



Gene Advocate

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Special Points of Interest:

- About Our Members: Neurofibromatosis.
- GSCWA AGM.
- 2004 World Congress for People who Stutter.
- Wheat Free Alternative For Gluten Free

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About Our Members

Neurofibromatosis

The disease takes its name from the tumours, or neurofibromas, that develop on the nerves and look like lumps under the skin. They can also develop in other parts of the body, causing problems such as bone deformities. There are two forms of neurofibromatosis –1 (NF-1) and neurofibromatosis-2 (NF-2).

NF-1 is also known as peripheral neurofibromatosis or von Recklinghausen's syndrome, and is the more common form of NF, affecting about 1 in 4000 people. Symptoms of NF-1 include the presence of multiple large brown spots on the skin (known as "café au lait" spots because they are the colour of milky coffee), two or more neurofibromas, freckling in the armpit or groin, benign growths on the iris of the eye, tumour of the optic nerve and abnormal curving of the spine. An individual affected with NF-1 will not necessarily have all of these symptoms, but the presence of at least two is required for a clinical diagnosis of NF-1.

NF-2 is also known as central neurofibromatosis. It is much less common than NF-1, affecting about 1 in 40,000 people. NF-2 is characterised by the development of benign tumours on the nerve responsible for hearing. These tumours affect nerves going to both ears and eventually result in deafness. In some cases these tumours cause further serious problems as they increase in size and press on other nerves in the brain. The disease is also character-

ised by the development of malignant tumours in the brain.

The symptoms of NF-1, particularly the skin spots, are sometimes seen at birth, but almost always by the age of 10. The neurofibromas also appear around 10-15 years of age. For most people, the symptoms of NF-1 are mild. However, the severity of symptoms varies widely, and other problems can occur. The symptoms of NF-2 (deafness) start in the early teens. Other symptoms associated with increasing size of the tumours include ringing in the ears, poor balance, headache and pain or numbness of the face may develop over time.

Treatment of both forms of NF is aimed at managing the symptoms. Surgery to remove the tumours causing problems is an option, but the tumours may grow back in increased numbers. Surgery can also help improve the bone and spine abnormalities. Generally, the tumours are monitored routinely to check their growth, and if needed can be treated with surgery, radio- or chemotherapy.

NF-1 and NF-2 are autosomal dominant disorders - only one mutated copy of the gene is needed to develop the disease. Therefore, a child has a 50% chance of inheriting the disease from an affected parent, although the severity of the symptoms may not be the same. However, about half of all new cases arise from unaffected parents – that is, the mutation causing the disease has arisen

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

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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
Mobile: 0411 585 113
Email: 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*

spontaneously sometime before birth for unknown reasons. This mutation can then be passed on to the children of that individual.

Genetic testing has proved to be difficult for NF-1 and NF-2. NF-1 is a very large gene with many types of mutations reported. This means that for each new family, the entire gene must be analysed in detail. The same argument applies to genetic testing for NF-2. However, a new method of mutation detection in NF-1 and NF-2 has recently been developed, which allows analysis of much larger sections of the gene at one time, making genetic testing more efficient. It should also be noted that genetic testing for prenatal diagnosis is only of use in families where the mutation has been identified in other affected family members. It is not useful for cases where the mutation has arisen spontaneously.

Current research into NF is focussed on developing improved methods of diagnosis, identifying factors that account for the variability in symptoms and finding new drugs and therapies that will improve the quality of life for NF sufferers.

Neurofibromatosis Association of WA Inc.

The Centre for Neurological Support
Suite B The Niche, 11 Aberdare Rd
NEDLANDS WA 6009
Ph: (08) 9346 7488 Fax: (08) 9346 7534
Email: ncwa@cnswa.com
Website: www.cnswa.com

Source: This article was provided by the Genomics Branch WA.

Genetic Support Council Annual General Meeting

Time: 5.30 to 7.30

Date: 30th of September 2003

Location: Grace Vaughan House, Shenton Park

**Guest Speaker: Professor Charles Watson,
Executive Dean, Health Sciences Curtin
University WA, Chair GSCWA Inc. Board.**

**Nominations for Office Bearers close:
23/9/2003**

For nomination forms and further information please ph: (08) 9389 6722

***We hope you are able to join us in what is
a very important event.***

Current Issues and Matters of Interest

'The 2004 World Congress for People who Stutter'

Every three years somewhere around the globe a conference occurs with the primary aim of helping to expose new treatments for those people in our world who stutter and support this minority group who are often discriminated against in the workplace or schoolyard. It is estimated that about 1-2% of all people have some form of stuttering problem.

