AGSA
THE ASSOCIATION OF GENETIC SUPPORT OF AUSTRALASIA INC.
NEWSLETTER

APRIL 2008 ISSUE 83 ISSN1033-8624 FUNDED BY NSW HEALTH

CONTENTS

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

EDITORIAL

Dear All,
I hope your year has started well and you have enjoyed the school holidays. I only wish all the rain in the last week had rained in the areas that badly need it.

What has been happening since Christmas? Mandy has been busying organizing our Rural Carers Seminar to be held in Tamworth on 3rd – 4th May. We are pleased to report we have already 6 children registered for the sibling workshop on the Sunday.

AGSA is participating in the IDEAS Expo to be held in Queanbeyan on 22nd -23rd May. Already many people have registered for AGSA's seminar on 22nd May from 1 – 4.15pm. Telegroup counselling commences on 5th May and we have 5 Mums registered. If you wish to be the lucky last to register just give me a call. The TGC will be held on a Monday commencing at 2pm till 3pm.

On 16th March AGSA held a Noonan's syndrome seminar. 15 people attended. Dr Felicity Collins kindly gave up her Sunday to present and we are very grateful to her. Matt Burgess stepped in and gave Gayathri's presentation for her as she was sick. This is not an easy task but he did a great job. Meryl Harris's moving personal story presentation was well received and many in the group volunteered to be support people for new families. The group now has an older parent, two parent's reps and a sibling representative which is great.

In March 2008, the members of the Australasian Genetic Alliance (AGA) and myself, attended the first never symposium on Involving People in Research, presented by the University of Western Australia School of Population Health and the Telethon Institute for Child Health Research, in Perth. The National Health and Medical Research Council (NHMRC) provided funds to bring consumers and researchers together at this inaugural symposium. Over 300 people work together on workshop concepts and problems. The outcome of this symposium was:-

Communiqué
Call to Action

**This symposium recognizes the enormous value and potential for consumer involvement to improve Australian health and medical research outcomes.*

**We recognize that there are tens of thousands of consumers already formally involved in groups, networks and non-governmental agencies.*

** We strongly advise that consumers are meaningfully involved at all stages and all levels of the research process.*

Members of Australian Genetic Alliance (AGA) took this opportunity to hold a meeting. The outcome of which AGA members are working together to comment on the consultation draft issued by the NHMRC – "Disclosure of genetic information to a patient's genetic relatives under Section 95AA of the *Privacy Act 1988* - Guidelines for health practitioners in the private sector" due in April 2008. Future plans for the Alliance, are to hold a meeting alongside the HGSA in Glenelg, South Australia in August this year.

Recently I was invited to present at the Joint 7th Human Genome Organisation (HUGO) – Pacific Meeting 8th Asia Pacific Conference on Human Genetics, Cebu City, Philippines in the concurrent session, *Genetics in Society.* The conference provided an excellent opportunity to meet our neighbours in the Asian/Pacific region e.g. Professor Dr. Thong Meow-Keong, and Juliana Lee Mei Har who founded the Malaysian Society of Rare Disorders Society (MRDS) in 2004, a non-profit organization that acts as a support network for individuals and families affected with rare disorders and Dr Stephen Lam, Consultant Clinical Geneticist, who is setting up a rare diseases group in Hong Kong. It is anticipated these groups along with the Taiwan Rare Diseases Foundation will develop close partnerships to further raise awareness of genetic conditions in the Asian/Pacific Region.

