AGSA
THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.
NEWSLETTER

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SYDNEY IVF

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CONTENTS

Battens Disease Centre of Excellence
Contact Corner
Hypophosphatasia
Profile A-Z Genetic Conditions
Gorlin syndrome
Personal story
FOP
Rare Treasures Support Group
BioNews
Conferences
Microarray Testing

MISSION STATEMENT

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

EDITORIAL

The next State election is now less than a year away so now is the time for us to raise awareness of the needs in the genetic community. The list is endless; respite care, group homes, better educational integration programs and more special schools to name a few. NGO's and support groups need to be listened to by local and federal politicians. In the next couple of months AGSA will be inviting support group's leaders and committee members to meet at AGSA to network and work together and decide how best to lobby the government for a fair go for people with a disability.

I must apologise the last newsletter did not have the usual issue number month etc. It was a case of the template having a mind of its own. Please note it was March 2010 Issue 90.

DATES TO REMEMBER

Genetic Disorders Awareness Week Launch will be on 15th September 2010 venue and speaker to be confirmed.

Orange
Caring for the Carers Seminar and Sibling workshop, 28-29th August 2010.

Williams Syndrome Picnic Day 29th August 2010 on the grounds of the Mater Dei School, Camden

Here is a website of one of our members whose baby was born without a pancreas - http://katmick.blogspot.com/2009/08/what-is-pancreatic-agenesis.html

A moving story so have a read.

AGSA is in the process of launching our new website so we will keep you posted.

We are also organizing with NCOSS a meeting for support groups on all the important issues of paperwork e.g. the Constitution as the Incorporation Act is being changed with big implications for groups.

Until next time

Best Wishes for the school holidays

DIANNE PETRIE OAM

CHIU LING LAU
PROJECTS MANAGER

I am sorry to say Chiu is leaving AGSA on 30th June and we will miss her and all her great ideas. Chiu has contributed greatly to AGSA and we wish her well in her new position at the Wellbeing Clinic as a Registered Psychologist. She wishes all the families she has worked with well and hopes to see them again at AGSA/FTV events. Congratulations Chiu.

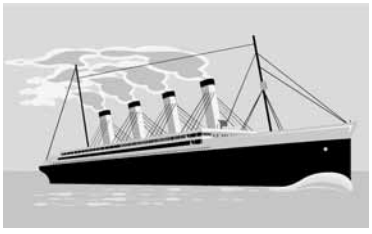
Elizabeth Pinkerton

11.07.1984 -13.04.2010

Elizabeth (Lizzie) was born with Tuberous Sclerosis Complex and Polycystic Kidney Disease long before genetic information was available to assist with diagnosis. She was severely disabled but enjoyed a great quality of life, participating in a wide variety of community activities despite her many challenges. She made an impact on all who met her, with her beautiful red hair, engaging smile and quirky sense of humour.

She is the inspiration for the commitment of her mother, Sue and her sister Clare to the Australasian Tuberous Sclerosis Society, a volunteer support group improving the lives of families affected by TSC.

Lizzie attended a number of the AGSA functions and her life was the topic of the occasional presentation by Sue! On these occasions you could hear a pin drop. Lizzie was a very determined young lady and she will always be remembered by those who had the pleasure of meeting her. Everyone at AGSA and our members send our love and support to Sue, Ross and Lizzie's wonderful brothers and sisters and grandma in their sad loss.



Batten Disease Centers of Excellence

BDSRA is proud to announce the development and implementation of two Batten Disease Centers of Excellence. Batten Disease Centers of Excellence are available to all families interested in receiving treatment from a team of professionals who have experience dealing with and treating all forms of Batten Disease. Children will be seen by a lead neurologist as well as other healthcare professionals like a nurse, social worker, genetic counselor, etc. A child may also have the chance to receive a consultation from other medical professionals like a cardiologist, pulmonologist, gastroenterologist, physical therapist, etc.

The first two Centers of Excellence are located at Massachusetts General Hospital in Boston, MA and Nationwide Children's Hospital in Columbus, OH.

Each Center of Excellence has a designated "Clinic Day" where appointments are only available to individuals with Batten Disease. These Clinic Days are available each month. Massachusetts General Hospital will have their Clinic Day on the SECOND MONDAY of each month. Nationwide Children's Hospital will have their Clinic Day on the THIRD THURSDAY of each month.

BDSRA is also working with the University of Rochester Medical Center to finalize the plans for their Center of Excellence. Information will be announced when it becomes available. BDSRA has hopes to expand this program to other hospitals around the country.

For more information about the Centers of Excellence, please contact BDSRA

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.

Hypophosphatasia

A lady with this condition would like contact with others. She knows of 4 other people in Australia. This is a soft bone disease due to a low level of the enzyme alkaline phosphatase.

Gorlin syndrome

Please read A-Z of genetic conditions.

AGSA's Rare Treasures

A Chromosomal Disorders Support Group

Represents 250 people who have a child with a rare chromosomal abnormality. AGSA works closely with Unique UK who provide AGSA with fact sheets and regular newsletters. Please contact AGSA if you are trying to find another family with the same or similar chromosomal abnormality or would like information on your child particular condition.

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.

First synthetic cell created in a laboratory

24 May 2010

By Lux Fatimathas

Appeared in BioNews 559

For the first time artificial life has been created in a laboratory, in the form of a bacterium. US researchers have chemically synthesised the DNA (deoxyribonucleic acid) of the simple bacterium Mycoplasma mycoides. This entirely man-made genome was transferred into a different bacterium and resulted in the creation of new Mycoplasma mycoides cells controlled solely by the artificial genome.

'It is pretty stunning when you just replace the DNA software in a cell and the cell instantly starts reading that new software and starts making a whole new set of proteins, and within a short while all the characteristics of the first species disappear and a new species emerges.

That's a pretty important change in how we approach and think about life', said Dr Craig Venter, who led the study at the J Craig Venter Institute, US.

