



Gene Advocate



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**November
2003**

Special Points of Interest:

- Klinefelter Support Group of WA
- Link Line
- GSCWA News Corner: Directory of Genetic Support Groups 2003

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About Our Members

KLINFELTER SYNDROME **Klinefelter Support Group Western Australia**

What is Klinefelter Syndrome?

The typical characteristics of this syndrome were first described by Drs Klinefelter and Albright in 1942. Dr Harry Klinefelter and his co-workers at the Massachusetts General Hospital in Boston published a report about nine men who had enlarged breasts, sparse facial and body hair, small testes, and an inability to produce sperm.

By the late 1950s, researchers discovered that men with Klinefelter syndrome, as this group of symptoms came to be called, had an extra X chromosome, (an XXY pattern instead of the usual male arrangement, XY). This is caused by a complex fault in the cell division of one of the parent's sex cells, and can occur in either the ovum or the sperm. Only males can be affected by Klinefelter syndrome.

In the early 1970s, researchers around the world sought to identify males with the extra chromosome by screening large numbers of newborn babies. One of the largest of these studies, sponsored by the National Institute of Child Health and Human Development (NICHD), checked the chromosomes of more than 40,000 infants. Based on these studies, the XXY chromosome arrangement appears to be one of the most common genetic abnormalities known, occurring as frequently as 1 in 500 to 1 in 1,000 male births. Although the prevalence of XXY genotype is relatively common, the characteristics of the syndrome are variable. Many men live out their lives without ever even suspecting that they have an additional chromosome.³

There is also a 'mosaic' form of Klinefelter syndrome where only some of the body cells have an XXY chromosome pattern; the remainder have the normal XY count. Generally, the severity of the

characteristics is related to the number of cells in the body which have the abnormal chromosome pattern.

Characteristics.

Because they often don't appear any different from anyone else, many XXY males may never learn of their extra chromosome. However, if they are diagnosed, it is likely to happen before or shortly after birth, during early childhood, adolescence, or in adulthood (as a result of testing for infertility).

XXY babies differ little from other children their age. They tend to be what many parents call "good" babies-quiet, undemanding, and are perhaps even a little passive. As toddlers, they may be somewhat shy and reserved. They usually learn to walk later than most other children, and may have delays in learning to speak. In some, the language delays may be more severe, with the child not fully learning to talk until about age 5. Others may learn to speak at a normal rate, and not meet with any problems until they begin school, where they may experience reading difficulties. A few may not experience any problems in learning to speak or to read.

XXY males usually have difficulty with expressive language, which is the ability to put thoughts, ideas, and emotions into words. In contrast, their faculty for receptive language (understanding what is said) is close to normal.

A female type of breast development also sometimes occurs in boys with Klinefelter syndrome. Tall stature is also one of the consistent features of the syndrome. The body proportions are unusual with disproportionately long legs compared with the trunk. Body hair is usually scanty in boys with Klinefelter syndrome. Normal pubic, axillary and chest hair fails to appear and daily shaving is rarely necessary.

DISCLAIMER

The views expressed in this newsletter are those of the individual authors and not necessarily those of the GSCWA. 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/treatment.

The GSCWA Board of Management

CHAIR

Professor Charles Watson: *Executive Dean,
Health Sciences Curtin University*

DEPUTY CHAIR

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Cystic Fibrosis WA Inc.*

TREASURER

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Kerry Gavey

Convenor

Klinefelter's Support Group

Susan Herrmann

Health Consumers Council WA

Julie Ireland

Family Support Worker

Down Syndrome Association of WA

Diane Lunt

Committee Member

Heart Kids WA Inc

Dr Peter O'Leary

Director

Genomics Branch

Department of Health

Del Weston

Executive Officer

*Australian Huntington's Disease Association WA
Inc.*

Our location and contact details:

Level 1, Oasis Lotteries House

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Office hours:

9.00am to 5.00pm.

Monday to Friday.

Staff:

Terry Keating, *Executive Director*

Anja Hermann, *Administrative Officer*

Kristina Johns, *Resource Officer*

Mental abilities are often below average. Diabetes and thyroid problems are also more common than usual in males with the syndrome.¹

Management Implications.

The most serious effects of Klinefelter syndrome are often the psychological trauma felt by many boys at puberty when secondary sexual characteristics do not develop along normal lines. The help of a clinical psychologist can be of great value in helping males with Klinefelter syndrome come to terms with their genetic inheritance.