Sue Manass – Genetic Counsellor, Sydney Children's Hospital, Randwick.
For many years Sue Manass has been AGSA's silent newsletter editor. She has given me tremendous support. Sue has been on the AGSA committee, helped with the preparation of Genetic Awareness Weeks, listened to the day to day problems and or course, edited the newsletter for many years. Sadly Sue is retiring in 9 weeks time. **She will be greatly missed by AGSA, her colleagues and the families. Thank you very much Sue for always being there.**

Looking forward to catching up our rural carers seminars
Best wishes to you,

Dianne Petrie

Welcome to Mandy

PROJECT OFFICER – FILLING THE VOID

A big hello to all AGSA families and friends! My name is Mandy Newton, the newly appointed Project officer of AGSA. Over the past month or so I've been steadily familiarising myself with AGSA and the Filling the Void project, previously managed by Laurie Taylor.

I have come to AGSA after 2 years with the CJD Support

Group Network where I worked with families who were caring for loved ones with the genetic variant of Creutzfeldt Jakob Disease (CJD), an extremely rare and invariably fatal neurological disease presenting in adults, and in point of fact, a condition which affects my own family. I am also a social work student...*still*...feels like it's been forever!

My personal life is extraordinarily chaotic, to say the least! I am Mum to Cooper (8) and Gus (5) who could very easily match the energizer bunny's energy levels any day of the week. When I'm not playing chauffer to the boys and running "mum's taxi" to football training and matches, you can usually find me escorting them to various other extra curricular sporting and artistic activities through the week. I slot these commitments in around my 3 day working week at AGSA and my study schedule. When time allows – *so hardly ever* - I enjoy seeing live music, I love the theatre and my house is full of fanatical AFL fans, so any Sydney Swans matches are entirely compulsory viewing.

I'm looking forward to the coming months and for the opportunity to meet many of you at our upcoming seminars in Tamworth and Canberra. I have already spoken with many of you recently and it's certainly very obvious to me that Laurie has left some rather enormous shoes to fill. I can certainly assure you that the hard work and commitment to AGSA's families demonstrated by Laurie so enthusiastically over the past few years will continue and I'm very pleased to have the opportunity to work with AGSA.

Mandy Newton

Reference - Website

"Genetics Home Reference
(Your guide to understanding genetic conditions) provides consumer friendly information about the effects of genetic variations on human health'.
Website: http://www.ghr.nlm.hih.gov

Recreation Rendezvous Inc.
support for adults with disabilities
- admin@recrend.ngo.org.au
Phone: 02 9420 8555
http://www.recrend.org.au

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.

Contacts requested for these conditions:-

Ehlers Danlos syndrome

Sandra Simms email shardan@tpg.com.au would like to establish some sort of contact base in Queensland and meet some families. Please email Sandra if you would like to be involved.

Floating Harbor Syndrome and Autism

Glucose GP1 transporter defect

Idiopathic Peripheral Neuropathy I am 65 and I have had PN for about 6 years. I do not know of a single soul that has it – other than me! So it is pretty lonely out here. Contact please.

Incontinentia Pigmenti

A lady with this condition would really like contact especially with someone affected by this condition and who has had children.

Lebers Optic Atrophy

Milroy's Disease a lady aged 63 with this condition would like contact.

Pallister Killian syndrome

A family with a 12 year old girl would like contact with families with the same challenge from Ipswich, Qld.

Pyknodysostosis

X-Linked hydrocephalus

A mother of a nine year old boy with this condition would like contact with others.

Letter to AGSA

I am looking to make contact as we have recently had our son diagnosed with ADA (Adenosine Deaminase deficiency.) He is termed ADA-SCID. I believe there to be only a few families with this in Australia and so I am also seeking people who have children suffering other forms of SCID too. I am happy to make contact or be contacted via email or alternately via telephone on 0412 403 739

SUPPORT GROUP NEWS

We have had interest in starting a new group for the followin issue. If you would like more information contact **Christine a cdo@connectgroups.org.au**

Hereditary Hemorrhagic Telangiectasia (HHT)
HHT is a genetic disorder that causes abnormalities of blood vessels and involves nosebleeds and characteristic red spot that are distinctly different from haemophilia. Most blood vessels in the body of someone with HHT are normal. However, a small percentage of the blood vessels in a person with HHT have a specific type of abnormality