The study, published this month in Science, details how the artificial genome of Mycoplasma mycoides was designed using smaller stretches of DNA sequence ordered from a company. 'Watermark' DNA sequences were added in order to identify this genome as synthetic. The DNA sequences were then assembled together using yeast. The newly synthesized genome was transplanted into Mycoplasma capricolum, which went on to divide. Daughter cells were then identified that were exclusively using the artificial genome and had been converted into Mycoplasma mycoides.

This cell we've made is not a miracle cell that's useful for anything; it is a proof of concept. But the proof of concept was key; otherwise it is just speculation and science fiction. This takes us across that border, into a new world', said Dr Venter. The long-term goals of this research are to create cells with specific functional applications, such as the mass production of pharmaceutical drugs and the ability to clean up oil spills.

Although the future applications of such artificially designed organisms could be beneficial, there are concerns over the misuse of this technology. Professor Julian Savulescu, an ethicist at Oxford University, commented that this technology '...could be used in the future to make the most powerful bioweapons imaginable'.

In an interview aired by ITN Dr Venter said: 'It is clearly a dual use technology, as most modern technologies are, that can be used for negative purposes or positive ones. In all our reviews of this with government officials, the view is that this is maybe a linear increase in the negative potential but an exponential increase in the tools to do good and ensure a healthy, long-term survival for our planet'.

SOURCES & REFERENCES

Artificial life: Have scientists created a monster?
The Mail | 21 May 2010
Scientists create artificial life in laboratory
The Times | 21 May 2010
Scientists create artificial life: World first as genome pioneer makes designer microbe from scratch
Mailonline | 21 May 2010
Technology allows humans to sidestep evolution
The Times online | 21 May 2010

RELATED ARTICLES FROM THE BIONEWS ARCHIVE

The perils of creating synthetic life
24 May 2010 - by Dr Gabrielle Samuel
As an ex-genetic researcher I was incredibly excited to hear in last week's news that researchers at the J Craig Venter Institute, US, have successfully constructed the first self-replicating, synthetic bacterial cell....[Read More]

First synthetic biology code of conduct launched
21 December 2009 - by Rachael Panizzo
There is a risk that advances in synthetic biology and low-cost DNA sequencing and synthesis could lead to the misuse of genetic technologies for bioterrorism purposes, where sequences of DNA could be ordered from a commercial gene synthesis provider and genetically engineered into a biological warfare agent....[Read More]

Birth certificates: a new era?

30 April 2010

By Louisa Ghevaert
Louisa Ghevaert, partner at Gamble and Ghevaert LLP (www.gambleandghevaert.com)

Appeared in BioNews 556

Birth certificates have been a hot topic in the UK in recent weeks.

There has been much controversy, confusion and misunderstanding, aptly shown by Caroline Gammell's article in The Daily Telegraph newspaper and Colin Fernandez's article in the Daily Mail on 19 April incorrectly hailing the advent of the first lesbian couple to both be named as parents on their baby daughter's birth certificate, born 31 March this year.

Lesbian couples have not in fact had to wait until the beginning of April this year to take advantage of new laws allowing them both to be named as the parents on their child's birth certificate. Legally, they could both be named as the parents on the birth certificate of any child they conceived on or after 6 April 2009, when the new female parenthood provisions of the Human Fertilisation and Embryology (HFE) Act 2008 were implemented. In practice, as from September 2009 lesbian couples could be named on their child's birth certificate following a viable delivery,

albeit at that stage heavily premature. Natalie Woods and Betty Knowles were not therefore the first in the UK to take advantage of this change in the law (just the first to speak to the press) and we have many clients who have preceded them.

This misunderstanding is undoubtedly the result of increasingly complex laws on parentage and the recent press coverage of the final staged implementation of the HFE Act 2008 on April 6 this year, allowing same sex couples (most notably two men) for the first time to be named as the parents on their surrogate-born child's birth certificate following the grant of a Parental Order which then triggers the re-issue of their child's birth certificate.

The Daily Telegraph article went on to quote Baroness Deech, The Chairman of the Bar Standards Board, saying: 'This is not a moral issue; it is about disguising true facts, and it is about confusing biological parenthood, legal and social parenthood.' The article further quoted Josephine Quintavalle, from Comment on Reproductive Ethics (CORE), saying that 'birth certificates should reflect how a baby is generated' and 'in a culture that is obsessed with genetics, it is strange that when it comes to birth certificates we are prepared to forget all that?' and 'as much as you try to play around with the terminology, the biology reflects what has happened and one day the child will ask about their father.'

Birth certificates are in fact a legal document recording the legal parentage of a child at birth. Birth registration procedures are governed by law, not biology, and birth certificates have never been in practice a definite record of a child's biological parents. In non-assisted reproduction cases, a married mother's husband is presumed to be the father and recorded as such on the child's birth certificate unless this is rebutted by DNA (deoxyribonucleic acid) evidence of another's paternity. If the child's mother is unmarried, she can choose whether to name the father or to leave the father's details blank.

Since the introduction of the HFE Act 1990, the UK's first legislation to regulate parentage following assisted reproduction treatment and the precursor of the HFE Act 2008, men and women have also been routinely named as parents on the birth certificates of their children conceived with donor eggs or sperm. Birth certificates of surrogate-born children also continue to record the surrogate mother as the legal mother at birth and her husband as the child's legal father, even when the intended parents are the biological parents and which then requires them to obtain a post-birth Parental Order to re-assign legal parentage (which can only be done with the surrogate's consent) and trigger the re-issue of the birth certificate.

With the growth of alternative family structures and recent changes in the law, increasing demand for assisted reproduction treatment using donor gametes and more people building their families through surrogacy, birth certificates will increasingly reflect legal parentage rather than biology. Whilst birth certificates were in the past perceived as representing a child's biological parenthood, this has not necessarily been the case. What has changed over the last year is the stark realisation amongst some campaigning groups that birth certificates can now in law name a wider range of people as parents on the face of a child's birth certificate, most notably same sex parents, challenging traditional values and perceptions of family life.

