THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.

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N E W S L E T T E R

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MISSION STATEMENT

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

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Contents

EDITORIAL

Happy Easter! I hope you had a pleasant break with friends and family.

It is Easter Monday as I type this column sitting in a house in the country overlooking the maple tree with its gold and red autumn leaves. Autumn brings not only cooler weather but also a period of great celebration for AGSA. I am very happy to announce The Hon Minister for Health, Craig Knowles has signed off approval for AGSA's Enhancement Funding effective immediately and recurrent. There are many people who have lobbied their local MP, written letters and made phone calls on AGSA's behalf and I would like to thank you very much for your effort and continued support of AGSA. AGSA has a strong following of people who believe AGSA's work is invaluable and it is through the support of these people and successive AGSA committees that AGSA has continued to survive and grow. This year AGSA has received donations from members, many of whom who wish to remain anonymous, assistance with the website, assistance with fundraising and assistance with public relations. TKT USA has once again donated funding towards AGSA's Genetic Awareness Week. This is a very exciting time for AGSA and I hope you will be able to join us in celebrating this Genetic Awareness Week in June.

Best wishes

Dianne Petrie

Advocacy Discussion Forum Phone in

Introduction

The Disability Council of NSW, as the Official Adviser to the New South Wales Government, has been requested to convene consultation sessions to discuss Systemic Advocacy and how it is, or could be, linked to Individual Advocacy throughout New South Wales. The discussions are part of the Minister for Disability Services' direction for an increase in the level of Individual Advocacy, particularly for people in rural and regional areas as well as from culturally diverse backgrounds.

Purpose of the Consultation

To obtain your views on:

- What the focus of advocacy services should be?
- How to ensure the effectiveness of advocacy in NSW?
- Determining the linkages between individual and Systemic advocacy functions?
- How to ensure the equitable provision of advocacy across NSW?

If you are unable to attend these sessions, we invite you to use our free phone-in line to have your say.

TTY/Phone 1800 -044 - 848

If you find it more convenient please feel free to fax, email or post your views to us, before May 11 We value your feed-back.

Fax 9211 2271

Email info@discoun.nsw.gov.au

Address: Level 21, 323 Castlereagh St., Sydney NSW 2000

CONTACT CORNER

AGSA will publish requests for contacts 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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LIPOMYELOMENINGOCELE
SPINA BIFIDA

A mother of a 19-month-old boy would like contact with others. Contact AGSA for details.

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STEVEN JOHNSON SYNDROME

A 33-year-old man would like contact with anyone who has had experience with this condition. Contact AGSA for details.

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FRIEDREICHS ATAXIA

Contact is wanted for a 10-year boy living in Queensland.

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Support Group News
as at 19th April 2001

Unique
Website address is: www.rarechromo.org

Australian Primary Immunodeficiency Association
A.P.I.A are requesting newsletter contributions from parents and professionals with information or experience with these issues – overseas travel with an immune deficiency, subcutaneous infusions, portacaths, related health issues.
Please call (02) 4942-1841.

Disability and Aged Information Service Inc
DAISI which is a resource and information service have opened a new office at 109 Molesworth St, Lismore on 7th February 2001. They can be contacted on 1800 800 340, www.daisi.asn.au or info@daisi.asn.au. They also provide various fact sheets, which include Rural Friendly Telephone Numbers, Complaints, Advocacy and Finance.

Commonwealth Carelink Centres have been established across Australia to provide a single point of contact to access information on agencies providing community care services within their region. They can be contacted on 02 6289-5577 or visit http://www.health.gov.au/acc

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Self Help, Queensland
Costello Syndrome - Australian contact for this syndrome can be contacted on (02) 6653 2428.

Aussiemito: - United Mitochondrial Disease Foundation can be contacted on (07) 3299 2607

FPA Health
Formerly known as Family Planning NSW is holding training workshops in May and June on intellectual disability, HIV/AIDS and sexual health. Topics include, Triple Taboo: HIV, Sexuality and Intellectual Disability, Policy Development in HIV and Intellectual Disability, Translating Duty of Care into Workplace Practice and Design and Deliver Training Programs. Contact them by phone (02) 8752 4335 or email stephaniek@fpahealth.org.au or more details.