WEBSITE MOWAT-WILSON SYNDROME *Dave Curry runs a website dedicated to Mowat Wilson Syndrome. He also manages an email support group for MWS with about 40 members from many different parts of the world.*

The website can be found at www.mowatwilson.org. It contains a Community Page listing people, by country and profiles. There is also a Links Page with information gathere relating to MWS and a map showing the location of many o the families. *You can also find the results from two different surveys conducted, one regarding associated conditions anc the other addressing behavior and developmental issues. People can find contact information for the email support group there also.*

New contact details for
The PKU (Phenylketonuria) Association NSW Inc. Registered Charity C.C 23085 ABN: 78 410 895 701
Jolanda Jarman
26 Vincent Road
Phone: 02) 4729 0423 Mobile: 0413 404 644
Cranebrook NSW 2749
Email: thejarmies@optusnet.com.au

Take 5 - Enduring Guardianship
ACON is partnering with the Office of the Public Guardian to produce a new resource about guardianship for members of the GLBT community. Called, *Take Five,* the booklet is designed to increase awareness about the importance of planning ahead for a time when you may not be able to make lifestyle decisions for yourself.

At the session, representatives from the NSW Office of Public Guardian will provide information on the

following topics:
Who can appoint an enduring guardian and how is it done?
What decisions can an enduring guardian make?
How does Power of Attorney and enduring guardianship differ?
What is available for a person who already has incapacity?
What provisions does the Guardianship Act make for same sex couples?
What happens when a patient can't consent to medical or dental treatment?
Who is the person responsible?
What happens if a patient is objecting to treatment?
For further information, and to RSVP, please contact Ian Down on 9206 2009 or idown@acon.org.au

Further information sessions will be held at ACON branches:
ACON Illawarra 6.00pm Thurs 8 May
ph. 4226 1163
ACON Northern Rivers 5.30pm Thurs29 May
ph. 4927 6808

For further information about the role of the Public Guardian and guardianship in NSW contact the Information and Support branch at the Office of the Public Guardian on (02) 8868 6070 or toll free on 1800 451 510.

Media release

26 March 2008

National recognition for top scientist

Joining the ranks of Australia's most esteemed scientists is **Professor Patrick Tam** from the **Children's Medical Research Institute**, (CMRI) who has just been elected as a Fellow of the Australian Academy of Science.

Peter Doherty, Ian Frazer and Fiona Stanley are also among only 400 Australians to have been honoured by election to the academy. Professor Tam is one of just 16 scientists elected this year.

Professor Tam and his team have recently completed a 'fate map' of early embryo development. His 28-year exploration mapped the movement of cells and tissues in the developing mouse embryo between the 5th and 10th days after conception, the human equivalent of one week to one month after conception. This research revealed how cells are directed to form specific parts of the body.

A 'fate map' reveals the precursors of different bod parts and organs - where they are in the early embry and how they mature and are assembled into the fina body plan. Professor Tam's 'fate map' is expected t be used as a reference by scientists around the worl investigating developmental problems such a neurological defects, and malformations of the head face and eyes.

The Australian Academy of Science recognise Professor Tam's research in Embryology by electin him as a fellow. "He is a world leader in th understanding of early mammalian embryoni development," said Richard Bray, the Academy' spokesman. "And his research has put Australia o the global map of mammalian developmental biology."

"It is indeed a great honour that my contribution to th world's knowledge in Embryology is recognised b peers both within and outside my field," said Professo Tam. "I would like to stress that the achievement is no mine alone. I am eternally grateful to present and pas members of my team for their dedication and effor and to the CMRI for the support of my research."

CMRI Director Professor Roger Reddel said tha "Professor Patrick Tam is an exceptionally generou colleague who gives his time and expertise to hel other scientists here, and in many top researc centres throughout the world. The CMRI i tremendously proud of Patrick and his research team and all they have achieved."