Testosterone therapy is often prescribed to young adolescents. This can aid the development of sexual characteristics, and although it will have no effect on infertility, it can help to reduce the psychological problems encountered during adolescence and early adult life.¹ Testosterone can also improve mood, school performance, and work capacity. For gynecomastia (large breasts), plastic surgery is an option.²

Attendance at a mainstream school is usually possible for most boys with Klinefelter syndrome, although some with more restricted intellectual abilities will need special facilities. Life expectancy for those with Klinefelter syndrome is thought to be the same as that of the general population.¹

References

¹ Gilbert, P. (1993) "The A-Z Reference Book of Syndromes and Inherited Disorders." Chapman & Hall, London.

² Magalini S I, Magalini S C: Dictionary of Medical Syndromes 4th ed, pp 456-457. Philadelphia, New York, Lippincott-Raven, 1997

¹ Gilbert, P. (1993) "The A-Z Reference Book of Syndromes and Inherited Disorders." Chapman & Hall, London.

³ National Institute of Child Health & Human Development, <http://www.nichd.nih.gov/publications/pubs/klinefelter.htm>

Klinefelter's Support Group Western Australia

Kerry Gavey

Convenor

14 Shetland Drive

HEENLEYBROOK WA 6055

kerrygavey@optusnet.com.au

Other Klinefelter Support Groups:

Victoria: Klinefelter Support Group

3/1 Anthony Street

CROYDON VIC 3136

PH: 03 9723 6148

New Zealand Klinefelter Association

96 Hair Street

WAINUIOMATA NZ 6008

PH: 04 9715 404

mwhope@ihug.co.nz

Information on the Web:

American Association for Klinefelter Syndrome Information and Support (access) <http://www.aaksis.org/>

Current Issues and Matters of Interest

Telecross – Australian Red Cross WA Services.

Telecross recruits and trains volunteers to provide a daily telephone call to people who live alone and are at risk of sudden health deterioration. If the volunteers' call is not answered, a Red Cross staff member arranges for someone to visit the client's home and check on their well-being. The Home and Community Care (H.A.C.C.) program in WA provides funding to assist Red Cross in the operation of this service.

Telecross currently operates in Perth, Rockingham, Mandurah, Bunbury, Denmark, Geraldton, Mullewa, Narrogin and Kalgoorlie. Currently over 170 frail aged or people with disabilities who live alone, receive a reassuring phone call from their Telecross volunteer at the same time each morning.

To find out more about Telecross telephone (08) 9325 5111 or email info@wa.redcross.org.au

Source:

Australian Red Cross Western Australia Services: Telecross. (30/10/2003) [Online], Available: http://www.redcross.org.au/wa/services_community.htm

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).

Designer Mice!

Some of the world's most serious diseases may be cured with the help of genetically modified (GM) mice from Perth. A WA-based company, Ozgene, has become a major supplier of these "designer mice" to renowned research laboratories around the world. Ozgene was founded by Dr Frank Koentgen and his wife Dr Gabi Suess in 1999.

Whilst we normally consider them a furry nuisance, GM mice are a worthwhile investment for researchers, because there is at least 90% similarity between each human gene and its corresponding gene in the mouse.

In order to discover the role of a particular human gene, scientists can "knock-out", mutate or add the gene to the mouse's DNA. Using these techniques, Ozgene has bred a particular strain of GM mouse which has symptoms similar to the human disease spina bifida. These mice have enabled an Australian research group to study the disease in more detail and it is hoped that this research could lead to a greater understanding of the factors that lead to spina bifida and other neural tube defects.

Other strains of mice generated by Ozgene are being used to study diseases such as Alzheimers, breast cancer and Huntington disease. Ozgene has won the Emerging Export Business Award at this year's WA Industry and Export Awards.

Source:

Genomics Directorate, Department of Health Western Australia.
Level 8, East Point Plaza
233 Adelaide Tce, Perth
Phone: (08) 9323 6628
Fax: (08) 9323 6611

Coming Events

International Volunteer Day

Date: 5 December

In 1985, the United Nations recognised the first International Volunteer Day to celebrate the important role that volunteers play in communities around the world. At the conclusion of the International Year of Volunteers (IYV) in 2001, the Premier of Western Australia announced in future years, 5 December would become 'Thank a Volunteer Day' in Western Australia.

For further information contact

Volunteering WA on 08-9420 7288

Email: community@volunteer.org.au

Web: <http://www.volunteer.org.au/index.html>

Or

Volunteering Australia on (03) 9820 4100

Web Site: www.volunteeringaustralia.org

E-mail:

volaus@volaus@volunteeringaustralia.org

Motor Neurone Disease Street Appeal

Motor Neurone Disease Association

Date: 5 December

This street appeal is organised to raise funds and promote awareness of Motor Neurone disease.

