AGSA
THE ASSOCIATION OF GENETIC SUPPOR T OF AUSTRALASIA INC.

NEWSLETTER

December 2007 ISSUE 82 ISSN1033-8624 FUNDED BY NSW HEALTH

CONTENTS

MISSION STATEMENT

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

EDITORIAL

Dear All,
It is the end of the year and a time to reflect on all the activities undertaken, and it struck me that not everyone may be aware what AGSA does. So, here is a quick overview. AGSA provides support at the end of a phone line, followed up with information and where possible, contact with others with the same or similar condition. One call to AGSA can generate up to 6 hours work. AGSA provides ten free face to face counselling sessions, three telegroup counselling sessions, three rural seminars and two sibling workshops per year. This year the regional seminars were in Ballina and Orange. Other activities were: - a National Conference on Williams syndrome, Klinefelter & Double Y syndrome seminar and two free weekend carer camps at Yarrabin, Bathurst, for a parent and sibling without the genetic condition.

AGSA has established a Rare Treasures support group representing over 200 chromosomal abnormalities and this group has its own bi-annual newsletter. (See report Swansea Seminar with Chromosome 18 Registry and Research Society Australia).

For the last 7 years, AGSA has held an annual Breast Cancer Information Day for the carriers of BRC1/2 and unknown gene faults. This year it was sponsored by the Cancer Institute NSW.

Each year we hold our Genetic Awareness Week launch kindly sponsored by Genzyme.

AGSA holds a number of coffee mornings and metro seminars on specific genetic conditions.

The AGSA newsletter is now produced quarterly and goes out to a total of 350 people around Australia and overseas.

AGSA liaises with overseas support groups and patient organisations through the International Genetic Alliance of which I am the Acting President, and the World Alliance of Organisations for the prevention of birth defects. This year AGSA presented on "*The Australasian experience and lessons learnt*" at the 3rd International Birth Defects Conference held in Rio. This was an excellent conference and we were able to meet the members of the newly formed Brazilian Genetic Alliance plus many families who have established a support group for Gauchers, Williams syndrome, Fragile X and FOP – to name a few. I was very impressed with what they have achieved in such a short time because it is an enormous job as Brazil is a huge country.

At present, I am catching up on all my call sheets and administration work. Kathy has been busy up dating the website and working on our new database with her husband Rick. It is ready for loading onto the computer. This will make a huge difference to AGSA's administrative work.

I would like to introduce you to Mandy Newton, our new Projects Officer for Filling the Void. Mandy has a Bachelor of Arts Degree and is currently studying Social Work.

Her interests are theatre, singing, music and reading. Mandy's recent work has been with the Creutzfelt Jakob Disease Network where she produced an educational DVD on CJD and organised a fundraising dinner for 300 people raising $25,000.

I am excited to have been chosen as a consumer representative on The Prince of Wales and Sydney/Sydney Eye Hospitals Consumer Advisory Committee (CAC). This is a great honour and I am looking forward to my orientation session next week where I will receive an overview of the Area Health Service.

The AGSA office will be closed from 21st December to 14th January 2008.

I would like to wish you a very **Merry Christmas and a Happy New Year.** I hope you have a safe and enjoyable festive break.

Until next year,
Best wishes
Dianne Petrie

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 many 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.

Hallevorden-Spatz syndrome
A family with a 28 year old son with this condition would like to meet others.
Emery- Dreifuss Muscular Dystrophy
If you know of others with this condition please contact AGSA.

Multiple Epiphyseal Dysplasia
AGSA has a couple of families with this condition who would like contact with others.

SUPPORT GROUP NEWS

Bardet-Biedl Syndrome Support Group Queensland
Email: bbs.australia@yaghoo.com.au or Please phone between 11am- 2.30pm Kathryn Murphy 07 3279 0763.

Kidney Support Network of Queensland is launching a new support group for people affected by kidney disease due to genetic disorders.

KIDNEY Support Network
C/- Kathryn Murphy
PO Box 16
The Gap Qld 4061
Phone: 07 3300 0906

Addison's disease (adrenal insufficiency) is a rare disease in a Western country. Prevalence is estimated between 40 – 110 per million. http://addisons.org.au

Sturge Weber Support Group
aswsg@alphalink.com.au
03 9762 4630

Australian Wolf-Hirschhorn syndrome Support Group Inc (AWHSSG) Inc. Meet monthly for lunch at Armidale Community preschool, Allingham Street, Armidale 2350. First Monday of each month from 12.30 until 2pm. Lunch is provided so RSVP to Anne Chaffey, 6775 2402 or email bectamumbi@bigpond.com

Booklet
Understanding Pompe Disease From diagnosis to Action produced by Genzyme.

Reference: www.athealth.com Excerpt from SAFDA (Vic) June 2007 newsletter.

"most of the literature on parental bereavement still tends to focus on the mother's grief. Often, men are not acknowledged as experiencing grief; or more importantly, men are not taught that it's necessary to grieve and are discouraged from demonstrating signs of grief openly. Bereaved fathers frequently feel that they are the forgotten mourners and are often referred to as "second class grievers". Fathers are expected to be strong for their partners, to be the 'rock" in the family. All too often fathers are considered to be the ones who should attend to the practical but not the emotional aspects surrounding the death; they are expected to be the ones who should not let emotions show or tears fall outwardly, the one who will not and should not fall apart. Men are often asked how their wives are doing, but not asked how they are doing.

Such expectations place an unmanageable burden on men and deprive them of their rightful and urgent need to grieve. This need will surface eventually if it is not expressed. It is not unusual for grieving fathers to feel overwhelmed, ignored , isolated, and abandoned as they try to continue to be caregivers and breadwinners for their families while their hearts are breaking. "Fathers' feelings (often) stay hidden under layers of responsibility and grim determination". Bereaved fathers often say that such strong emotions are very difficult to contain after their child's death. Fathers often fear that they will erupt like volcanoes if they allow themselves to release these feelings and so, too often, fathers try to bury their pain with the baby who died.