Professor Tam also leads research projects on th gene mutations causing Rett syndrome, cleft lip an palate, eye development, cataracts and glaucoma.

Professor Tam's election to the Fellowship of th Australian Academy of Science is especially timely This year, CMRI celebrates its 50th anniversary an reflects on the achievements of its world-clas researchers.

Humanitarian Scientific Achievement Award Presented To Dr. Yu Yamaguchi

Burnham Institute for Medical Research Professor Yu Yamaguchi, M.D., Ph.D., was recently awarded The Humanitarian Scientific Achievement Award by the MHE Research Foundation. The focus of the foundation is to find a cure for Multiple Hereditary Exostoses, a rare genetic bone disorder. The disorder causes people to grow exostoses (bone tumors) on their bones. MHE patients can also suffer from non-skeletal medical issues including mental and neurological issues.

At this time, there is no treatment or cure. Surgery, physical therapy and pain management are currently the only options in management of the condition.

Mutations in a gene known as EXT1 cause MHE. Dr. Yamaguchi created a mouse model, in which the EXT1 gene can be experimentally disrupted in tissues and organs of interest. His research has provided insights into why MHE patients suffer from non-skeletal medical problems. Dr. Yamaguchi has recently shown that mutations in the EXT1 gene can cause dysfunction of nerve cells, providing a clue as to why MHE patients sometimes associate mental and neurological symptoms. Today, the mouse model is being used in more than 20 laboratories around the world, helping researchers explore the function of the EXT1 gene in a variety of organs and tissues.

"Dr. Yu Yamaguchi has expanded the frontiers of understanding of MHE," said Sarah Ziegler, MHE Research Foundation Vice President. "These insights also suggest potential novel approaches that can be explored in order to make the dream of a treatment into a reality." Ziegler is also the mother of a son with MHE.

A crystal plaque commemorating the award was presented to Dr. Yamaguchi during the Foundation's FUNTASIA Research Banquet held on September 30, 2007 in Brooklyn, New York. Dr. Yamaguchi was also presented citations and proclamations from U.S. Congress, New York State Senate, and the Borough of Brooklyn.

About Burnham Institute for Medical Research: Burnham Institute for Medical Research conducts world-class collaborative research dedicated to finding cures for human disease, improving quality of life, and thus creating a legacy for its employees, partners, donors, and community. The La Jolla, California campus was established as a nonprofit, public benefit corporation in 1976 and is now home to three major centers: a National Cancer Institute-designated Cancer Center, the Del E. Webb Center for Neuroscience, Aging, and Stem Cell Research, and the Infectious and Inflammatory Disease Center. Burnham today employs nearly 800 people, ranks consistently among the world's top 20 organizations for the impact of its research publications, and rates fourth among all research institutes in the United States for obtaining grant funds from the National Institutes of Health. In 2006, Burnham established a center for vascular mapping and bionanotechnology in Santa Barbara, California. Burnham is also establishing a campus at Lake Nona in Orlando, Florida that will focus on diabetes and obesity research and will expand the Institute's drug discovery capabilities, and employ over 300 people.

For additional information about Burnham and to learn about ways to support its research, visit http://www.burnham.org/.

For more information about The MHE Research Foundation, visit http://www.mheresearchfoundation.org/.

Source: Andrea Moser

2008 Genetic Alliance Conference *Transformational Leadership*

On July 11-13, Genetic Alliance will hold its 2008 Annual Conference at the Bethesda North Marriott in Bethesda, Maryland. Like last year's conference, which was attended by more than 200 advocates, health professionals, policymakers, industry professionals, and community leaders, the 2008 conference will provide an outstanding opportunity to participate in cutting-edge workshops and insightful discussions.

The conference kicks off with five concurrent daylong symposia — Industry Partners, Leadership Development, Nonprofit Management, Policy Training, and Research Translation.

