



Gene Advocate

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2005

Special Points of Interest

- ▶ The Androgen Insensitivity Syndrome (AIS) Support Group Australia

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About Support Groups

The Androgen Insensitivity Syndrome (AIS) Support Group Australia Inc.

Our Mission

The Androgen Insensitivity Syndrome (AIS) Support Group Australia Inc. (A0041398U) is a peer support, information and advocacy group for people affected by AIS and/or related intersex conditions, and their families.

We support members (both in Australia and overseas) that have any grade of Androgen Insensitivity Syndrome, and support any issues relevant to living with AIS. These issues include infertility, disclosure, hormone therapy, gender identity, surgical intervention of children with intersex conditions, sexual intimacy etc.

We also provide support to those with related intersex conditions like Partial and Complete Gonadal Dysgenesis, MRKH (also known as Vaginal Agenesis), 5 α -Reductase Deficiency, 3 β -Hydroxysteroid Dehydrogenase Deficiency, 17-Ketosteroid Reductase Deficiency and 17 β -Hydroxysteroid Deficiency. (The previous name for AIS was Testicular Feminisation Syndrome).

We acknowledge that the gender of people with intersex conditions (including AIS) range from female to male and anywhere in between. The AISSGA believes in a holistic model of health as per the World Health Organisation definition of 'health':

"Health is a state of complete physical, mental and social well-being and not merely the absence of disease and infirmity".

What is AIS?

Androgen Insensitivity Syndrome (old name Testicular Feminisation Syndrome) causes an interruption of the foetal development of the reproductive system. In AIS the child is conceived with typical male (XY) sex chromosomes. Embryonic testes develop and start to produce androgens but the body is unable to respond to these androgens to a varying degree.

This means that the genitals can vary from completely female if there is no response through to almost completely male if there is a slight insensitivity to androgens or anywhere in between. So the external genital development continues along *female* lines (the 'backup' route) but the development of *female internal* organs has already been suppressed by a hormone (MIF or Mullerian Inhibitory Factor) from the foetal testes.

A gene on the X chromosome controls tissue androgen sensitivity, and AIS is an X-linked recessive condition, inherited down the maternal line (or, in an estimated one third of cases, results from a spontaneous mutation).

A mother who carries the variant gene has a 1:2 chance of any XY child having AIS and a 1:2 chance of any XX child being a *carrier* of the variant gene like herself, but XX individuals can be tested to Syndrome, Testicular Feminisation Syndrome, Feminising Testes Syndrome, Male Pseudo-hermaphroditism,

The views and opinions 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

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Executive Dean, Health Sciences Curtin University

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Information and Development Officer, Down Syndrome Association of WA

Del Weston
Executive Officer, Australian Huntington's Disease Association WA Inc.

Our location and contact details:

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Telephone: (08) 9389 6722
Mobile: 0411 585 113
Email: 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

Goldberg- Maxwell Syndrome (CAIS), Morris's Syndrome (CAIS), Lub's Syndrome (PAIS), Reifenshtein Syndrome (PAIS), Gilbert-Dreyfus Syndrome (PAIS), Rosewater Syndrome (PAIS).

Other XY conditions with some AIS-like features: 5 alpha-reductase deficiency, 17 keto-steroid reductase deficiency, XY gonadal dysgenesis (Swyer Syndrome), Leydig cell hypoplasia, Denys-Drash Syndrome, Smith- Lemli-Opitz Syndrome. *XX conditions with some AIS-like features:* Mayer Rokitansky Kuster Hauser (MRKH) Syndrome, Mullerian dysgenesis.

Incidence

The most accurate figure currently available for CAIS comes from an analysis (1992) of a Danish patient register, suggesting an incidence of 1 in 20,400 XY births (hospitalised cases only, so true incidence probably higher). PAIS may be only about 1/10 as common as CAIS.

Effects (CAIS)

Even in the complete form there will be no ovaries, Fallopian tubes or uterus, and the vagina will be blind ending and spontaneous feminising puberty to occur, which may have physiological and psychological advantages over one induced by exogenous hormones. Note that girls with CAIS cannot be *masculinised* by hormone administration, because of their complete insensitivity to androgens.

Although the pre-adult risk of cancer is too small to justify it, gonadectomy is sometimes done in infancy or childhood, usually with the intention of avoiding a psychological crisis arising from explaining the need, later on, for such an operation.

