THE · ASSOCIATION OF·GENETIC·SUPPORT OF · AUSTRALASIA

FUNDED BY THE NSW HEALTH DEPARTMENT

OCTOBER 1994 ISSUE 14

ADVANCE NOTICE
ANNUAL GENERAL MEETING

AGSA will be holding its Annual General Meeting on Sunday 27th November 1994 at 10 am at the office in Surry Hills. This will be followed by a Christmas Celebration Lunch. Put this in your diary, it will be a day not to be missed.

MISSION STATEMENT

To facilitate support for those affected directly or indirectly by genetic conditions throughout Australasia.

Registered Charity No.C.C.27702

EDITORIAL

Many of you will be aware that the AGSA Committee has been addressing ways of streamlining many aspects of administration. With this in mind, the AGM is to be brought forward to coincide with the end of our financial year, 30th September 1994 and not, as previously the case, held over until the following March. Hence, two AGM's this year! Further information regarding the AGM and combined Christmas luncheon will be forwarded shortly.

HELP!

AGSA receives many requests to attend disability expos, conferences etc. and we believe there is an advantage in displaying a banner, including a slogan. Here is the catch. The committee needs your help in creating this slogan. We have discussed possible suggestions at many meetings but have been unable to reach a consensus, therefore it was decided to ask for suggestions from our membership. We would appreciate any suggestions, either by phoning the office or in writing. Ideally we would like this by the AGM on 27th November however any inspirations following this date will be considered.

We hope by now that many of our members will have made contact with Dianne. We believe that she would appreciate hearing from you, as keeping in touch with our members is the best way to meet your needs and find out how we can be of assistance.

Ros Smith

ISSN 1033-8624

SUPPORT/ INFORMATION OFFICER REPORT

AGSA's "How to start a support group" seminar was very successful and I would like to thank the excellent speakers, Glenn and Geoff Fisher and Alison Gatt from Turner's Syndrome, Dr. Jacqueline Morgan from the Muscular Dystrophy Association, Mandy O'Reilly of NSW Genetic Education and David Fennell from Charcot-Marie-Tooth Support Group. Members from six new groups and seven people representing existing groups attended. Naturally the new groups found the information overwhelming and the existing group representatives used the opportunity to clarify the new charity laws, catch up on constitutional requirements and learn about privacy laws. The video of the seminar will be available for hire. A special thank you to all the committee for helping with preparations for the day.

There was a Rare Choromosomal Support Group coffee morning on 23rd October at 10.00 a.m. at AGSA's offices. It was decided to have a picnic day on 12th March 1995. Check further Newsletters for final details.

On 6th November a second informal Klinefelter Support Group meeting will be held at 10.00 a.m. and lunch will be provided by AGSA. Dr. Robert Leitner, Developmental Paediatrician at St. George Hospital Assessment Centre has agreed to return for this meeting and Professor Stuart Einfeld, Psychiatrist, Prince of Wales Hospital, will also be in attendance to answer any queries unresolved from the first meeting.

Margaret Sahhar, Social Worker at Royal Children's Hospital, Melbourne, is willing to assist in establishing a Klinefelter group in Victoria.

The Inner West Disbility Forum is holding an information expo on November 5th 1994 at 10 am. AGSA will be participating and we would like to display your support group brochures. Please give me a call if you are interested.

It has been a busy couple of months and I thank you for your many calls. I look forward to celebrating with you at our Christmas party.

Dianne Petrie

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GENETIC SERVICES

Judith Elber, Genetic Counsellor previously of Wollongong has now moved to Liverpool. Judith covers the area from Bankstown to Bowral and can be contacted at the:
Liverpool Health Service
P.O. Box 103,
Liverpool.
Phone: 828 5694

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CAN I HELP ?

Support Groups meet many of your needs. However, if you want to talk things through with a psychologist/ genetic counsellor with wide ranging hospital experience, I am now available for private consultations. Individuals or families welcome. For further information please contact:

Bronwyn Butler
(02) 419 6782

CONTACT CORNER

AGSA will publish requests for contact and letters from people searching for families with similar experiences, from those seeking or contributing specific information as well as other resource information.