CONFERENCES

11th International Symposium on Mucopolysaccharide and Related Diseases

Adelaide South Australia
June 23-25th 2010.
"Translating Research into Clinical Reality"
www.mps2010.com.au

Mucopolysaccharide & Related Diseases Society Aust. Ltd, Lysosomal Diseases Australia and Lysosomal Diseases New Zealand are hosting the 11th International Symposium on Mucopolysaccharide and Related Diseases in Adelaide on 23 – 27 June 2010. The organising committee warmly invites you to join us at the Adelaide Convention Centre for this event.

The theme for the symposium is "Translating Research into Clinical Reality" Our scientific and family programmes will be exciting, and relevant with a focus on the areas of newborn screening, prognostics, understanding pathology and therapeutic options. Genuine opportunities for thorough discussion and debate will be a feature of the program - not only for the academics but also the families.

The organising committee hope you will be able to join us for five exciting days of cutting edge science, exciting family experiences and an enjoyable cultural experience.

For more information and registration details please visit www.mps2010.com.au or email wendy@mpssociety.org.au

The Annual Australasian Tuberous Sclerosis Seminar Day

will be held at Sydney Children's Hospital, Randwick
on Sunday August 22, commencing at 9am
with the 29th Annual General Meeting.

Guest speakers will present Seminars covering a range of relevant TSC topics including: An Australian study: are surveillance guidelines being met?, Establishing a Clinical Trial, the Impact of TSC on Adult Learning and Behaviour, the Impact of Epilepsy on Learning and a session for Carer's on Looking after Yourself.

Further details from Hayley on 99200261 or www.atss.org.au for registration details.

XIth Meeting of the International Patient Organization for Primary Immunodeficiencies (IPOPI) –

Istanbul, Turkey – 6-9 October 2010

The XIth Meeting of the International Patient Organization for Primary Immunodeficiencies (IPOPI) will be held in Istanbul in

October in conjunction with the XIVth Meeting of the European Society for Immunodeficiencies (ESID) and the IXth Meeting of the International Nursing Group for Immunodeficiencies (INGID).

IPOPI would like to invite submissions for abstracts in a variety of areas including quality of life and long term management. The deadline for abstract submissions is 1 June 2010.

PROFILE
A – Z OF GENETIC CONDITIONS

This fact sheet is kindly provided by the Centre of Genetics Education, Sydney

GORLIN SYNDROME

Also referred to as:
Nevoid Basal Cell Carcinoma Syndrome, Basal Cell Nevus Syndrome, Gorlin-Goltz Syndrome

Gorlin syndrome is a genetic condition that is characterised by the development of multiple jaw cysts and non-cancerous skin cancers called basal cell carcinoma (BCC) that are not usually due to external causes.

Gorlin syndrome is highly variable and affects people in different ways. The condition has many different symptoms. Life expectancy for people with Gorlin syndrome is not significantly different from other unaffected individuals.

The main features associated with Gorlin syndrome are:

- **Multiple jaw cysts** (odontogenic keratocysts). These are usually painless swellings but if left untreated can cause disruption of tooth development and fracturing of the jaw. These do not usually appear until after 20 years of age.
- **Multiple skin lesions called basal cell carcinoma** (BCC). These lesions sometimes called nevus or nevi are usually found on the skin or in the mouth and are not usually due to external causes. They typically appear around 20 years of age.
- **Hardening of the soft tissue between the hemispheres of the brain** (calcification of the falx). Approximately 90% of affected individuals develop this symptom.

Other features of Gorlin syndrome, some of which are very rare, include:

- **Larger than average head size** (macrocephaly)
- **Abnormalities of the skeleton such as split ribs** (bifid ribs), wedge-shaped vertebrae and protruding forehead and/or jaw bone
- **Pits** that appear as pink "pin prick lesions" or white dents on the palm of the hand or the sole of the foot (Palmar or plantar pits)
- **Development of other tumours** such as ovarian tumours or cardiac (heart) tumours
- **Brain tumour** called childhood medulloblastoma. This rarely occurs with only 5% developing this tumour
- **Eye problems** such as cataracts
- **Cleft lip or palate**
- **Additional thumb or finger** (polydactyly)

WHAT CAUSES GORLIN SYNDROME?

Gorlin syndrome is caused by a variation (mutation) in the PTCH1 gene, which provides instructions for making proteins that are involved in the development and suppression of cell growth. It is located on chromosome 9q22.3 and follows an autosomal dominant pattern of inheritance. About 20%-30% of affected individuals are the first to be affected in their family. When this happens, it is referred to as sporadic. Recently researchers have identified that in a few cases, Gorlin Syndrome may also be caused by a variation (mutation) in the PTCH 2 gene (located at 1p32) or the SUFU gene (located at 10q24-q2). These genes work together with the PTCH1 gene and many others, in the development and suppression of cell growth.

The PTCH gene is highly penetrant, which means that if you inherit a gene mutation you will have some form of the condition. However the gene also has variable expressivity which means that the symptoms of the condition will vary in the same family.

Genes, chromosomes and genetic conditions In all the cells of our body, our genes are found on chromosomes (long strings of genes). We have many thousands of genes that provide information for our body to grow and develop, and remain healthy.

There are usually 46 chromosomes in each cell that are arranged into 23 pairs. One of each pair is passed on from our mother and the other from our father. 22 of these chromosome pairs are numbered. These numbered pairs are known as the ***autosomal chromosomes***. The 23rd pair is made up of the ***sex chromosomes***. If the person is a male, there will be an X and a Y chromosome or if they are female, there will be two copies of the X chromosome.

Everyone has variations (changes) in their genes, which is why we are all unique. Most gene variations don't cause problems however; some variations in the genetic information make the gene faulty and are called ***mutations***. We all have a small number of faulty genes but that does not mean we will develop a genetic condition as a result of this.

Faulty genes do not work as they should in the body and are unable to provide the correct information to our cells. A faulty gene can either be inherited from one or both parents or can occur during the creation of an egg or sperm or at conception. Once a faulty gene is present in an individual, it can be passed on to future generations. This is referred to as ***genetic inheritance***.

