

## About Our Members

### Rett Syndrome - WA Rett Syndrome Support Group

Rett Syndrome is a neurological condition which occurs almost exclusively in females, and results in a variety of physical and mental impairments<sup>1</sup>. In affected individuals, development appears normal until 6 to 18 months, and it is followed by stagnation and regression in development<sup>2</sup>. The most common of these include impairment of speech, loss of purposeful hand use, and difficulty walking. Additional problems may include seizures, breathing irregularities, and scoliosis.

Research conducted by Dr Huda Zoghbi in 1999 identified the cause of Rett Syndrome as a defect in the MECP2 gene, sometimes referred to as the 'Rett gene'<sup>3</sup>. This gene is located on the X chromosome at Xq28, and so the condition overwhelmingly affects females. Mutations in the MECP2 gene are lethal to the male fetus<sup>4</sup>.

The occurrence rate of Rett Syndrome ranges between 1:10, 000 to 1:23, 000 female births<sup>4</sup>. It is estimated that 99.5% of cases of Rett Syndrome occur only once in a family<sup>4</sup>. This indicates that whilst Rett Syndrome is a genetic condition, it is not a hereditary one, and may be attributed to sporadic mutation, that is mutations that occur now and again. Rett Syndrome is not predominant in any particular ethnic group.

Genetic research has been an important mechanism to advance what is known and understood about Rett Syndrome.

Prior to the identification of the MECP2 gene Rett Syndrome was often misdiagnosed as autism or cerebral palsy. Genetic testing procedures are now capable of confirming the diagnosis in 80% of cases<sup>2</sup>.

However, there is still a great deal more to learn about Rett Syndrome. Whilst it is known which gene is primarily responsible for the development of the condition, there is still only limited knowledge about the exact mechanisms that underlie it. This information is important to shed light on possible treatments for the condition<sup>3</sup>.

Future research on this condition will focus on areas such as developing new approaches to the pharmacological treatment of the disorder, and to probing further into the mechanisms that underlie the MECP2 mutations<sup>3</sup>.

### Contact details:

WA Rett Syndrome Support Group Telephone: (08) 9259 1516  
Email: sanafay@hotmail.com

### References

- Disability Online. *Rett Syndrome affects communication and movement*. Available at: [INSERT]
- Gibson, J., Bennetts, B., Slobedman, B., Armati, P., & Christodoulou, J. (2002). *Gene expression in Rett syndrome*. Available at:  
<http://www.chs.usyd.edu.au/conf2002/minipost/av-gibson.pdf>
- Rett Syndrome Research Foundation. (2002). *The RSRFlash Newsletter*, 1 (7).
- International Rett Syndrome Association. *Rett Syndrome Fact Sheet*. Available at:  
[http://www.rettsyndrome.org/main/fact\\_sheet.htm](http://www.rettsyndrome.org/main/fact_sheet.htm)

## The GSCWA Board of Management

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Executive Dean, Health Sciences Curtin University

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Cystic Fibrosis WA Inc.

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Australian Huntington's Disease Association WA Inc.  
Carmel Wright  
Client Services Coordinator  
Spina Bifida Association of WA Inc.

### **Our Location and contact details:**

Level 1, Oasis Lotteries House  
37 Hampden Road  
Nedlands WA 6009  
Telephone: 08 9389 6722  
Mobile: 0411 585 113  
Email: [admin@geneticsupportcouncil.org.au](mailto:admin@geneticsupportcouncil.org.au)  
Web: <http://geneticsupportcouncil.org.au>

### **Office hours:**

9.00am to 12.00pm and 1.00pm to 5.00pm.  
Monday to Friday.  
12.00pm to 1.00pm by appt.  
Monday to Friday.

### **Staff:**

Terry Keating, *Executive Director*  
Anja Hermann, *Administrative Officer*  
Kristina Johns, *Resource Officer*

## *Current Issues and Matters of Interest*

### **GSCWA Services.**

Providing support to Genetic Support Groups through:

- information
- advocacy
- education
- referral

Services to Genetic Support Groups include:

- *Increasing awareness of genetic conditions in the community by:*
  - o Participating in forums and member Genetic Support Group awareness events
  - o Providing an up-to-date website
  - o Widely circulating a monthly newsletter
  - o Seeking opportunities to engage the media, including Community and Regional newspapers and Access radio
  - o Establish and maintain an annual "Genetic Disease Awareness week"
- Publish a Directory of Genetic Support Groups in WA
- Providing administrative support and facilities by:*
  - o Assisting member Genetic Support Groups in the establishment and maintenance of sound administrative processes
  - o Making available the GSCWA office and staff to assist in these processes
  - o Assisting with the development and maintenance of budgets
  - o Assisting with the development of strategic and business plans
  - o *Providing forums for policy development and matters of common interest by:*
    - o Conducting forums for members on issues of interest or concern
    - o Provide a forum to enable members' views to be represented to Government and others in the field of genetics
    - o Written and verbal submissions to

Government and others in the field of genetics

- o Actively pursue opportunities to ensure that the views of the Genetic Support Council and its members are considered

- o The GSCWA providing a vehicle for recommending Consumer Representatives to committees and the like, considering matters concerning the field of genetics and genetic support

Participating with like bodies nationally to further represent the Genetic Support Groups and their members

- *Specialised support to smaller member Genetic Support Groups by:*

- o Providing an office base for smaller member Genetic Support Groups

- o Making available seminar facilities

- o Assisting with the development of information packages relating to their specific genetic conditions or predispositions

- o Printing of material and pamphlets

- o Providing opportunity to these groups to publish articles, advertise and promote awareness of their Support Group in the GSCWA newsletter.

