

# Gene Advocate



## EDITION 22

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2004

### Special points of interest:

- About our members: Motor Neurone Disease
- Grants

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## About Our Members

### Motor Neurone Diseases

“I may have had a tough break, but I have an awful lot to live for”  
ALS sufferer, Lou Gehrig, in his farewell speech to his baseball fans at the Yankee Stadium, 4<sup>th</sup> July, 1939.

**Motor Neurone Disease (MND)** was first identified by French physician Jean Martin Charcot in 1874. MND covers a spectrum of related diseases in which the nerve cells in the brain (called neurones) and spinal cord, that enable us to move around, breathe, speak and swallow, fail to function properly. MND is a fatal disease which causes muscular weakening and eventual paralysis in most sufferers; however, progression of the disease varies between individuals. Sufferers of MND include a list of considerable notorieties, including scientist Stephen Hawking, actor David Niven, Chinese dictator Mao Tse-Tung, composer Dmitri Shostakovitch and baseballer Lou Gehrig, after whom the most common form of the disease **amyotrophic lateral sclerosis (ALS)** has been informally named.

### Prevalence

Forms of MND are classified as either sporadic or genetic, according to prevalence of the disease in an individual's family history, but there is little difference between the course and symptoms of the two. About 5-10% of cases are considered to have a genetic basis,

where usually more than one family member have been identified with the disease.

There is currently no genetic test for genetic MND but the disease is thought to be associated with the gene responsible for overseeing the production of an enzyme called **superoxide dismutase 1 (SOD1)**. Because SOD1 is an enzyme used by the body to help destroy toxic substances known as free radicals, researchers have speculated that MND may be associated with the excessive accumulation of free radicals in the body.

Another theory is that the presence of excessive levels of a neurotransmitter known as **glutamate**, may over stimulate neurones and increase free radical production. Autoimmune factors or premature ageing, viral agents or environmental toxins have also been considered as playing a possible role in the disease. However, despite the numerous theories, there is no certain cause for MND and the actual pathway of destruction remains unknown. The disease affects nearly twice as many men as it does women and usually occurs in those over 40, with onset usually occurring between 50 and 70 years of age. It is suggested that about 2 per 100,000 people are diagnosed by MND world-wide. The disease is not infectious or contagious.

### Forms of MND

In MND neurones die, leading to the progressive loss of the ability

*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.*

## ***The GSCWA Board of Management***

### **CHAIR**

Professor Charles Watson  
Executive Dean, Health Sciences Curtin University

### **DEPUTY CHAIR**

Mitch Messer  
Executive Director  
Cystic Fibrosis WA Inc.

### **TREASURER**

Vicki Mort  
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Holyoake Institute

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Acting Director  
Genomics Branch  
Department of Health  
Del Weston  
Executive Officer  
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  
Fax: 08 9389 9377  
Mobile: 0411 585 113  
Email: [admin@geneticsupportcouncil.org.au](mailto:admin@geneticsupportcouncil.org.au)  
Web: <http://geneticsupportcouncil.org.au>

### **Office hours:**

9.00am to 5.00pm.  
Monday to Friday.

### **Staff:**

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

to move any muscles in the body. Disease progression is generally about 2–5 years from the onset of symptoms, but this can vary considerably in different individuals.

### **Amyotrophic Lateral Sclerosis (ALS) or Lou Gehrig's Disease**

The most common form of MND is amyotrophic lateral sclerosis, or ALS. *Amyotrophic* refers to the loss of muscle bulk, *lateral* refers to the region of the spinal cord affected and *sclerosis* refers to the hardened tissue that develops in place of healthy nerves. About two thirds of MND cases are ALS. The disease is manifested by weakness and wasting of muscles and early symptoms usually manifest as involuntary tripping and dropping things. ALS generally occurs after the age of 55. Well known baseballer, Lou Gehrig, died from the disease in 1939 and, as a tribute, the disease was informally named after him.

### **Progressive Bulbar Palsy**

Progressive bulbar palsy (PBP) occurs in about a quarter of diagnosed MND cases. *Bulbar* refers to the bulb at the base of the spinal cord. In PBP, the neurones associated with the head and neck are damaged, leading to difficulties chewing, swallowing and speaking

### **Progressive Muscular Atrophy**

Progressive muscular atrophy (PMA) affects about 10% of people diagnosed, 80% of whom are males. Onset is often earlier than in other forms (under 50) and is usually first noticed in weakness or clumsiness of the hands.