The three most common patterns of genetic inheritance are known as ***autosomal recessive inheritance, autosomal dominant inheritance and X-linked recessive inheritance***. Gorlin syndrome follows an autosomal dominant pattern of inheritance.

WHO IS AFFECTED BY GORLIN SYNDROME?

Gorlin Syndrome affects males and females in equal numbers. There have been only a few studies estimating the prevalence in some parts of the world. For example, Gorlin syndrome is estimated to occur in at least 1 per 57,000 individuals in the United Kingdom. An Australian study in 1994, found that the incidence of Gorlin syndrome was at least 1 in 164,000 births. The true figure may be higher as milder cases may not have been recognised.

HOW IS GORLIN SYNDROME DIAGNOSED AND TREATED?

Diagnosis

In most people, the diagnosis of Gorlin syndrome is based on whether an individual has specific clinical symptoms associated with the condition.

Treatment

The treatment of individuals with Gorlin syndrome will be directed towards the needs of each individual. Just like each individual will be different, the treatment plan will be unique and best discussed with the health professionals involved in their care.

Specific treatment, management and prevention strategies for Gorlin syndrome include:

- Surgical removal of jaw cysts
- Early treatment of skin cancers (basal cell carcinomas). Chemical peel or surgery is often used to treat basal cell carcinomas.
- Management of symptoms eg monitoring tooth eruption.
- Sun protection - this is a priority as it is highly likely that basal cell carcinoma will develop
- Avoidance of radiation therapy where possible - it is recommended that all original x-rays are kept and that x-rays are performed only when essential.
- Physical and developmental assessment routinely for anomalies and tumours
- Monitoring head circumference through childhood

Genetic Testing Options

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

Gorlin Syndrome Group

11 Blackberry Way. Penwortham. Preston Lancashire PR1 9LQ. United Kingdom.
(website): www.gorlingroup.org
(ph): +44 1772 496849
BCCS Life Support Network.
PO Box 321 Burton, OH 44021. United Kingdom.
(website): www.bccns.org
(ph): 440-834-0011 (email): info@bccns.org

A PERSONAL STORY

Dear Fellow Gorlin Syndrome Traveller

My name is Anna and I now live in Melbourne. In my early teen age years I was found to have several large jaw cysts. Over the next few years I had quite a few removed in Eastman's Dental Hospital, or the National Temperance Hospital in London. I came to Australia in l965 and had a few more removed by Specialists in Melbourne. In l968 I started to have "spots" attended to by a Dermatologist. He did a good job but I didn't tell him about the ones he could not see! In l977 he tried radium on a difficult spot on my nose. The area did not heal so a year later he referred me to a plastic surgeon who warned me that I had to be very careful as he found cancer under the area which had been treated with radium. A friend advised me to go to Peter Mac Callum Hospital.

Fortunately for me Mr. Simon Donahoe was on duty the day I first attended and he connected the jaw cysts I had as a child and the multitude of bccs which I presented with. The late Mr. Peter Gibbs was the Registrar on duty. They decided to wait until Mr. Graeme Southwick returned from his studies under Prof. Gorlin to operate early in May l979. Thus began my journey under the banner "**Gorlin Syndrome Patient**".

Since l979 I have had regular treatment. In my case, three different types of creams were unsuccessful in treating the BCCs as was cryotherapy. Surgery and Photo Dynamic Therapy (PDT) have been more successful. Whilst taking part in the trial of Isotretinoin I met many Gorlin Syndrome Patients but only kept in touch with one named Jill. We did think at the time of starting a support group but I was not in a position to do so. There is no doubt that the condition affects each of us in different ways, and our response to the different treatments can also vary, so each patient has to find out what works best for them. We can share tips and stories of coping with this challenging condition.

Late last year I contacted Jill to see if she was interested in starting a group with me. We are not offering any professional support. This is best obtained from your own practitioners. The type of Support Group we have in mind is one "which allowed people in similar circumstances to share experiences". Professor Georgia Chenevix-Trench of Queensland institute of Medical Research helped us get in touch with patients around Australia. At this stage we intend staying in touch by email as the people on my mailing list are from Cairns, Newcastle, Brisbane, Adelaide and Melbourne.

My email address is brianna@netspace.net.au. Please put "Gorlin Syndrome" as the subject heading on any email. Naturally I would love to hear from anybody who is interested in joining us.

We are also considering trying a type of chat session on the internet. Dianne Petrie of The Association of Genetic Support of Australasia Inc. (AGSA) has kindly offered to run a workshop with Professional Guest Speakers in Sydney some time in the future.

Thank you for taking the time to read my story.

Kindest regards

ANNA

AGSA will be organising a seminar on this condition in the near future.

RARE DISEASES

GSK launches new specialist unit to research and develop medicines for rare diseases

Issued: Thursday 4 February 2010, London UK

GSK today announces the formation of a new standalone unit specialising in the development and commercialisation of medicines for rare diseases.

Over 5,500 rare diseases have been identified1 of which less than 10% are currently being treated2, presenting a significant unmet medical need. Despite the rarity of each condition, the number of diseases means that between 6-8% of the population3 may be affected by a rare disease. Many are genetic in origin, start in childhood and cause lifelong debility and premature death.

Operating under a lean structure, Marc Dunoyer, GSK's President of Asia Pacific and Chairman of Japan, will lead this new operation, working closely with Patrick Vallance, GSK's Senior Vice President of Drug Discovery. The new unit will seek to leverage existing capabilities and partnerships and establish further in-licensing opportunities. During 2009, GSK entered into strategic collaborations with two specialist companies, Prosensa and JCR Pharmaceuticals.

The alliance with Prosensa, announced in October 2009, focuses on nucleic acid based therapeutics, correcting gene expression in diseases with large unmet medical needs. The scope of the alliance includes four RNA-based compounds intended to treat specific, but different, subpopulations of patients suffering from Duchenne Muscular Dystrophy (DMD).

