AGSA
THE ASSOCIATION OF GENETIC SUPPOR T OF AUSTRALASIA INC.
NEWSLETTER

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**MISSION STATEMENT**

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

# EDITORIAL

Hello everyone. As I write this I feel very sad as on 27th February I along with 600 others attended Matt Laffan's funeral service. I feel very privileged to have met him and his father when he kindly presented at Genetic Awareness Week Genetics: An Adult's Perspective *"Beyond the muffled crisis"* at the Powerhouse Museum in 2004. the modest bio which he gave me for the program did not mention he had diastrophic dysplasia read as follows: **Matt Laffan** – is a Lawyer with the New South Wales Office of the Director of Public Prosecutions and Member of the New South Wales Disability Council since 2003. He is regularly engaged as a speaker and writes a travel column for http://www.accessibility.com.au/ detailing domestic and international sojourns and how they were achieved. Matt ran as an Independent in the Lord Mayoral elections for the City of Sydney in 2004. Peter FitzSimons who wrote his obituary in the Sydney Morning Herald on 3rd March sums it up very well *"An extraordinary battle to live an ordinary life".*

I still remember that evening and his very powerful, spiritual and encouraging speech. My son, who is now 29, still remembers that evening too and as well as having an impact on many; it had a huge impact on him. On meeting Matt for the first time he immediately put me at ease and was very engaging.

It is an enormous loss and I send our heartfelt condolences to his Mum, Jenny and Dad, Dick family and friends.

Ironically the weekend Matt died was International Rare Disease Day and AGSA held a seminar to celebrate this day and to raise awareness of the 8000 rare Diseases worldwide. Rare diseases affect 1.5 million Australians compromising 300,000 children. AGSA organized the day for Australia in conjunction with Smile Foundation (see press release). Included in the newsletter there will be excerpts from presentations made on the day. I would like to thank all our members who came along and who strongly agreed to support this movement. It will be a big job and hopefully next year will see a united national approach.

Also included is an article from the Australian Paediatric Surveillance Unit who on 9th February held a meeting with support groups, AGSA, New Zealand Rare Diseases Organisation and a representative from EURORDIS plus health professionals and eminent researchers to discuss a national approach to rare diseases.

AGSA is part of a world wide movement to identify and classify rare diseases, to promote research into rare diseases and to work productively with researchers and families to develop treatments.

While our primary role is to provide support to families, carers and individuals with information, contact and counselling we believe that participation in advocacy for promotion of research and therapies is also part of our role.

Some of the ways we do this is through consumer participation in organisations such as NSW Genetics Advisory Committee, NSW Newborn Screening Committee, Australasian Genetic Alliance, International Genetic Alliance and other groups who lobby government for support for carers and individuals for better services and support for research diagnosis and therapies.

On 15th March AGSA held a Noonan syndrome seminar at AGSA and we thank Dr Anne Turner and Dr Maria Craig to coming along to present.

On 21st March AGSA held a Carers Seminar and a Sibling workshop the following day in Newcastle. The response to this was very good.

Until next time,
Best Wishes to you all for a Happy Easter.

## Dianne Petrie

### EURORDIS AND THE RARE DISEASE PATIENT MOVEMENT IN EUROPE

Eurordis was founded in 1997, on the initiative of the French Muscular Dystrophy Association (AFM), French Cystic Fibrosis Association, French National Cancer League (LNCC) and the French National Aids Federation, on the model of the North American NORD (National Organization for Rare Disorders) to advocate for the adoption of the EU Orphan Drug Regulation and for EU Rare Disease Public Health and Biomedical Research policies.

These founding organisations realised that the patients they represented shared common characteristics, such as delay in diagnosis, lack of information, knowledge and research on their diseases, heavy social consequences for patients and their families, and difficulty of access to treatment and care.

Today Eurordis represents more than 600 rare disease patient organisations (directly or indirectly) in nearly 40 countries, covering more than 1,500 rare diseases. It is therefore the voice of the 30 million patients affected by rare diseases throughout Europe.

Among Eurordis' most visible achievements are the contribution to the adoption of the EU Orphan Drug Regulation in 1999; the contribution to the maintenance of rare diseases as a EU public health and research priority; the contribution to the adoption of the Paediatric Drugs Regulation in 2006; and the contribution to the Advanced Therapy Regulation in 2006 (which includes gene therapy).

At the end of 2008, the EU issued a Communication and a Recommendation on Rare Diseases. These important political instruments set up the landscape for rare diseases in Europe and recommend a list of steps to put in place for the 27 Member States of the Union. These steps include the establishment of national plans for rare diseases and the development of cooperation on rare diseases at the European level.

The aforementioned political achievements have had some practical implications for the lives of rare disease patients, including the designation of more than 400 orphan drugs, the dissemination of knowledge and the raising of awareness on rare diseases, the development of social services such as respite care centres, and the identification and networking of centres of expertise, to which patients can be referred to for treatment and care.

In Australia, there are many excellent patient organisations and research institutes for rare diseases. There is currently a worldwide movement for rare diseases and it is important that Australia be part of it. The International Rare Disease Day epitomises this movement. Australia is also in a position to benefit from lessons learned elsewhere; it can copy, adapt and develop tools and strategies used in other countries such as Europe and the USA to improve the lives of people affected by rare diseases in Australia.

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***"Optimism is the faith that leads to achievement. Nothing can be done without hope and confidence."* Helen Keller**

SMILE Foundation's CEO Karen Gair reminded us that the historical meeting was merely the start of a long journey where optimism, hope and confidence need to be our constant companions.

The SMILE (Supporting Medical Innovation for Life Enhancement) Foundation was established in July 2007 to fund medical research into rare diseases and to help families with the financial burden of having a child with a rare disease. Through its Family Relief Program SMILE makes $1,000 grants to struggling families through social workers in children's hospitals across the country. Each month SMILE receives a dozen applications from families whose needs range from medical equipment and hospital bills to petrol, mobile phone calls and electricity bills to ensure their fridge at home housing medications is not turned off.