Saturday and Sunday will be organized around workshop and panel discussions centered on cutting edge topics of international significance. Conference topics will cover issues such as clinical research, delivery of services, education and information, family health history, health disparities, health information technology, and newborn screening.

Throughout the conference, participants will have ample opportunity during long meals and breaks to network and socialize.

Saturday evening will feature our signature awards banquet celebrating outstanding individuals and organizations in advocacy, reporting, industry, and healthcare. The conference ends on July 13 following panel discussions and a dynamic concluding keynote address.

ThalNSW 30th Anniversary Charity Dinner Dance at the Grand Roxy Brighton-Le-Sands
30th August 2008
Please contact Jane Lampitsi on P: 02 9550 4844 F: 02 95193517 or send an email to coordinator@thalnsw.org.au

GENETIC ALLIANCE BIOBANK

Founded: October, 2003

Board members

Sharon F. Terry, Genetic Alliance and PXE International
Joan Scott, Center for Genetics and Public Policy
Patrick F. Terry, International Genetic Alliance and PXE International
Claire Driscoll, National Human Genome Research Institute
Liz Horn, PhD, Research Director, National Psoriasis Foundation
Owen Johnson, CEO, Inflammatory Breast Cancer Research Foundation

BioBank functions as a cooperative - in other words, the Board, representing the \members, configures the best possible infrastructure, and continually improves the options available to the members as science and technology improve. In addition, as the number of members of the BioBank increases the BioBank has more resources and is able to offer more and more options.

Our Vision

Individuals and Advocacy Organizations Partnering with Researchers & Industry to drive Translational Research Leading to New Safe and Effective Treatments."

Members:

- Angioma Alliance
- CFC International
- Inflammatory Breast Cancer Research Foundation
- Joubert Syndrome Foundation
- National Psoriasis Foundation
- NBIA Disorders Association
- PXE International

Challenges –

Rare Genetic Diseases Research

- Limited subjects
- Variable disease phenotyping
- Limited bio- sample & data repositories
- Fragmentation/lack of scale
- Limited privacy & data security
- Limited funding → limited research
- Poor feedback to participants

Genetic Alliance

Biological Samples and Clinical Data Bank

- **Recruitment** → trust, highest privacy & empowerment, and ongoing education
- **Informed Consent** → educated, informed & dynamic process
- **BioBank Informatics** → Encodes identifiers in a centralized database maintained by the advocacy organization
- **Research Focus** → disease and treatment research via academic collaborators and industrial partnerships

Genetic Alliance

Bio-data Bank -- Functioning

- Centralized, standardized collection and archiving
- Maintains the integrity of advocacy's organizations collections & data
- Enable IRB approved investigator research that otherwise would not happen
- Ensure appropriate use of samples & data [patients & advocacy organization]
- Enable ethical re-contact and follow-up for phenotype/genotype correlations, natural history and longitudinal studies
- Regular communications to key constituents
- Advocacy organization control & benefit sharing with the advocacy organization

Subject: Fw: Message from NSW Ambulance Service

We all carry our mobile phones with names & numbers stored in its memory if we were to be involved in an accident or were taken ill, the people attending us would have our mobile phone but wouldn't know who to call. Yes, there are hundreds of numbers stored but which one is the **contact person in case of an emergency? Hence this "ICE" (In Case of Emergency) Campaign.**

The concept of "ICE" is catching on quickly. It is a method of contact during emergency situations. As cell phones are carried by the majority of the population, all you need to do is store the number of a **contact person or persons who should be contacted during emergency under the name "ICE" (In Case Of Emergency).**

The idea was thought up by a paramedic who found that when he went to the scenes of accidents, there were always mobile phones with patients, but he didn't know which number to call. He therefore thought that it would be a good idea if there was a nationally recognized name for this purpose. In an emergency situation, Emergency **Service personnel and hospital Staff would be able to quickly contact the right person by simply dialing the number you have stored as "ICE".**

Please forward this.