Centrelink – Disability & Carer Connections
Dial '106' for Text Emergency Calls.
Australians who are deaf, or have a speech or hearing impairment, can now ring Police, Fire or Ambulance via the toll-free Text Emergency Call Service '106'. This is the world's first national text emergency call service, provided by the Australian Communication Exchange as part of the National Relay Service. The service can be accessed via TTY or computer with modem. The '106' text emergency call service is available 24 hours, 7 days a week and is equivalent to the '000'emergency call service.

Neurofibromatosis
May will their Awareness month. They are presently putting together a Teacher's Information Kit to address the needs of children with NF in the classroom.

Alliance
This USA based umbrella genetic support group has updated its website.
www.geneticalliance.org

Australian Leukodystrophy Support Group
The support group is raising funds for it's inaugural conference this year. Graham Cadd, who has a son who is affected by this neurological disorder, is cycling from Perth to Sydney in May 2001 and hopes to complete it in 10 days. He his looking for sponsors to help raise the funds needed for the conference. For details please contact the group at:
10 Mitchell St, Mentone, Vic 3194, Fax: (03) 9583 4379 or email: leuko@vicnet.net.au

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Geriaction Incorporated (NSW)
Video available with a discussion guide on "The Heart Had No Wrinkles" a video about ageing

and sexuality. Cost $66.00 plus p & h. In this newsletter there are abstracts on "The care giving burden of relatives with dementia - experiences of Chinese-Australian families", "Grief and loss experienced by patients with Alzheimer's disease and their caregivers", "Resident abuse – an unacceptable reality". For more information on this organization, contact Freecall 1800 308 282

Contact-a Family (UK)
Contacts wanted for families affected with – Infantile Fibromatosis Tumor and Cowden Syndrome. New support groups established – Pendred Syndrome, Aarskog Syndrome and Necrotising Fasciiltis.
Email: info@cafamily.org.uk .
CaF's new address is:_
209-211 City Road, London EC1V 1JN.
Phone (020) 7608 8700

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 Lowe syndrome. The following fact sheet is reprinted with permission from the NSW Genetics Education Program.*

LOWE SYNDROME

Also known as:
- Cerebro-Oculorenal Dystrophy
- Lowe's Disease
- Lowe-Bickel Syndrome
- Lowe-Terry-MacLachlan Syndrome
- LS
- OCRL
- Oculocerebrorenal Dystrophy
- Oculocerebrorenal Syndrome
- Renal-Oculocerebrodystrophy

General information about Lowe syndrome:
Lowe Syndrome, also known as oculo-cerebro-renal syndrome, is a rare inherited *(genetic*) metabolic disease that affects males.

The symptoms of this disorder are:
- lack of muscle tone *(hypotonia*);
- multiple abnormalities of the eyes and bones;
- the presence at birth of clouding of the lenses of the eyes *(cataracts*);
- mental retardation;
- short stature;
- kidney problems

Other symptoms may include:
- protrusion of the eyeball from the eye socket *(enophthalmos*);
- failure to gain weight and grow at the expected rate;
- weak or absent deep tendon reflexes;
- multiple kidney problems (e.g. renal tubular dysfunction, renal hyperaminoaciduria, etc.).

Lowe Syndrome follows a pattern of inheritance in families known as X-linked genetic inheritance and symptoms develop due to lack of the enzyme phosphatidylinositol 4,5-biphosphate 5phosphatase.

Symptoms of Lowe syndrome in more detail:
In males, the symptoms and physical features of Lowe Syndrome typically appear during early infancy. Symptoms may include a rare form of congenital glaucoma known as hydrophthalmos. Excessive pressure in the eyes results from the obstruction of fluid flow around the eyes. All infants have clouding on the lens of the eyes *(cataracts*).
Physical characteristics may include:
- lack of muscle tone *(hypotonia*);
- the absence of normal muscle reflexes *(areflexia*);
- excessive mobility of the joints *(hypermobility*);
- abnormally soft bones *(rickets*);
- underdeveloped testes;
- mental retardation and behavioural problems.

The average IQ of affected boys is in the moderately intellectually disabled range, although about one quarter of the boys have IQs in the normal range. About 80% of affected boys have difficult behaviours including stubbornness, temper tantrums, irritability and obsessions.

Female carriers of the Lowe syndrome gene sometimes have opacities or cloudy regions in the lens of the eye, even though they do not have the disorder. These opacities are small, irregularly-shaped and off-white. Examination of the eyes of suspected carrier women using a special instrument called a slit lamp, is a very accurate means of detecting those women who are carriers for Lowe syndrome. Some children with Lowe Syndrome may also have abnormalities in kidney function *(renal tubular acidosis*) that results in the excessive loss of water from the body *(dehydratio*n), fatigue, and kidney damage.