Over the past week and in the coming week, SMILE – with the assistance of pro-bono PR firm Ogilvy – have had radio and press reports in most capital cities highlighting Rare Disease Day.

Karen said the SMILE Foundation was supportive of an Australian Organisation of Rare Diseases similar to the European and US models. The power of a large group – that represents the estimated 1.5 million Australians with a rare disease – can have enormous advocacy potential. A loud and unified voice brings change.

This started discussion amongst the attendees about how and if a new body should be formed. It was noted that AGSA represented the rare disease community effectively already but needed greater advocacy, PR and policy-making resources to become a more broad-based organisation representing all rare diseases.

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**BIONEWS**

*Reprinted with permission from BioNews (www.bionews.org.uk)"a free web-and-email-based news service on human genetics and assisted reproduction, published by the UK charity Progress Educational Trust.*

**RARE DISEASE DAY 2009 HAILED A SUCCESS:**

By Melissa Hillier,
Genetic Interest Group:

Rare Disease UK (www.raredisease.org.uk) and the Genetic Interest Group (www.gig.org.uk) are organising activities around International Rare Disease Day on 28th February 2009, including three parliamentary receptions to raise the profile of rare diseases and to campaign for the development of a National Plan. A national plan for rare diseases would improve the care, support and treatment for the over 3.5 million people currently living in the UK with a rare condition, according to the two organisations.

The first event was held on 25th February at the House of Commons and over 200 patients, clinicians and industry representatives as well as over 20 MPs and Lords filled the room. As Ann Milton MP noted in her speech, it is not often that so many various stakeholders gather in a room over a Wednesday lunchtime! It was a true measure of the support and commitment from so many that ensured the room was full, she said.

Rare Disease UK is a cross-sector campaign organisation planning to work with the NHS in the four home nations to create plans and strategies for systematic delivery of high quality integrated care and support to patients and families.

This will help to stimulate research and to promote effective horizon scanning so that new possibilities for intervention can be quickly incorporated into clinical practice. With over 6000 rare conditions identified it is vital that strategic measures are taken to deal with this growing issue, according to the Genetic Interest Group (GIG).Many families with a rare condition struggle to get a diagnosis and when they do it is often not clear what the next steps should be, leaving them without information and adequate support. Rare Disease UK and GIG feel that all families have the right to care and support that reflects current scientific understanding and best clinical practice and that the most appropriate way to do this for rare conditions is to create a national, coordinated plan. It will also enable the NHS to make the best use of its limited resources, say the organsiations.

Researchers will be able to use their skills in addressing unmet health needs arising from rare conditions and industry must also play its part within a clear framework that encourages genuine innovation and speedy transfer from lab to into clinical practice. National Plans are already being implemented in other European Countries such as France, Bulgaria and Portugal.

It is important to build on the current areas of good practice that are evident and ensure that these

pockets of best practice can be extended to all those living with rare conditions in the UK, stress GIG and Rare Disease UK. In the coming months Rare Disease UK will be working with all stakeholders in the development of a national plan that can support the NHS in the healthcare it provides.

Rare Disease UK membership is open to anyone with an interest in this area and is free. To sign up please go to our website www.raredisease.org.uk

## PRESS RELEASE

smile
supporting medical innovation for life enhancement

**Urgent Call for Support on International Rare Disease Day 26th February, Sydney:** Australians are being urged to recognise the needs of the many children and young adults across the country suffering from rare diseases.

The 28 February marks International Rare Disease Day which will be celebrated for the first time in

Australia to draw attention to the need for a better quality of life for those living with rare diseases and the importance of medical research to develop new treatment, providing hope for those recently diagnosed with a rare disease.

Around 1.5 million Australians suffer from rare diseases, which is similar to the number with diabetes, however in comparison funding levels are significantly less.

As part of world-wide activities to mark the occasion, local experts will meet in Sydney at a conference organised by The Association of Genetic Support of Australasia (AGSA) to discuss the latest research in the field and next steps for a recently formed national taskforce, the SMILE Foundation, set up to provide a united approach and voice for rare diseases.

"Australia has many excellent groups researching rare genetic diseases, and continues to find new insights and treatments for those who suffer from them", said Professor Bob Williamson, a member of the Board of the SMILE Foundation and Professor of Medical Genetics at the University of Melbourne.

"Sadly, because each disease is rare, these children are orphans of the health system. There are few doctors who are expert in rare disorders in Australia, support systems for children with chronic illness are poor, and pharmaceutical companies are not interested because not enough people are affected," he added.

This year's campaign entitled 'Patient Care: A Public Affair' will be promoted around the world in an attempt to gain the attention and compassion of key decision makers and a diverse range of communities.

International Rare Disease Day is organised by the European Organisation for Rare Diseases (EURORDIS) which represents over 600 patient groups in Europe.

We are very excited about working with our colleagues in Australia to help them develop a national plan for rare diseases that will not only provide a central support network for patients but also bring key decision makers together, increasing visibility of rare diseases and improving funding for care and treatment," said Jerome Parisse-Brassens from EURORDIS.

There are more than 8000 rare diseases, many of which present in childhood and many are chronic, some life-threatening and others associated with significant disability.

**Media Interviews:**
Media interviews are available with Professor Bob Williamson, a member of the Board of the SMILE Foundation and Professor of Medical Genetics at the University of Melbourne and the families of children suffering from rare diseases.
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**About SMILE Foundation:**
The SMILE Foundation is a charity set up to support research into the diagnosis, treatment and/or cure for rare childhood diseases or conditions and to help afflicted families. For more information,
Visit: www.smilefoundation.com.au
or call (02) 8986 5395.