Anyone who wishes to reply to a request or a letter should write direct to the individual or group concerned where an address is provided. The AGSA office may be contacted for the information to be passed on in the case of anonymous requests. Privacy and anonymity will be ensured if requested.

While AGSA aims to facilitate contacts between families it is unable to assess the suitability of these in individual cases.

It should be remembered that a shared genetic condition does not mean an equally shared value system between families. Different degrees of acceptance and different mechanisms for coping will be encountered and a non-judgmental approach is recommended in establishing contact.

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CHROMOSOME 18

A request has been made to set up a Chromosome 18 Support Group similar to the USA model so if you know of anyone who would be interested please contact the AGSA office.

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NON KETOTIC HYPERGLYCINAEMIA

Joanne would like to speak to another family who have had experience with this condition.
Please contact her at:-
Joanne Hunt
45 Lake Drive
Narrandera 2700
Phone: 069 591818

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OPITZ TRIGONOCEPHALY

A mother of a seven month old baby would like contact with another family. Contact the AGSA office.

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O C U L O D E N T O D I G I T A L SYNDROME

A family with a daughter with this condition would like contact with another family. Please phone the AGSA office for details.

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MAPLE SYRUP URINE DISEASE - INTERMITTENT FORM

A lady in Queensland has two daughters with this condition and she is seeking contact with another family. Please contact AGSA for further details.

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V.A.T.E.R ASSOCIATION

A family would like contact with another family. Please contact the AGSA office for details.

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DOUBLE Y SYNDROME (XYY Syndrome)

A family is seeking contact with another family. Contact AGSA.

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ERTHROPOIETIC PROTOPORPHYRIA PROPHYRIA

Gillian Shannon, Genetic Counsellor Bathurst, would like contact for a mother and two sons with this condition. Telephone 063-315533

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PELIZES-MEZBACHER DISEASE

Kerry would like contact with another family.
Please contact:
Kerry Prosser
37B Glencairn Way
Parkwood WA 6147

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RENDU OSLER WEBER SYNDROME OR HEREDITARY HEMORRHAGIC TELANGIECTASIA

Lorinda would like contact with another family. For contact write or ring:

Lorinda Beveridge
P.O. Box 18
Nowa Nowa 3887
Ph: 051-557 307

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SUPPORT GROUPS

KLINEFELTER SYNDROME

Robyn in Kingaroy Queensland would like to meet up with other families with the view to starting a Queensland Klinefelter Support Group.

Contact:-
Robyn Staniland
6 Duke Street
KINGAROY QLD 4610
Telephone: (071) 62 2823

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THE GLAUCOMA FOUNDATION OF AUSTRALIA

Notice of the next Support Group Meeting for Sydney.
Date: Saturday 26th November 1994
Time: 2.00 - 4.30 pm
Place: Ferguson Hall
St. Stephen's Uniting Church
197 Macquarie Street
SYDNEY

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SUPPORT GROUP UPDATE

Australian Huntington Disease Association (Qld) Inc.

43 Cordelia Street
SOUTH BRISBANE QLD 4101
Tel: (07) 844 5688
Fax: (07) 846 2100

Mailing Address:
P.O. Box 3152
SOUTH BRISBANE QLD 4101

PROFILE A - Z GENETIC CONDITIONS

It is the intention of AGSA to profile, in each issue, a particular Support Group/Disorder, thus increasing awareness within our membership of the range of genetic conditions. Also it hopes that where overlaps occur in conditions, Support Groups may liaise with each other and thus gain a broader understanding of facilities, aids, etc. that may be of value to your individual membership.

Please ensure that all support group information is recent and reliable. It is of paramount importance that you let us know your group is 'Alive and Well' and happy to take referrals.

For your information we profile.....