Two years ago members of the *Speak Easy Association of WA (SEAWA)* attended the '2001 World Congress for People who Stutter' in Belgium and delivered a fantastic bid, winning the rights to host the Seventh 'World Congress for People who Stutter' here in Perth at the Esplanade Hotel in Fremantle, from the 15th to 20th February 2004.

SEAWA originated back in 1980 and has since acted as a support organisation for those in Western Australia who stutter.

The 2004 Congress will include the presentation of papers and workshops by professionals in the field of stuttering treatment and related personal development issues. The theme of the conference is *'Fluency and Beyond: A holistic look at Stuttering'*.

Dr David Shapiro, the Professor of Communication Disorders at Western Carolina University has spent 30 years providing clinical services for people who stutter and their families and will be presenting several papers at the conference. Marilyn Langevin works at the 'Institute for Stuttering Treatment and Research' at the University of Alberta and is a world expert in stuttering programs to alleviate bullying and teasing in the school environment. She will be presenting a paper on *'Teasing and Bullying: Unacceptable Behaviour'*.

A locally based researcher Dr Michelle Byrnes will also be presenting at the Congress on her research using functional Magnetic Resonance Imaging in examination of the brain patterns of people with verbal communication disorders, including stuttering.

Additionally of particular interest there will be workshops and sessions on topics including *'Fluency for Fun – A workshop for Children who Stutter'*, *'Stuttering Innovations – A workshop for Teenagers'* and *'Supporting the Child who Stutters – A workshop for Parents and School Teachers'*, just to name a few.

The Conference will provide valuable information for School Teachers, Psychologists, Parents,

Speech Pathologists as well as support for Children and People who Stutter. The selection of invited speakers mixed with workshops, concurrent sessions and an active social program will ensure that this is a conference to remember.

The Congress Committee has also organised four workshops to be held prior to the conference on Sunday 15 February:

- "Teasing and Bullying in Schools" – Presented by Ms Marilyn Langevin
- "Recent Treatment Research in Stuttering" Presented by Professor Mark Onslow
- "Living with Prolonged Speech – Developing your bag of tricks" – Presented by Ms Angela Cream
- "Overview of the Lidcombe Programme" – Presented by Ms Christine Lewis

For more information on the conference (including speaker profiles) and the pre-conference workshops you can visit the website at www.2004worldcongress.speakeasy.org.au or for a registration form and information program please contact Pippa McCreery of SEAWA on (08) 9225 4111 or email at seawa@bigpond.net.au

The Wheat Free Alternative For Gluten Free - Alternative Bites Pty. Ltd.

Alternative Bites has developed a process for producing palatable gluten Free cakes, confectionary & savoury products. Alternative Bites products look & most importantly, taste like ordinary food. However, for a gluten-intolerant person it represents not only a 'treat' but is also a welcome respite from sometimes-boring & repetitive home cooking.

All Alternative Bites products have been tested by an accredited nata laboratory: results prove that Alternative Bites products contain no measurable gluten content.

This is vitally important to CD sufferers as even minute traces can cause considerable discomfort & ill health.

An added benefit is that this process also delivers low fat, gluten free and some lactose - free lines, which fortunately, are secondary but substantial markets, & ones that will continue to be developed. Alternative Bites currently operates one factory that is fully operational & of high quality, with state of the art equipment and fit out adhering to strict HACCP principles.

Currently Alternative Bites are in the process of developing a franchise group to bring these products closer to the public domain. Alternative Bites is coming to a shopping centre near you soon.

Article: Herbert — Alternative Bites.
Ph: (08) 9258 8700

Coming Events

Spina Bifida Awareness Week Spina Bifida Association of WA

Dates: 1 – 7 September 2003

This week helps to promote the awareness of spina bifida within the community. Sneakers for a Day will be held on 5 September, and people are encouraged to wear sneakers and make a donation for services and research.

Contact: (08) 9346 7527

Website: www.sbawa.asn.au

National Gynaecological Awareness Week G.A.I.N Gynaecological Awareness Information Network

Dates: 8 - 12 September 2003

This week promotes gynaecological disease awareness, with gynaecological displays at hospitals, and joining medical professionals and community women to work as a team in raising the profile and access to information.