Providing industrial relations advice

- o Assisting in researching information regarding particular genetic conditions or predispositions

Keeping members aware of matters of interest, changes of legislation that may impact on the area or opportunities for them to participate in making comment on matters of concern

- *Maintaining a Resource Centre that:*

- o Includes information on the services provided by member Genetic Support Groups

- o provides information on a range of associated topics such as online resources, families living with a family member with a genetic condition, parenting and education in association with a genetic condition are some of these

- o Maintains a wide range of website

links on genetic conditions and organisations and groups that may provide support

- o Maintains a database of Genetic Support Groups throughout Australia and with some international links

## **Coming Events**

### **Cancer Meeting the Challenge Seminar**

#### **Cancer Support Association of WA Inc.**

Date: 7 March

Primarily for people with cancer and their carers, the seminar is open to any interested person. An introduction to a wellness and holistic model of health and well being and an overview of complementary and self-help therapies which can be used in conjunction with mainstream therapies. This seminar runs on the first Friday of each month.  
Contact: (08) 9384 3544

### **Hat Frenzy Friday Activ Foundation**

Date: 7 March

Wear your favourite hat, cap or bonnet for a day (or buy a Lapel Pin) to help raise money for children and adults with intellectual disability in Western Australia.

Contact: (08) 9387 0555

Website:

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

### **Activ Awareness Week Activ Foundation**

Dates: 8 – 14 March

An awareness week to find out what services Activ Foundation provides to individuals with intellectual disability and their families in Western Australia.

Contact: (08) 9387 0555

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

### **Dogs' Big Day Out Association for the Blind of WA**

Date: 9 March

Time: 8am to 1pm

An awareness and fund raising event to support guide dogs. Includes a 'Walk for Guide Dogs', dog displays and exhibits.

Contact: (08) 9311 8202

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

### **Brain Awareness Week**

#### **Brain Foundation**

Dates: 10 – 16 March

A week to promote awareness of the brain as well as brain, spine and nerve disorders and diseases.

Contact: (02) 9437 5967

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

### **Muscular Dystrophy Week**

#### **Muscular Dystrophy Association of WA (Inc.)**

Dates: 10 – 14 March

The week is held to raise awareness of Muscular Dystrophy and raise funds for research.

Contact: (08) 9382 2700

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

### **Red Cross Calling**

#### **Australian Red Cross**

Dates: 10 - 22 March This is the annual door knock to raise funds for the Australian Red Cross.

Contact: (08) 9325 5111

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

### **Coeliac Disease Awareness Week**

#### **Coeliac Society of WA**

Dates: 13 - 20 March

The week is designed to promote awareness of coeliac disease to the general community.

Contact: (08) 9444 9200

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

### **The Freo Doctor Festival**

#### **Fremantle Hospital Medical Research Foundation**

Dates: 14 - 23 March

The festival includes ten days of community events to raise funds for medical research at Fremantle Hospital. All profits will go to the Fremantle Hospital Medical Research Foundation. Events include an

Art Exhibition Opening, the 96FM Orient X-Press, a Columbia Tristar Movie Premier, the East vs South Fremantle Legends Football Game, St Patricks Day Celebrations, a Dragon Boat Regatta, the 14th Annual Corporate Triathlon, a Family Walk, a Fashion Parade and the Fremantle Ports Open Day.

Contact: (08) 9431 2133

### **The Worlds' Greatest Shave for a Cure Leukaemia Foundation**

Dates: 14 - 15 March

A major fundraising event which includes participants shaving or colouring their hair to raise funds for leukaemia research and patient support.

Contact: 1800 500 088

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

### **Walking with the Stars**

#### **Muscular Dystrophy Association of WA (Inc.)**

Date: 16 March

A fundraising walk around the Narrows bridge and the Perth Causeway at night. Starting at 6pm at Taylor's Reserve, South Perth, walking around Riverside Drive and over the Narrows bridge to finish back at Taylor's Reserve around 8.30pm.

Contact: (08) 9382 2700

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

### **Kidney Street Appeal**

#### **Australian Kidney Foundation**

Date: 28 March

The annual fundraising street collection day will be held in Perth. The regional dates may vary.

Contact: (08) 9322 1354

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

## **Relay For Life Perth & Geraldton Cancer Foundation of WA**

Dates: 29 - 30 March

Relay for Life is a team event to raise funds for cancer research and services. It's 24 hours of fun, with the challenge of completing a walk or run type of relay within a team of 10-15 people. First lap is for survivors and carers and the Candle-light Ceremony is to honour those going through treatment and to remember loved ones lost to cancer. Relay for Life is for 24 hours because cancer never sleeps.