It is most important that a father's grief be verbalized and understood by his partner, other family members, professionals, co-workers, friends, and by anyone who will listen. Fathers need to try to free themselves of stereotypes and societal expectations about men and grief; they must be able to tell others that their grief is all they have from their child's brief life. Fathers repeatedly say that for their own peace of mind, they need to move away from this mindset and allow them to grieve, as they are entitled.

LISMORE

Lismore City Council – on-line service for users of it Community Services Directory. Community Services Directory
http://www.lismore.nsw.gov.au/csd
Grand-carer & Youth Initiative 02 6622 8002.
www.daisi.asn.au

Annual Health Assessment

People with an intellectual disability can now have an annual health assessment by their GP covered by Medicare. This is under the new Medicare health assessment items 718 and 719.

The following is reprinted with permission from daisi the DAISI Link August 2007

Major Investment in Disability Support

Additional services and supports announced by the Australian Government

- $23.6 million over 5 years for existing specialist services for children with a disability which both support the developmental needs of children and also provide suitable care so that their families can have a break or work.
- Parent carers who are aged 65 years or more, who have provided care for a significant period of time for their adult son or daughter will have priority access to new supported accommodation services. Over five years, there will be around 1750 new places in 175 new facilities.
- Older parent carers will be able to access additional in-home and centre-based respite services to assist them to continue in their caring role. About 71, 500 carers aged 60 years or more, and their children aged 25 years and older will benefit from this measure through access to additional respite services – around 800 new respite places over five years.
- $115.3 million over five years will provide a new in-home support service for people with a disability who are being cared for by their parent carers aged 65 and over. Services will also be provided to enable a son or daughter

previously cared for by their parent to continue to live in the family home after the parent can no longer provide care.
- New transition support services with funding of $13.8 million over five years will help older carers and their adult son or daughter with a disability, work through their options and plan their future care needs.
- $100 million dollars over five years to ensure additional support is available to people in disability business services.
This will include 500 additional supported employment places in disability business services ($21.6 million); continuing support for people participating in non-vocational activities ($31 million); increased funding to disability business services ($26.5 million); temporary assistance to disability business services facing short-term financial difficulties in rural and regional areas ($21.8 million).
-To address the longer term, the Government will provide $1.5 million to set up an inquiry to explore ways to provide greater choice and flexibility in delivery of supported accommodation, including the potential for support from corporate and philanthropic sources to develop a market for private disability accommodation.

Disability Assistance Inquiry Line:
1800 101 888
http://www.facsia.gov.au/internet/facsinternet/nmsf/disabilities/assistance_package.htm

OVERSEAS NEWS

Joint 7th Human Genome Organization (HUGO)- Pacific Meeting and 8th Asia Pacific Conference on Human Genetics
2 to 5 April 2008
Shangri-la's Mactan Island Resort, Cebu, Philippines
Website: http://www.HUGO-AP2008.ph
Contact name: Carmencita D. Padilla
"Genomics for Better Health in the Asia Pacific" is expected to bring together an outstanding scientific program on the latest clinical trends & advances in the field of genetics and genomics.
Organized by: HUGO and Asia Pacific Society of Human Genetics

40th European Society Human Genetics Conference (ESHG 2008) in Barcelona, Spain, 31 May – 3 June 2008

The XX International Congress of Genetics (ICG) will be held in Berlin from July 12-17, 2008. The International Genetics Federation (IGF) has elected the German Genetics Society (GfG) to organise the biggest and most influential forum in the field of genetics worldwide. After more than 80 years, this prestigious genetics world congress congregates in Germany again. The ICG has taken place once every five years in many major cities around the world. Next year, in the German capital of Berlin, more than 300 of the most renowned international geneticists will report on the latest insights in genomic research. The lectures on the up-to-date status of research in the field of genetics are expected eagerly by the experts.

2008 European Scientific Conference on NF to be held in Killarney, Ireland 30 October - 2 November 2008

A-Z PROFILE OF GENETIC CONDITIONS

This fact sheet is kindly supplied by the Centre for Genetics Education.

EMERY-DREIFUSS MUSCULAR DYSTROPHY
Includes: EDMD
Emery-Dreifuss muscular dystrophy (EDMD) is a rare, often slowly progressive genetic condition affecting the muscles of the arms, legs, face, neck, spine and heart. The condition consists of the clinical triad of weakness and degeneration (*atrophy*) of certain muscles, joints that are fixed in a flexed or extended position (*contractures*), and abnormalities affecting the heart (*cardiomyopathy*). Major symptoms may include muscle wasting and weakness particularly in arms and lower legs

(*humeroperoneal* regions) and contractures of the elbows, Achilles tendons, and upper back muscles. In some cases, additional abnormalities may be present. In most cases, EDMD is inherited as an X-linked recessive or autosomal dominant condition. In extremely rare cases, autosomal recessive inheritance has been reported.

WHAT IS EMERY-DREIFUSS MUSCULAR DYSTROPHY?

Emery-Dreifuss Muscular Dystrophy (EDMD) belongs to a group of rare genetic muscle conditions known as the muscular dystrophies. These conditions are characterised by weakness and atrophy of various voluntary muscles of the body. There are approximately 30 different muscular dystrophies and these may affect different muscles, depending on the type of muscular dystrophy.

The age of onset, severity, and progression of EDMD varies greatly from one individual to the next, even among affected individuals from the same family. Some affected individuals may experience symptoms from childhood with rapid disease progression and severe complications; others may experience adult-onset symptoms and a slowly progressive course.

EDMD is associated with the clinical triad of:

1. Contractures
2. Muscle weakness
3. Heart disease

Contractures

A *contracture* occurs when thickening and shortening of tissue causes deformity and restricts movement of affected areas, especially the joints. The elbows and Achilles tendons are the most common sites for contractures. Contractures are often the first sign in the X-linked form of EDMD and may occur early during childhood. In autosomal dominant EDMD contractures usually develop after the onset of muscle weakness.