Contact: (08) 9279 7773

Website: www.gynsupport.com

Brain Injury Awareness Week Brain Injury Australia Inc

Dates: 15 - 21 September 2003

Brain Injury Awareness Week aims to raise awareness about acquired brain injury and to develop a focus on acquired brain injury as an important social, economic and political issue.

Contact: (02) 6290 2253

Home and Play Safety Day Wound Foundation

Date: 17 September 2003

Home and Play Safety Day is a national first aid awareness campaign. It has been set up by the Wound Foundation of Australia to help focus on the need to be better prepared for accidents in the home.

Contact: (02) 9904 0666

Website: www.safetyday.com.au

ADHD Awareness Week Learning and Attentional Disorders Society of WA

Dates: 22- 28 September 2003

This week is designed to raise the awareness of ADHD via media releases with the latest information on ADHD and community events to raise awareness of learning and attentional disorders.

Contact: (08) 9346 7544

World Retina Week WA Retinitis Pigmentosa Foundation Inc.

Dates: 22 - 27 September 2003

This week promotes awareness of retinal dystrophies. There will be fundraising with badges for sale with the motto "Give two hoots about blindness".

Contact: (08) 9227 7585

Website: www.retinitispigmentosa.com.au

Community Health Nurses Week Community Health Nurses Special Interest Group

Dates: 21- 27 September 2003

A celebration of Community Health Nursing in WA. The events will include professional development, networking and social events in country and metro areas.

Contact: (08) 9391 2283

Leukaemia Week Leukaemia Foundation

Dates: 15 – 21 September 2003

The week is designed to raise the awareness of leukaemia and other related blood disorders in the community.

Contact: (08) 9272 9332

Website: www.leukaemia.com

International Physiotherapy Day Australian Physiotherapy Association

Date: 8 September 2003

This day promotes physiotherapy and the role of the practitioner.

Contact: (08) 9389 9211



Interesting Websites

LD Online

<http://www.ldonline.org>

Website on learning disabilities for parents, teachers and other professionals (USA).

Band-Aides and Black Boards

<http://www.faculty.fairfield.edu/fleitas/contents.html>

An informational site about chronic medical conditions, provides stories, poems, tips, and coping strategies for children with chronic illnesses, as well as information and insights for their families, friends, and classmates (USA).

Education World

http://www.educationworld.com/a_lesson/lesson115.shtml

Contains online resources for educators on the Internet where teachers share ideas. Site contains information and articles relating to children with disabilities (USA).

GSCWA SPECIAL PROJECT FUND GRANT

The Genetic Support Council WA has a limited one off fund that is available to 'full' members (located in WA) 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.

› Contact GSCWA for further information and criteria
Telephone: 08 9389 6722

Email:

admin@geneticsupportcouncil.org.au

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:

Telstra Community Development Fund

Closes: 12 September 2003

Fund charitable organisations in the area of children and young people: Disability, Environment, Research, Health, Education and Training, Cultural diversity and Arts.

For further information contact:

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

Free Call: 1800 208 378

Friends of ACE Support (FACES) Program

Closes: 22 September 2003

Target Australians who have a speech, communication or hearing impairment. Projects which benefit the wider Australian community will also be considered

For further information contact:

Website:

<http://www.aceinfo.net.au/Resources/Sponsorship/FACES/index.html>

Ph: (07) 3815 7610 - TTY: (07) 3815 7610

- ☑ GSCWA can assist members with grant applications. Ph: (08) 9389 6722

Advertise in '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.

(see page 2 for GSCWA contact details).

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
- Trisomy and Related Disorders
- 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
- Professor Charles Watson, Curtin University
- Terry Keating, GSCWA

GSCWA News Corner

The Genetic Support Council currently has a Social Work student: Paul on placement from Curtin University.

Paul will be contacting all GSCWA members in regards to privacy statement about organisations/support groups details.

CHANGE OF ADDRESS

**To help the GSCWA keep our records up to date please email us :
admin@geneticsupportcouncil.org.au
or complete this section and return to:**

Level 1, Oasis Lotteries House
37 Hampden Road
Nedlands WA 6009

Name (Block letters)

Address

.....State.....Postcode.....

Ph:.....

Fax:

Email.....

Website.....