As part of the agreement with JCR Pharmaceuticals, a Japanese developer and manufacturer of bioactive products, GSK has obtained global rights to a number of enzyme replacement therapies that could, upon approval, be used to treat orphan diseases such as Hunter syndrome, Fabry disease and Gaucher disease.

Marc Dunoyer, GSK's President of Asia Pacific and Chairman of Japan, said: "In addition to our existing discovery effort, alternative opportunities need to be explored to make treatments available for rare diseases. This complementary approach will combine our existing global expertise with specialist partners .Overtime, this new unit has the potential to deliver multiple therapies responding to high medical needs of underserved populations of patients."

Patrick Vallance, GSK's Senior Vice President of Drug Discovery, said: "The entry into this new therapeutic area forms part of GSK's strategy to deliver more products of value and improve returns in R&D through a focus on areas with a higher probability of success. The risk associated with product discovery and development in rare diseases is generally lower than other disease areas as disease definitions are very clear and clinical trials tend to be small with robust endpoints. In most cases the molecular target is known, making it easier for specialised physicians to diagnose patients"

GlaxoSmithKline – one of the world's leading research-based pharmaceutical and healthcare companies – is committed to improving the quality of human life by enabling people to do more, feel better and live longer.

For further information please visit:- http://www.gsk.com

PRESS RELEASE

http://www.ox.ac.uk/media/news_stories/2010/100415_1.html

Donation enables research into a disease that turns muscle to bone Health
15 Apr 2010

A UK research group dedicated to work on a rare genetic condition that turns muscle to bone will be established thanks to a donation that takes the University of Oxford's fundraising campaign, Oxford Thinking, past the £800m mark.

Richard Simcox chose to donate to Oxford University because it is the only institution in the UK which researches fibrodysplasiaossificans progressiva, or FOP.

FOP is a progressive disease in which new bone forms in muscles around bones and joints in the body. It is a very rare condition with no cure. There are about 45 known cases in the UK. Patients become gradually more disabled as extra bone forms across joints, restricting movement and locking joints in place – sometimes in uncomfortable positions.

In FOP, the formation of new bone often follows inflammation as a result of injury or trauma. No one knows why inflammation or 'flare-ups' in FOP patients leads to bone formation. And immobilised joints can't be released - any surgery would exacerbate the condition. The only treatment - other than trying to avoid injury - is corticosteroids, which appears to help in the early stages of FOP. It doesn't stop progression of the disease but may ameliorate the condition.

Richard Simcox's donation, via his company Roemex Ltd, will fund the first dedicated UK programme to study the gene responsible for FOP and how drugs can be developed to halt or prevent the disabling bone formation. He has given £110,000 towards two postdoctoral positions working on FOP, and has guaranteed to underwrite the remaining £220,000 cost of the three-year research posts.

Marian Granaghan, mother of Seanie Nammock, 14, who has FOP, said: 'The donation is great news. Every morning on the news there are unbelievable breakthroughs in the field of genetics.

The research at Oxford offers a chance there might be one in FOP in the future to help Seanie and others affected by FOP.'

Marian, Seanie, and their family and friends have separately raised £15,000 for Oxford research.

Richard Simcox is founder, managing director and main shareholder of Roemex, a firm based in Aberdeen that supplies specialty chemicals to the oil and gas industry. He is also president of Action FOP UK, a forum for those with the condition.

He hopes that his donation will raise awareness of the rare condition and stimulate further gifts and fundraising efforts. He says: 'I am delighted to see something happen in FOP on these shores. At Oxford, we'll be able to get something going, something real and tangible. Now that the gene for FOP is known, researchers can focus on looking for a cure.'

Professor James Triffitt of the Botnar Research Centre at Oxford University, who researches FOP, says: 'It is very difficult to get research grants to study rare diseases, so the generosity of individual donors is what keeps work like this going.

'This donation will make a great difference. It will create a research nucleus in Oxford to understand the changes that occur in FOP and search for potential therapies. Without it, it would be impossible to have this intense research activity on FOP, one of the most disabling conditions that any patient can get.'

In 2006, Professor Triffitt and colleagues at Oxford were part of an international collaboration that identified the gene responsible for FOP. In FOP, a mutation turns the gene on which leads to the unwanted bone formation.

Marian Granaghan and her daughter Seanie Nammock, who suffers from FOP.Since the gene was identified, there has been significant research progress. The gene encodes a protein which is involved in a number of processes in cells in the body that eventually lead to bone formation. It has been shown in mice that it is possible to stop the action of the protein. The two new researchers at Oxford will start projects to understand the structure of the protein, how drugs can be best designed to stop its action, and carry out the cell biology necessary to test whether any new, early-stage drug candidates are working in the right way.

Dr Alex Bullock of the Structural Genomics Consortium at Oxford University, who will be supervising the new research projects along with Professor Triffitt, explains: 'The difficulty is that the protein involved is only one of 500 similar proteins in humans, and we want to affect this one and only this one. That's the challenge.'

Oxford Thinking: The Campaign for the University of Oxford is the largest fundraising campaign in European university history, with a goal of a minimum of £1.25 billion to support world-class research, teaching and facilities. The donation takes the Oxford Thinking campaign over £800m, under six years after fundraising began.

Professor Andrew Hamilton, Vice-Chancellor of Oxford University, says: 'Richard Simcox - like so many other generous donors - identified Oxford as the best place to address the challenge that mattered to him: finding a cure for a devastating disease.

'His generosity takes our fundraising campaign past the £800m milestone, and this funding will allow us to invest in a whole range of research that tackles the pressing needs of the 21st century and advances human knowledge. It will also help support and teach the brightest minds of the next generation.'

Those wishing to donate to FOP research at the University of Oxford can go to

http://www.giving.ox.ac.uk/academic_departments/medical_sciences/fop_research.html

It is very difficult to get research grants to study rare diseases, so the generosity of individual donors is what keeps work like this going.

Professor James Triffitt, Botnar Research Centre Research could benefit those with FOP

The FOP research now being funded could end up benefiting people like Seanie Nammock, 14, of West London.

