



Gene Advocate

EDITION 11

March 2003

Special points of interest:

- About our members: Down Syndrome
- Neuroscience Conference

Inside this issue:

About Our Members	1
GSCWA Board of Management	2
Current Issues and Matters of Interest	3
Coming Events	4
Interesting Websites	5
Grants	5
GSCWA Members	6
GSCWA News Corner	6

About Our Members

Down syndrome

Down syndrome is the most common form of intellectual disability, it occurs in one in every 460 births in Western Australia¹. What causes the presence of the additional chromosome 21 at the time of conception is still unknown.

It is almost certain that there have always been people with Down syndrome. Children with Down syndrome are seen in paintings from 1505. However, the first person to recognise Down syndrome as an entity was Dr John Langdon Down (1828-1896) an English physician working in Surrey. The syndrome, therefore, bears his name⁴.

Normally, each cell in the human body contains 23 pairs of chromosomes, which contain the genetic material that determines each person's inherited characteristics. Every person receives one of each chromosome pair from their mother and the other half from their father. The most common form of Down syndrome, has an extra copy of chromosome 21: this is the origin of the name Trisomy. People with Down syndrome have learning disabilities and characteristic physical features².

Although babies with Down syndrome are born at the same rate into families from all social, economic and racial backgrounds, there is an increased risk of having a child with Down syndrome as

maternal age advances. At age 21, a woman has a one in 1,507 risk of having a child with Down syndrome. The risk increases to one in 796 at age 31 and to one in 85 at age 41³. However, the majority of children born with Down syndrome are born to mothers under the age of 35, because 85% of pregnancies occur in women in this age group.

Down syndrome is due to an alteration in the replication of genetic material and is not a hereditary condition, this means it can not be passed down from one generation to the next. Other less common forms of Down syndrome include:

Mosaic Down syndrome - Early in development, some cells divide abnormally, resulting in the extra chromosome being present in a proportion, but not all cells; and

Translocation Down syndrome - approximately 3-4% of individuals with Down syndrome have cells containing 46

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The GSCWA Board of Management

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Carmel Wright
Client Services Coordinator
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Our Location and contact details:

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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*

chromosomes, but still have the features associated with Down syndrome.

In such cases, material from one chromosome 21 gets “stuck” or translocates to another chromosome, at conception. In such situations, cells from individuals with Down syndrome have two normal chromosomes 21, but also have additional chromosome 21 material on another chromosome.

Thus, there is still an extra chromosome 21, resulting in the features associated with Down syndrome. It will confirm a diagnosis of Down syndrome. Other health problems associated with Down syndrome include

increased risks of hearing and vision defects, heart abnormalities, infection, leukaemia, thyroid disorders, and of developing Alzheimer-type dementia in later life⁴.

Though there is no cure for Down syndrome, children born with the condition can lead productive lives. All children born with Down syndrome exhibit some level of learning disability, but this usually falls within the mild to moderate range.

Just like other children, babies with Down syndrome will learn basic skills - such as sitting, walking, talking, and self-care (such as toilet training and bathing) - but they will do so at a delayed pace. Early intervention with physical therapy, begun shortly after birth, can help strengthen muscles for these basic motor skills.

Some adults with Down syndrome live with their families; some live in group home accommodation; and others live independently.

Many people with Down syndrome hold productive jobs. The opportunities for people with Down syndrome are great, and these individuals can make lasting, positive contributions to society².

For more information and or support please contact either:

Down Syndrome Association of
Western Australia

PO Box 1388
 East Victoria Park WA 6981
 Ph: (08) 9472 8566
 Freecall: 1800 623 544
 Fax: (08) 9472 8577
 Email: dsawa@upnaway.com
 Website: www.dsawa.asn.au

References:

¹ Bower C, Ryan A, Rudy E & Cosgrove P (2002) Report of the Birth Defects Registry of Western Australia 1980-2001. King Edward Memorial Hospital, Centre for Women's Health, Subiaco. Number 9. (*In 460 is the average number of Down syndrome births - live and stillborn - during 1997-2001 in Western Australia*)

² Grayson C. My Web MD Website (2003): http://my.webmd.com/content/article/8/1680_54175

³ Morris JK, Mutton DE & Alberman E (2002) Revised estimates of the maternal age specific live birth prevalence of Down's syndrome. *J Med Screen.* (9):p4.

⁴ Down Syndrome Information Network Website (2003): <http://www.down-syndrome.info/topics/keyfacts/key-facts-EN-GB.htm>

⁵ Victorian Better Health Website (2003): http://www.betterhealth.vic.gov.au/bhcv2/bharticles.nsf/pages/Down_syndrome_is_a_common_genetic_condition?OpenDocument

⁶ National Institute of Child Health and Human Development Website (2002): <http://www.nichd.nih.gov/publications/pubs/downsyndrome/down.htm#DownSyndrome>

O'Leary PC, Bower C, Crowhurst J, Murch A, Goldblatt J. The impact of maternal serum screening for Down syndrome in Western Australia: 1980-1994. *Aust NZ J Obstet Gynaecol* 1996; 36:385-8

Current Issues and Matters of Interest

Researchers focus on helping brain to repair itself from range of degenerative diseases. From the Neuroscience Society Conference 29/1/2003.

Researchers focus on helping brain to repair itself from range of degenerative diseases. Researchers have discovered the adult human brain contains stem cells which have the ability to multiple and repair the brain in a range of degenerative diseases that affect millions of people worldwide.