Muscle Weakness

Progressive muscle weakness and degeneration (*atrophy*) usually develops during late childhood or early adolescence, and is generally in the upper arms and lower legs (*humero-peroneal* regions). Weakness and atrophy of leg muscles may cause children with EDMD to walk on their toes and may result in an abnormal, waddling *gait* (manner of walking). Muscle weakness affecting the arms may cause various problems such as difficulty in raising the arms above the head.

Eventually, the muscles of the thigh and hips may become involved, making it difficult to climb stairs. The neck, shoulder girdle, and forearms may eventually become involved and the spine may become rigid. As affected individuals grow older, they may experience limited mobility of the neck. Mild weakness of facial muscles is possible and abnormal curvature of spine (*scoliosis*) may also occur.

Muscle weakness and atrophy is usually slowly progressive during the first three decades of life. Eventually, it may become more rapid. Some individuals with the autosomal dominant form of EDMD may eventually lose the ability to walk (*ambulate*) and may require a wheelchair. Loss of ambulation is rare in the X-linked form of EDMD.

Heart abnormalities

Heart abnormalities are the third prominent feature of EDMD, and may result in serious complications. Although onset may vary, heart abnormalities usually develop after the second decade of life. Disease of the heart muscles (*cardiomyopathy*) may develop, potentially resulting in palpitations, fatigue, poor exercise tolerance, and an impaired ability for the heart to pump blood. Some individuals may experience conduction defects resulting in irregular heartbeats (*arrhythmias*) or heart block.

Heart block is characterised by interference with the transfer of the electrical nerve impulses (*conduction*) that regulate the normal, rhythmic, pumping action of the heart muscle. The normal heart has four chambers. The two upper chambers are the atria and the two lower chambers are the ventricles. Within the right atrium of a normal heart is a natural pacemaker that initiates and controls the heartbeat. The electrical stimulus travels from the 'pacemaker' (*sinoatrial* or *SA node*) to the ventricles along a very specific path consisting of conducting tissue and known as the AV (*atrioventricular*) node. As long as the electrical impulse is transmitted normally, the heart behaves normally. If the transmission of the signal is impeded, the blocked transmission is known as a heart block or an AV block.

Heart block may be categorised according to the degree of impairment. The severity of such conduction abnormalities varies among individuals with EDMD. In the mild form of heart block, the two upper chambers of the heart (*atria*) beat normally, but the contractions of the two lower chambers (*ventricles*) lag slightly behind. In the more severe forms, only a half to a quarter of the atrial beats are

conducted to the ventricles. In complete heart block, the atria and ventricles beat separately. In some cases, heart block may lead to blackouts (*syncope*), breathlessness, and/or irregular heartbeats (*arrhythmias*). In severe cases, sudden death is possible.

WHAT CAUSES EMERY-DREIFUSS MUSCULAR DYSTROPHY?

There are different forms of EDMD, depending on the type of inheritance pattern that is involved.

X-Linked Recessive Inheritance

In most cases, EDMD is inherited as an X-linked recessive trait.

This type of inheritance refers to the inheritance of a 'recessive' gene change (*mutation*) located on the X chromosome (one of the sex chromosomes). Since females have two X chromosomes and males have one X chromosome and one Y chromosome, faulty X-linked recessive genes affect them differently.

Females have two X chromosomes. They consequently have two copies of each gene on the X chromosome. A woman will be a *genetic carrier* for an X linked recessive condition if one of her X chromosome gene copies is faulty, and the other gene copy is working. She is able to send the right gene message from the working gene copy, and therefore would not usually show symptoms of the condition.

An X-linked recessive genetic carrier female may pass on the faulty copy of the X chromosome to her children. In each pregnancy, there is:

- A 25% chance of having a daughter who is also a genetic carrier
- A 25% chance of having a daughter who inherits the working copy of the gene
- A 25% chance of having a son who will only have the X chromosome with the faulty gene copy, and will therefore be affected by the condition (since he does not have a working copy of the gene)
- A 25% chance of having a son who has the working copy of the gene.

Males have one copy of the X chromosome and are therefore affected differently when there is an X-linked recessive faulty gene present. Males who have a faulty copy of the X chromosome are not able to send the right gene message since they do not have a working copy of the gene. For this reason, they will almost always show symptoms of the condition and are described as being 'affected'.

If an affected male has children, he may pass on the X chromosome with the faulty gene copy. In each pregnancy, there is:

- A 50% chance of having a daughter who is a genetic carrier for the X-linked recessive condition
- A 50% chance of having a son who does not inherit the faulty gene copy

Autosomal dominant inheritance

EDMD may also be inherited as an autosomal dominant trait.

This type of inheritance refers to the inheritance of a 'dominant' gene change (*mutation*) located on an autosome (one of the numbered chromosomes). There are two copies of every autosomal gene. Both copies of the gene send a message to the body, but a dominant faulty gene copy will override the message sent from the working gene copy.

If a parent has an autosomal dominant condition, there is a 50% chance that they will pass on the dominant faulty gene (and therefore the genetic condition) to each of their children. This chance is the same for each pregnancy and is the same for males or females.

Autosomal recessive inheritance

Autosomal recessive inheritance is extremely rare, but has been reported in a few families.

This type of inheritance refers to the inheritance of a 'recessive' gene change (*mutation*) located on an autosome (one of the numbered chromosomes). There are two copies of every recessive gene. Both copies of the gene send a message to the body, but a recessive faulty gene will usually be overridden by a message sent from a working copy of the gene.

An individual will be *affected* by an autosomal recessive condition if *both* gene copies are faulty. They do not have a working gene copy and therefore their body will not receive any of the messages required to override the effects of the recessive faulty gene copy.

An individual will be a *genetic carrier* for an autosomal recessive condition if one of their gene copies is faulty and the other copy is working. They are able to send the right message from their working gene copy and therefore do not usually show symptoms of the condition caused by the mutation.

If two parents are genetic carriers for the same autosomal recessive genetic condition, there is a chance that they may have children affected with the condition. There is a 25% chance in each of their pregnancies for having a child with two faulty gene copies, who is therefore affected with the condition. There is a 50% chance of having a child who is a *genetic carrier* (like the parents) and not affected by the condition and 25% of having a child who has two working copies of the gene for that condition (is not a genetic carrier or affected).

