- Information, advice and guidance
- Funding opportunities
- Advice and assistance in areas such as governance, policy and mediation.

### **Resources/publications**

Various in the areas of:

- Governance
- Operations
- Delivery
- Community engagement and development.

## Office of Population Health Genomics

Public and Aboriginal Health Division  
Department of Health  
3rd Floor, C Block  
189 Royal Street  
EAST PERTH WA 6004

PO Box 8172  
Perth Business Centre  
PERTH WA 6849

Phone: 08) 9222 6888  
Fax: (08) 9222 6820

Email: [genomics@health.wa.gov.au](mailto:genomics@health.wa.gov.au)  
Website: [www.genomics.health.wa.gov.au](http://www.genomics.health.wa.gov.au)

The Office of Population Health Genomics (OPHG) is a directorate within the Public and Aboriginal Health Division of the Department of Health, Western Australia.

### Objective

The aim of OPHG is to optimise health benefits by leading the integration of genomic knowledge and innovation into the WA health system.

### Activities and services

OPHG develops system-wide and service-specific public policy. This help guide Government action on issues related to rare diseases, genomics and population-based screening programs at state and national levels to promote health and wellbeing, and prevent disease and disability where possible. OPHG also ensures that health services delivered to people living with rare and genetic diseases, and population-based screening programs are high quality, safe, equitable, accessible and sustainable.

Key areas of focus include: rare diseases; health service planning, support, monitoring and evaluation; stakeholder engagement; and population-based screening programs and policy.

### Publications and resources

OPHG resources for the public include pamphlets on prenatal screening and diagnosis tests, folate: reducing the risk of spina bifida and other neural tube defects, and newborn bloodspot screening.

To order these pamphlets, please use the Online Publication Ordering System <http://dohquickmail.com.au> or contact OPHG.<http://dohquickmail.com.au> or contact OPHG.

Visit [www.genomics.health.wa.gov.au](http://www.genomics.health.wa.gov.au) for more information about OPHG.

## People with Disabilities (WA) Inc (PWdWA)

Suite 1, Oasis Lotteries House  
37 Hampden Road  
NEDLANDS WA 6009

Phone: (08) 9485 8900  
Country Callers: 1800 193 331  
TTY: (08) 9386 6451  
Fax: (08) 9386 1011

Email: [info@pwdwa.org](mailto:info@pwdwa.org)  
Website: [www.pwdwa.org](http://www.pwdwa.org)

### Objectives of the group

PWdWA is run by and for people with disabilities and strives to be the voice for all people with disabilities in Western Australia.

Since 1981, PWdWA has been the peak disability consumer organisation representing the rights, needs and equality of all Western Australians with a physical, intellectual, psychiatric or sensory disability via individual and systemic advocacy.

### Activities and services

- **Individual advocacy**– Information and issue based one to one advocacy for Western Australians with a disability.
- **PEEL**- Based in Mandurah- information and individual advocacy for the Peel Region.
- **Disability First Stop (DFS)** - An advocacy service for adults with a newly diagnosed disability. Assist individuals and families to navigate the care system.
- **Systemic advocacy**- Campaigns to influence change, remove barriers and discrimination for people with disabilities.

## **Rare Voices Australia**

PO Box 138  
MENTONE VIC 3194

Phone: 0497 003 104

Email: [admin@rarevoices.com.au](mailto:admin@rarevoices.com.au)

Website: [www.rarevoices.org.au](http://www.rarevoices.org.au)

Facebook: [www.facebook.com/RareVoicesAustralia](http://www.facebook.com/RareVoicesAustralia)

Rare Voices Australia (RVA) is Australia's non-profit, national peak organisation advocating for all who live with rare disease. RVA provides a strong common voice to advocate for health policy and a healthcare system that works for those with rare diseases. Patient-centred, RVA works with all stakeholders including patients, key peak bodies, governments, researchers, clinicians and industry to promote rare disease research, diagnosis, treatment and services.

Rare Diseases are not formally or officially recognised by the Australian Government and there is no Australian rare disease policy in place. The data collected on rare diseases is based on international conservative estimates of 6-8% of the total Australian population.

### **Objectives of the group**

- Providing leadership and advocacy on national rare disease issues for the benefit of those who live with a rare disease.
- Raise awareness of issues faced by the rare disease community to promote understanding, motivation and empowerment.
- Collaborate and connect with all Australians sharing the experience of living with a rare disease.