The 28th of February is Global Rare Diseases Awareness Day. ***Did you know that rare diseases are actually common?*** There are about 8000 rare diseases affecting an estimated 1.5 million Australians, including about 300,000 Australian children.

The Australian Paediatric Surveillance Unit convened a rare diseases meeting in Sydney on the 9th of February to discuss the need for a more co-coordinated approach to providing health care and support for Australians affected by rare diseases.

The meeting was attended by a group of eminent researchers, health professionals and family support groups, including representatives from the European Rare Diseases Organisation www.eurordis.org and the New Zealand Rare Diseases Organisation www.nzord.org.nz We discussed the need for an integrated patient support organisation for Australians affected by rare diseases, improved access to information and education for health professionals, and new models of healthcare.

Discussions highlighted the striking similarities among the ~8000 rare diseases:

- Most begin in childhood (usually diagnosed in children aged < 2years) and continue throughout life
- Obtaining a definitive diagnosis is often difficult and delayed
- Many rare diseases have no cure or require new high-tech and often expensive treatments
- Neurological and intellectual disabilities occur in about half of all cases regardless of disease type and lead to reduced independence and opportunities
- Families experience isolation, psychological and financial stress
- Health professionals have inadequate access to information, education and resources
- It is estimated that about 35% of deaths in children aged < 1year are due to rare diseases.

The European Union, Canada, the UK and New Zealand have recognised the complex problem of rare diseases and are responding by establishing national support organisations for rare diseases, by making rare diseases a public health and a research priority and by developing integrated clinics that pool expertise to enable quicker diagnosis and better access to treatments and interventions.

A Creswick Foundation Fellowship will allow Dr Zurynski to study health and community services for rare diseases operating overseas and to implement this knowledge in Australia.

The second Rare Diseases Global awareness day will be held on the 28th of February 2009 and Australian organisations such as the Smile Foundation www.smile.org.au and Association of Genetic Support of Australasia (AGSA) (www.agsa-geneticsupport.org.au will be holding an event to highlight the importance of rare diseases and the Australian Paediatric Surveillance Unit www.apsu.org.au has joined the global effort to raise awareness of rare diseases: www.rarediseases.org


# OVERSEAS NEWS

## EEOC Requests Public Comment on GINA Regulations

On Wednesday, February 25, the Equal Employment Opportunity Commission (EEOC) released a notice of proposed rulemaking (NPRM) for Title II of the Genetic Information Nondiscrimination Act (GINA). Title II of the law protects individuals from genetic discrimination in employment, and these provisions will be in effect in November 2009. The NPRM opens a 60-day comment period that will allow the public to provide input regarding the implementation of GINA and the regulations proposed by the Commission. Throughout the next few weeks, the Coalition for Genetic Fairness (CGF) will draft comments for submission to the NPRM with the help and input of the genetics community. We strongly encourage individuals and organizations to engage in this policymaking process by considering the NPRM and its impact on you and your community. Let's shape the future for GINA together.

To learn more about the NPRM on Title II of GINA, please visit:
http://www.eeoc.gov/press/2-25-09.html

## *A – Z GENETIC CONDITION PROFILE*

# DIASTROPHIC DYSPLASIA (DD)

*Also sometimes known as diastrophic dwarfism or diastrophic nanism syndrome*

**WHAT IS DIASTROPHIC DYSPLASIA (DD)?**
Diastrophic dysplasia (DD) is a condition that is present at birth (congenital). The range and severity of symptoms and physical findings associated with DD may vary greatly from person to person. The most commonly seen symptoms are short stature; short arms and legs; changes in the development of bones (skeletal dysplasia) and joints (joint dysplasia) in many areas of the body; curvature of the spine (scoliosis and/or kyphosis). DD is an autosomal recessive genetic condition which can affect males or females.

**WHAT ARE THE SYMPTOMS OF DIASTROPHIC DYSPLASIA (DD)?**
The symptoms and physical findings associated with diastrophic dysplasia (DD) may be extremely variable, differing in range and severity even among affected family members. However, in all individuals with the condition, there are changes in development of bones and joints of the body (skeletal and joint dysplasia).

During development before birth and during early childhood, cartilage in many areas of the body is gradually replaced by bone (ossification). In addition, a layer of cartilage (epiphyseal cartilage [growth plate]) separates the shafts (diaphyses) of long bones (e.g., bones of the arms and legs) from their ends (epiphyses), allowing long bones to grow until the cartilage is no longer present. In those affected by DD, however, there is delayed growth before and after birth. The development of the ends of the long bones (epiphyses) is also irregular and the ossification of the epiphyses is delayed. Thus, newborns and children with DD typically have markedly short, bowed arms and legs and short stature (short-limbed dwarfism).

Due to abnormalities of skeletal development, infants and children with DD also have additional distinctive changes in bones of the hands, feet, and other areas of the body. For example, the first bone of each hand (first metacarpals) may be unusually small, short, and "oval shaped." As a result, the thumbs deviate away from the body and are described as "hitchhiker thumbs". In addition, other fingers may be unusually short (brachydactyly) and joints between particular bones of the fingers (proximal interphalangeal joints) may become fused (symphalangism), causing limited flexion and restricted movement (reduced mobility) of the finger joints. In some cases, bones of the wrists may also be involved and have irregular development.

Infants with DD also typically have severe foot problems (talipes or "clubfeet") due to fusion and deviation of bones within each foot (metatarsals). This usually makes the heels turn outward (talipes valgus) while the fore part of each foot deviates inward (metatarsus adductus). In other infants, the soles of the feet may be flexed (talipes equinus) and, in some cases, the heels may also turn inward (talipes equinovarus). The great toes, like the thumbs, may also deviate away from the body.

In addition to having limited flexion of finger joints, many affected infants and children also experience partial dislocation (subluxation) and/or complete dislocation of particular joints of the body. For example, in many cases, dislocations of the knees and hips occur upon weight bearing. Affected individuals may also have loose and/or stiff joints; experience limited extension of joints at the elbows and/or knees; and/or develop permanent flexion and immobilisation (contracture) of certain joints (e.g., knees).