Seanie Nammock is 14 years old. She likes make up, clothes and loves cooking. She listens to Lady GaGa, Beyonce, and Lily Allen and is to be a bridesmaid at her cousin's wedding. She's done one of her Science GCSEs two years early and dreams of being a vet.

In the summer of 2008, Seanie had a minor accident on a trampoline. A couple of weeks later, a large lump developed on her back. After a lot of going back and forth to the hospital, she was diagnosed with FOP in September 2008.

Seanie says, 'When I was eventually told what my condition was and what the outcome could be, I just thought "Oh well, I just have to get on with my life and I won't let it stop me from doing what I want."

Her neck now has little movement and her left and right arms have locked, restricting what she is able to do.' I can't brush my hair and style it anymore, and my mum is pretty rubbish at doing it!' says Seanie. 'My mum also helps me to get dressed.

'Sometimes I do get fed up when I can't do things but it normally doesn't last too long. You just have to find ways around things or just ask for help.'

Marian Granaghan, Seanie's mum, describes how she felt when they received the diagnosis of FOP: 'I felt numb. I just went onto autopilot. The hurt and grief was indescribable - it was a pain worse than anything I'd ever felt in my life. As time has passed, she says she's gained more acceptance of the situation. 'Our lives have changed, so we have to get on with it as best we can. We live for each day and I try not to think too much about the future.

"Seanie on the other hand has been amazing and accepts this has happened to her. She feels she should be able to do whatever she can for herself, and she keeps telling me "You have to let me get on with things, if I want help I'll ask for it.""

Like many people affected by FOP and their families and friends, Seanie and Marian have focused on raising awareness of FOP and raising funds for research at Oxford University. They talked about their story in the media in April last year and they have succeeded in raising £15,000 for Oxford.

Marian says, 'I am constantly thinking of things we can do or people we can contact to raise money for research, because this condition is so rare it is down to the families and friends to help raise funds so research can be ongoing.

'This is my dream: one day there will be a cure or, if not a cure, a tablet that stops bone growth. This would mean that Seanie and everyone affected with FOP can be operated on and the excess bone taken away to get their movement back.'

Resources

www.ifopa.org
Fundacion FOP
Rare disease in Argenentina: a social, sanitary, legal and scientific research approach.
To promote research and the improvement of people with Fibrodysplasia Ossificans Progressiva.
Email: moira.liljesthrom@gmail.com

Microarray-based comparative genomic hybridisation (array CGH)

What are chromosomes?

Chromosomes are the structures in each of the body's cells that carry the genetic information (DNA) that tells the body how to develop and function. They come in pairs, one from each parent, and are numbered 1 to 22 approximately from largest to smallest. Each person has another pair of chromosomes, called the sex chromosomes. Girls have two Xs (XX), whereas boys have an X and a Y chromosome (XY). Each chromosome has a short (p) arm and a long (q) arm.

Looking at chromosomes (chromosome analysis)

Chromosomes cannot be seen with the naked eye, but if you stain them and magnify them many hundreds of times under a microscope, you can see that each one has a distinctive pattern of light and dark bands. By looking at your child's chromosomes in this way, often referred to as karyotyping, it is possible if the change is large enough to see if there is a chromosome imbalance (loss or gain of chromosome material) or if the chromosome is rearranged in any way. However, because the amount of material gained (duplicated) or lost (deleted) can often be very small and impossible to see on a routine chromosome test, you may have been told your child's chromosome analysis was normal. A new more enhanced test now available for looking at chromosomes is called a microarray-based comparative genomic hybridisation (array CGH) test.

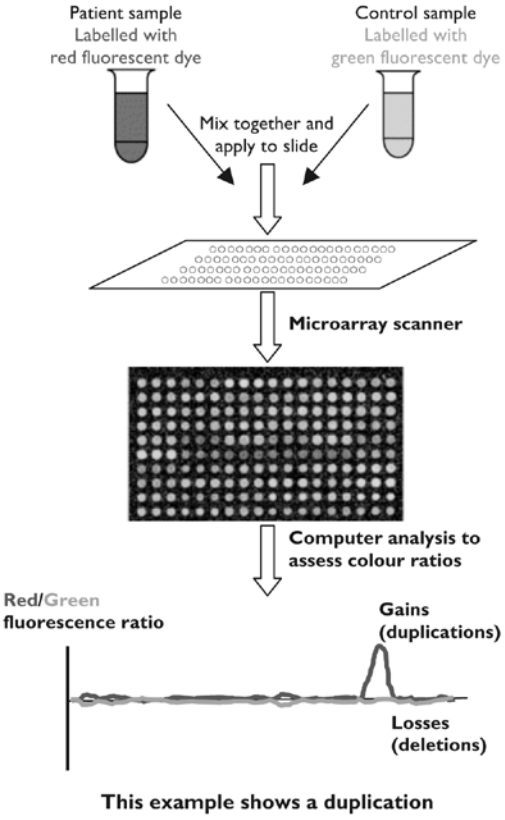
What is array CGH?

Array CGH is an advance in technology that allows detection of chromosome imbalances that are smaller than can be detected through looking down the microscope. Karyotyping is only as good as the resolution of the microscope and is not able to detect subtle chromosome changes. These smaller alterations, often called submicroscopic alterations because they cannot be seen through the microscope, can still disrupt development. These very small changes are often called microdeletions and microduplications. Array CGH is also sometimes called CGH array or simply microarray.

Array CGH compares your child's DNA with a control DNA sample and identifies differences between the two sets of DNA. In this way, deletions or duplications (imbalances) in your child's DNA can be identified. From this, the gene content of any such imbalance can be established.

Principles of array CGH.

The patient and control DNA are labelled with different coloured fluorescent dyes (red and green below) and applied to an array (a solid surface, usually a microscope slide on to which a number of short stretches of DNA are spotted). The samples bind to the DNA on the slide. Where there is no change between the patient and control sample there will be equal binding and therefore equal amounts of red and green fluorescence. For regions where there is a duplication in the patient sample there will be more red fluorescence than green; conversely deletions will result in reduced red fluorescence and more green fluorescence.



