

Gene Advocate

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Special Points of
Interest:

- Thalassaemia Association of WA *Inherited Haemoglobin Disorders & Carer WA*
- VATER Syndrome
- Link Line: 'Frontonasal dysplasia condition'.

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About Our Members

Thalassaemia Association of WA *Inherited Haemoglobin Disorders*

Introduction

In August 2004 there is going to be an exciting international conference on Genetics and Population Health at the Esplanade Hotel in Fremantle. The conference will incorporate the Second Australasian Thalassaemia Workshop, with world expert guest speakers such as Dr Suthat Fucharoen from Thailand and Professors Bernadette Modell and Sir David Wetherall from the UK.

Thalassaemia is an important global health issue and the purpose of this article is to provide you with information about this inherited condition.

What is Thalassaemia?

The term thalassaemia encompasses a group inherited disorders of haemoglobin. There are two main types of inherited haemoglobin disorders, the thalassaemias and structural haemoglobin variants.

Current World Health Organisation data indicate that at least 5% of the world's population are healthy carriers of an inherited haemoglobin disorder. Our population is becoming increasingly diverse, with approximately 20% of West Australians identifying their ancestry as originating from regions of endemic haemoglobinopathies, such as Southern and Eastern Europe, North Africa and Middle East, South East Asia and Sub-Saharan Africa.

Haemoglobin

Haemoglobin is the oxygen carrying protein in red blood cells that gives blood its red colour. Haemoglobin A, which constitutes over 95% of the

haemoglobin in persons older than one year, consists of two alpha and two beta globin proteins, each attached to an iron containing pigment (haem). Haemoglobin is a complex and delicately balanced molecule and its oxygen carrying capacity is impaired when globin production is faulty due to genetic mutations in either alpha or beta globin genes.

Haemoglobin disorders are inherited in an autosomal recessive manner, which means that a person must inherit a defective gene from both parents to be affected. The name thalassaemia is derived from the Greek word "thalassa" which means "the sea" as the condition was first described in populations living near the Mediterranean Sea; though thalassaemias are also prevalent in Africa, the Middle East and Asia.

In thalassaemia, gene mutations result in deficient synthesis of either the alpha or beta chains of haemoglobin. The type of thalassaemia is named according to which globin is affected so the two main types are alpha and beta thalassaemia. Thalassaemias vary in severity, from a barely detectable anomaly in the blood to severe anaemia requiring lifelong blood transfusions and daily iron chelating therapy, depending on which globin gene is affected and which mutation/s are inherited.

Alpha Thalassaemia

In alpha thalassaemia, there is deficient synthesis of alpha globin and inefficient oxygen is transported to the body tissues. The genes that regulate alpha globin production are located on the tip of the short arm of chromosome 11. The inheritance pattern can be complex because there are four copies of the alpha globin gene, one pair on

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The GSCWA Board of Management

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Our Location and contact details:

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Monday to Friday.

Staff:

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

each chromosome 11. In general, people who carry one and two gene deletions are asymptomatic, those with three gene deletions have anaemia and may require periodic blood transfusions and the deletion of all four genes is not compatible with life.

Thus it is important for those who carry one and two gene deletions to be aware, as they have a 25% (one in four) chance of having an affected child if their partner is also a carrier. In a first step to examine the genetic variation of alpha thalassaemia in WA, a recent laboratory study identified over three hundred healthy carriers and twelve different alpha gene mutations.

These results illustrate that alpha thalassaemia is prevalent in WA and appropriate community awareness strategies need to be considered. Sickle Cell Disease is an example of a genetic mutation resulting in the production of abnormally shaped red blood cells. These "sickle" shaped cells can become trapped in small blood vessels and block them, causing pain and potentially damaging organs.

Beta Thalassaemia

In beta thalassaemia, beta globin production is impaired, causing these cells to die. The genes that regulate the production of beta globin are on the short arm of chromosome 16 and there are just two copies, one on each chromosome.

Carriers of beta thalassaemia, who have one effective copy and one defective copy of the beta globin gene, are generally healthy. As with alpha thalassaemia, it is important for carriers to be aware of their genetic status as they have a 25% (one in four) chance of having an affected child if their partner is also a carrier.

It is also important to remember that the different types of haemoglobin disorders can be inherited together, potentially impacting on health.

References

World Health Organisation 2002, Updated Estimates of the Frequency of the Haemoglobin Disorders in Each Country, WHO Human Genetics, 10 December 2002.

Australian Bureau of Statistics 2002, A Snapshot of Western Australia, 2001 Census Basic Community Profile and Snapshot.