What causes Lowe syndrome?: Lowe syndrome follows a pattern of inheritance in families called X-linked genetic inheritance.

The gene that is responsible for Lowe Syndrome is located on the long arm (q) of the X chromosome in the region called Xq26.1. The Lowe's Syndrome gene controls production of a specific enzyme, phosphatidylinositol 4,5-biphosphate 5 phosphatase and symptoms may develop due to a lack of this enzyme. Thisdiscovery has opened the door to further investigations into the metabolic defect and how it relates to the various medical problems associated with this disorder.

Who is affected by Lowe syndrome?: Lowe's Syndrome is a very rare metabolic disorder which affects only males. However, females are carriers of the gene.

Is there any treatment for Lowe syndrome?

Diagnosis

Laboratory testing may detect the abnormal presence of amino acids *(aminoaciduri*a), phosphate *(phosphaturi*a), potassium, and carnitine in the urine as well as decreased production of ammonia in children with Lowe Syndrome. A definitive test may be made by testing a small skin sample for the presence or absence of the enzyme associated with Lowe syndrome. Microscopic studies of tissue samples may show abnormalities in the kidneys, testes, eyes, and/or brain. A specialised eye examination may determine if a female is a carrier for the disease.

Prenatal testing is also available to determine if a developing fetus has Lowe Syndrome. Cells called amniocytes from the amniotic fluid which surrounds the baby in the womb can be tested for levels of the enzyme phosphatidylinositol 4,5-biphosphate 5 phosphatase.

Treatment

Treatment of Lowe Syndrome consists of appropriate medications necessary to reduce symptoms. Other treatments may attempt to reduce behavioural problems. Kidney problems related to low phosphorus are treated with oral replacement of phosphorus alone, or phosphorus in combination with Vitamin D to prevent rickets. Sodium bicarbonate or other alkaline solutions are often prescribed to correct the abnormal accumulation of acid in the blood and urine *(renal acidosi*s).

Surgery or drugs may be considered for treatment of eye problems such as cataracts and glaucoma associated with Lowe's Syndrome. Infants with congenital cataracts may have surgery to remove the impaired lens of the eyes. Males with this disorder may need to wear eyeglasses or contact lenses.

We would like to thank the Lowe Syndrome Association in Indiana, USA, for granting AGSA permission to reprint the following from their website http://www.lowesyndrome.org

Frequently Asked Questions

What is Lowe syndrome?

Lowe syndrome (LS) is a rare genetic condition affecting males that causes physical and mental handicaps and medical problems. Also called the oculo-cerebro-renal syndrome of Lowe (OCRL), it was first described in 1952 by Dr. Charles Lowe and colleagues.

What causes Lowe syndrome?

Lowe syndrome is caused by a defective gene that results in the deficiency of an enzyme called phosphatidylinositol 4,5-biphosphate 5 phosphatase. This enzyme is essential to normal metabolic processes that take place in a small part of the cell called the Golgi apparatus. Because of the enzyme deficiency, cell functions that are regulated by the Golgi are abnormal, leading to various developmental defects including cataracts and problems in the brain and kidneys. How the enzyme deficiency leads to these defects is not yet completely understood.

Why can't the missing enzyme just be replaced?

Scientists must first better understand the subtle imbalance caused by the biochemical defect. It is possible that over correction could be just as harmful as the original lack of the enzyme. In addition, there is currently no method available to target therapies to the Golgi apparatus, the small sub-compartment of the cell where the LS enzyme is located.

How is LS inherited?

The LS gene is located on the X-chromosome. Only males can actually have the condition. Females who have the LS gene are carriers. In some cases, LS is the result of an original mutation and the mother is not a carrier.

Can LS be prevented?

In families in which a case of LS has occurred, a slit-lamp eye examination can help determine carrier status of at-risk females. Research currently underway may lead to a more definitive genetic test for carrier status. Various family planning options are available, including prenatal testing. Families should consult with a geneticist to learn more about their options.

Where are diagnostic tests done?

To diagnose LS, a small skin sample is taken and sent to the Biochemical Genetics Laboratory at Baylor College of Medicine in Houston, Texas. Prenatal diagnosis is also provided at this lab. Physicians may make arrangements for these tests by calling 1-800-246-2436 or 713-798-4982 or through e-mail at: bioc@bcm.tmc.edu.