Due to joint and bone changes such as those affecting the feet, many individuals with DD have a tendency to walk on tiptoe. In addition, affected individuals may be predisposed to degenerative changes (osteoarthrosis) of particular joints (e.g. of the hips), resulting in pain with use of the joint, tenderness, stiffness, and, in some cases, deformity.

Many infants with DD also have changes in the bones within the spinal column (vertebrae). For example, there may be incomplete closure of the vertebrae (spina bifida occulta) within the neck area and the upper portion of the back (lower cervical and upper thoracic vertebrae) and/or abnormal narrowing of portions of the vertebrae of the lower back (interpedicular narrowing in lumbar vertebrae). During the first year of life, some infants may begin to develop progressive sideways curvature of the spine (scoliosis). In addition, during adolescence, individuals with DD may also develop abnormal front-to-back curvature of the spine (kyphosis), particularly affecting vertebrae of the neck region (cervical vertebrae). In severe cases, progressive kyphosis may result in difficulties breathing (respiratory distress).

Some individuals with DD may also be prone to experiencing partial dislocation of joints between the central areas of cervical vertebrae (cervical subluxation), potentially resulting in compression of the spinal cord. (This cylindrical structure of nerve tissue extends from the lower portion of the brain and is located inside the central canal within the spinal column [spinal cavity].) Such spinal cord injury may result in muscle weakness (paresis) or paralysis and/or life-threatening complications.

Most newborns with DD also have or develop fluid-filled sacs (cysts) within the outer, visible portions of the ears (pinnae). Within approximately two to five weeks after birth, the pinnae become swollen and inflamed. When such swelling and inflammation subside, the pinnae remain thick, hard, and changed in shape. The areas of tissue involved may gradually accumulate deposits of calcium salts (calcification) and eventually be replaced by bone (ossification). As a result of this, affected infants may experience narrowing (stenosis) of the external ear canal which in a few people will cause hearing problems. Another cause of hearing problems in some people who have DD is fusion or absence of the three tiny bones (auditory ossicles) in the middle ear that conduct sound to the inner ear.

Some infants with DD also have characteristic features of the head and facial (craniofacial) area, such as a high, prominent forehead; small jaws (micrognathia) and a broad, highly arched roof of the mouth (palate) or incomplete closure of the palate (cleft palate). If cleft palate is present, it may cause difficulties with feeding and/or breathing. DD can also cause changes in the supportive connective tissue (cartilage) within the windpipe (trachea), voice box (larynx) and air passages in the lungs (bronchi). This can lead to narrowing and collapse of such airways. If left untreated, this can be a life-threatening complication. People with DD can also develop dental problems such as small teeth and dental crowding.

## WHAT CAUSES DIASTROPHIC DYSPLASIA (DD)?

DD is inherited as an autosomal recessive genetic trait. 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.

A gene responsible for DD, known as SLC26A2 (DTDST) [***S***o***L***ute ***C***arrier family ***26***, member ***2*** (***D***iastrophic ***D***ysplasia ***S***ulfate ***T***ransporter)] gene has been located on the long arm (q) of chromosome 5 (5q32-q33.1). Chromosomes are found in the nucleus of all body cells. They carry the genetic characteristics of each individual. Pairs of human chromosomes are numbered from 1 through 22, with an unequal 23rd pair of X and Y chromosomes for males and two X chromosomes for females. Each chromosome has a short arm designated as "p" and a long arm identified by the letter "q." Chromosomes are further subdivided into bands that are numbered. For example, 5q32 refers to band 32 on the long arm of chromosome 5.

The symptoms and findings associated with DD are thought to result due to abnormalities in the formation of cartilage, thus affecting skeletal development. Early on, during normal embryonic development, the skeleton mainly consists of cartilage that is gradually replaced by bone (ossification). After birth, many bones of the skeleton still consist primarily of cartilage that will eventually

ossify. However, researchers suspect that certain mutations of the DTDST gene result in abnormalities of cartilage cells (chondrocytes) and the substance (matrix) that lies between such cells, ultimately causing the symptoms and findings associated with DD. For example, in individuals with DD, the growth plate of long bones may contain an abnormal distribution of cartilage cells (chondrocytes) and abnormal fibrous and cystic areas within its matrix.

## WHO IS AFFECTED BY DIASTROPHIC DYSPLASIA (DD)?

DD affects males and females in equal numbers. Although DD is considered to be a rare condition, the percentage of genetic carriers in certain ethnic groups is high. In Finland, 1-2% of the general population are carriers and there is a prevalence ratio of 1 in 30,000.

DD has been observed in many different ethnic and cultural groups and the exact frequency of the condition in Australia is not known.

### HOW IS DIASTROPHIC DYSPLASIA (DD) DIAGNMOSED AND TREATED?

***Diagnosis***

In some families with a previous child affected by DD, it is possible that the condition may be detected before birth (prenatally) during early pregnancy During an ultrasound, a diagnosis of DD may be considered due to detection of certain characteristic findings such as marked shortening of bones of the fingers (phalanges), arms, and legs; deviation (abduction) of the thumbs ("hitchhiker thumbs") and great toes and/or other findings.

In most cases, DD is diagnosed and/or confirmed at birth based upon a thorough clinical evaluation, identification of characteristic physical findings, and a variety of specialising tests, such as advanced imaging techniques. For example, specialised x-ray studies such as computerised tomography (CT) scanning and magnetic resonance imaging (MRI) may be used to detect, confirm, and/or characterise certain skeletal features that may be associated with DD.

Specialised diagnostic testing (i.e., auditory tests) may also be performed to help detect hearing deficits that may occur in some children with DD.