De Silva S, Fisher C, Premawardhena A, Lamasbadusuriya S *et al* 2000. Thalassaemia in Sri Lanka: implications for the future health burden of Asian populations, *The Lancet*, 355:786-791.

For further information please contact:
Thalassaemia Association of WA Inc
PO Box 879 MORLEY, WA 6943
Ph: (08) 9276-1380 Fax: (08) 9276-1380

Current Issues and Matters of Interest

Carers WA – Carer Counselling Line (CCL) Media Release 28th August 2003

By the simple expediency of a toll free phone call I am in touch with others in my world and no longer having to bottle up problems.

Luke Lazich, Carer

Carers WA have launched their new state-wide, 24 hour, 7 days per week, Carer Counselling Line.

Many carers need to talk for a number of reasons. One reason is that carers may feel unable to discuss their feelings and issues with other members of their family or friends, who may not understand or have the time to listen. Carers WA aim is to provide carers with a “safe place” to express their emotions, needs and wants any-time, day or night.

The Carers Counselling Line is the first 24 hour professional and confidential counselling service available free to carers in Western Australia. As well as providing individual counselling this service will be providing telephone group counselling. Email and web based counselling options will be explored, in particular to address remote and rural carers needs.

Carers WA is recognised as the peak body for carers by both state and federal governments, representing over 200,000 family carers in WA.

A family carer is a person such as a family member, friend or neighbour who provides regular and sustained care to another person with a disability, who is frail aged or has a mental or chronic illness. The assistance is provided without payment other than pension or benefit.

After two years of providing counselling to carers at Carers WA it is very gratifying to finally address the emotional support needs of carers by providing free and convenient access to a dedicated carer specific 24 hour counselling service.

Pamela Siu, Counselling Coordinator, Carers WA

The Carers Counselling Line was officially launched last week by The Hon. Sheila McHale MLA, Minister for Community Development, Women's Interests, Seniors and Youth; Disability Services; Culture and the Arts.

The Carers Counselling Line is proudly funded by the Department of Community Development, Senior Interests and Volunteering. Carers WA would like to thank all the carers and service providers for their support in our endeavours to establish this much needed service for carers.

I have had the good fortune to call on the carers phone counsellor and found empathy and a sense of calming followed by valuable guidance so I was back in control of the situation. The experience left me feeling validated, reassured and able to take action. I would thoroughly recommend this service.

Ellen Walker, Carer

Tina Williams, Manager – Community Relations

Direct: 9202 8623 Mobile: 0416 095561

Email: tina.williams@carerswa.asn.au

28th August 2003

For further information please contact: Carers WA

Direct: 9202 8623 Mobile: 0416 095561

Fax: 9444 8966 Freecall: 1800 242 636

Coming Events

Australia's Breast Cancer Day - Cancer Foundation of WA

Date: 27 October

This day promotes awareness of breast cancer, fundraising for research and support for women with breast cancer through the sale of pink ribbons and enamel pins.

Contact: (08) 9212 4333

Website: www.cancerwa.asn.au

Asthma Week - Asthma Foundation of WA

Dates: 5- 11 October

This is the national week promoting awareness, education and self management. A range of education and support activities will be held throughout the state.

Contact: (08) 9481 1234

Website: www.asthmawa.org.au

Children's Week - Meerilinga Young Children's Foundation

Dates: 19 – 26 October

Children's week is a celebration of children and children's rights. It is a national week built around Universal Children's Day. It holds many free events, lectures, summits and family activities.

Contact: (08) 9424 5210

Website: www.meerilinga.org.au/childweek.cfm

Breast Cancer Awareness Month - BreastScreen WA

Dates: 1 – 31 October

The major focus for Breast Screen WA (BSWA) this month is the "Health Promotion in the Practice" event, an initiative to further develop partnerships between BSWA and Divisions of General Practice in the metropolitan area. This approach recognises the large numbers of women who pass through general practices and the importance of health promotion in a general practitioner's population health role.

Contact: 9237 6900 TTY: 1800 241 708

Website: www.breastscreen.health.wa.gov.au

Down Syndrome Awareness Week - Down Syndrome Association of WA

This week will include activities to help promote awareness of people with Down syndrome as valued members of the community. It is also an opportunity to fundraise to enable the support of people with Down syndrome, their families and the community.

Awareness week starts on Sunday 12th of October with a Family Fun Day and official launch by Professor Fiona Stanely. 17th of October is 'Dress up for Down syndrome' day – dress up or going casual throughout schools and businesses around the state for the donation of a gold coin.