What are the common features of LS?

Cataracts in both eyes, found at birth or shortly after

Glaucoma (in about half the cases)

Poor muscle tone and delayed motor development

Mental retardation, ranging from borderline to severe (in a few cases intelligence may be normal)

Seizures (in about half the cases)

Significant behaviour problems (in many, but not all, cases)

Kidney involvement ("leaky" kidneys, or renal tubular acidosis)

Short stature

Tendency to develop rickets, bone fractures, scoliosis, and joint problems

Maximum life span of about 35-40 years due to progressive kidney failure, although deaths have occurred at earlier ages due both to renal failure and to other causes. Life expectancy may increase as knowledge increases and new treatments are developed.

How is LS treated?

There is no cure, but many of the symptoms can be treated effectively through medication, surgery, physical and occupational therapies, and special education.

What about research?

In 1992, the gene that causes LS was found. In 1995 researchers discovered that the gene defect causes an enzyme deficiency. Researchers continue to investigate the function of the gene and the complicated biochemistry and cellular mechanisms of LS. Other areas of research in recent years include behaviour problems and clinical care.

What are boys with LS like?

Generally, they are affectionate and sociable, love music, and have a great sense of humour.

IV. Genetics

Lowe syndrome is a genetic disease, which means that the condition is always caused by an altered gene. In many families,

Lowe syndrome is passed from a carrier mother (which means she carries the altered gene) to her affected son. In that situation, female relatives of the mother may also have the altered gene and may be at risk of having boys with Lowe syndrome. In other situations, the mother of the affected boy is not a carrier of the altered gene. In that case, the mother and her female relatives are not at risk of having more children with Lowe syndrome.

Families in which Lowe syndrome occurs should understand whether the condition is inherited in their family, the likelihood of reoccurrence of the disease in subsequent children of the same couple, who else in the family could be a carrier and how to determine this, and what resources are available to clarify those risks and to assist with decisions about personal family planning.

A. Understanding how Lowe syndrome is inherited

*1. **Genes.** Lowe syndrome is due to an "inborn error of metabolism." Metabolism refers to the complex physical and chemical processes involved in the*

maintenance of life and the function of every cell in our bodies. All these processes are controlled by genes, the basic units of life that we inherit from our parents. When a mutation (an alteration or a change) occurs in a gene, an imbalance or "error" in metabolism may result, causing a disease or disability.

Most gene mutations occur spontaneously, usually for unknown reasons. They are not caused by being exposed to drugs or alcohol or emotional events during pregnancy. Once a mutation occurs, however, it becomes "fixed" in a person's genetic material. If that individual survives and grows up, he or she then has the potential to pass or transmit the altered gene to subsequent generations. Thus the disorder becomes hereditary in that family.

2. ***Chromosomes****. Genes are grouped together in packages called chromosomes. Except for red blood cells and the reproductive cells, every human cell contains 23 pairs of chromosomes, or a total of 46 chromosomes. One member of each pair of these chromosomes comes from the father and the other member of each pair comes from the mother. The reproductive cells, the ovum (egg) and the sperm, each contain 23 chromosomes. At the moment of conception, the sperm and the ovum fuse, thus creating a cell with 23 pairs of chromosomes and all the genetic material for another human being.*

Twenty-two pairs of chromosomes contain genes that determine general body characteristics, like eye colour, stature, and growth. The twenty-third pair is different because it determines the sex of the offspring. This pair includes the "X" and "Y" chromosomes. Females (XX) have two X-chromosomes (one from each parent). Each of the female's ova or eggs will carry either one or the other X-chromosome (but not both). Males (XY) have one X-chromosome (from the mother) and one Y-chromosome (from the father). Therefore, each sperm in the male will carry either an X-chromosome or a Y-chromosome (but not both). If the father passes on a sperm that carries his X-chromosome, the resulting offspring will be female (XX); if he passes on a sperm that carries his Y-chromosome, the offspring will be male (XY).

3. ***X-Linked inheritance****. Since the early 1960's, physicians have known that the gene causing Lowe syndrome is located on the X-chromosome. Diseases caused by genes that are located on the X-chromosome are called "X-linked" and have a distinctive pattern of inheritance. X-linked diseases typically affect only males but are "carried" and passed on by females.*

Women who are Lowe syndrome carriers have one X-chromosome with the normal gene and one X-chromosome with the faulty gene.

Lowe gene.