KLINEFELTER SYNDROME

kindly supplied by NSW Genetic Education Program October 1994

Synonyms:
Chromosome XXY
Chromosome 46,XY/47,XXY (Mosiac)
Chromosome 48 XXXY

Klinefelter syndrome is a chromosomal abnormality which is characterised by the presence of an extra X chromosome in the cells of the body. Males usually have 46 chromosomes including one X chromosome and one Y chromosome which make up the pair of sex chromosomes (XY). The classic form of Klinefelter syndrome has one extra X chromosome in each cell (XXY). The presence of the Y chromosome, regardless of the number of X chromosomes present makes a person a male.

There are variants of the syndrome which are characterised by whether there is more than one extra X chromosome, or whether all the cells of the body are affected by the chromosomal abnormality (mosaicism). In cases where there is more than one extra X chromosome, the symptoms are more severe. The symptoms are milder when not all the cells in the body have the extra X chromosome. The major feature in Klinefelter syndrome is impaired function of the testes, also called **primary hypogonadism.**

The signs and symptoms of Klinefelter syndrome can be very subtle. It is common that Klinefelter syndrome may not be diagnosed until puberty, or until adulthood.

Symptoms

There is a range in the kind and severity of the symptoms that boys and men with Klinefelter syndrome exhibit. The most common symptoms of Klinefelter syndrome may include abnormally small testes, a small penis (although still with the normal range), and the diminished development of secondary sexual characteristics.

Within the testes, sperm is not produced (axzoospermia) as the seminiferous tubules are often hardened (sclerosed). Most males with Klinefelter syndrome are infertile.

Males with Klinefelter tend to be slightly taller than expected, given the heights of their parents and other siblings. There may be less muscular development and there may be a slightly feminine distribution of body fat. Some males with this disorder may have abnormally

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enlarged breasts (gynecomastia).

Although puberty occurs at the usual time, the development of secondary sexual characteristics may be diminished resulting in scant body hair and a high-pitched voice.

Gross motor coordination may be poor. However, males with Klinefelter syndrome usually have motor development which is within normal limits. Language development may be delayed in Klinefelter syndrome.

Males affected by Klinefelter syndromemay tend to be shy, aprehensive, and passive. Coupled with the delayed language development, boys with Klinefelter may experience learning problems in school.

Approximately 80% of males with Klinefelter syndrome have only one **extra X** chromosome. The presence of two or three **extra X** chromosomes is very rare and the symptoms are much more pronounced.

Mosaicism describes the presence of at least two distinct cell types (populations) within the affected individual. In males who have Klinefelter syndrome **mosaicism,** there is one cell population that has one or more extra X chromosomes. Generally, males who are mosaic for Klinefelter syndrome have fewer symptoms.

Causes

Klinefelter syndrome is a chromosomal disorder that occurs for unknown reasons (sporadically). It results from an error that occurs during the division of the sex chromosomes in either the egg or the sperm. In other words, the extra X chromosome (or chromosomes) may be derived from either the mother or the father.

Affected Population

Klinefelter syndrome is a chromosomal disorder that occurs in approximately 1 in 800 newborn males.

Treatment

In Klinefelter syndrome, the testes are not functioning normally therefore the male hormone **testosterone** is not produced. Males with Klinefelter syndrome are started on testosterone therapy at the start of puberty, around 12 years of age. The testosterone promotes the development of secondary male characterisitcs including growth of the testes. **The function of the testes is not restored, however, with testosterone therapy.** The testosterone therapy, either in tablets or injection, enhances muscular development and a more masculine physical appearance (habitus). The use of testosterone therapy has been demonstrated to have an impact on development of self-esteem, to increase sexual libido and potency, and to reduce some behavioural problems in males with Klinefelter syndrome. Generally the hormone replacement therapy continues until late adulthood.

Testosterone therapy for males with Klinefelter syndrome must be managed by a physician who is familiar with the hormone replacement treatment of the syndrome.

Genetic counselling will be of value to individuals with Klinefelter syndrome and their parents and/or spouses. Also supportive counselling is of benefit in conjunction with the hormone replacement therapy.

For more information on Klinefelter syndrome, please contact:-

National Organization for Rare Disorders (NORD)
P.O. Box 8923
New Fairfield, CT 06812-1783 USA
(203) 746-6518
(203) 746-6927 (TDD for the hearing impaired).