### **Primary Lateral Sclerosis**

A rarer form of MND is called primary lateral sclerosis (PLS).

### **Symptoms and Testing**

Motor Neurone Disease is not a particularly common disease. The early symptoms are often involve clumsiness, weakness in the legs or mildly slurred speech. The muscles first affected tend to be those in the hands, feet or mouth and throat, depending on the form of the disease. It does not generally affect the senses, so sight, smell, touch and hearing tend to remain unaffected, as do the sphincter muscles that control the bladder and bowel. Other areas that are not usually affected are sexual function and intellect.

### **Treatment**

There is no known cure for motor neurone diseases, although scientists have been experimenting with various enzyme replacement

therapies such as superoxide dismutase injections, hoping that it may help to arrest the disease process in genetic forms of the disease. Stem cell replacement therapy is considered a potential treatment option, but there are controversial ethical issues surrounding the use of stem cells. Amino acid supplements such as L-threonine have been trialled in the hope of offering symptomatic relief from the disease, but there has been no conclusive evidence regarding the efficacy of such trials.

### **Medication: Rilutek Information**

There is no cure for Motor Neurone Disease (MND), but a medication has now been approved in Australia for the treatment of amyotrophic lateral sclerosis (ALS) – the most common form of MND.

This drug is Rilutek (riluzole), which is available at a subsidised price on the Pharmaceutical Benefits Scheme. There are some criteria governing which patients can receive subsidised Rilutek, and it is best to discuss these with a neurologist.

Rilutek is the first effective treatment for people with ALS, and can be prescribed by a Neurologist or General Practitioner. It may:

- Extend patient survival time by an average of 9-12 months
- Slow disease progression
- Keep patients in the milder stages of disease for longer, thus contributing to quality of life.

Patients started on Rilutek soon after diagnosis show the greatest benefits. As with all drugs, some patients may have a better response to Rilutek than others, and it is impossible to predict the benefits that each individual will gain. Neurologists will be able to provide guidance on the suitability of this treatment.

The most common side effects of Rilutek are weakness, nausea, a mild liver dysfunction and headache. Doctors may arrange for patients to have regular blood tests to ensure that Rilutek isn't causing side effects of which the patient is not aware.

For more information please contact your neurologist or telephone the Rilutek Hotline on 1800 00 58 58.

### **Motor Neurone Disease Association of Western Australia Inc.**

The Motor Neurone Disease Association of Western Australia provides a list of facts and services for those who are living with the disease or for those who are interested in learning more about MND. The website address is: <http://www.mndawa.iinet.net.au/index.html>

Contact details:

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

Ph: (08) 9346 7355

Fax: (08) 9346 7332

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

Website: <http://mndawa.iinet.net.au>

## ***Current Issues and Matters of Interest***

### **COFFEE CUP COUNSELLING**

by David Woodroffe. B.A. Hons., P.S.W., MSW.

Everything you need to know to help friends and family in distress. Coffee Cup Counselling is for the lay person seeking to help a friend or relative suffering from the pain or anxiety of a personal problem.

It is written in an easy, accessible style that will help you to develop skills in listening with empathy and understanding. The book provides insights into the nature of emotional pain so you can develop the confidence to offer constructive support and help to your friends or relatives to work out suitable pathways to manage issues and problems.

The book will be soft cover and a little larger than A5+ size RRP

Skills and information covered includes:

- \* Non directive listening and empathy
- \* Mirrored listening
- \* Non verbal communication
- \* The nature of emotional pain and appropriate responses
- \* How to successfully help another person to achieve good outcomes
- \* Dealing with grief
- \* The suicidal person
- \* Support for the caring friend

Cost: \$19.95 Aus each (incl GST) + \$5 mail (Australia)

You can download the order form from:

<http://members.iinet.net.au/~dwoodroffe/orderform.pdf>

## *Coming Events*

### **Western Australian Council of Social Service Inc. (WACOSS)**

WORKSHOP - Mar 5th 2004

An Issue of Risk and Insurance: This popular WACOSS seminar will cover the essentials of risk management and overview the types and range of insurance considerations for Not-For-Profit-Groups.