***Treatment***

Treatment of individuals with DD should be directed towards the needs of each individual. It may be necessary for a team of specialists to work together and plan for the best strategy to enable each individual to reach their full potential. Such specialists may include paediatricians; physicians who diagnose and treat changes in the skeleton, joints, muscles, and related tissues (orthopaedists); surgeons; physiotherapists; dental specialists (orthodontists); specialists who assess and treat hearing problems (audiologists); and/or other health care professionals.

Specific therapies for the treatment of DD are symptomatic and supportive. Doctors may carefully monitor affected infants to ensure prompt detection and appropriate preventive or corrective treatment of respiratory obstruction and distress that may result due to certain abnormalities potentially associated with the condition (e.g., breathing difficulties). In addition, special supportive measures may be used to help ensure an appropriate intake of nutrients in infants who experience feeding difficulties due to cleft palate. In some cases, surgical procedures may be performed to correct malformations resulting in breathing and/or feeding difficulties. The specific procedures performed will depend upon the location, severity, and combination of such anatomical problems.

In addition, various orthopaedic techniques, including surgery, may also be used to help prevent, treat, and/or correct certain skeletal problems associated with DD. In some cases, physiotherapy in combination with surgical and supportive measures may be helpful in improving an affected individual's ability to walk and perform other movements (mobility). Early, persistent therapy for foot problems such as talipes or clubfeet may be helpful in achieving beneficial results. In addition, because particular skeletal changes associated with DD are progressive (e.g., kyphosis) and, in some cases, may lead to severe complications (e.g., respiratory distress, compression of the spine, potential paresis or paralysis), health professionals may perform ongoing monitoring to ensure prompt detection of and appropriate preventive and/or corrective measures for such abnormalities.

In affected children with dental abnormalities, braces (orthodontics), dental surgery, and/or other corrective procedures may be undertaken to correct such malformations. Steroid injections and/or other measures may also be used to help decrease the ear deformity that often affects infants with the disorder.

Just like each individual will be different, the treatment plan will be unique and best discussed with the health professionals involved in the care plan.

Genetic counselling will be of benefit for affected individuals and their families.

**REFERENCES**

National Organization for Rare Disorders (NORD) www.rarediseases.org

Online Mendelian Inheritance in Man

www.ncbi.nlm.nih.gov/Omim

GENE TESTS Gene Reviewswww.geneclinics.org

# Genetic Awareness Week 2004 Presentation by Matt Laffan at the Powerhouse Museum

## *"Beyond the Muffled Crisis"*

Ladies and Gentlemen,
It's a great honour to be here, and Your Excellency, it's wonderful again to see you and of course, meet some new people and new friends. We started off tonight, of course with the National Anthem. "Australians all let us rejoice for we are young and free" and there is no time quite like the one that I am about to mention, which sent a shiver down my spine, as on the occasion that I sang that at the Para-Olympics when Louise Savage was being given a gold medal. But it wasn't just because Louise Savage was being given a gold medal. At that time I was standing next to a good friend of mine, Nick Farr-Jones who is a former Wallaby and Captain of the Wallabies. I had known Nick for many, many years and I got to know him extremely well in about 1985 or '86. He, of course, having been a captain of the Wallabies had often been on the field when I was watching the rugby and just prior to kick off was singing the National Anthem, so it was a great honour for me to be standing beside my friend, who had sung it so many times before going out to represent Australia and play hard, to be actually singing the National Anthem.

And it also, I think, symbolises something extra special for AGSA, "Australians all let us rejoice for we are young and free", because the whole idea about this organisation is indeed to set people free, as it were. When you are born with a genetic disorder or when you become aware that you have one, I think the big thing is that you feel you are locked in, to something about which you have no control. You are unable to get free. But what AGSA is about is releasing you and allowing you to acquire the knowledge and work out for yourself the journey that you might be able to take.

It was in September 1970, a beautiful Spring day, so I'm told, that I was born. I was born at King George Hospital there in Missenden Road, Camperdown. Mum and Dad had no idea that I would be born with a genetic disorder by the name of Dystrophic Dysplasia. Once again one of those titles that is almost impossible to say and even more difficult to spell.

Prior to my arrival, Mum and Dad and my grandparents both maternal and paternal, had no idea that their first born son and first grandchild would indeed have such complications, as I was about to reveal to them. So when I was delivered via Caesarian, it became quickly obvious to the Obstetrician as she held me in her gentle hands, that all was not right, if you like. It took my maternal grandfather, who was an Obstetrician and Gynecologist, to be the one who actually diagnosed me with Dystrophic Dysplasia.

Dystrophic Dysplasia has many traits. Obviously, I have shortened limbs, that means that no matter how many Vita Brits I might eat for breakfast I was never going to grow up to be tall and strong and a robust lad. It also meant that I was born with cauliflower ears, and for any of you who are aficionados of sport, you will know that boxers acquire cauliflower ears, and prop forwards acquire cauliflower ears. So I always felt as I was growing up that I belonged to the tough tribe of men; there were boxers, there were prop forwards and there was me.

Also one tends to have a spine which twists and turns with scoliosis cyphosis?? So as I grew up the spine deteriorated and curved, and that meant that I needed an operation in 1980 which meant that I became a paraplegic and therefore relied on an electric wheelchair for mobility.

But during that time when I was first diagnosed and the doctors only gave me a week or so to live, as I say, 'I was given time to live, a very short time,' but I turned that around. But during that crisis time and it's the thing I refer to as 'the muffled crisis.' There had been this expectation, of course, and this excitement; with my family, maternal and paternal and Mum and Dad especially, with their first child

an excitement awaiting for the kid to be born. And then when I arrived all that excitement was still there and the love was poured into my heart and that's why I'm here today. But there was a muffled crisis: where do we go, what happens now? And for Mum and Dad, as most of you people understand, not all of you, the path wasn't so certain.