Klinefelter's Syndrome Association of America
P.O. Box 93
Pine River, Wl 54965

Klinefelter Syndrome and Associates
P.O. Box 119
Roseville, CA 95661-0119 USA

Klinefelter's Syndrome Support Group of Canada
P.O. Box 5000
Pentanguishene, Ontario, LOK 1PO

For a copy of the brochure "Understanding Klinefelter Syndrome: A Guide for XXY Males and their Families" contact:-

NICHD
P.O. BOX 29111
Washington DC 20040 USA

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FAMILY STORY

OUR SON MARK

Our son Mark is afflicted with Klinefelter Syndrome. In the following few paragraphs, we have tried to provide an insight into Mark's condition and his development for information purposes.

Mark was born on Monday 13th December 1976 at the Nepean District Hospital, near Penrith, New South Wales. He was 6 weeks premature at birth and weighed in at the reasonably healthy 5 pounds 4 ounces. For the first few days, Mark was kept in a humidicrib because of breathing difficulties. He also suffered from a slight case of jaundice and was immediately diagnosed as having an undescended testicle and extreme case of Hypospadias (the opening of the urethra was under the base of the penis).

Between February 1980 and March 1987, Mark underwent a series of operations for repair and reconstruction of his Hypospadias condition and the correction of the undescended testicle. Because Mark's older brother Paul (born 1974) also had a minor genital disorder, in October 1984 we arranged an appointment with Dr J Brown from the Royal Alexandra Hospital for Children (RAHC) at Camperdown to discuss the possible causes for the deformities in our children. He organised a number of blood tests which resulted in Mark being diagnosed as "XXY", ie Klinefelter Syndrome. This was the first time we had ever heard of Klinefelter Syndrome and Dr Brown briefly explained what that meant, ie "the inability to father children". Following his diagnosis, Mark was referred to Professor Silink, Endocrinologist at the RAHC where he visited regularly for chromosome study, X-Rays to check for his bone growth (osseous age) and to keep him informed of his overall development. Professor Silink has always been very helpful in ensuring we were well advised on anything relating to Klinefelter Syndrome.

In July 1983, Mark developed the first stages of pneumonia which was later diagnosed as a mild case of Whooping Cough. This developed into Bronchial

Asthma. These Asthma attacks occurred over a number of years and through treatment they slowly decreased as he entered his teens. His last attack was in September 1991.

Formal medical treatment began in May 1991 when Mark (aged 14 1/2 years) started to go through puberty. He was prescribed testosterone therapy using "Halotestin " tablets twice daily. In September 1993 (aged 16 3/4 years) at the peak of puberty, Mark was prescribed "Andriol" capsules three times a day. Mark's final visit to the RAHC took place in May 1994 (aged 17 1/2 years), when once again his treatment was changed because the tablets were no longer effective. The new treatment was injections of "Sustanon" which he is currently having every 3 weeks for the first 12 months. At the end of this time, he will require the injections every two weeks. The injections are administered intramuscular by our local GP who bulk bills his visits. Because they are oil-based, Mark has indicated that the injections are quite often very painful with soreness around the injection site lasting up to several days.

From about September 1991, Mark began to suffer from severe muscular cramps in the legs. This was during a period when his growth rate was quite rapid. Since 1991 when Mark measured 5 foot 11 inches, he has grown 1 inch each year to his present height of 6 foot 2 inches. This rapid growth caused stretch marks to appear in his pelvic (hips) area, and upper arms. The cramps were frequent and some were quite severe and occurred over a period of about 12 months. Over the past 18 months, the frequency and severity of the cramps has lessened which seems to be in line with the slow down in his growth rate. He was prescribed "Quinate" tablets as a treatment for the cramps although we were unsure as to their effectiveness because the cramps persisted. Massaging the affected muscle helped relieve some of the pain however we found that spraying with "Bosisto's Eucalyptus Spray" seemed to work wonders!