Obviously, gonadectomies should **not** be performed on males with PAIS, as they produce the testosterone needed for secondary sexual characteristics, although additional testosterone treatment may be required.

HRT/Osteoporosis

When the testes are removed from women with AIS after puberty, immediate long-term female hormone replacement therapy (HRT) is needed to prevent menopausal symptoms and osteoporosis (bone thinning) and protect against cardiovascular disease. In the case of gonadectomy in infancy/childhood, HRT is often started at 11, in order to initiate puberty. Low bone density seems to be more common in AIS women.

Contact Information

P.O. Box 1089 Altona Meadows
Victoria 3028 Australia
International: +61 3 9 872 4884
Australia: 03 9872 4884

For Support and general enquiries

President - Ms. Andie Hider -
ashider@alphalink.com.au or 03 9872 4884
Vice-President - Phoebe (Queensland) -
Government and Advocacy contact -
Vice-President - Mr. Tony Briffa -
aissg@iprimus.com.au or 03 9315 8809.

Information Source: AIS Support Group Australia Inc. AIS Fact sheet. [Online]. Available:
http://home.vicnet.net.au/~aissg/fact_sheets.htm

Document reviewed by the AIS Support Group Australia Inc.

Current Issues and Matters of Interest

More Help For GPs: New Prescription Shopping Hotline Opens. Tony Abbott - Minister for Health & Ageing 14/02/2005.

The Minister for Health and Ageing, Tony Abbott, and the Minister for Human Services, Joe Hockey, announced a new hotline to prevent prescription shopping.

The Prescription Shopping Information Service gives doctors 24-hour access seven days a week to a free phone call service. The service will help doctors identify if their patients have been supplied PBS medicines within a three-month period that may be in excess of therapeutic need. Media Release available online.

Information Source:
http://www.aushealthcare.com.au/news/news_details.asp?nid=5009

Privacy Commissioner Makes Health Related Determinations. Privacy Commissioner - Karen Curtis 14/02/2005.

The Privacy Commissioner, Karen Curtis, announced that she has made two health related determinations.

These determinations are made under section 80A and section 80B(3) of the Privacy Act 1988.

The determinations have effect from 10 February 2005 and expire on 9 February 2006. The determinations were made in response to an application from a general practitioner who was concerned that accessing the Information Service provided by the Health Insurance Commission (HIC) under its Prescription Shopping Project may have been in breach of elements of the National Privacy Principles of the Privacy Act. Media Release available online.

Information Source:
http://www.aushealthcare.com.au/news/news_details.asp?nid=5027

Regulator Takes Tough Action On Arthritis Drug. The Therapeutic Goods Administration 14/02/2005.

The Therapeutic Goods Administration has introduced tough new measures on the prescribing of anti-arthritis drugs known as Cox-2 Inhibitors following the findings of a review into the safety of this family of medicines. Media Release available online.

Information Source:
http://www.aushealthcare.com.au/news/news_details.asp?nid=5013

Mesoblast Contracts Cell Processing Facility for Pilot Clinical Trials – Mesoblast 15/02/2005

Mesoblast Contracts Cell Processing Facility for Pilot Clinical Trials Mesoblast Australia's adult stem cell company, Mesoblast Limited (ASX:MSB), today announced that it had signed an agreement for production of specialised adult stem cells, known as Mesenchymal Precursor Cells (MPCs), to be used in human pilot clinical trials in patients with orthopaedic and cardiovascular diseases. Media Release available online.

Information Source:
http://www.aushealthcare.com.au/news/news_details.asp?nid=5033

Advertise in the 'Gene Advocate' or on our website!!!

Have you got anything you want to be published?
Either in our newsletter or our website , it can be included to display your information.

So contact us via e-mail or phone and we can arrange it for you.
Email: admin@geneticsupportcouncil.org.au or Telephone: (08) 9389 6722

Coming Events

"Hat Frenzy Friday" - Activ Foundation

Date: 4 March 2005

Wear your favourite or craziest hat on Hat Frenzy Friday (or buy Hat Frenzy Friday merchandise) to help raise money for people with intellectual disability in Western Australia.

Contact: Hat Frenzy Friday Hotline on 1300 13 72 31.