All individuals carry a few gene mutations. Parents who are close relatives (*consanguineous*) have a higher chance than unrelated parents to both carry the same gene mutation. This therefore increases their chance of having children with a recessive genetic condition.

Genes causing Emery-Dreifuss Muscular Dystrophy
It has been shown that the X-linked form of EDMD is caused by disruption or changes (*mutations*) of the *EMD* gene, located on the long arm (q) of the X chromosome (at Xq28). The *EMD* gene produces a muscle protein known as Emerin. Emerin is found in most cell types of the body, but occurs in greatest quantity in bone and heart muscle cells.

The autosomal dominant and autosomal recessive forms of EDMD are caused by mutations within the same gene, located on the long arm (q) of chromosome 1 (at 1q21.2). This gene is known as the *LMNA* gene, and encodes the proteins lamin A and lamin C. These proteins are thought to have a role in cardiac and skeletal muscle function. Interestingly, faulty copies of the *LMNA* gene also cause a variety of other human diseases, including limb-girdle muscular dystrophy, dilated cardiomyopathy, Dunnigan-type familial partial lipodystrophy, and the premature aging disease Hutchinson-Gilford progeria syndrome.

For more information about genes, chromosomes and the autosomal dominant, autosomal recessive and X-linked recessive forms of genetic inheritance, please refer to Genetics Fact Sheets 1, 2, 4, 8, 9, and 10.

WHO IS AFFECTED BY EMERY-DREIFUSS MUSCULAR DYSTROPHY?

The overall prevalence of EDMD is unknown. The X-linked form is estimated to affect 1 in every 100,000 people in the general population.

X-linked EDMD is fully expressed in males only. Approximately 10-20% of female carriers for X-linked EDMD will develop heart conduction defects and/or muscle weakness.
The autosomal dominant and recessive forms of EDMD affect males and females in equal numbers.

HOW IS EMERY-DREIFUSS MUSCULAR DYSTROPHY DIAGNOSED AND TREATED?

Diagnosis
A diagnosis of EDMD is based upon a thorough clinical evaluation, a detailed patient history, identification of characteristic symptoms (contractures, myopathy, heart defects, etc.), surgical removal and microscopic study (*biopsy*) of affected tissue, and specialised tests such as DNA testing and immunodetection (particularly in X-linked forms).

Immunodetection describes the technique used to determine the presence, and levels of certain proteins such as Emerin in tissue samples obtained from affected individuals. These tests involve the use of certain antibodies that react to certain proteins. Samples taken from tissue biopsies are exposed tested with these antibodies. In approximately 95% of individuals with X-linked EDMD, Emerin is absent.

DNA testing involves the examination of deoxyribonucleic acid (DNA) to identify a specific gene mutation. A blood sample may be used for this test.

Additional tests that may be used to diagnose EDMD in an individual include a test called an *electromyograph*. This test measures the electrical potential generated by muscle cells when these cells contract, and also when the cells are at rest, thus assessing the health of muscles and nerves. Blood tests may reveal elevated levels of the creatine kinase (CK), an enzyme that is often found in abnormally high levels when muscle is damaged. The detection of elevated CK levels may confirm that muscle is damaged or inflamed, but cannot confirm a diagnosis of EDMD.

Treatment
There is no specific treatment for EDMD. Treatment is aimed at the specific symptoms present in each individual. Options may include physical therapy and active and passive exercise to build muscle strength and prevent contractures. Surgery may be

recommended in some cases to treat contractures or scoliosis. The use of mechanical aids (e.g., canes, braces, and wheelchairs) may become necessary to aid walking (ambulation).

Children diagnosed with EDMD should be monitored regularly for potential heart involvement. In the case of serious heart problems, cardiac pacemakers may be implanted and treatment with antiarrhythmic drugs may become necessary.

Genetic counselling is recommended for individuals with EDMD and their families.

Genetic Testing Options

Genetic testing may be available for this condition. Genetic testing may be carried out on individuals, on an unborn baby (*prenatal testing*) or on an embryo (*preimplantation genetic diagnosis*). For the most appropriate and accurate information, contact a genetic counselling service to find out whether genetic testing is available for this condition and discuss your specific options and questions.

For further information on genetic counselling, prenatal testing and preimplantation genetic diagnosis please refer to Genetics Fact Sheets 3, 17 and 18.

RESOURCES

For information regarding local genetic counselling services, please contact the Centre for Genetics Education (CGE) on 9926 7324 or contact@genetics.com.au or www.genetics.edu.au

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Muscular Dystrophy Campaign
7-11 Prescott Place London SW4 6BS
E-mail: info@muscular-dystrophy.org
Home page:
http://www.muscular-dystrophy.org

A PERSONAL STORY
Emery-Dreifuss MuscularDystrophy

A MOTHER'S HEARTBREAKING JOURNEY

(AGSA would like to acknowledge our appreciation of the emotional effort taken to write this story and in so doing, we all will remember Keira and her family. Thank you Christine.

Keira - In the beginning.

I still remember vividly my dear Dad coming to see me in hospital when his first grandchild, our Keira, was born. His words to me were, "Well Christine you now have your own "real live doll" and she was. She was the baby girl I always wished for. Keira was petite with glorious black hair and deep brown eyes. I couldn't take my eyes off her and I remember I wouldn't let the nurses take Keira to the nursery so that I could get some rest. No-one was taking my precious daughter away from my side.

Keira learnt to play the piano and was a wonderful artist (she won the Parents & Citizen Award for her Year 12 major). She obtained her HSC, learnt to drive and was hoping to become an Early Childhood Teacher. We, her parents are so proud of our daughter's achievements in life.

Keira loved tap dancing and swimming when younger. Keira was an avid reader and loved music (something she shared with her Dad and girlfriend Anna), they enjoyed many concerts together. Keira was also keen on architecture and was always drawing houses. Keira's Year 10 schoolwork experience was with a local Draftsman and he said she had an amazing ability to succeed in this career, but because of her physical disability, Keira felt that this wasn't possible for her.