### **Publications/resources**

Helpful information and resources can be found on their website  
[www.rarevoices.org.au/page/43/patient-information](http://www.rarevoices.org.au/page/43/patient-information)

# Rocky Bay (and Spina Bifida and Hydrocephalus Association of WA)

60 McCabe St  
MOSMAN PARK WA 6102

Phone: (08) 9383 5111

Email: [admin@rockybay.org.au](mailto:admin@rockybay.org.au)

Website: [www.rockybay.org.au](http://www.rockybay.org.au)

Rocky Bay is a leading provider of disability services offering choice and independence to people of all ages living with a disability in WA. With a focus on quality and service excellence, we work with individuals and their families to deliver relevant therapies, the latest equipment and technology, innovative leisure and social programmes, training and employment opportunities, in the home, at our sites or in the community.

## Main conditions services are offered for

Muscular dystrophies

Physical, intellectual and sensory disabilities<sup>[OBJ]</sup>

## Objectives of the group

To provide a broad, innovative, quality-focused disability service that leads the industry. We will seek growth in order to ensure sustainability and further our aim of being a provider of choice for all of our services.

- Customer service – optimise the customer experience
- Governance – processes and decisions that define actions, grant authority and verify performance
- Financial capacity – maintain operations support growth and raise capital
- People – spirit of the organisation
- Brand – distinguish ourselves from competitors in the market.

## Activities and services

- Therapy and professionals services: physiotherapy, occupational therapy, speech therapy, psychological support, social work, hydrotherapy pool
- Leisure and independence: explore the possibilities and create your own plans for leisure and learning, social activities and financial control
- Accommodation support, respite, residential- home and community.

## Publications/resources

Annual Report.

## **Self Help Queensland Inc (SHQ)**

PO Box 353  
SUNNYBANK QLD 4109

Phone: (07) 3344 6919

Email: [info@selfhelpqld.org.au](mailto:info@selfhelpqld.org.au)

Website: [www.selfhelpqld.org.au](http://www.selfhelpqld.org.au)

Self Help Queensland Inc (SHQ) is a not for profit community organisation formed in 1983. Our aim is to support people in taking responsibility for improving their own health and wellbeing. We do this by carrying out a range of activities that help establish, develop, promote and sustain Self Help and Support Groups in Queensland. We also act as a point of contact for individual community members, organisations and professionals to locate self help groups for themselves, their clients or patients.

### **Objectives of the group**

- To develop, promote and sustain the self help sector in Queensland.

### **Activities and services**

- Assistance to start self help and support groups
- Promotion of groups and assistance to build the capacity of groups to better help their members and to sustain the group
- Free, online searchable Directory of Queensland Self Help and Support Groups
- Referrals to self help and support groups across a broad range of health conditions and wellbeing issues.

### **Publications/resources**

Fact sheets available.

## **SHOUT (Self Help Organisations United Together) (ACT)**

PO Box 717  
MAWSON ACT 2607

Phone: (02) 6290 1984

Email: [admin@shout.org.au](mailto:admin@shout.org.au)

Website: [www.shout.org.au](http://www.shout.org.au)

Facebook: [www.facebook.com/pages/SHOUT-Inc/358679317537976](https://www.facebook.com/pages/SHOUT-Inc/358679317537976)

### **SHOUT (ACT)**

SHOUT operates as an umbrella organisation for member and affiliate self-help groups providing:

- A point of contact for clients and member/affiliate organisations
- A referral point for the public
- An information resource centre.

### **Objectives of the group**

SHOUT works with its members and the community:

- To promote the philosophy, practice and value of self help
- To provide support and resources which develop and empower self-help groups
- To enable individuals to find support to meet their needs by connecting them to appropriate groups
- To facilitate member groups to meet their goals through resources and connections.

### **Activities and services**

By actively promoting the values of cooperation and unity, SHOUT enables member organisations to continue to function effectively.