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

"Relay For Life Merredin" - The Cancer Council Western Australia

Date: 5 - 6 March 2005

Relay is a fun 24-hour team event to raise funds to further the fight against cancer. The highlights of this event are the Survivor's Walk and the Candlelight Ceremony. The team members fundraise prior to the event.

Contact: 1300 65 65 85

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

"Orthoptic Awareness Week" - Orthoptic Association of Australia Inc.

Date: 7 - 11 March 2005

To educate the community about what orthoptics do and how to contact them.

Contact: (03) 9521 9844

Facsimile: 03 9598 9499

Email: admin@orthoptics.org.au

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

"Red Cross Calling" - Australian Red Cross

Date: 7-26th March 2005

This is the annual door knock to raise funds for the Australian Red Cross.

Contact: (08) 9325 5111

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

To become a volunteer collector telephone:
1800 253 744.

"World's Greatest Shave to Care and Cure" Leukaemia Foundation

Date: 10 - 12th March 2005

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>

"Home Freeway Bike Hike"- Asthma Foundation of WA

Date: 20 March 2005

A freeway bike ride from Belmont, through the tunnel and up to Joondalup to raise awareness and funds for Asthma Foundation research and education.

Contact: (08) 92893600

For registering in the event go to website:

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

"Walking with the Stars" - Muscular Dystrophy Association of WA (Inc.)

Date: 19 March 2005

Walking with the Stars is a fundraising event organised by the Muscular Dystrophy Association of WA to raise funds to help researchers bring hope to affected children & their families.

Location: Victoria Gardens, East Perth.

A 7.5kms twilight walk around bridges to help find a cure for Muscular Dystrophy. Fun starts at 4pm, registration at 5pm, walk starts at 5.30pm.

Contact: Jude Newberry

Ph: (08) 9346 2022

Email: newberry@cyllene.uwa.edu.au

Website: <http://www.anri.uwa.edu.au> or

<http://www.mdaawa.asn.au>

Institute of Public Administration Australia WA Division (IPAA) - "Contracting for the Delivery of Community Services"

Date: Tuesday 15 March at Burswood

Major awareness seminar - the flyer and registration form is available on the website. Half-day seminar with a range of government and non-government speakers. Presented in conjunction with the State Supply Commission.

Queries: Telephone IPAA WA on 9221 1177

Email: info@ipaawa.org.au

Website:

<http://www.wa.ipaa.org.au/ipaa/about.aspx>

"International Women's Day" The Office for Women's Policy

Date: 8 March 2005

International Women's Day (IWD) is a global celebration of the economic, political, environmental and social achievements for women.

Website:

<http://womenwa.communitydevelopment.wa.gov.au/content/events/order.asp>

"AHPA 15th National Health Promotion Conference" Australian Health Promotion Association

Date: 13-16 March 2005

A Health Promotion Conference reflecting on the past and creating a vision for the future.

Venue: National Convention Centre, Canberra.

For further information,

Email: ahpa@confco.com.au

Website: <http://www.healthpromotion.act.gov.au>

"Health Promotion Courses"

Date: 20 May and 3 and 17 June 2005

Details of 3 one-day courses in planning, methods and evaluation in health promotion to be held at Curtin University. For further information,

Website: <http://www.man.org.au/newsletter>

Flyer:

<http://www.man.org.au/newsletter/32/HealthPromotionCourses.pdf>

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 following conditions/syndrome:

- A mother of an eight year old son living with **Hereditary Multiple Exostosis** in the Canberra region would like contact with other families around Australia with a child with the same condition.
- A mother with an eight year old son living with **CAPOS syndrome** in the Victoria region would like contact with other families around Australia with a child with a similar condition.
- A mother with a 16 month old child living with **Bilateral periventricular nodular heterotopia** in the VIC region would like contact with other families in any part of Australia with a child with a similar condition
- A Perth family would like contact with other families living with **XYX Syndrome** in the expectation of forming a support group, online & or in person. Health professionals with an interest or knowledge are welcome to contact us

also

- ☞ Please contact Kristina at the Genetic Support Council if seeking contact with this family, telephone: 08 9389 6722 or email info@geneticsupportcouncil.org.au



Grants

Support Fund For Adults With Physical Disabilities ANZ Charitable Trusts

Closes: 1 April 2005

Aim: To provide assistance to physically disabled adults. Organisation must have both an Income Tax Exempt Charity (ITEC) and a Deductible Gift Recipient.