The entire period of Mark's schooling took place in the public education system, all of which were co-educational except for his High School. Mark's first school was Winmalee Primary (1982) and in 1986 he transferred to Berowra Primary School. In 1989, he started at Asquith Boys High School leaving at the end of 1992 in Year 10 (aged 15) after attaining his School Certificate. In all of his school days, Mark was a slow but persistent learner needing extra parental help with his school work. In primary school, Mark had some minor temper outbursts through frustration as he found some of the subjects difficult to understand. Nothing specific was advised to the various schools about Mark's condition, other than to indicate on the High School enrolment form that Mark suffered from 'a hormone imbalance which could cause some behavioural problems.' Mark did not repeat any classes and received passes on all subjects which he sat for in the School Certificate examination.

Between 1988 and 1991, Mark attended Speech and Drama Classes in an attempt to overcome his shyness. He has also worked as a stage hand with a local music society. Mark has shown little interest in most sports activities other than 10 pin bowling which he does regularly in a league at Hornsby Bowl. He is very good artistically and frequently came first in art classes with drawing and pottery in school. He is very interested in motor cars and plans to apply for a Learners Licence within the next few months. His main worry is that because of his size he may not be able to

buy the type of car that he wants!

Just prior to leaving school, Mark commenced working at Coles Supermarket as a casual shop assistant. He has been in the same shop for three years and is currently working part-time 5 days a week. He has applied for a full time position and hopes to undertake management training in the next couple of years. Mark is very happy working at Coles and has done most of the jobs in the store. He is well liked by his co-workers and has been given a number of very responsible jobs.

The following list shows some of the special characteristics of Mark which again, may or may not be related to his condition:-

* Writes right handed but eats left handed.
* Has stretch marks due to rapid growth.
* Insatiable appetite.
* Frequent nose bleeds.
* Crooked toes.
* Shyness around people he does not know.
* Large hands with short, stumpy fingers.
* Tends to be loud in volume when speaking.
* Temper outbursts. Although rare now, they were more frequent whilst he was going through puberty.
* Has very easily hurt feelings (he is very tender hearted).

Mark's temper outbursts were a worry for us for some time, however with constant confrontation of the problem and with reasoning from both of us, he has managed to control and understand his anger.

Physically, Mark is a big boy for his age (17 3/4 years). He has a very broad neck and weighs 87 kilograms. He has a fair complexion, blue eyes and what would be best described as a young looking face. He has light beard growth and only shaves about once a week however he does have a reasonable distribution of hair on his legs and torso. He has a small acne problem which may be related to his medication. Over the last few years he has become very conscious of his personal appearance and is showing considerable interest in girls.

In conclusion, Mark's views on Klinefelter Syndrome is that each person should be treated as a normal, everyday individual and their condition should not be common knowledge to everyone, otherwise there is the danger they will be given a 'tag' and treated as a 'disability'. We have always been a close knit family and have never treated Mark as being 'different' - he has always been a normal child going through 'stages'. We have also abided by Mark's wishes in that only close family members be told of his full medical condition, and as far as school, work and friends are concerned, he is completely normal.

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The Association of Genetic Support of Australasia (AGSA) Inc.

66 Albion Street
SURRY HILLS
NSW 2010

Tel: (02) 211 1462

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Support and Information Officer -
Dianne Petrie
Office Hours: 10.00am - 2.00pm
Monday - Friday

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President
Ros Smith
may be contacted on:
Tel: (047) 51 5872

Regional Contact
Judy Rands
10 Roosevelt Avenue
WAGGA WAGGA
NSW 2650
Tel: (069) 26 1560

ANNUAL SUBSCRIPTION

Individual	$15.00
Group/Organisation	$30.00

Subscription Year
1st October - 30th September

AGSA aims to:

* provide a contact point for families who are affected by genetic conditions so rare that they do not have their own support group

* facilitate access to individual support groups for those families with a particular genetic disorder

* provide a forum for the exchange of information between support groups regarding available community services

* educate the medical and allied health professionals and the community about genetic disorders

* lobby government bodies, both Federal and State, for appropriate funding for genetic services

The views expressed in this Newsletter are not necessarily those of AGSA.