# Speak Easy Association Western Australia

Claisebrook Lotteries House  
33 Moore St  
EAST PERTH WA 6004

Phone: 0419 930 903  
Email: [seawa@speakeasy.org.au](mailto:seawa@speakeasy.org.au)  
Website: [www.speakeasy.org.au](http://www.speakeasy.org.au)

## Stuttering

Stuttering affects the verbal communication abilities of a significant proportion of the population. It is characterised by the halting, repetition and disruption of speech. Stuttering is a neurological, cognitive disorder. Current research suggests that stuttering has a neurological basis and that genetics play a part resulting in a loss of coordination of the speech muscles.

## Objective of the group

To provide advocacy and support to people who stutter and their families.

## Activities and services

- Education services
- Advocacy
- Support group meetings
- Research assistance
- Training workshops and seminars

## Publications/resources

- Liaison with Government departments
- General information services
- Membership \$35 per annum

The Association has a library for members with resources suitable for the community and health professionals. The Association publishes a quarterly newsletter.

## **TADWA (Technology Assisting Disability WA)**

371 Collier Road  
BASSENDEAN WA 6054

Phone: (08) 9379 7400

Email: [enquiries@tadwa.org.au](mailto:enquiries@tadwa.org.au)

Website: [www.tadwa.org.au](http://www.tadwa.org.au)

TADWA is a not for profit organisation delivering services to people with a disability, the aged and their carers in metropolitan and regional WA. We custom design solutions to meet mobility, daily living, recreation and communications needs including computers, assisting clients to live independently and in the community.

### **Objectives of the group**

To improve the quality and enjoyment of life of people with disabilities, older people and those caring for them, through the application of technology and the skills of volunteers and staff.

### **Activities and services**

- Customised Aids and Equipment- bathroom modifications, daily living equipment, freedom wheels customised bicycles and tricycles, repair and service wheelchairs, walkers and scooters, postural seating, ramps and rails
- Computer Services- supply and support for refurbished and new computers, including help desk, mobile technicians, workshop technicians, computer training.

### **Publications/resources**

See website as above.

# **The Neurological Council of WA Inc**

Centre for Neurological Support (CNS)  
Suite B, The Niche  
11 Aberdare Rd  
NEDLANDS WA 6009

Phone: (08) 6457 7533  
Free call: 1800 645 771  
Fax: (08) 9346 7534

Website: [www.ncwa.com.au](http://www.ncwa.com.au)  
Email: [admin@ncwa.com.au](mailto:admin@ncwa.com.au)

The Neurological Council of WA (NCWA) provides care, support and information services to people with neurological conditions in the community. The NCWA is also an umbrella organisation to several neurological and associated organisations with whom it works towards improved services and understanding in the community. The NCWA also supports and provides leadership in professional networking and educational opportunities for neurological nurses in the community.

## **Objectives of the group**

- To provide quality care, support and information services to people in the community with neurological conditions;
- To work with member and other associated organisations towards improved services for people with neurological conditions;
- Professional networking and educational opportunities for neurological nurses in the community.

## **Services and activities**

The NCWA provides a comprehensive range of services to people in the community with neurological conditions. These include support services, counselling, information and advocacy in all areas of its operation. The NCWA auspices a strong support group network for adults of all ages.

The NCWA has a community neurological nurse service which operates within the Bunbury, Manjimup, Albany and Geraldton areas and their surrounds. The NCWA also works professionally with other organisations and associated groups to promote education and collaborative working opportunities in the neurological community.

## **Therapy Focus**

5/1140 Albany Hwy  
BENTLEY WA 6102

PO Box 20  
BENTLEY WA 6982

Phone: 1300 135 373  
Fax: (08) 9451 5480

Email: CEO@therapyfocus.org.au  
Website: www.therapyfocus.org.au  
Facebook: www.facebook.com/therapyfocus

Therapy Focus is Western Australia's leading provider of therapy and support services for children and young people with disabilities and their families. We cover all types of disability conditions by delivering comprehensive therapy services.

### **Objectives of the group**

By 2017 Therapy Focus will lead innovation in therapeutic practice in Australia.

### **Activities and services**

Predominantly speech, occupational therapy, physiotherapy, psychology, social work, behavioural support in schools, community and home environments. Additional programs include Sunflower Sunday event, state-wide art competition for school children.

### **Publications/resources**

In Focus newsletter, Family Focus newsletter, annual reports, art competition, story books, multiple resources on Complete Advantage website ([www.therapyfocus.org.au/resources.aspx](http://www.therapyfocus.org.au/resources.aspx)).