It won't take too many "forwards" before everybody will know about this. It really could save your life, or put a loved one's mind at rest. For more than one contact name simply **enter ICE1, ICE2 and ICE3 etc.**

Malaysian Rare Disorder Society (MRDS)

Founded in 2004, the Malaysian Rare Disorders Society (MRDS) began as an informal grouping for families with rare genetic and metabolic conditions treated at the Genetics Unit, Department of Paediatrics at the University of Malaya Medical Centre, Kuala Lumpur, Malaysia. In 2007, MRDS was registered as a non-profit organization that acts as a support network for individuals and families affected with rare disorders in Malaysia. Till date, MRDS has almost 30 families registered. The main objectives of MRDS are to create a network among individuals and families with rare disorders, to serve as a contact and resource centre and to promote awareness on rare disorders among the Malaysian community. Our past activities included organizing a Family Day for MRDS members that consists for parent group discussions and children's art competition, 1st MRDS Annual General Meeting together with talks presented by invited speakers on special education and disability benefits and collaboration fundraising project with Kiwanis USJ on "Lend a Hand" notepads. As a new support group, we look forward to network with other rare disorders societies to learn the ropes and exchange ideas. **(Email**: info@mrds.org.my, **Website:** www.mrds.org.my). Currently the President of MRDS is Dato' Hatijah Ayob and she is assisted by her committee members. Professor Dr. Thong Meow-Keong, a clinical geneticist who proposed the setting up of the society continued to serve as MRDS advisor.

Face Shape in Williams Syndrome

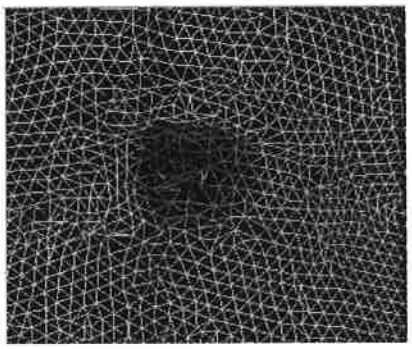
Peter Hammond
UCL Institute of Child Health, London

At the 2007 Williams Syndrome Conference in Sydney, a room was set aside for taking 3D photographs of the faces of children and young adults with Williams syndrome, and also unaffected family members. For many of the children, it was great fun for them to see their faces spinning like a top and from the inside out. There was, however, an underlying serious reason for taking these photographs. Children and adults with Williams syndrome have face shape differences from the general population, as do individuals with many other conditions with a genetic basis. The face and the front part of the brain develop in intricate harmony so that in conditions where development is atypical, facial features and brain function are often simultaneously affected. In such situations, the face can be an important clue in the initial diagnosis. In Williams syndrome, it is often the recognition of the facial features which first suggests the diagnosis to doctors. For many years before we had genetic tests for Williams syndrome, and even now, the presence of the characteristic facial features was an important pointer towards the diagnosis.

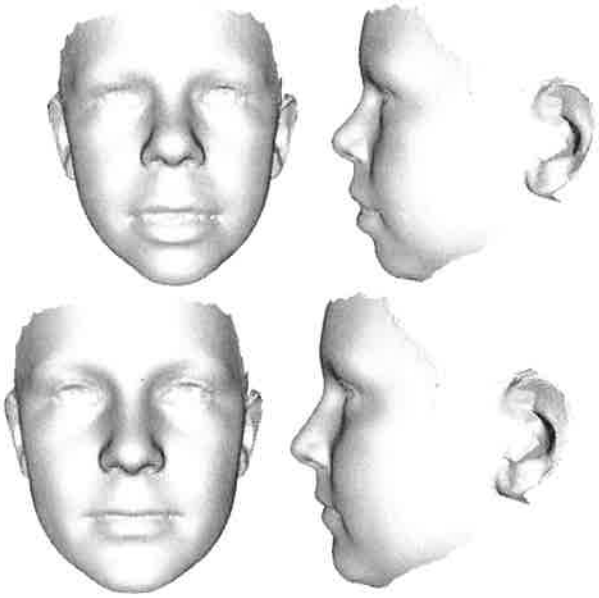
We (along with other medics and computer scientists) have already shown how 3D face photographs and the special computer software we have developed can identify face shape differences very accurately. Our aim is to develop models of face shape to support genetic screening and to encourage appropriate genetic testing in situations where a diagnosis is unclear. In addition, the models will be useful in the training of pediatricians and clinical geneticists to help improve skills in identifying facial features that are characteristic of many genetic conditions.