They are now planning to extend their research to study the potential of adult stem cells to help the brain strengthen its defences against diseases such as Huntington's, Parkinson's, Alzheimer's and epilepsy.

The regenerative potential of the human brain was the focus of a keynote address today to the Australian Neuroscience Society Conference in Adelaide.

The conference has attracted medical leaders and researchers from around the world to probe new frontiers in human brain research and factors affecting sensation and perception, learning, memory, movement, sleep, stress and ageing.

Richard Faull, Professor of Anatomy at the University of Auckland School of Medicine, said groundbreaking research has shown clear evidence the human brain attempts to repair itself from disease. "We believe it is possible to help the brain in this process by using a patient's own stem cells for tissue repair," he said. "This would open the door to new opportunities to treat people with neurodegenerative diseases and stroke.

"By tapping into and genetically engineer-

ing the adult stem cells from the diseased adult brain, we would overcome major ethical, immunological and technical problems associated with the more controversial area of embryonic stem cell technology."

Professor Faull said the studies were supported by a very successful human brain donor program in New Zealand that allowed researchers to probe the degenerative processes associated with various diseases.

"What we have found in the diseased brains is that adult stem cells proliferate to form replacement neurons as well as other repair cells.

"By accessing these stem cells and introducing growth factors to generate new neurons, it may be possible to help the brain repair itself and for sufferers of neurodegenerative diseases to regain health and normal functions.

"There is still a lot of work to be done to understand the genetic and chemical mysteries of the brain before this type of approach can be used to help patients.

"But there is reason for excitement about the potential of neuron replacement in the treatment of patients that suffer from neurodegenerative diseases."

Reference: Media Release 29 January 2003
Australian Neuroscience Society Conference, Adelaide.

Coming Events

Motor Neurone Awareness Week Motor Neurone Disease Association

Dates: 6 – 12 April

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.

Contact: (08) 9346 7355

E-Mail: www.mndawa@cnswa.com

World Health Day - Theme: Healthy Environment for Children United Nations Association of Australia

Date: 7 April

The association wishes to raise the level of community awareness of the United Nations work on health through speakers and seminars.

Contact: (08) 9221 9455

Website: www.unaa-wa.org.au

World Parkinson's Day and Parkinson's Western Australia Annual Street Appeal.

Parkinson's Western Australia

Date: 11 April

This day aims to raise awareness of Parkinson's and to raise funds for research into the disease.

Contact: (08) 9346 7373

Great Australian Bite Diabetes Australia WA

Date: 23 April

The national fundraising event for Diabetes Australia

Contact: (08) 9325 7699

Website: www.diabetesaustralia.com.au

International Dance Day Ausdance WA

Date: 27 April

A free afternoon of multicultural dance in the city centre

Contact: (08) 9322 6101

Heart Week

National Heart Foundation

Dates: 27 - 4 April

Heart Week 2003 - A Heart for life, will be promoting heart healthy living across the lifespan. The Heart Foundation encourages healthy eating and physical activity with smoke-free living to help people live longer and better through prevention and control of heart disease and stroke. The WA branch will focus on rehabilitation and will launch of new materials for patients

living with cardiovascular disease.

Contact: (08) 9388 3343

Website: www.wa.heartfoundation.com.au



Interesting Websites

Internet Resources for Special Children (IRSC)

<http://www.irsc.org:8080/irsc/irscmain.nsf>

The Internet Resources for Special Children (IRSC) web site is dedicated to children with disabilities and other health related disorders worldwide.

The IRSC is the world's premier gateway to disability and other health related resources on the Internet, featuring thousands of the best web sites conveniently arranged by category, online communities, latest news articles, and access to books.

RUCSN

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

The Resource Unit for Children with Special Needs: facilitate the inclusion of children with special needs in child care in Western Australia. RUCSN's library has about 4,000 items covering child development, child care and disability.

The collection includes books, videos, journals and audio tapes. There is also a comprehensive collection of toys and equipment suitable for children with disabilities.

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:

Ian Potter

Provide grants for organisations with Deductible Gift Receipt (DGR). Funding amounts vary between \$2000 and \$50,000. Further information can be obtained at: <http://www.ianpotter.org.au>

Kodak Corporate Contributions

Provide grants for education, employment & training. Funding amounts vary, see their website for further information: <http://www.kodak.com> (see community relations)

* GSCWA can assist members with grant applications

GSCWA SPECIAL PROJECT FUND GRANT

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.

* Contact GSCWA for further information and criteria

GSCWA Grants

The Australian Huntington's Disease Association WA received a grant of \$847.00. The grant consisted of \$190 to provide a trainer from St John's Ambulance and \$657.00 towards the cost of a member of the Huntington community attending the Huntington conference in Tasmania.

GSCWA Members

FULL MEMBERS

Acoustic Neuroma Association of Australia
 Alzheimer's Association of WA
 Angelman Syndrome Association
 Arthrogyrosis 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
 Thalassaemia 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.

GSCWA News Corner

The Genetic Support Council is currently updating the 'Directory of Genetic Support Groups'. Our student Sophie is undertaking this project and has mailed out the details to organisations. Please feel free to contact us for further information.

So far we have had 21 responses out of 88, please keep them coming in.



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.

"Were on the Web"

www.geneticsupportcouncil.org.au