I was fortunate enough that Mum and Dad thought that the best track to put me on was, to use that horrible phrase, the 'normal' track, so I was able to enjoy a normal education, if you like. I grew up in Coffs Harbour where the winters were never too cold and the summers never too hot and I had a splendid co-education through the local Catholic primary school and high school until I went to the state school in years 11 and 12. But along the journey they had to make some big choices, and once again, we didn't have an AGSA.

When Mum and Dad looked, as to what perhaps was my potential and how we'd go about ensuring that that was reached, they had no real direction. There was no map for us, there were no people to whom we could turn. There was certainly an association for little people, but with the Dystrophic Dysplasia there was something different, of course. The spine brought about different complications, breathing difficulties, and the like.

Growing up in Coffs Harbour we were isolated I guess, so Mum and Dad just set about doing what they could. And I found, going back to the rugby association, that I was very lucky. The old man was involved in rugby for many years, still is, off to Ireland shortly for 3 months to do some coaching, and that enabled me to get in contact and to meet some of Australian rugby's greatest: Nick Farr-Jones, Simon Poidevin, and Peter Fitzsimons. And those fellows; what they taught me, as a young fellow growing up, was that what they could do on the paddock, I could emulate, if you like, off the paddock. They taught me that I had to turn to my own skills, my own abilities and use them and that's what I endeavoured to do. And they gave me the courage, as did Mum and Dad, to set about going to things like the local eisteddfods. So sure there was a flight of stairs that I had to get up, and Dad got me up there. And sure I had to go out to the middle of the stage to an audience, such as yourselves, who I had never met before, and say my piece, but it was about getting out there and getting exposed. And I guess that was me coming to terms with what I had, this genetic disorder.

And of course it's on your shoulder. It's always haunting you a little as you're growing up. You start to wonder what it all means for you, I think you're always aware of your mortality. We like to think that we're bigger than ourselves, especially me, I tend to think I'm 6 foot 10 and absolutely bullet proof, and if any one tries to tell me differently I usually tell them their absolutely wrong. I think when you have a genetic disorder that you are aware of that vulnerability, if you like, that inability to be in total control. You're tied to something which is outside yourself but which is also within you. But as I say, turning back to 'Australians all let us rejoice for we are young and free,' it's about being aware that we are in control of who we are, and the way in which we take the direction, and how we are going to cope with this genetic disorder or disability remains with us.

So I turn to AGSA and I look at what I have been able to enjoy on my journey and I remember in 1988, and it was a specific year because that was when I was finishing my HSC and at the end of that I was about to head to Sydney and begin my university career, which I would hope would allow me to enjoy law and participate in the roller coaster of life. And I was interviewed on a radio programme because I had been made school captain and won a public speaking competition. And the interview went very well on this particular radio station, 2UE. And sometime after that, a short time after that, Mum received a call from a mother who had recently given birth to a young fellow with Dystrophic Dysplasia.

Now she didn't have an AGSA to whom she could turn, but she was delighted to hear that I'd been able to achieve some things and was moving on as an 18 year old, if you like, robust and ready to take on the world. But when I heard that this mother had called and had been delighted by my story and been able to feel some hope and inspiration for her own little fellow, I, myself was wounded. I felt a little sad. I didn't think it was fair that another young fellow had to tread the path, take the journey that I had taken. It's not that I am not pleased with my life, I am. It's not as if I didn't want someone else to have the opportunities that that I have had, I certainly do. But I felt, Gee, you know, to think of another young fellow having to take it all on and be part of it and experience it. Wouldn't it be great if he was just of slender limb, strong and able to do it without any

cares. But after a time I thought well that's the dice that is thrown your way. I cope with it as others do. Fortunately AGSA is there to ensure that the path is a little more understood and the journey can take place.

In 1996 I was in my second year at the NSW DPP (Dept of Public Prosecution) and I received a phone call from a woman who'd seen me in the newspaper or in the courts or something and she wanted to catch up and have a chat because she had a son with dystrophic dysplasia. And sure enough she came in, and that son happened to be the same boy that had been mentioned to me by my Mum all those years ago when I was 18. And I was able to find out that he had gone to school, he was going extremely well, he was finding his own pathway through life and he was out to achieve some goals. She just wanted to know the simple things. How did I cope in high school and becoming aware, I guess, about adolescence and the difference between boys and girls and how you cope with those shifting shadows when things occur? So we had a good chat about that. She came to realize as I did, that it doesn't matter what you might be born with, it is all about what's within and the support around you and how you are able to take it on and achieve whatever goal it is you want to set.

AGSA has some huge goals. AGSA has set a great goal for themselves in being able to offer support, understanding and knowledge to the community who turn to them. So I also wish and echo her Excellency's words in wishing AGSA well for Genetic Awareness Week.

I think what we all have to be aware about with regards to our genetics is that sure we are made up of it, we have to make big decisions as to whether we will have other children or we have to do particular things to ensure that we can get the most out of our life and the most potential. But we should not feel as if we are tied to a history or a future which is already preordained. We are the making of our own future. We are young, we are free. So next time you sing that National Anthem regard it as really a personal anthem for yourselves. And with AGSA, I thank you for inviting me and I wish you all extremely well in your journey and lets all be aware of Genetic Awareness Week and aware of what we can achieve ourselves.

## VALE

## MATT LAFFAN

*Reprinted with kind permission of the author Peter FitzSimons and the Sydney Morning Herald. Tuesday March 3 2009.*

MATT LAFFAN, the disabled activist, lawyer with the Department of Public Prosecutions, Sydney lord mayor aspirant and well-known man about town, died peacefully in the intensive care ward of Royal Prince Alfred Hospital at 3pm on Sunday, with his devoted parents Dick and Jenny at his side.
It was the conclusion of a fiercely intense two-month struggle, and a lifetime's battle against the ravages of diastrophic dysplasia, which twisted his spine from a young age and restricted his breathing.
He was 38.

I was proud to call Matt a dear friend, and feel blessed that my children grew up around him.