Download Registration Form:

[http://www.wacoss.org.au/idu/pdf/Risk\\_Mngt\\_Flyer\\_March\\_5\\_2004.PDF](http://www.wacoss.org.au/idu/pdf/Risk_Mngt_Flyer_March_5_2004.PDF)

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

Date: 5/3

Don 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: 1300 13 72 31

Email: [hatfrenzy@activ.asn.au](mailto:hatfrenzy@activ.asn.au)

Website:

<http://www.hatfrenzyfriday.activ.asn.au>

### **Kidney Street Appeal Australian Kidney Foundation**

Date: 26/3

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

Contact: (08) 9322 1354

Website: <http://www.kidney.org.au>

### **Motor Neurone Disease Association**

Dates: 28/3 - 3/4

This week aims to increase awareness in WA of Motor Neurone disease. Displays will be held at hospitals, shopping centres and libraries. Donation boxes will be available in shops, and there will be a schools participation concert and art show. Schools and companies are encouraged to Be Blue for Someone with MND by selling cornflowers (the Association's symbol) or arranging blue-themed events".

Contact: (08) 9346 7355

E-Mail: [mndawa@cnswa.com](mailto:mndawa@cnswa.com)

Website: <http://www.mndawa.iinet.net.au>

### **Multiple Birth Awareness Week Australian Multiple Birth Association (WA)**

Dates: 7/3 - 14/3

This is a week designed to raise awareness in the community about multiple birth families.

Contact: (08) 9487 8825

Website: detail also on

<http://www.mbawa.org.au>

### **National Pregnancy Loss Awareness Week**

#### **Bonnie Babes Foundation**

Dates: 21/3 - 28/3

This week aims to raise awareness grief counselling services available to Australian families who have lost a baby.

Contact: (03) 9758 2800

Website: <http://www.bbf.org.au>

### **Walking with the Stars Muscular Dystrophy Association of WA (Inc.)**

Date: 21/3

This day will be filled with entertainment, prizes, refreshments and planet and star gazing with the Perth Observatory.

A fundraising twilight walk around two courses. The longest route is around the Narrows bridge and the Perth Causeway at night. Starting at 5.30pm at Taylor's Reserve, South Perth finishing back at Taylor's Reserve around 7.30pm.

For those interested in star-gazing of a different kind, some of Perth's local 'stars' will also be participating".

Cost:

\$10 Adults Children (5-15yrs): \$5 Concession: \$8 Family (2 adults + 2 children): \$25

*Group discounts available. Everyone invited.* Call 9382 2700 to find out more or to volunteer.

Contact: (08) 9382 2700

Website: <http://www.mdawa.asn.au>

### **World's Greatest Shave for a Cure Leukaemia Foundation**

Dates: 12/3 - 13/3

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

Contact: 1800 500 088

Website: <http://www.worldsgreatestshave.com>

### **Coeliac Disease Awareness Week Coeliac Society of WA**

Dates: 13/3 - 20/3

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

Contact: 1800 449 200

Website: <http://www.wa.coeliac.org.au>



## Interesting Websites

### CareforKids.com.au

<http://www.careforkids.com.au>  
(last Reviewed: 10/02/04)

CareforKids.com.au is designed to help Australian parents find the right care for their families and is an extension of traditional child care services, empowering parents with greater ability to choose child care.

CareforKids.com.au has designed and developed a powerful tool using the latest internet technology, to offer the only service in Australia dedicated to bringing parents, agencies, carers and child care centres together in a secure on-line environment.

Source:

<http://www.careforkids.com.au/about.asp>

Information accessed through the World Wide Web is of varying levels of quality and accuracy. The material supplied is for information purposes only, and is not to be used for diagnosis or treatment.

## Grants

### The Australian Government's Volunteer Small Equipment Grants 2004.

The closing date is 5 March

These grants will provide funding of up to \$5000 to organisations involving volunteers for equipment or other one off costs, which will make the volunteer's work easier, safer and more enjoyable.

In recognition of the valuable work that volunteers do in local communities, the Australian Government has made additional funding available from the Stronger Families and Communities Strategy for the Volunteer Small Equipment Grants 2004.

If you are interested in applying I encourage you to read the attached Guidelines and Application Form carefully to determine if your organisation is eligible to apply for funding. The Guidelines also provide detailed information on the type of equipment that will be funded by these grants.