Contact: (08) 9346 7355

Website: www.mndawa.iinet.net.au

Hemophilia World Congress 2004

XXVI International Congress Of The World Federation Of Hemophilia

Date: October 17 to 21, 2004

Location: Bangkok, Thailand

Contact Hemophilia Foundation in your state. WA residents please contact Michelle on 9420 7294 - Fax: 9486 8966

E-mail: office@hfw.org

The Institute of Public Administration Australia (IPAA) WA

IPAA's small group skills training courses for the period *December 2003 to May 2004* is now available.

Full details about each course, and registrations forms, are available on IPAA's website at:

<http://www.wa.ipaa.org.au/training.html>

* Queries to IPAA's Training Officer on 9426 8629 or ipaa@wt.com.au

Please note: IPAA's October and November 2003 seminars are not included in the Calendar as they are already fully booked. IPAA seminars fill quickly on a 'first come' basis.

Grants

Information and Communication Technology Grants –Lottery West WA, Community Funding.

To assist community organisations improve their efficiency and effectiveness through the use of technology. To help community organisations purchase essential equipment and software.

✓ Non-profit community groups can apply for funding.

Further information and contacts:

Ph: (08) 9340 5270

Toll Free: 1800 655 270

TTY: (08) 9340 5236

Email: grants@lottery.wa.gov.au

Guideline documents maybe downloaded from website:

http://www.lotterywest.wa.gov.au/corporate/grantsguide/guid_012.html

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



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 groups for the conditions; *XYX Chromosome, Frontonasal Dysplasia, Vitelli Form Macular Dystrophy (Degeneration) & Brachydactyly*.

A Western Australian Man is seeking linkage with others living with a condition called 'Vitelli Form Macular Dystrophy' (Degeneration).

.....

A Perth Family is seeking linkage with other people living with 'XYX Chromosome' condition.

.....

A Western Australian family is seeking linkage with others living with a condition called 'Brachydactyly'.

.....

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.

.....

The Genetic Support Network of Victoria have someone looking for another person living with 'Essential Thrombocythemia'.

Please contact Kristina at the Genetic Support Council if seeking contact:

☎ Telephone: (08) 9389 6722

✉ Email: info@geneticsupportcouncil.org.au

Interesting Websites

Green PC

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

(Last reviewed 30/10/2003)

Locations: Western Australia, Queensland & Victoria.

Green PC refurbishes used computers received from government and business organisations and makes them available to those who might not usually be able to afford a new computer system.

To be *eligible* to purchase a Green PC, you must be holders of a current *Health Care Card, Aged Pension Card, Disability Card* or some *other* form of documentation relating to low-income or disadvantage.

Green PC also offers computer systems and IT services at affordable rates to *registered charities and community groups*.

Contacts - Melbourne: (03) 9486 9355, Brisbane (07) 3407 2054, Perth (08) 9434 0530

Ehlers-Danlos National Foundation (USA) **EDNF Guide for Teachers and Parents**

(Last reviewed 1/10/2003)

<http://www.ednf.org/ptguide.php>

Part of the guide is available online, the EDNF wanted to make the information available to people as soon as possible so that children can benefit from it this school year.

* Please *be aware* that the completed guide will include pictures and several appendixes. The completed guide will be available soon to download from the web site and also in a small printed booklet that will be practical for inclusion in the students folder.

Guidelines for Public Sector Agencies working with Volunteers - Department of the Premier & Cabinet Government of Western Australia Published April, 2003.

<http://www.dpc.wa.gov.au/psmd/pubs/psrd/volunteer/index.html>

This second edition of the guidelines has only been published electronically (Online).

GSCWA Members



FULL MEMBERS

- Acoustic Neuroma Association of Australia
- Alzheimer's Association of WA
- Angelman Syndrome Association
- Arthrogryposis 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
- Professor Charles Watson, Curtin University
- Terry Keating, GSCWA

GSCWA News Corner

The Genetic Support Council now has the 'Directory of Genetic Support Groups 2003' available online at our website under 'publications'.

Web: <http://geneticsupportcouncil.org.au>
The directory lists the full members & associated members of the GSCWA.

➔ *Our online PDF documents are encrypted for 'Accessibility' purposes, you may need to download the latest version to read the PDF 'Directory of Genetic Support Groups 2003'.*

GSCWA 'Christmas Morning Tea'
Date: Tuesday 9th of December
Location: GSCWA Office



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.....