He arrived in Sydney in l989 to study arts/law at Sydney University at the same time as his father Dick was coaching the NSW rugby team. He was a frequent and welcome presence around the NSW dressing rooms. Just before the Wallabies played the All Blacks in the crucial semi-final of the l991 World Cup, the Australian Captain Nick Farr-Jones read an inspirational letter to the team, penned by Matt. By that time, Matt was well on his way to becoming the Senior Student of the highly prestigious St John's College. No, he could never play rugby for John's but one of his proudest moments came at the end of a rather drunken soiree when his closest friends from John's used him as the ball to score a wonderful try at the southern end of St John's Oval in the week hours of a wintry morning. *Be* the ball, Matt.

After securing his law degree, he quickly found work with the Department of Public prosecutions and prospered, becoming a familiar sight in Sydney's legal district. As Matt's box, Nicholas Cowdery QC, said in a statement yesterday, "Matt was intelligent, quick-witted, industrious and possessed with both a keen sense of justice and an appreciation of the pressures that could lead to wrongdoing."

Though Matt needed help at the beginning and end of every day to get into and out of bed, and all the rest, he was able to do just that – moving into his own apartment in Castlereagh Street and living a fully independent life, which included a long stint with his attractive girlfriend. When he saw me chatting to another attractive female friend at the conclusion of the Stepping Stone charity function at the Westin Hotel in July last year, his wheelchair instantly became a Ferrari, so quickly did he surge

forward and carry her away. That strong charisma he had shone through when *Australian Story* featured him, in one of their most popular shows ever, and later when Andrew Denton interviewed him on *Enough Rope.*

Beyond his legal activities, he served on the judiciary of the NSW Rugby Union, was an activist on all things to do with disabilities, a passionate advocate of social justice and lived a full social life with family and friends. He was a wonderful godfather to Nick and Angela Farr-Jones's oldest daughter, Jessica.

Matt was a powerful orator. Six years ago, in the presence of Quentin Bryce, now our Governor-General, I heard him tell a wonderful story to 500 spellbound Queenslanders, where he recounted how on one occasion he had had what he considered to be a great legal triumph and had left the Supreme Court building feeling a million dollars. "And then," he continued, "I was rolling along Elizabeth Street, when I caught sight of my reflection in the window at David Jones and stopped….and stared. A funny-looking bloke in a wheelchair, three-foot high, with a massive chest….and I thought, "Who are you kidding?" Look at you. *Look at you.* Buy then what could I do? I kept rolling along, and crossed a street, and then I crossed another street, and kept rolling and going along the road…until I got to you here tonight."

And that was Matt all over. Yes, he had the odd down time, but mostly he just got on with it. Even as he lay dying over these past two wretched months, not once did he complain or even be seen to feel sorry for himself. Which is not to say he didn't dream that things might have been different.

"I once used to say to some mates of mine that all I really wanted was 24 hours without a disability," he told *Australian Story* in 2001. "I just wanted 24 hours so that I could do certain things. Since the, I've got a little greedy and now I want one week. And during that one week there'd be hell to pay, because Sydney just would not be big enough! I think the nightclub scene would be in a world of trouble, because dancing is something I'd really like to do. There'd certainly be a rugby match I'd have to get involved with. I'd go running with the old man. I think I'd pick Mum up and put her over my shoulder and run down the end of the block with her just to stir her up."
And that was Matt, too – fun, family oriented and with an extraordinary zest for life. Physically, life had dealt him a pair of red twos and a couple of black fives, but he always played like he was holding four aces. I think Matt would want his own epitaph to read: "No man ever had finer parents than Dick and Jenny Laffan." Which is true. But I think it should read: "No parents ever had a finer son than Richard Matthew Laffan.
**Vale, Matt.**
**Peter FitzSimons.**

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## *From AGSA's Contact Register – a personal contribution by Tarja Kelly.*

# My Worm

In 1875, Charles Darwin described a peculiar disorder that appeared in each generation of one family's male members, affecting some, sparing others. The mysterious condition became apparent in the very young, manifesting itself with "...small and weak incisor teeth...very little hair on the body...excessive dryness of the skin..."

128 years later my husband and I discovered these same symptoms and a few more in our son. After just over a year of trying to understand our newborn, who cried and screamed when swaddled, went limp in our arms on a warm day, grew his first pointy tooth at 15 months old and slept like an angel when stone cold, we finally found out he belongs to an 1 in 100,000 group.

Hypohidrotic Ectodermal Dysplasia affects one boy child in every 100,000 births. It is so rare that it only rates a paragraph this big (*fingers approx 3cm apart*) in our paediatrician's medical tome.

We learned most of the details about Ectodermal Dysplasia from researching it ourselves on the Internet. After seeing so many, often confusing explanations, I decided to come up with a simple way to explain it to my children. I found two ways. One came to me by talking to my eldest Sebastian. His innocent view of himself inspired this booklet, "I have HED". Hypohidrotic Ectodermal Dysplasia from the point of view of a 5 year old. The other was apples and bananas.

Remember them? Let me re-cap.

Women are made with two X chromosomes, men are made with one X inherited from their mother and one Y chromosome inherited from their father. For ease of understanding, X = Apple and Y= Banana

So it came that in 2003 I was told "Mrs Kelly, you have a worm in one of your apples and you passed it on to both your sons". My worm is the reason my boys have pointy teeth, no sweat, white fluffy hair and thin skin. Now let me explain how my worm was passed on to all 3 of my boys.

I have two apples, one of my apples has a worm in it but, but the symptoms of damage were masked by my normal apple. Every child I have has a 50/50 chance of being given the apple with the worm. There is a genetic test to find the worm but ours didn't show up at the most common address and remains unfound to this day. As a consequence this will also leave the question of where it began in our family unanswered. Effectively that means we may well be an, as yet, unclassified variety of the 170 variations of Ectodermal Dysplasia which have been identified to date.