Over the past few years, Keira developed a passion for handbags. She nearly drove her father crazy one day. It took Keira four hours to choose a handbag. He ended up walking off and doing his own thing. Keira's good

friend Julie was also frustrated to no-end when they went shopping, as they always ended up in a store selling handbags. There was then no time to shop for anything else!

Keira and Anna, her girlfriend, would often go to a nightclub and have a cocktail or two. They would dress themselves up with the latest fashion and we would drop them off and they would phone us in the early hours of the morning still bright and chirpy. Needless to say we weren't! Each time they just sat and chattered all night and all the young men who tried to intrude on their time together would soon be rebuffed if they asked for a dance. Keira was madly in love with a dream man on paper and had all her baby's names chosen which was so beautiful. She also had her wedding all planned, "a frangipani theme". The loss of Keira's hopes and dreams causes us much pain and grief. These are the joys that we, Keira's parents, no longer can look forward to.

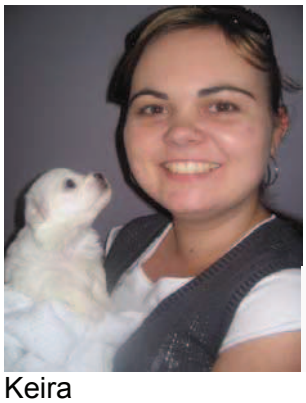
Keira and her good friend Julie had many happy times together. Julie introduced Keira to scrap booking and they would spend the day doing this instead of chores. Keira was a perfectionist with her work and she nearly drove the others who participated to distraction with her perfectionism.

I miss coming home to the sound of music and the smell of Keira's oil burner going. So many sights and sounds bring memories flooding back of our precious Keira. Some are joy and others to painful to bear.

Keira loved photographing flowers and made beautiful cards to share with others.

Keira also loved animals; she shared this love with another girlfriend Naomi. Keira was hoping to have a puppy when her health issues resolved.

We are so blessed for the happiness Keira brought to our lives for 22 years. Not a day goes by that I am not grateful for the time we shared.



Keira
1985 – 2007

Keira - The Story - The Life – the courage and the frustration.

We would like to tell others about our precious daughter Keira who passed away suddenly in August 2006 aged 22 years from heart failure.

Keira had suffered from an undiagnosed muscle condition from the age four.

Keira was a much longed for baby because we were told we were unable to have children. To find that I was pregnant three years later was an absolute joy. Our daughter was certainly our greatest gift in life. Keira was our only child and we have always felt so blessed.

During the first three years of life, Keira accomplished all her milestones as other children of the same age. At about three and a half years of age, Keira's physical abilities seem to decline after a prolonged bout of illness (constant diarrhoea) during which she lost a considerable amount of weight. She seemed to trip more easily and began having difficulty climbing stairs over the next twelve months. She was assessed by two Paediatric Neurologist's and underwent a muscle biopsy on her arm and leg. This biopsy was unable to make a diagnosis and it was subsequently forwarded to a Professor in West Germany who was also unable to define Keira's muscle condition. We were advised to bring Keira to Sydney for six monthly check ups and to treat her as a "normal child".

At ten years of age, Keira began experiencing contractures of her elbows which caused her much pain and anguish. Event though Keira continued to be monitored at six monthly

intervals by a Neurologist until her passing, her muscle condition remained undiagnosed until recently.

Keira's disability did make it more difficult for her to partake in some physical activities and she suffered emotional bullying from some children about her disabilities. She was often frustrated with herself. In spite of this, Keira never let her disability stop her from doing her best at whatever she chose. Over the years Keira tired more easily and she didn't have the physical strength of someone the same age.

Keira had a wonderful way with children, they seemed to be drawn to her and she gave of herself in a loving and caring manner. She seemed to have an amazing ability to transform herself into "their world". It was because of this love for children that she was planning to become an Early Childhood Teacher. Keira did exceeding well and was three weeks from completing her degree when she developed a bad flu. This illness seemed to bring on heart palpations, something Keira had not previously experienced. Keira subsequently developed atrial fibrillation and was referred to a local Cardiologist who then referred her to Newcastle to see a Cardiac Electrophysiologist. Keira was admitted to hospital in Newcastle and underwent cardiac examination and was found to have AV conduction disease. She was sent home on medication for the atrial fibrillation and Warfarin to prevent clots. This meant weekly blood tests for Keira. The taking of blood was very painful for Keira because of the contractures in her elbows which made it very difficult for the collectors to insert the needle into the veins adequately. There was always more than one attempt necessary. Keira never complained her strength and courage was amazing.

Keira was not herself during 2006. She became continually short of breath and was unable to complete her studies. Because of increasing concerns, Keira underwent a test for her heart in Newcastle in May 2006. Keira was to have CT scan mapping of her heart but unfortunately the machinery broke down and after a week of hospitalisation and further investigations of her heart she was sent home without the procedure being performed. It was felt that she needed biventricular pacing. Keira was very anxious about this. She was not provided with any counselling or ongoing support except from us and we knew very little. We were informed that Keira would be recalled shortly for the heart mapping to be performed. Keira was still waiting to be recalled for this procedure when she passed away!

The Cardio Electrophysiologist in Newcastle was keen to have a more definitive diagnosis made of Keira's muscle condition and suggested that Keira make arrangements to consult with a Geneticist.

In June 2006, we accompanied Keira for her consultation with a Geneticist in Sydney The Geneticist felt that Keira had a muscle condition called "Emery-Dreifuss Muscular Dystrophy" because of her newly diagnosed cardiac condition and the contractors of her elbows - these are significant in the diagnosis of this condition.