Contact: 1300 656 585

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



## **Interesting Websites**

### **Microsoft**

<http://www.healthinsite.gov.au>

Gain knowledge about built-in accessibility features in Microsoft products, so that persons can adjust programmes to suit their needs and preferences. For example, enlarging text, or hearing tones when pressing specific keys on the keyboard

### **WebABLE**

<http://www.webable.com/>

WebABLE is the Web site for disability-related internet resources. The WebABLE site goal is to stimulate education, research, and development of technologies that will ensure accessibility for people with disabilities to advanced information systems and emerging technologies.

## **Grants**

The GSCWA maintains a list of grants that can be applied for by community organisations such as genetic support groups. Members can contact our office for further information. Several relevant grants this month are:

### **Volunteer Small Equipment Grants 2003**

[http://www.facs.gov.au/sfcs/funding/sm\\_l\\_equip\\_grants.htm](http://www.facs.gov.au/sfcs/funding/sm_l_equip_grants.htm)

Funded through the Stronger Families and Communities Strategy, \$3 million will be provided for Volunteer Small Equipment Grants 2003. The grants will provide up to \$5,000 to help organisations purchase equipment in the direct support of volunteers whose work contributes to benefiting families and communities.

All enquiries should be directed to the Volunteer Small Equipment Grants 2003 Hotline 1800 018 312.

Guidelines and application forms are available online in Pdf format (See web address above).

\* GSCWA can assist members with grant applications.

### **GSCWA SPECIAL PROJECT FUND**

The Genetic Support Council WA has a limited one off fund that is available to members for grants of up to \$1000 to assist with special projects such as:

- Assisting with publications
- Conducting seminars and forums
- Compilation of information base for consumers
- Attendance at conferences
- Assistance in engaging expert speakers at conferences, seminars and forums.

*Other projects that have a direct benefit to Genetic Support Groups and their members may be considered*

## CRITERIA

The following criteria is considered essential:

- The applicant must be able to demonstrate that the project is of benefit to a particular genetic support group or genetic support groups in general or individual/families with genetic conditions or predispositions and/or is consistent with the aims of the GSCWA.
- The primary applicant must be a member of the GSCWA, but may be working in partnership with another organisation.
- The applicant may need to demonstrate that other sources of funding have been pursued unsuccessfully.
- Contribution by the applicant to the project whilst not essential, is considered highly desirable.
- The applicant agree to provide a report on the completion of the project on the acquittal of the grant and how the funds were used to achieve the aims of the grant.
- providing ongoing research for developments and up to date information on genetic conditions
- Assisting in sourcing funding by:
  - o Providing information on possible funding/grant sources
  - o Giving assistance with applying for grants
- The GSCWA providing some limited funds for special grants to members for special projects. These grants are, however, limited and small
- Provide a direct support service to individuals for whom there is no Genetic Support

The views expressed in this newsletter are those of the individual authors and not necessarily those of the GSCWA. The material supplied is for information purposes only, and is not to be used for diagnosis/treatment.

## GSCWA Members

### FULL MEMBERS

Acoustic Neuroma Association of Australia  
 Alzheimer's Association of WA  
 Angelman Syndrome Association  
 Arthrogryposis Support Group  
 Australian Huntington's Disease Association  
 Australian Pituitary Foundation WA Branch  
 Australian Tuberous Sclerosis Society Inc.  
 Coeliac Society of WA  
 Cushings Disease Support Group  
 Cystic Fibrosis WA  
 Down Syndrome Association of WA  
 Dyslexia SPELD Foundation WA Inc  
 Epilepsy Association of WA  
 Even-Keel Bi-Polar Support Association (Inc)  
 Haemophilia Foundation WA Inc.  
 Heart Kids WA  
 Klinefelters Support Group  
 Learning and Attention Disorders Society of WA (LADS)  
 Lupus Group of WA  
 Motor Neurone Disease Association of WA Inc.  
 Mucopolysaccharide & Related Diseases Society (MPS)  
 Muscular Dystrophy Association of WA  
 Neurofibromatosis Association of WA.  
 Parents of Children with Disabilities  
 Periodic Paralysis Society of Australia  
 Raynaud's Syndrome Support Group  
 Rett Syndrome Association of WA  
 Schizophrenia Fellowship of Western Australia Inc.  
 Short Statured People's Association WA Branch  
 SIDS and Kids Western Australia  
 Sjögren's Syndrome Support Group  
 Spina Bifida Association of WA  
 Thalassemia Association of WA  
 Trisomy and Related Disorders  
 Western Australian Retinitis Pigmentosa Foundation  
 WA Tourette Syndrome Organisation (WATSO)

### CORPORATE ASSOCIATE MEMBERS

ARAFMI Western Australia  
 Australian Kidney Foundation  
 Carers WA  
 Health Consumers Council WA  
 Genomics Branch, Dept. of Health  
 The Kalparrin Centre  
 The Neurological Council of WA Inc  
 Western Australian Deaf Society Inc.

### INDIVIDUAL ASSOCIATE MEMBERS

Professor Charles Watson, Curtin University  
 Terry Keating, GSCWA



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