This example shows a duplication

What samples are needed for array CGH testing?

Array CGH can be performed on a blood sample from an adult or child. It can also, less frequently, be performed on prenatal samples, for example amniotic fluid from an amniocentesis or the chorionic villus (placental tissue) from chorionic villus sampling (CVS).

Why has array CGH been offered for our child?

Your doctor or geneticist may consider array CGH testing if your child has problems with learning, physical development,

behaviour or birth defects or medical concerns such as seizures. Recent studies have shown that around 15 per cent of children with unexplained learning and/or developmental disability will have chromosome changes that could not be detected by conventional chromosome analysis but can be detected through array CGH.

How will we be given the results?

The results are likely to be given to you by your geneticist who will talk you through your child's results. You will almost certainly then receive a follow-up letter. Alternatively, you may receive a preliminary result from the doctor doing the test and then refer to geneticist (if appropriate).

How long do the results take?

Results are usually available in 6-8 weeks. Testing a newborn baby with multiple problems is considered a priority and therefore results may be available slightly sooner.

What are the advantages of array CGH?

- All 46 chromosome can be examined in a single test
- More sensitive and accurate than conventional karyotyping
- A diagnosis from array CGH may avoid your child having to undergo many other tests
- It can reveal which specific genes are included in the deletion or duplication
- It can be useful to further define breakpoints in imbalances that are already known

What are the benefits of array CGH?

- It may help you and your doctor watch for common health problems associated with your child's chromosome imbalance
- It may help to predict what to expect as your child gets older
- It may show which specific genes are included in your child's deletion or duplication. If the gene(s) has been associated with a particular feature or health problem it may help to guide management or treatment for your child
- It can help you to obtain specialist services for your child
- You can choose to join a support group to meet other parents facing similar challenges
- Parents and other family members can be tested to see if they are carriers of changes in their DNA that put them at risk of having more children with a chromosome change

What are the limitations of array CGH?

- Some chromosome or DNA changes cannot be detected by array CGH (for example very tiny changes to the DNA or rearrangements that do not result in any loss or gain of DNA material)
- It may identify chromosome changes known as copy number variants (CNVs). These changes are common in the general population and are often completely harmless. But sometimes a CNV can affect health or development. CNVs can make interpreting an array CGH difficult so the parents may need to be tested to help interpret the results. This will be discussed by your local paediatrician or genetics specialist

Families say …

"Our geneticist used an analogy which made things clearer for us. He said that previous test results were like an old-fashioned map of the world which showed just a wide overview (country level) and that doing an array is more like using Google earth which allows us to zoom in much more closely, even down to street level, to give a closer and clearer idea of which genes, if any, are missing or duplicated.

"Without array CGH we wouldn't know what my son had. Now that we know, it has made us look forward and get on with our lives.

Unique

Support and Information
Rare Chromosome Disorder
Support Group
PO Box 2189, Caterham,
Surrey CR3 5GN, UK
Tel/Fax: +44 (0) 1883 330766
info@rarechromo.org
www.rarechromo.org

EuroGentest

Produced with the support of EuroGentest (www.eurogentest.org)

This leaflet is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication. It was compiled by Unique and reviewed by Dr Shehla Mohammed and Dr Caroline Ogilvie, Guy's Hospital, London. UK and Professor Maj Hultén BSc, PhD, MD, FRCPath, Professor of Reproductive Genetics, University of Warwick, UK. 2010