We were very alarmed by what we read on the internet regarding Emery-Dreifuss. Keira printed out several copies of the information available on Emery-Dreifuss and gave one to her local Cardiologist to read and on to our local GP. Keira had a Colonoscopy and Gastroscopy in August 2006. Keira had been experiencing regurgitation and continually diarrhoea which was restricting her life. Keira's GP referred her to a local Gastroenterologist who was fully informed about Keira's health concerns and the suspected diagnosis of Emery-Dreifuss. The Gastroenterologist consulted with Keira's local Cardiologist regarding the safety of Keira undergoing this procedure and he felt that there were "no problems for Keira". Keira and I were reassured that this was only a minor Day Procedure and that only a local anaesthetic would be administered. I had grave concerns about Keira undergoing the procedure. I now wish I had listened to my inner instinct.

Keira had continually vomiting, tiredness, shortness of breath and subsequently developed stomach, feet and hand swelling over the ensuring 17 days after the colonoscopy and gastroscopy were

performed. We took Keira back to the local hospital where the procedure was performed, twice in the following week, to the After Hours Medical Centre and to her local GP on three occasions during the second week of her illness. On each consultation, Keira was given an anti-vomiting medication or injection and told that she would recover soon. I really felt that she should be hospitalised but was reassured that this wasn't necessary.

Keira's condition seemed to be deteriorating rather than improving and we were beginning to feel very frustrated and concerned, Keira by this time was beginning to express that "no one is listening to what she is trying to tell them about her health concerns".

I took Keira back to the local hospital on Monday 28th August 2006 because of my increasing concerns about her health. She had been ill for too long – 17 days! In emergency Keira's full medical history was given including her muscle and heart condition, details about the recent hospital procedure and the recent diagnosis of suspected Emery-Dreifuss, which was later confirmed in December 2006 after her passing. I was informed that Keira was in heart failure but not to be concerned as there were many degrees of heart failure and she was fine to go home and see her cardiologist in his rooms tomorrow. I did not want my daughter to be discharged and expressed this opinion. I reluctantly took my precious daughter home. I felt very bewildered and concerned as it was very clear to me that Keira was not well. We lived thirty minutes from the hospital.

At six o'clock two hours after being discharged from the hospital Keira passed away. We tried desperately to resuscitate Keira to no avail. The wonderful ambulance officers were also unsuccessful in their attempts to save our precious daughter.

We cannot believe that our precious daughter, who lit up our world every day with her smile and unconditional love, has been taken from us. She touched so many lives with her strength and courage. Family and friends cannot believe that she has gone.

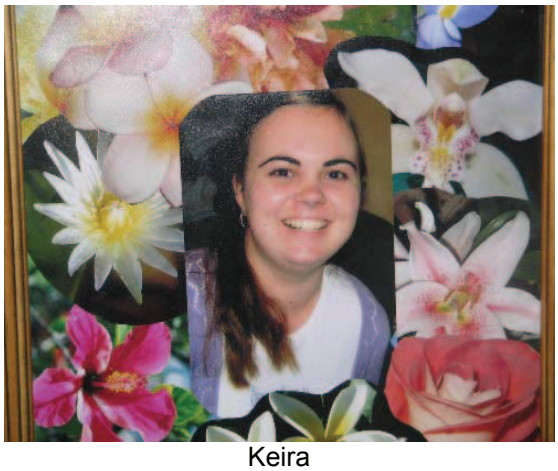
We her broken parents feel greatly privileged to have shared our lives with our one and only child. Our lives will never be the same again.

My daughter, my best friend, in life you were my world. You will always be remembered. You fought so bravely. Words cannot come close to how I feel.

There are many lessons for all to learn from our daughter's passing.

As Keira's parents we can appreciate that technology has brought about better communication for all, but sometimes this is not enough. We are very fortunate for the advancements in genetic diagnosis of medical conditions.

We feel very strongly that there must be a more proactive way to managing young adults with serious medical conditions who do not have ready access to the specialists in times of crisis.



Keira

PERSONAL STORY TMAU

Australian Magazine, Edition 1, SAT 30 JUN 2007, P 14

GOOD TIMES, BAD TIMES

Ron Green lived in Sydney as a social outcast before he Googled "body odour" and discovered what his doctors had failed to diagnose.

I knew there was something wrong with me from a very young age, but no one knew what it was. I remember being a six-year-old at school and my teacher sending me home and telling my mother to bathe me because the other students shouldn't have to put up with the smell. My mother didn't seem aware of it or understand what they were talking about.

My life was an absolute disaster. I was treated as a pariah and no one would tell me what was wrong; they just avoided me. I was always made to feel that I was not welcome and it was very embarrassing. People made comments like: "What died?" I couldn't detect the scent on my skin, but I could smell it on my clothes after I had worn them or if I opened a suitcase after I had been away. I went to doctor after doctor and had tests but they couldn't find anything wrong. They even suggested it was psychological. It used to make me so angry because I knew there was something wrong. I started to doubt my sanity.

As a young man I had a lot of problems trying to make my way in the world. I was always on the outside and I was the one who was never involved. I resorted to activities I could do alone, like fishing, and I had my dogs. I think I did okay despite the problem. I did a university degree and became a teacher. But the kids and the other teachers would complain.

When the internet came out I did a search. I just typed in "body odour" and the word "trimethylaminuria" came up. Also known as Fish Odour Syndrome, it is a genetic disorder that prevents the body from breaking down the compound trimethylamine. There are between 600 and 700 people diagnosed with the condition around the world. There is no cure but there are ways to reduce the odour.

As I read about it and the symptoms I thought, "That's me - that's it." I went back to my GP and after two referrals I found a geneticist who had heard of the condition. He tested me for the disorder and it came back positive. Finally I had validation - I wasn't mad. All these years I had known there was something wrong and it was just my internal strength that had stopped me from going crazy.

It was a great relief knowing there was something I could do about it. I was advised to go on a low-choline diet, which means no organ meats, fish or seafood, egg yolk, nuts, beans, peas or pulses. I also take probiotics and supplements and drink lots of water to flush out my system. I found the diet worked for me. What I am angry about is that it took the best part of 50 years to find out what was wrong with me. It was so unfair.

Since taking steps to alleviate the problem, my life has turned around. I've nearly finished a law degree and I'm in the process of doing a graduate diploma. I am hoping to be a criminal lawyer, and I hope to be arguing cases in court before long. I'm confident it will happen. I've spent the past four years in stuffy theatres and other places and I've not had a negative comment. My life is nothing like it used to be. People don't want to jump out of planes and leave cinemas when I'm around any more.