## A-Z Index of Synonyms of Genetic & Rare Conditions

18p deletion syndrome .....	32	Autoimmune disease.....	17
18p syndrome .....	32	Autoimmune disorder .....	17
18q deletion syndrome .....	32	autoimmune liver disease .....	76
18q syndrome .....	32	Bacterial/viral arthritis .....	17
45 X syndrome.....	113	Batten disease.....	22
5p minus (5p-).....	39	Bertolotti's syndrome.....	23
A1AD.....	13	Bilateral acoustic neuroma.....	11
AATD.....	13	Bipolar.....	52
Absence seizures.....	51	Bipolar disorder .....	52
Acoustic neurilemoma .....	11	Bipolar I.....	52
Acquired long QT syndrome .....	77	Bipolar II.....	52
Adrenal cortex adenoma.....	40	Birthmarks .....	90
Adrenal hyperfunction .....	40	Bonnevie-Ulrich syndrome .....	113
Affective disorder .....	52	Borreliosis .....	79
Albinism.....	12	Brachmann-de Lange syndrome.....	38
Alopecia Areata.....	20	Brittle bones.....	89
Alpha-1 .....	13	C1-esterase inhibitor deficiency .....	61
Alpha-1 antitrypsin deficiency .....	13	C1-inhibitor deficiency .....	61
ALS.....	79, 83	Cardio-facial-cutaneous syndrome .....	25
Alzheimer disease.....	14	Cat eye syndrome .....	104
AMD .....	96	CDH .....	26
Amsterdam syndrome.....	38	Cerebellopontine angle tumour.....	11
Amyoplasia.....	18	CFC syndrome .....	25
Amyoplasia congenita.....	18	Characteristic Ear abnormalities .....	29
Androgen insensitivity syndrome.....	15	Charcot-Marie-Tooth disease.....	28
Angelman syndrome .....	16	CHARGE syndrome .....	29
Ankylosing Spondylitis .....	17	Chiari .....	30
Anorectal Malformation .....	24	Choanal Atresia .....	29
Anorectal Malformation Hirschsprung's disease.....	24	Chromosomal triplication.....	104
anxiety .....	19, 136	Chromosome 18.....	32
Arnold Chiari malformation .....	30	Chromosome X.....	113
Arthritis .....	17	Chronic motor tic .....	111
Arthrogryposis .....	18	CJD.....	33
arthrogryposis multiplex congenita .....	18	Cleft lip.....	34, 90
AS.....	16	Cleft lip and palate.....	34
Asthma .....	119	Cleft palate .....	34
Ataxia .....	57, 58, 98, 105	CMT .....	28
Atrophic .....	17	Coeliac disease .....	35
Attention deficit disorder (ADD) .....	74	Coloboma .....	29
Attention deficit hyperactivity disorder .....	74	Complex partial seizures.....	51
autism.....	19, 98, 112, 146	Congenital Diaphragmatic Hernia .....	26
		Congenital heart defects .....	64