When my husband handed over one of his bananas to make each of our boys, he counted on me to hand over an apple to make them whole. I lucked out and gave them the 'wrong' apple, which, in turn gave them Hypohidrotic Ectodermal Dysplasia.

The only way Sebastian, Matthew and Nathanael will avoid passing the wormy apple to their future children, is to have their embryos screened for gender. If they have only sons, the faulty gene in our family will end with their generation. If however they want to leave it to chance, every daughter they have, will be a carrier of the apple with the worm.

The worm is a malfunction in the Ectoderm. This is what forms the Skin, Hair, Teeth, Sweat glands and Nails. Even in my three boys the effects of the malfunction varies. For example, Sebastian's skin seems problem free but Matthew and Nathanael are plagued by severe eczema.

As for teeth, they had become a bit of a competition between my boys. Sebastian has 8 teeth. Matthew, who is 15 months younger and has 9 teeth, maintains that the extra tooth makes him the eldest. They won't get any more teeth, the average is 6 to 9. This is Matthew's smile (*pass photos*). He and Sebastian are fiercely proud of their 'shark teeth'. Last January however, Sebastian agreed to have his four front teeth capped to protect the tips. He'd had a mishap where one of his precious shark teeth had the tip snapped off. He has the tip to this day, locked secure in his toy safe so the tooth fairy won't take it.

The Educator magazine we receive from the American support group has stated that dentures won't necessarily prevent the loss of bone density in the jaw. That is one reason we aren't pursuing dentures anymore. The other reason is the distance and time it took to travel to a paediatric dental specialist at the Coffs Harbour outreach clinic. The mould would be made and by the time the denture got fitted at the next appointment the mouth would have grown a bit. The denture ended up being a wobbly fit. Would you wear that?

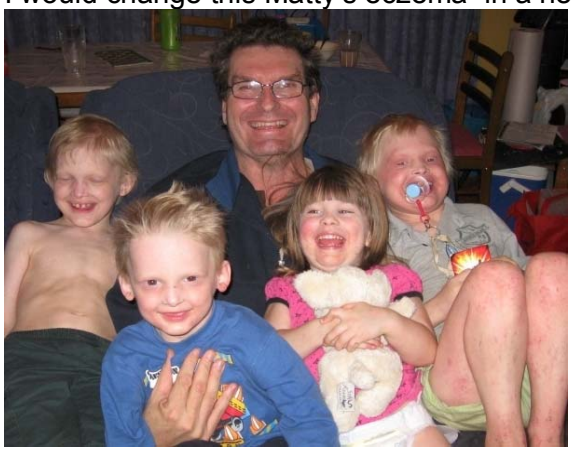
So once the boys are old enough we will look into the dental implant options available, but for now my priority is trying to find the one 'magic potion' to help with eczema.

Eczema has as yet not been officially linked to Ectodermal Dysplasia, however given the frequency and severity with which it shows up in ED children we strongly suspect it belongs.

We have two bookshelves worth of creams and lotions, pills and powders, liquids and oils. All of which can be taken or applied to this and that, but without the patients consent, cannot be tested. But that's a whole other lot of research and a whole other story!

I won't say the last 6 and bit years have been a walk in the park, but neither have they been filled with unmentionable hardship. I am always asked if I would change anything, well…

I would change this Matty's eczema in a heartbeat.



But I would NEVER change this -

## Soapbox

I have a bone to pick. A rather large one, so bear with me.

My family is big, I have three boys (on a bad day I have 4 but he doesn't like that. I suppose he *is* married to me) and a girl.

Lately, when I take all my children shopping with me, I get "attacked" by people trying to tell me what I can do better or could have avoided all together.

For example...
" Oh your poor boys. My little one used to have eczema like them. Oh no, nowhere near as bad as yours have it. They do look like they are in so much pain" (Note: Both Matthew and Nathanael, the boys in so much pain, are chasing each other around a play ride, squealing and laughing) "I used to use blah blah and it went overnight. Tell me, do you.....?"

In my head the resulting answer from me is something like this.

"Well, if your little one had eczema like mine get, and blah blah took it away overnight, how come I don't see your name on a jar of miracle cream? I just want to get milk and bread! Not a cure for something we have the upper hand on right now. I am happy, my family is happy! Bugger off and let me chase my kids to Woolies."

To the do-gooder I say,

"I wish I had brought a pen and paper. Sounds interesting, I might give it a go. Thank you so much, have a nice day."

Now, why would I do this to myself? Why do I practically invite progressive do-gooders to "assault" me every time I go out with the kids?

Yes, I kid you not!
Well, guess what...I *DID*!

Why should my family even attract them? Are we sending out "please help us for the love of god" signals? Can anyone tell me how to turn them off?

I have even been told that it is my fault the boys have Hypohidrotic Ectodermal Dysplasia.

I did not, however, poison my DNA by washing with soap, eating off plastic plates, using laundry detergent and dishwasher tablets. I did not break it by eating processed foods, brushing with Colgate or by using shampoo. I didn't drink, I've never smoked and I hold my breath when I use bug spray.

I gave my boys Hypohidrotic Ectodermal Dysplasia, because my mum gave it to me, *her* mother gave it to her and her grandmother got it from **her** mother!

So *PLEASE,* I am a mother of four, taking my kids to the shops for food, not for 101 miracle cures for eczema and HED!

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**The Australian Government** has announced a package called Making Ends Meet - Federal Labor's Plan for Older Australians, People with Disabilities and Carers.

Eligible Single Customers will receive a Telephone and Internet allowance of $33 every 3 months and $16.50 for each eligible couple.
Contact Centrelink for more information.

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

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**Subscription Year lst July – 30th June**

**ANNUAL SUBSCRIPTION**

| | |
|---|---|
| Individual: | $22.00 |
| Group/Organisation: | $44.00 |

**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****