**Contact Details:** Dept of Family and Community Services

Freecall: 1800 050 199

TTY: 1800 055 001

Website: <http://www.facs.gov.au/volunteergrants>

GSCWA can assist members with grant applications

### Heart Foundation Research Grants-in-Aid

**Provider:** Heart Foundation of Australia  
**Closes:** 2 April 2004

Grants to support research in the cardiovascular field at institutions in Australia with the requisite facilities. Research projects may be pursued as an investigation in Basic, Clinical or public health (including education/health promotion, epidemiology, behavioural/social sciences) research.

For further information:

Website:

<http://www.heartfoundation.com.au>

Email: [grants@heartfoundation.com.au](mailto:grants@heartfoundation.com.au)

### A Healthy Start to Life for Aboriginal and Torres Strait Islander Children

**Provider:** National Health and Medical Research Council (NHMRC)  
**Closes:** 26 March 2004

To improve the health of Aboriginal and Torres Strait Islander people - especially children and young people - through the participation of many sectors, often outside the mainstream health sector.

For further information:

Prof Aileen Plant NHMRC

Website:

<http://www.nhmrc.gov.au/funding/hlthstrt.htm>

Email: [A.Plant@curtin.edu.au](mailto:A.Plant@curtin.edu.au)

Telephone: (02) 6289 9187

### Multi-State Research Grants

The Cancer Council Western Australia

**Closes:** 16 April 2004

Research into the diagnosis, prevention and treatment of cancer, leukaemia and allied disorders in people.

For further information:

Website:

<http://www.cancerwa.asn.au/grantscourses/researchgrants/intending/>

Email: [education@cancerwa.asn.au](mailto:education@cancerwa.asn.au)

Telephone: (08) 9212 4354

Fax: (08) 9212 4399

"Were on the Web"  
Directory of Genetic Support Groups.  
<http://www.geneticsupportcouncil.org.au>

## ***GSCWA Members***

### ***FULL MEMBERS***

Acoustic Neuroma Association of Australia  
 Alzheimer's Association of WA  
 Angelman Syndrome Association  
 Arthrogyrosis Support Group  
 Australian Huntington Disease Association  
 Australian Pituitary Foundation WA Branch  
 Australian Tuberous Sclerosis Society Inc.  
 Coeliac Society of WA  
 Cornelia De Lange Syndrome Support Group  
 Cushings Disease Support Group  
 Cystic Fibrosis WA  
 Diabetes Australia -Western Australia  
 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  
 Mental Illness Fellowship WA (Formerly: Schizophrenia Fellowship of Western Australia Inc.)  
 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  
 Senses Foundation Inc.  
 Short Statured People's Association WA Branch  
 SIDS and Kids Western Australia  
 Sjögren's Syndrome Support Group  
 Spina Bifida Association of WA  
 Thalassaemia Association of WA  
 Support Organisation for Trisomy and Related Disorders of WA (SOFTWA)  
 Western Australian Retinitis Pigmentosa Foundation  
 WA Tourette Syndrome Organisation (WATSO)

### ***CORPORATE ASSOCIATE MEMBERS***

ARAFMI Western Australia  
 Association of Genetic Support of Australasia  
 Australian Kidney Foundation  
 Carers Association of WA Inc  
 Genomics Branch, Dept. of Health  
 Health Consumers Council WA  
 Lone Parent Family Support Service (LPFSS)  
 The Kalparrin Centre  
 The Neurological Council of WA Inc  
 Western Australian Deaf Society Inc.

### ***INDIVIDUAL ASSOCIATE MEMBERS***

Anja Hermann, GSCWA  
 Bonita Anne Eaton, LPFSS  
 Darren Webb, ECU – Human Biology Student  
 Kristina Johns, GSCWA  
 Linda Bovill  
 Mark Bovill  
 Professor Charles Watson, Curtin University  
 Terry Keating, GSCWA

## **GSCWA News Corner**

*The Genetic Support Council  
 'Directory of Genetic Support Groups  
 in Western Australia 2004' is avail-  
 able at our website under  
 'publications'.*

**Website:**

<http://geneticsupportcouncil.org.au>

*Please feel free to contact us  
 for further information, or if you  
 are having difficulties accessing  
 this document*



**Advin 'Gene Advocate' or  
 on our website!!!**

Have you got anything you want to be published?

Either our newsletter or our website can be the

place to display your information. So contact us via e-mail or phone and we can arrange it for you.