Congenital multiple arthrogyriposis .....	18	Gastric adenocarcinoma and proximal polyposis of the stomach.....	59
congenital rubella syndrome.....	126	Gelastic paroxysm.....	60
Congenital word blindness.....	48	Gelastic seizure.....	60
connective tissue disorders .....	37	Genetic counselling.....	9, 134
Connective tissue dysplasia .....	37	Genomics .....	143
Convulsions.....	51	Gilles de la Tourette's disease.....	111
Cooley's anaemia .....	108	glomerulonephritis .....	141
Cornelia de Lange.....	38	Gonadal dysgenesis (45, X).....	113
corpus callosum .....	19	Grave's disease.....	109
Cortico basal degeneration.....	94	Guerin-Stern syndrome.....	18
Cranial nerve abnormalities .....	29	HAE .....	61
Craniofacial syndromes/conditions.....	90	Haemochromatosis .....	62
Cri du chat syndrome.....	39	Haemophilia.....	63
Cushing's disease.....	40, 79	Hair loss.....	20
Cushing's syndrome .....	40	hamartoma .....	60, 91
cyclothymia .....	52	Happy puppet syndrome.....	16
Cystic Fibrosis.....	42	Hare lip .....	34
Cystinosis.....	43	HCU .....	66
de Lange syndrome .....	38	Heart defects.....	64
Deafblind .....	44, 100	Hereditary .....	28, 61, 62, 65
Deafness .....	126	Hereditary Angioedema .....	61
Degenerative arthritis.....	17	Hereditary Hemorrhagic Telangiectasia .	65
Degenerative joint disease .....	17	Hereditary motor and sensory neuropathy .....	28
Del (18p) syndrome .....	32	Heterozygous familial hypercholesterolaemia .....	56
Dementia.....	81	HHT .....	65
Dementia praecox.....	81	Hirschsprung's disease .....	24
depression.....	52, 136	Homocystinuria.....	66
developmental disability.....	127	Homozygous familial hypercholesterolaemia .....	56
Developmental reading disorder.....	48	Hunter syndrome.....	84
Diabetes .....	46	Huntington's disease.....	9
Discoid lupus erythematosus.....	78	Huntington's disease .....	80
Disorder of sex development.....	15	Hurler .....	84
Disseminated lupus erythematosus .....	78	Hyaluronidase deficiency .....	84
Distal arthrogyriposis .....	18	Hyperkinetic disorder.....	74
Down syndrome .....	47	Hyperlipoproteinemia .....	56
Drug-induced lupus erythematosus.....	78	Hyperthyroidism.....	109
Dyskinesia.....	94	Hypothyroidism.....	109
Dyslexia.....	48	Idiopathic hypersomnia .....	68
eating disorders .....	136	Idiopathic Hypersomnolence.....	68
Ectodermal.....	50	immune deficiencies.....	70
Ectopic ACTH syndrome .....	40	intellectual disability.....	9, 16, 47
Edward's syndrome .....	104	Intermediate cystinosis.....	43
Epidermdysis bullosa .....	45	Intersex.....	15
Epidermolysis Bullosa.....	45	Intestinal failure .....	93
Epilepsy.....	51	Intravenous nutrition.....	93
Ethnic people with disabilities.....	132	Isochromosome 12p.....	92
Fabry disease.....	53	Jervell and Lange-Neilson type.....	77
Facial difference.....	90	Juvenile idiopathic arthritis .....	17
Facio-cardio-cutaneous syndrome .....	25	Kawasaki disease.....	71
fibromyalgia.....	17, 79	Kidney disease .....	141
Fibrous ankylosis of multiple joints .....	18	Killian syndrome .....	92
Fragile X syndrome.....	57	Klinefelter's syndrome.....	73
Fragile X-Associated Primary Ovarian Insufficiency .....	57		
Friedreich ataxia .....	58		