Rare Chromosome Disorder Support Group Charity Number 1110661

Registered in England and Wales Company Number 5460413

AGSA's SUPPORT GROUP & ORGANISATIONAL MEMBERS as at March 2010

Actelion Pharmaceuticals
ACT Muscular Dystrophy Association Inc
Ageing and Disability Department
Androgen Insensitivity Assoc. Support Group of Australia
Alagille Syndrome Support Group
Albino Support Group
Genetic Alliance USA.
Angelman Syndrome Assoc.
Alzheimer's Aust ACT
Immune Deficiencies of Australia)
Assoc. for Children with a Disability, Vic.
Assoc. for the Wellbeing of Children in Healthcare (AWCH)
AUSSIE FOLKS
Australian Addison's disease Assoc Inc.
Aust. Arthrogryposis Group (TAAG) Inc.
Aust Assoc for Families of Children with Disability
Australian Chapter of the BDSRA
Aust. CHARGE Association
Aust. Crohn's & Colitis Assoc.
Aust. Huntington's Disease Assoc. (NSW) Inc. Aust. Huntington's Disease Assoc (Qld) Inc.
Australian Kabuki Syndrome Assoc Inc
Australian Leukodystrophy Support Group Inc.
Aust. Speak Easy Assoc.
Australasian Tuberous Sclerosis Society Inc.
Aust. Leukodystrophy Support Group
Australian Thyroid Foundation Ltd
Autistic Spectrum Australia (ASPECT)
Foundation, Aust
Beckwith-Weidemann Syndrome Support Group
Bundaberg SEDU Norville Special School
CAH Support Group
Cardiomyopathy Assoc of Aust. Limited
Catholic Education Office
CdLS Association Inc.
Centacare Early Intervention.
Central Coast Genetic Counselling Service
Cancer Institute NSW – Hereditary Cancer Registry
Charcot Marie-Tooth Assoc. of Aust Inc.
Charcot Marie-Tooth Disease, USA
CHARGE Syndrome Assoc of Australasia
Chatswood Assessment Centre
Child Health & Safety Vic
Children Hospital at Westmead
Children's Medical Research Institute (CMRI)
CJD Support Group Network
CLIMB Children Living with Inherited Metabolic Diseases UK
Community Support Team
Connected, Connective Tissue Dysplasia Clinic
CONTACT A FAMILY U.K.
Coorinda Family Support Group
Cleft Pals, The Cleft Palate & Lip Society
Coeliac Society of NSW Inc.
Congenital Adrenal Hyperplasia Support Group
Cornelia de Lange Syndrome Support Group
Cri du Chat Syndrome Support Group of Australia
CVS Support Group (WA)
Cystic Fibrosis NSW
Cystic Fibrosis Assoc of Qld Ltd.
Cystic Fibrosis Assoc. Vic
Cystic Fibrosis Assoc of ACT
Cystic Fibrosis Foundation, North Ryde.
Darling Point Special School
DEBRA A NSW Inc.
Dept of Ageing Disability & Homecare, Albury, ACT
Dept of Clinical Genetics, Westmead, Liverpool Health Service
DIAL (Qld)
Disability Information Resource Centre (SA)
Donor Conception Support Group
Depressive & Manic Depressive Assoc.
Dystrophic Epidermolysis Bullosa Research Association (DEBRA) NSW Inc.
Early Education Clinic, North Sydney
Early Childhood Intervention Program, Coffs Harbour
Early Learning, Devonport,Tasmania
Eastern Suburbs Services for the Developmentally Delayed
Ehlers-Danlos Syndrome Support Group
Exceptional Parent (USA)
Fabry's Support Group Australia Inc.
Family Planning Assoc.
FAP Register (NSW Cancer Council)
Fragile X Assoc of Australia
FRANS
Friedreich Ataxia Assoc of NSW
Gaucher Assoc. of Australia
Genetic Support Network of Victoria
Genzyme Australasia Limited
Genetic Interest Group (GIG)
Greater Southern Area Health Service
Haemochromatosis Information Service & Support Group QLD
Healthlink
Haemophilia Foundation NSW
Haemophilia Foundation, Qld
Hastings Early Intervention - Coffs Harbour
Health Consumer Council (WA) Inc
Hereditary Haemorrhagic Telangiectasia
Hereditary Fructose Intolerance
HSP Research Foundation
Hunter Orthopedic School
Huntington's Disease Assoc. (NSW)
Huntington's Disease Assoc. (QLD)
Huntington's Disease Assoc, Wellington
IDEAS Inc
Information Centre, RCH, Vic
Kidney Kids of NZ Support Group Inc.
Klinefelter Syndrome Support Group
Ku Children's Services
Kurrajong Early Intervention
Maternity Alliance
Leukodystrophy Foundation (USA)
Leighs Disease Support Group
Library/Disability Information and Resource Centre (DIRC)
Liverpool/Fairfield Disabled Persons
Lowe's Syndrome Assoc. Inc.(USA)
Lower North Shore Community Support Team
Lucas Gardens School
Lysosomal Storage Disorders
M.P.S. Society
Marfan Syndrome Support Assoc. NSW
Medical Library, RHW
Meniere's (NSW) Support Group
Metabolic Dietary Disorders Association (MDDA
Mid North Coast Area Health Service)
Motor Neurone Disease Assoc. of NSW Inc.
Multiple Epiphyseal Dysplasia Assoc.
Australian MPS Society
Muscular Dystrophy Assoc of NSW
Muscular Dystrophy Assoc (NZ) Inc.
National Council of Intellectual Disability
Neurofibromatosis Assoc of Aust Inc.
Noonan Syndrome Support Group
Northcott Society
North Coast Area Health Service
NZORD, Wellington
Osteopetrosis Support Group
Osteogenesis Imperfecta Society of Aust
OzED-Australian Ectodermal Dysplasia Support Group
Parents Bereavement Support Group
Parent to Parent (NZ)
Pen-Parents of Aust. (ACT)
Physical Disability council of NSW
PKU Assoc of NSW
Post Adoption Resource Centre
Pseudohypoparathyroidism Support Group
Port Macquarie Community Health Centre
Pseudoxanthoma Elasticum Support Group
Prader-Willi Syndrome Assoc
Pyruvate dehydrogenase deficiency.
Rare Chromosomes Disorders Support Group (Unique UK)
Society of NSW Inc.
Rett Syndrome Assoc. of Aust.
Retina Australia (NSW) Inc.
Royal Blind Society of NSW
SANDS (Qld)
Schizophrenia Fellowship NZ
Self Help Qld.
Short Statured People of Aust (NSW)
Short Statured People of Aust (Vic)
Short Statured People of Aust. (SA)
Spinal Muscular Atrophy
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
Stillbirth & Neonatal Death Support Qld Inc.
Tasmanian Clinical Genetics Service
Thalassaemia Society of NSW
The Centre for Genetics Education
The Coeliac Society of NSW Inc
The Chromosome 18 Registry & Research Society (Aust) Inc.
The Lupus Association of NSW
The Northcott Society – Coffs Harbour, Dubbo, Tamworth, Wagga
Thalassaemia Society of NSW
The Centre for Genetics Education
TS+
Turner Syndrome Assoc. of Aust. Ltd. (NSW)
Uncontrolled Epilepsy Support Assoc (Vic)
United Leukodystrophy Foundation (USA)
VCFS & 22q11 Foundation.
Wellington Huntington's disease Assoc. (Inc.) (NZ)
West Syndrome Support Group
Williams Syndrome Association of Aust. Inc.
WISH
Wolf-Hirschhorn 4p- Syndrome Support Group
Women with Disabilities Australia
Yeerongpilly SEDU

Members of the Australasian Genetic Alliance (AGA) formed 2003 –
Assoc of Genetic Support of Australasia
Genetic Support Council of WA
Genetic Support Network of Victoria
NZORD
SHOUT ACT

AGSA is on the Board of the
International Genetic Alliance (IGA)
World Alliance of Organisations (WAO)

NB: This list represents support groups and associations only. In addition to this list of members AGSA has established a Contact Register of over 850 genetic conditions representing families and individuals seeking contact.

AGSA has established a Chromosomal Disorders Support Group, Rare Treasures representing over 250 chromosomal abnormalities. A Rare Treasures Newsletter is produced twice a year. AGSA works in partnership with Unique on this project and we thank them for their support.

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

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Medical and Professional Advisory Board

Subscription Year Ist July - 30th June

ANNUAL SUBSCRIPTION

Individual	$30.00 incl.GST
Group/Organisation	$48.00 incl.GST

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***