A male (XY) who happens to inherit his mother's X-chromosome with the Lowe gene will be affected with Lowe syndrome, since that X provides the only genetic information for that gene. (The Y-chromosome contains no equivalent information.)

4. ***Carriers.*** *A female who has a mutated Lowe syndrome gene on one of her X-chromosomes is called a "carrier" because she bears or carries this information, in the form of one copy of the mutated Lowe gene, on one of her two X-chromosomes.*

With each pregnancy, she has a 50% chance (one out of two) of passing on the X-chromosome with the Lowe gene to her child. (Since she has two X-chromosomes, she may pass either one or the other X-chromosome to her offspring, with equal probability.) If the child is a male (because he inherits his father's Y-chromosome), there is a 50% chance that he will inherit the mother's X-chromosome with the mutated Lowe gene, and thus he will have Lowe syndrome. If he inherits his mother's X-chromosome with the normal gene, he will be an unaffected (normal) male. If the child is a female (because she inherits her father's X-chromosome), there is a 50% chance she will inherit the mother's X-chromosome with the mutated Lowe gene and thus be a carrier just like the mother; similarly, there is a 50% chance she will not be a carrier because she will inherit the normal X-chromosome.

In summary*, with each pregnancy a carrier female has one chance in four (a 25% risk) of having a boy with Lowe syndrome; one chance in four of having a normal boy; one chance in four of having a carrier daughter; and one chance in four of having a normal daughter.*

Obviously, these probabilities apply only to women who are genetic carriers. As discussed in the following section, however, in some families the affected male Lowe child is the first individual in the family with the condition. Determining whether his mother is a carrier or not holds tremendous importance in predicting the outcomes of her future pregnancies and those of her female relatives.

5. ***New mutations****. A number of males with Lowe syndrome occur as the first and only affected individuals in their families.*

In such cases, it is possible that the mother is not a carrier; that is, her genetic makeup may not include the altered gene.

Rather, the gene in the single ovum (egg) which developed into the boy may have become altered. An alteration in a previously normal gene is called a mutation. Since the boy is the first person with this altered gene, he is affected as the result of a new genetic event, a "new mutation."

When Lowe syndrome is the result of a new mutation, the mother is not a carrier. Her risk of having another child with Lowe syndrome is no greater than that faced by non-carrier females in the general population. To determine if a boy with Lowe syndrome is the result of a new mutation, the mother must undergo carrier testing (see "Carrier detection" and "Non-carriers" in the following section).

6. ***Mosaicism****. In a few rare families, a child affected with Lowe syndrome may be neither a new mutation nor an inherited mutation coming from a carrier mother. In these cases, the mother may have had a new mutation occur during her own prenatal development. In this case, the mutation may be present in a minority of the cells, including those from which some of her eggs are derived. The cells of her body, other than the cells in the ovary, do not all carry the mutation and so she may not have any signs on her eye examination of being a carrier. Yet, more than one egg will carry the mutation, so she will be at increased risk for having a son with Lowe syndrome, or a daughter who is a carrier. This situation, in which a woman has some cells carrying a mutation and other cells that do not, is called "mosaicism".. More than one egg, but fewer than half, will carry a Lowe syndrome mutation. Mosaicism has been documented in Lowe syndrome.*

B. Genetic Counselling

Couples who are at-risk for having an affected child with Lowe syndrome should explore their family planning options with the guidance of their physician and/or a genetic counsellor. Geneticists and genetic counsellors can help determine the chances of having an affected child. Trying to determine whether or not a woman is a carrier is very important because it has substantial impact on the risks for having other affected sons (or grandsons). Her carrier status may also be relevant to the status of her female relatives.

1. Carrier detection. *Families at-risk for having a boy with Lowe syndrome may wish to investigate their chances of having an affected child. Women who are at-risk include the mothers and sisters of affected boys, as well as the boy's maternal aunts and their daughters. Thus, determining whether or not a woman is a carrier may also be critically important to extended family members who may wish to plan their own families. Geneticists and genetic counsellors can advise couples about the reproductive risks.*

In some cases, a woman's carrier status can be determined from her family history. For instance, the mother of a son with Lowe syndrome is presumed to be a genetic carrier if there has been a previous case of Lowe syndrome in her family. If there were no other known cases of Lowe syndrome in the family, however, the mother of an affected boy might not be a carrier, since her son's condition could be the result of a new gene mutation or mosaicism. However, even in the absence of any history of Lowe syndrome in a family, one cannot assume that the mother is not a carrier. The carrier status of the mother must be determined by examination.