Laughing seizures.....	60	neural tube defect.....	9
LDL receptor disorder.....	56	Neurinoma of the acoustic nerve.....	11
Learning and attention disorders.....	74	Neurofibroma of the acoustic.....	11
learning disability.....	48	Neurofibromatosis.....	31
Lejeune's syndrome.....	39	Neurofibromatosis Type 2.....	31
Leukodystrophy.....	75	Neurogenic arthrogryposis multiplex	
Lewy body dementia.....	94	congenita.....	18
Liver diseases.....	76	neurological conditions.....	151
Long arm deletion syndrome Del (18q)		Neurological disorders.....	94
syndrome.....	32	neuronal ceroid lipofuscinoses.....	22
Long QT syndrome.....	77	Neuronal ceroid lipofuscinosis.....	22
Lou Gehrig's disease.....	83	Non-nephropathic cystinosis.....	43
Lupus.....	78, 79	Noonan Syndrome.....	88
Lupus erythematosus.....	78	obsessive compulsive disorder.....	136
Lyme disease.....	79	ocular albinism.....	12
Machado Joseph disease.....	80	oculocutaneous albinism.....	12
Macular Degeneration.....	96	Osteoarthritis.....	17
Macular Dystrophy.....	96	Osteogenesis Imperfecta.....	89
Major affective disorder.....	52	Osteoporosis.....	17
Maladie de tics.....	111	Ovarian dwarfism.....	113
Manic depression.....	52	Ovary aplasia.....	113
Manic depressive illness.....	52	Pallister mosaic aneuploidy syndrome ...	92
Manic-depressive psychosis.....	52	Pallister-Hall syndrome.....	60, 91
Maroteaux-Lamy syndrome.....	84	Pallister-Killian syndrome.....	92
MD.....	96	Parenteral Nutrition.....	93
Mediterranean anaemia.....	108	Parkinson's disease.....	79, 94
Mental illness.....	136	Patau syndrome.....	104
micro deletion syndrome.....	114	Paulitis.....	111
Mitochondrial disease.....	82	Perineural fibroblastoma.....	11
MJD.....	80	Peroneal muscular atrophy.....	28
MND.....	83	physical disabilities.....	117
Monosomy 18p syndrome.....	32	Physical, intellectual and sensory	
Monosomy 18q syndrome.....	32	disabilities.....	146
Monosomy X.....	113	Pitt-Hopkins syndrome.....	32
Morgagni-Turner-Albright syndrome.....	113	pituitary conditions.....	40
Morquio syndrome.....	84	Pompe disease.....	95
mosaic 21.....	47	Pre-senile dementia.....	14
Mosaic tetrasomy 12p.....	92	Primary reading disability.....	48
Motor neurone disease.....	83	prion disease.....	33
MPSIII.....	99	Progressive pigmentary retinopathy.....	96
Mucopolidoses.....	84	Progressive supranuclear palsy.....	94
Mucopolysaccharide.....	84	Pseudo seizures.....	51
Mucopolysaccharidoses.....	84	Pterygolympangiectasia.....	113
Multi system infectious disease syndrome		rare chromosome conditions.....	21
.....	79	reading disability.....	48
Multiple congenital contractures.....	18	renal failure.....	141
Multiple Sclerosis.....	85	retinal dystrophies.....	96
Multiple system atrophy.....	94	Retinitis pigmentosa.....	96
multiplex congenita.....	18	Retinitis Pigmentosa.....	96
Muscular dystrophies.....	146	Rett syndrome.....	98
Muscular dystrophy.....	86	Rheumatism.....	17
Myasthenia gravis.....	87	rheumatoid arthritis.....	17, 40
Myasthenia Gravis.....	87	Ring 18.....	32
Myoclonic seizures.....	51	Ring chromosome 18.....	32
Myopathic arthrogryposis.....	18	Rod-cone dystrophy.....	96
Nephropathic cystinosis.....	43	Romano-Ward type.....	77

RTS .....	98	Tetrasomy short arm of chromosome .....	32
Sanfilippo syndrome .....	84, 99	Thalassaemia .....	108
Scheie syndrome .....	84	Thyroid hormone .....	109
Schereshevskii-Turner syndrome.....	113	Tics .....	111
Schizophrenia .....	81	Tonic clonic seizures.....	51
Schizophrenic psychosis .....	81	Tourette syndrome .....	111
Schwannoma of the acoustic nerve .....	11	translocation 21 .....	47
Schwannomatosis.....	31	Tremor Ataxia syndrome.....	57
scleroderma .....	17	Trisomy 1.....	104
Seizures .....	51, 60	Trisomy 18.....	104
Septic arthritis .....	17	Trisomy 21.....	47
Short arm 18 deletion syndrome .....	32	Trisomy 22.....	104
Short stature.....	37, 101	Trisomy 9.....	104
Simple partial seizures.....	51	Trisomy disorders.....	104
Sjögren's peripheral neuropathy.....	103	Tuberous sclerosis .....	112
Sjögren's syndrome .....	103	Tumours.....	115
Sly syndrome .....	84	Turner syndrome .....	113
special needs .....	140	Turner-Varny syndrome .....	113
Spielmeyer-Sjogren disease.....	22	undiagnosed genetic condition .....	106
spina bifida .....	9	Usher syndrome .....	44
Spinocerebellar ataxia .....	105	VCFS 22q11 .....	114
Sporadic CJD.....	33	Velocardiofacial syndrome .....	114
Stuttering.....	149	Vestibular schwannoma .....	11
sudden infant death syndrome (SIDS) ..	102	Vogt-Spielmeyer disease .....	22
Syringomyelia.....	30	von Hippel-Lindau syndrome .....	115
Systemic lupus erythematosus.....	78	Von Willebrand disease .....	63
Tarlov Cyst Disease.....	107	Whistling face syndrome .....	18
Teshler-Nicola.....	92	Williams syndrome .....	116
Testicular feminising syndrome .....	15	Williams-Beuren syndrome .....	116
tetrasomy 18p .....	32	XO syndrome.....	113