One area of special interest, for Williams syndrome, is in understanding face shape differences associated with particular alterations in the genetic material on chromosome 7. We are collaborating with Dr May Tassabehji of Manchester University in a study of so-called phenotype-genotype correlation. Dr Tassabehji was also present at the meeting and has written a separate explanation of the genetics of Williams syndrome and the motivation for identifying atypical individuals with Williams syndrome. Finally, another reason for taking the 3D images is to document the way that the face in Williams syndrome changes over time so that we know what to look for in children of different ages.

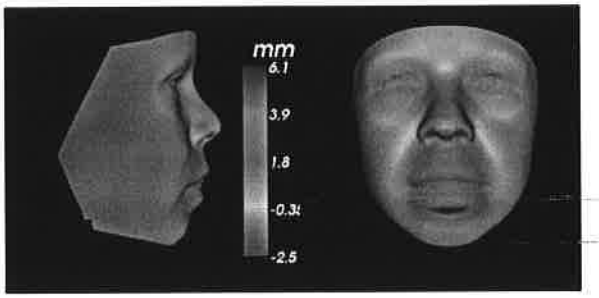
The picture above (on previous page) shows the special 3D camera we used in Sydney. It captures as many as 25,000 points on a face producing a very accurate map of its contours. Before the Sydney conference, we had collected 130 or so images of children and adults with Williams syndrome. The picture below shows the detailed mesh of points around the eye. Hold the image away from you to see the eye more clearly.



A collection of 3D face surfaces of children with the same genetic syndrome can be combined to find an average or typical face for that condition. The first pair of images immediately below shows the portrait and profile of the average face of 85 children with Williams syndrome aged between 1 and 18 years of age. The second pair shows the same views of the average face of 185 children with no known genetic condition and of about the same age range.



By eye, there are some obvious differences in the shapes of these two average faces. But a computer-based analysis can detect much more subtle differences, as the figure below demonstrates where green identifies regions that are essentially the same and other colors emphasize differences around the forehead, eyes, nose, lips and cheeks.



These differences enable special pattern recognition software to distinguish between the faces of unaffected individuals and those with Williams syndrome with an accuracy of over 94% even when the tested faces are not part of the face shape model. We need to collect a large number of images to improve the accuracy of the recognition of facial features in Williams syndrome in a diversity of ethnic backgrounds. This will take some time to achieve. High levels of accuracy are particularly important for the study of atypical individuals with May Tassabehji. But as we produce preliminary results we will report back to families through support group newsletters and future conferences when we hope more of those attending will take part. Every family who volunteered for the 3D photography in Sydney will be sent a CD containing their pictures and computer software that will let them view and manipulate the pictures in 3D on a computer. So, as the scientific analysis of the faces continues, your children (and maybe some of the adults) can have some fun spinning, colouring and playing with the 3D faces.

We are only able to complete this kind of research with the support of families with affected members and the co-operation of organizations such as the UK Williams Syndrome Foundation, the US Williams Syndrome Association, the Association of Genetic Support of Australasia and the Australian Williams Syndrome Association. The UK charity NewLife were generous in their funding of the purchase of the expensive 3D face scanners. So sincere thanks to all of you who took part and we look forward to meeting many of you again next time.