In most cases, especially in women after puberty, carrier status can be determined by an eye examination performed by an ophthalmologist (a physician specialising in diseases of the eye). The examination must be done with the pupils dilated (with eye drops). Approximately 95% of all carriers of the gene for Lowe syndrome have subtle changes in the lenses of their eyes, especially in the teenage and adult years. These changes, which appear as tiny dots and flecks in the lens in a characteristic distribution and pattern, typically cause no effect on vision and, if not looked for diligently, may be mistakenly dismissed as normal variations. Therefore, the geneticist should insist that the examination only be made by an ophthalmologist with substantial experience with the subtle variations of the lens opacities of Lowe syndrome carriers.

If a reliable examination detects these characteristic lens opacities in an at-risk female of any age, then she is a carrier. If the opacities are not present in an adult female, she is probably not a carrier. This conclusion cannot be made with absolute certainty, however, especially if she is less than 15 years old.

There is no chemical or laboratory test to determine carrier status for all at-risk females at this time. However, genetic analysis may be possible for some family members (see Research).

2. ***Non-carriers.*** *When a careful eye examination of the mother of a child with Lowe syndrome is normal (that is, the characteristic lens opacities are not present), there are three possible explanations for the Lowe syndrome in her child. First, and most*

commonly, the child is the result of a new genetic mutation in the egg that developed into him. In this case, the mother does not carry any other mutated Lowe syndrome genes in the rest of her eggs. She is not a carrier and she is no more likely to have another child with Lowe syndrome than any other non-carrier female in the general population. Second, and more rarely, is that she really is a carrier but falls into the group of 5% of all carriers who fail to show significant changes in their lenses. Her probability of having a child with Lowe syndrome is 25% with each pregnancy (50% for each male pregnancy).

Third, and most rarely, the mother may be mosaic for the mutation in her ovaries. In this case, she may have additional eggs carrying the Lowe syndrome mutation without showing signs in the lenses of her eyes. Her risk of having another child with Lowe syndrome or a daughter who is a carrier would be much greater than that of the general population, but not the full risk that a true carrier female has (see Carriers).

Unfortunately, there is no reliable biochemical or molecular test to diagnose mosaicism. For that reason, even women with normal eye exams can be offered prenatal testing because of the risk of unsuspected mosaicism. However, whether the mother of a child with Lowe syndrome is a non-carrier or represents a mosaic situation, her sisters or other female relatives (except for her daughters) are not at risk for being carriers because, in either case, the mutation occurred as a new genetic event in the mother herself.

*3. **Family planning options**. At-risk couples have a wide range of options available to them. Some may choose to "take their chances" with a pregnancy, while others may consider prenatal testing. Some couples may be interested in adoption or in techniques that would use a donor egg from a non-carrier female. Others may be interested in methods to increase their chances of conceiving a female since females, even if carriers, do not develop the disease.*

If prenatal testing is an option, testing to determine if the fetus is male or female is a well-established procedure and is widely available. If the fetus is male, prenatal enzyme analysis identical to the test used to diagnose Lowe syndrome in patients is available. Enzyme analysis can be used for prenatal diagnosis even if carrier status has not been established firmly. It can also be used whether or not gene analysis has succeeded in identifying the family's specific gene alteration.

Prenatal testing can be accomplished by one of two methods: CVS (chorionic villus sampling), which is done at 11-13weeks, or amniocentesis, which is done at 15 weeks onwards. Although the procedure itself can be performed in the local community, at the present time the analysis for the OCRL enzyme test can only be carried out at the Biochemical Genetics

Laboratory in the Department of Molecular and Human Genetics at Baylor College of Medicine in Houston, Texas. To make arrangements for the test, physicians may call 1-800-246-2436 or 713-798-4982 or e-mail: bioc@bcm.tmc.edu. As with any prenatal diagnosis, the testing should be planned well in advance of the pregnancy.

An experimental technique called preimplantation genetic diagnosis has been suggested as another option for some couples.

This technique involves in vitro fertilization and then testing the DNA in one cell of the fertilized eggs for the presence of a disease-causing gene alteration. Any embryo that is found to lack the disease-causing alteration (and by inference is normal) could be implanted into the womb. Preimplantation genetic diagnosis has been used successfully in a few genetic disorders but\ has not been tested rigorously in Lowe syndrome and therefore remains only a theoretical possibility. Preimplantation genetic diagnosis is a DNA test that would require accurate knowledge of the family's specific gene alteration. It cannot be used for the enzyme diagnosis for Lowe syndrome because the amount of material available for study is too small.