But I am the person I am because of the condition - I'm very much a loner and will probably always be that way. I don't go out a lot, and even though I don't get any negative comments about it anymore I am still afraid people are going to reject me. It's a constant battle to be with people because of the fear that developed over 50 years of shame. I don't have a partner and I don't think I ever will - I am happy as I am now.

***Ron Green** is a pseudonym.*

20 September, 2007

MEDIA RELEASE

The Hon Tony Abbott MP Minister for Health and Ageing
The Hon Ian Macfarlane MP Minister for Industry, Tourism and Resources

GENETIC CONDITIONS HANDBOOK FOR GPs LAUNCH

General Practitioners across Australia will soon have access to a unique resource enabling them to quickly and easily access information on genetic conditions for their patients.

Genetics in Family Medicine: The Australian Handbook for General Practitioners was released today by the Minister for Health and Ageing, Tony Abbott, and Minister for Industry, Tourism and Resources, Ian Macfarlane.

Mr. Abbott said the handbook had been developed in response to the demand for support material for GPs dealing with genetic medicine in their everyday practice.

"Genetic medicine is a constantly evolving field and this handbook, produced by Biotechnology Australia, addresses key genetic issues such as cancer in the family, cardiovascular conditions, diabetes and many inherited conditions," Mr. Abbott said.

"This handbook will help GPs provide information on genetic conditions to patients and make appropriate referrals." Mr. Macfarlane said the handbook was a definitive resource for GPs and one, which would prove invaluable in their daily practices.

"This is the only national resource of its kind available," Mr. Macfarlane said.

The handbook was developed by the Genetics Education in Medicine Consortium which consulted GPs, consumers and experts in genetics to produce a relevant and targeted handbook for everyday use by GPs.

"I am delighted that the National Health and Medical Research Council (NHMRC) is ensuring this handbook is widely available to GPs," Mr. Macfarlane said.

Copies of the handbook will be made available at the 50th annual conference at the Royal Australian College of General Practitioners in Sydney on 7th October, and will be also available online at www.gpgenetics.edu.au

For further information:
Craig Cormick, Public Awareness Manager, Biotechnology Australia, (02) 6213 6805
Claire Kimball, Media Adviser, Mr. Abbott's office, 0413 486926
Claire Wilkinson, Media Adviser, Mr. Macfarlane's office, 0419 840452

Rare Chromosome Seminar in Swansea

By Kathy Fitzgerald
AGSA
Rare Treasures Support Group

On Saturday 19th May 2007 the 'Rare Treasures' group from AGSA joined with the Chromosome 18 Registry and Research Society (Aust) to run a seminar targeting issues associated with rare chromosome disorders.

Associate Professor Matt Edwards explained the procedures involved in CGH genetic testing – hope for a new kind of diagnosis for children

with a developmental disability. His talk was very interesting and explained many of the difficult technical procedures in lay terms as well as addressing many issues raised from parents at the seminar.

Sister Patricia Wilson spoke on 'You can Train your Brain – using Neuro Therapy to assist students with learning difficulties'. Sister Patricia led us all through a series of exercises, some of which she would use in a therapy session and while we all looked pretty silly, Sister Patricia does seem to be able to get results.

Cathy Wilson and Jann Harrison from AGSA's Rare Treasures group contributed their own personal stories about their children. Cathy's child is 13, and Jann's son 22, so we had a broad range of issues to cater for the various stages of parenting as well as some lovely, entertaining and thoughtful insights into the roles of a carer for a child with a rare genetic condition. Jann also gave some helpful websites that I have copied at the bottom of the page.

Tim Steenson, who has a Chromosome 18 disorder, provided us with a very entertaining talk on his life so far. Tim has been attending Toastmasters and his talk provided ample evidence of his skill at public speaking.

Jenni and Bruce Purvis had travelled all the way from their property in central Queensland, and together told a wonderful story of how Bruce, who has Ring Chromosome 18, helps out with all the jobs of the farm, but also of some of the setbacks of being geographically distant .

Veronica Wain, who has a daughter with Chromosome 18 deletion, has produced a DVD on 18q deletion. Veronica used the seminar group to evaluate the DVD and to provide feedback.

Overall, it was a worthwhile day with lots of helpful advice between the carers. Many families found other families that they will continue to contact after the seminar both as friends and as sharing a common bond of caring for a child with a rare genetic condition.

Many thanks must go to Marlene Brightwell and Mary Steenson who planned the day so thoroughly.

Websites for Rare Chromosome Registration

(with thanks to Jann Harrison)

NORD (North American Rare Disorders)
http://www.rarediseases.org/
CDO (Chromosome Deletion Outreach)
www.chromodisorder.org/
UNIQUE: Rare Chromosome Disorder Support Group www.rarechromo.org
Contact a Family
http://www.cafamily.org.uk/index.html


Cathy Wilson and Jann Harrison


Kim Fidock and Juno Hearnshaw with Hannah

ANNUAL SUBSCRIPTION
Individual $24.00 incl.GST
Group/Organisation $48.00 incl.GST

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

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Fax: + 61 2 9211 8077

Office Hours:
Mon – Wednesday 9 am – 5 pm
Thurs – Friday 10 am – 4 pm

Subscription Year Ist July - 30th June

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.

* consult with government bodies, both Federal and State, for appropriate funding for genetic services.