Contact details:

Freecall: 1800 808 910

Website:

<http://www.anz.com/australia/charitabletrusts/guidelines.asp>

Advancing Participation in Community Life, LotteryWest

Closes: Ongoing

Aim: To support initiatives that encourage broad community involvement in social, cultural or recreation activities, particularly for people who face barriers to participation. Website:

<http://www.lottery.wa.gov.au/asp/index.asp?pgid=263>

Freecall: 1800 655 270 TTY: (08) 9340 5236

Healthway Funding

Closes: No date

Healthway has recently introduced a new type of health promotion project grant. Innovation grants of up to \$20,000 are available to encourage projects that focus on developing and trialling new innovative strategies for targeting Healthway's high priority health issues (tobacco, mental health, physical activity or nutrition), OR for reaching priority population groups such as Indigenous people.

There is no closing date - applications can be accepted at any time and with the usual 4 months lead time. There is more information on the Healthway web-site (link below), and in line with all other health promotion project funding, Healthway encourages people to contact them first to discuss their ideas before applying: <http://www1.healthway.wa.gov.au/internal.aspx?MenuID=575>

Information Source: M.A.N News 32, 21 Jan 2005. [Online]. Available: <http://www.man.org.au/newsletter/32.htm#10>



► GSCWA can assist **members** with grant applications & further resources!

Interesting Websites



➤ 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.

CAN YOU RISK IT?

RiskCover Insurance Commission of WA.

<http://www.icwa.wa.gov.au>

(Last Reviewed: 8/02/05)

A 22 page guide on risk management.

"An introduction to Risk Management For Community Organisations". A risk management initiative of the Government of Western Australia.

Available:

http://www.icwa.wa.gov.au/cifund/cif_risk_it.pdf

SHOCKING RESULTS FROM COMMUNITY SECTOR SURVEY - WACOSS

<http://wacoss.org.au/election2005/index.htm>

(Last Reviewed: 10/02/05)

WACOSS released the results of the Community Sector Survey which reveal alarming trend in service closures and roll-backs in the services provided by non-government organisations in WA. The details of the survey can be found by going to the WACOSS Election 2005.

MEN'S SERVICES

Department for Community Development

<http://community.wa.gov.au/Communities/Men/>

(Last Reviewed: 10/02/05)

The Department for Community Services has launched a website with a page devoted to men's services.

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
 Cushing's 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)
 Fragile X Support Group WA (Inc)
 Haemophilia Foundation WA Inc.
 Heart Kids WA
 Klinefelters Support Group
 Learning and Attention Disorders Society of WA (LADS)
 LQTS Support Group WA
 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
 PXE Support WA
 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
 Turner Syndrome Association of Australia (WA Branch)
 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
 Ectodermal Dysplasia Support Group - OzED
 Genomics Directorate, 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

Recently a number of the members of the GSCWA met to establish a Reference Group to assist the Council in planning its activities in 2005. In particular, holding a number of information sessions on matters and issues that they felt would be of interest to GSCWA members.

The Council in conjunction with the Reference Group will hold a series of information sessions and the following are the initial proposed topics:

- Genetic counselling, testing and current issues.
- Pre-implantation and genetic testing.
- Folate Fortification of certain foods.
- Parenting and managing challenging behaviour.
- Genetic links to other conditions, such as Autism, Down syndrome and Alzheimer's.

Speakers with particular expertise will be engaged to present the various sessions. At the moment it is envisaged that these will be held approximately monthly.

First session: Genetic counselling, testing and current issues.

Guest Speaker: Dr C.-E. Kiraly-Borri
 Clinical Geneticist, MD, PhD
 Genetic Services of Western Australia.

Time: 10.30am -12.00pm

Date: 2nd of March 2005

Venue: "The Niche"
 Aberdare Rd & Hospital Avenue, Nedlands.

Cost: Free

Morning Tea Provided!

For catering reasons, please RSVP by 28th of February 2005.

Please let us know if you have particular dietary needs.

Contact: info@geneticsupportcouncil.org.au
 Telephone: (08) 9389 6722

Flyer available online:

<http://www.geneticsupportcouncil.org.au/pages/publications.asp>