Many factors will affect a couple's family planning decisions, including their personal, family, ethical, and religious views, as well as financial, educational, and geographical considerations. Ultimately, the right decision for any individual or family is the one with which they are comfortable now and will remain comfortable with as they look back from the future.

Resources:

Lowe Syndrome Association Inc
222 Lincoln Street
West Lafayette, IN 47906 USA
(765) 743-3634
e-mail: info@lowesyndrome.org
Home Page: http://www.lowesyndrome.org
Lowe's Syndrome Association
29 Gleneagles Drive
Penwortham Preston
Lancashire PR1 0JT UK
Tel: 0772 745070

FAMILY STORY

The following story is reprinted from their Winter/Spring 2001 Newsletter. Vol 20, No 1. with the author's permission.

This is our first time writing for *On the Beam* We are very proud to introduce you to our son, Joel Ashley Gaut, born on January 24, 2000. Joel also has a 7 year-old sister, Breanna.

Our roller coaster ride started well before Joel's arrival. A 12week ultrasound showed an enlarged measurement of the nuchal translucency in the neck. At 13 weeks the ultrasound and CVS showed no abnormalities. At 19 weeks the ultrasound showed normal functions of the brain, heart and kidney. The fourth ultrasound at 27 weeks showed a build-up of polyhydramnios, but we were told this can also be normal. At 35 weeks an ultrasound showed hydrocephalus and a breech position. The sixth ultrasound at 36 weeks, we were faced with making a choice between a caesarean or natural birth. After all these ultrasounds and numerous blood tests, we were told our son was fine! When we asked about the original enlargement of the nuchal translucency, we were told "it's nothing to worry about. We often learn from this tract that a child develops asthma or eczema". And that's how they explained the enlargement. The only time I saw Joel's doctor again was 3 days after Joel was delivered by caesarean section. I bumped into him in the corridor. The only thing he said was "terrible news!" I knew then we were on our own. The only way Joel and our family were going to have quality of life was to accept the roller coaster ticket and get on with living.

Joel was born in Tweed Heads NSW, Australia. A week later, he was transferred to the Mater Hospital Brisbane for diagnosis. After numerous tests on Joel and an eye examination done on myself, it was confirmed that I was a carrier of the LS gene. We packed up our belongings and our business and moved to Melbourne to be with our family and to be closer to a hospital. When we left Brisbane we were told just to love him and hold him for the little time he would be with us. They did not hold much hope for Joel's future. They told us that he would not develop beyond 2 weeks of age and would not live beyond 3 months.

Joel is now 1 year old and we now see a much brighter future for him. At birth, Joel weighed 2880 grams (6.7 lb). Now he weighs 8500 gms (19 lbs) and he is 69.3 cm (27.2 inches) long. He's come a long way in 12 months. He is always happy and extremely placid. Joel can feed himself with his bottle, clap hands and is trying very hard to talk, although he communicates very well with us in other ways. Joel loves food and will eat anything dished up to him. He's never been sick and has just cut his 6th tooth in 4 weeks with no problems, just a blackened gum. His cataracts were removed when he was 6 months old. He developed glaucoma in one eye after his first operation. The glaucoma measures were in the high 50's. This condition reversed itself once we learned he had an allergic reaction to the steroid eyes drops. Joel wears 14+oo Aphakic glasses and the pressure in both eyes is around 11. We feel blessed with this result alone.

An ultrasound of Joel's kidneys showed nephrocalcinosis . He is currently being treated with calcium once a week, sodium bicarbonate 8.4/cc solutions 3 ml twice daily and iron 5 ml daily.

As for his brain, it doesn't look good on MRI, but who knows? He certainly has developed beyond 2 weeks of age and is showing signs of being able to progress. Slowly but surely Joel is getting there. We are extremely proud parents and are in no hurry. Joel is here for all the right reasons. He puts light in our lives and smiles on our faces. What more could we ask for?

We would love to receive letters from anyone who would like to write. We believe only special families receive special children. This belief helps us enjoy each precious day!

Susanne Gaut.

If you wish to write to Sue please contact AGSA for address details.

CONFERENCES

as at 19th April 2001

Unique - 9th Annual Conference 5th May 2001 will be held at Hanover International Hotel in Daventry, UK.