Other events include a cake and craft Stalls. For further information or for an Information Pack (to mount a display)

Contact: 1800 623 544 or 9358 3544

Website: www.dsawa.asn.au

Mental Health Week WA - Association for Mental Health

Dates: 5 - 11 October

Many events are happening across WA. The launch is to be held between 12.00 noon and 3.00 pm on Sunday 5th October at the Western Power Parklands, Kings Park. Come along for a fun filled afternoon. Bring along a picnic, enjoy a sausage sizzle, listen to music, watch the dancers. Best of all, it's FREE!

A calendar of events can be downloaded online at the web address below.

Website:

http://www.mental.health.wa.gov.au/one/whats_new_view.asp?NewsID=16

National Stuttering Awareness Week - Speak Easy Association of WA

Dates: 19 – 25 October

This week the awareness and understanding of issues and difficulties faced by people who stutter. It raises awareness of the need for early intervention in children and to outline what treatment options are available to adults who stutter.

Contact: (08) 9225 4111

Website: www.speakeasy.org.au

National Lupus Awareness Month - The Lupus Group of WA

Dates: 1 October - 31 October

It is planned that this month will raise the awareness of Lupus across the community with corporate displays, educational seminars, literature, support services and research activities.

Contact: (08) 9224 3144

Website: www.lupuswa.com.au

National Bandanna Day - Canteen WA

Date: 31 October

National Bandanna Day is the major fundraiser for Canteen, the Australian organisation for young people living with cancer. As treatment can cause people to lose their hair Canteen sells bandannas to increase awareness of its work with young people.

Contact: (08) 6380 1884

Website: www.canteen.org.au

Occupational Therapy Week - OT Australia WA

Dates: 19 - 25 October

This week promotes the role of occupational therapists in the community.

Contact: (08) 9388 1492

Email: info@otauswa.com.au

Website: www.otauswa.com.au

Red Rose Day - Cystic Fibrosis Association of WA (Inc.)

Date: 17 October

This day involves the sale of red roses or Tattoos to public with proceeds to Cystic Fibrosis Australia Research Trust.

Contact: (08) 9346 7333 Freecall: 1800 678 766

Website: www.cysticfibrosiswa.org/index.html

Grants

Website Development Grants - LotteryWest

Aim: To help not for profit community groups develop websites and assist organisations to electronically communicate information about their services and activities to clients and the general community. *Further Information:*

www.lottery.wa.gov.au/corporate/grantsguide/guid_015.html

Freecall: 1800 655 270 Telephone: (08) 9340 5270

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

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.
- 3) Contact GSCWA for further information and criteria Telephone: 08 9389 6722
Email: admin@geneticsupportcouncil.org.au
Website:
<http://geneticsupportcouncil.org.au/pages/publications.asp>

Link Line



The Link Line provides a supportive and confidential means of connecting individuals and families for whom no known genetic support group exists. If any individual is seeking contact with others in these circumstances, *The Link Line* is available to you for this purpose.

Genetic support groups are an important resource for families or people in a similar situation. To date, there appears to be no specific support group for the condition *frontonasal dysplasia*.

A mother with an 8 yr old child with 'frontonasal dysplasia condition' in the New South Wales region would like contact with other families in Australia with a child with a similar condition.

- 3) Please contact Kristina at the Genetic Support Council if seeking contact with this family, telephone: (08) 9389 6722
Email: info@geneticsupportcouncil.org.au

VATER Syndrome and Oesophageal Atresia Support Group

Have you got VATER Syndrome or know of someone close to you that has? There is support available for you, as well as:

- ☞ parents;
- ☞ family members;
- ☞ friends;
- ☞ medical personnel.

Everyone welcome.

If you wish to get involved or would like further information, please contact Julie or Mary on (02) 6025-9684 or julielee@bigpond.com.



Interesting Websites

LWW.com
www.lww.com

Site of Lippincott Williams & Wilkins, an international publisher of professional health information for physicians, nurses, specialised clinicians and students. More than an online store, LWW.com contains tools for faculty, students, librarians, and practitioners.

Yahoo groups—Australia and New Zealand
<http://au.dir.groups.yahoo.com/dir/1600094714>

Alpha-1 Antitrypsin Deficiency	Cystinosis
Gardner Syndrome	Haemochromatosis
Hemophillia	Huntington's Disease
Marfan Syndrome	Porphyria
Prader-Willi Syndrome	Rett Syndrome
Treacher Collins Syndrome	Williams Syndrome

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 a survey.

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:

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Website.....