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

AGSA'S SUPPORT GROUP & ORGANISATIONAL MEMBERS as at January 2003

Act Muscular Dystrophy Association Inc.
Androgen Insensitivity Assoc. Support Group of Australia
Alagille syndrome Support Group
Albino Support Group
Angelman syndrome Assoc. Inc.
Alzheimer's Assoc of Aust Inc.
A.P.I.A. (Aust.Primary Immune Deficiencies Assoc.)
Assoc. for Children With a Disability, Vic.
Assoc. for the Welfare of Child Health (AWCH)
AUSSIE FOLKS
Australian Addison's Disease Assoc. Inc.
Aust. Arthrogryposis Group (TAAG) Inc.
Australian Assoc. for the Welfare of Child Health (AWCH)
Aust. CHARGE Association
Aust. Crohn's & Colitis Assoc.
Aust. Huntington's Disease Association (Qld) Inc.
Aust. Huntington's Disease Assoc. (NSW) Inc.
Aust. Speak East Assoc.
Australasian Tuberous Sclerosis Society Inc.
Aust. Leukodystrophy Support Group
Aust. Society for Ectodermal Dysplasia
Autistic Assoc. of NSW
Batten's Disease Support & Research Assoc. Inc. (Australian Chapter)
Beckwith-Weidemann syndrome Support Group
Bunyip Special Needs Group Inc.
Cardiomyopathy Assoc of Aust. Ltd.
Centacare Early Intervention.
Centre for Developmental Disability Studies
Charcot Marie-Tooth Assoc. of Australia Inc.
Charcot Marie-Tooth Disease, USA
Child & Family Health Centre
Child Health Information Centre
Community Resource Team (Albury)
CONTACT A FAMILY U.K.
Cleft Pals, The Cleft Palate & Lip Society
CLIMB Children Living with Inherited Metabolic Diseases
Coeliac Society of NSW Inc.
Congenital Adrenal Hyperplasia Support Group
Cornelia de Lange syndrome Support Group
Cri du Chat syndrome Support group of Australia Inc.
CVS Support Group (WA)
Cystic Fibrosis Assoc of Qld Ltd.
Cystic Fibrosis Assoc. of Vic
Cystic Fibrosis New South Wales
Early Education Clinic, North Sydney
Early childhood Intervention Program
DIAL (Qld)
Donor Conception Support Group
Depressive & Manic Depressive Assoc.
Dystrophic Epidermolysis Bullosa Research Association (DEBRA) NSW. Inc.
Early Learning Tasmania
Ehlers-Danlos syndrome Support Group
Exceptional Parent (USA)
Fabry's Support Group Inc.
Family Advocacy
Family Planning Assoc.
Fragile X Assoc of Australia
Friedreich Ataxia Assoc of NSW
Gaucher Assoc. of Australia
Genetic Alliance (USA)
Genzyme Australia Pty. Ltd.
Genetic Interest Group (GIG) (UK)
I.D.E.A.S. Inc
Kidney Kids Support Group NZ
Klinefelter syndrome Support Group
Kurrajong Early Intervention
Haemochromatosis Society Inc.
Haemophilia Foundation NSW
Hereditary Cancer Registers (NSW Cancer Council)
Hereditary Haemorrhagic Telangiectasia
Hereditary Fructose Intolerance
Hunter Orthopaedia School
IDEAS Inc.
Kidney Kids of NZ Support Group
Maternity Alliance
NALAG
Leukodystrophy Foundation (USA)
Leigh's Disease Support Group
Lowe's syndrome Assoc. Inc. (USA)
Lower Nth Shore Community Support Team
Lupus Association of NSW Inc.
Lysosomal Diseases Australia
M.P.S. Society
Marfan syndrome Support Assoc. NSW
Marfan syndrome Assoc. Australia (S.A.Branch))
Meniere's (NSW) Support Group
Mental Illness Nervous Disorders Association
Metabolic Dietary Disorders Association (MDDA)
Mid North Coast Area Health Taree Genetics Service
Motor Neurone Disease Assoc. of NSW Inc.
Multiple Epiphyseal Dysplasia Assoc.
Muscular Dystrophy Assoc of NSW
Muscular Dystrophy Assoc (NZ) Inc.
National Council of Intellectual Disability
NCOSS (NSW Council of Social Services)
Neurofibromatosis Assoc.
Noonan syndrome Support Group
NSW Genetics Education Program
NSW Cancer Council
Osteopetrosis Support Group
Osteogenesis Imperfecta of Aust.
Parents Bereavement Support Group
Parent to Parent (NZ)
Pen-Parents of Aust. (ACT)
PKU Assoc of NSW
Polycystic Kidney Disease Association
Psoriasis Society
Pseudohypoparathyroidism Support Group
Pseudoxanthoma Elasticum Support Group
Prader-Willi syndrome Assoc. of NSW (Aust) Inc.
Pyruvate dehydrogenase deficiency.
Rare Chromosomes Disorders Support Group
Retina Australia (NSW) Inc.
Rett syndrome Assoc. of Aust.
Royal Blind Society of NSW
SAFDA (Support After Foetal Diagnosis of Abnormality)
SANDS
Short Statured People of Northern Qld
Short Statured People of Aust (NSW)
Short Statured People of Aust (Vic)
Short Statured People of Aust. (SA)
Spinal Muscular Atrophy
Schizophrenia Fellowship NZ
Smith Magenis syndrome Support Group Inc.
Spastic Society of Victoria
Spina Bifida Assoc. of NSW
Spina Bifida Assoc. of WA Inc.
Society of Ectodermal Dysplasia
SOFT Australia
Southern Child Care Support Program
Sotos syndrome Support Group
Steele Street Early Special Education Centre Devonport
St Paul's Special School
The Chromosome 18 Registry & Research Society
The Northcott Society
The Toybox Centre Inc.
Thalassaemia Society of NSW
Turner syndrome Assoc of Aust. Ltd. (QLD)
Turner syndrome Assoc of Aust. Ltd. (SA)
Turner syndrome Assoc. of Aust. Ltd. (NSW)
Uncontrolled Epilepsy Support Assoc (Vic)
United Leukodystrophy Foundation (USA)
Velo-Cardio-Facial syndrome Foundation of Australia.
Wellington Huntington's Disease Assoc. (Inc.) (NZ)
Western Institute for Self Help (W.I.S.H)
West syndrome Support Group
Wolf-Hirschhorn 4p- syndrome Support Group
Williams syndrome Association of Aust. Inc.

(NB: This list represents support groups and associations members only. In addition to this list of members AGSA has established a Contact Register over 550 genetic conditions representing families and individuals seeking contact.)