FRAGILE X SYNDROME

The Good, The Bad & Some Answers

A conference featuring Dr Marcia Braden
Monday 14th May 2001
Venue: Northern Sydney Education Centre
Wicks Road, North Ryde NSW (in the grounds of Macquarie Hospital) Ph (02) 9887 5601

Isodicentric 15 - First International Conference on Isodicentric 15, also known as Inv Dup or Marker 15,
will be held in Philadelphia, Pennsylvania, USA from 14th – 16th June 2001.
For more information contact the IDEAS group at www.idic15.org or write to Donna Bennet, 416 Big Mount Road, Thomasville, PA17364, USA or email ideas@craftech.com

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Australian Leukodystrophy Support Group – International conference will be held at the Royal Children's Hospital, Victoria, 17th and 18th August 2001.

Costello Syndrome Conference – Toronto Canada, August 2001.
Contact Cath & Colin Stone, 90 Parkfield Rd, New Mosten. Manchester. M40.3RQ, England, email: C.A.Stone@mmu.ac.uk

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Professional Development Course – "Managing Challenging Behaviours" – various locations around Sydney from 29th May to 1st June 2001.
Contact (08) 8288 7511 for details.

NCOSS Conference – "Building NGO's – Building Communities" conference will be held 31st May 2001 at the Furama Central Hotel, 28 Albion St, Surry Hills, Sydney. For more details contact (02) 9211 2599, info@ncoss.org.au, www.ncoss.org.au/conferences

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TURNER SYNDROME NATIONAL CONFERENCE – PERTH

5th-7th October 2001

Venue: C.W.A., 1174 Hay Street, West Perth

For further details contact
Jill Topzer (08) 9457 0337

Consensus Conference on the Diagnosis and Management of Osteoporosis in Osteogenesis Imperfecta
1st National Scientific Conference on Osteogenesis Imperfecta
Friday 27th July 2001 9 am – 5 pm
Centenary Lecture Theatre
Royal North Shore Hospital, Sydney, NSW

Focus on: *Evaluation of the Child with O.I.*
Evaluation of Bone Density in Children and Adults
Treatment of O.I. & Related Disorders

To celebrate The International decade of Bone & Joints.

7th National O.I. Carers & Adults Conference
Friday (evening) 27th – Sunday 28th July 2001
The Collaroy Centre, Homestead Ave, Collaroy, Sydney NSW

Topics: Pamidronate Treatment, support the early years, schooling, aspects for teenagers, special problems for adult "parenting skills". Medical up-dates; Rehabilitations, Allied Health & Nursing
For further Information or to Register please Contact:
Lynne Foxall (02) 9869 1486,
PO Box 4001 Epping NSW 2121
Email: petlyn@hotkey.net.au

ConnecTed Tissue Dysplasia Clinic

The Splash Swimathon
Saturday 16th June 2001
At the Sydney Aquatic Centre, Homebush
For information please ring Lynne Foxall (02) 9869 1486 if you wish to sponsor a swimmer or join in on the day.

ConnecTed Dinner /Dance

Epping RSL on

Friday 19th October 2001 at 7.30 pm for 8.00 pm
For information Contact:
Ros Smith (02) 47515872
Lynne Foxall (02) 9869 1486

The 8th National Conference of the Association for the Welfare of Child Health (AWCH)
11-12 October 2001

Powerhouse Museum, Sydney

Children on the Margin

The ideal forum for networking with parents, health professionals and other community members interested in psychosocial issues in child and adolescent health care.
For further information Please contact
Ms Emma Waygood
Conference Secretariat
Conference action Pty Ltd
(02) 9956 8333

The Australasian Tuberous Sclerosis Society Inc invites families affected by Tuberous Sclerosis Complex and interested professionals to attend.

The Fifth Annual Family Weekend Conference
On 11th and 12th August 2001

At The Royal Institute for Deaf & Blind Children, North Rocks Road, North Rocks. Our main Guest speakers will be Dr Debbie Yates who will speak about Lymphangioleiomyomatosis (LAM) and Lung Involvement in Tuberous Sclerosis. A variety of workshop topics on kidneys, genetics and neurology, teeth, eyes and skin will be offered, covering some of the many aspects of TSC. Phone Sue Pinkerton on (02) 9630 3147 (evenings) for further information.

Have a cuppa for Cancer Research Be a host Australia's biggest morning tea

Thursday 24th May 2001.