

# Association of Genetic Support of Australasia (AGSA)

## Rare Disease Day

Dianne Petrie OAM  
APSU Workshop,  
Saturday, 27<sup>th</sup> February, 2010



# AGSA

## *Making the Right Connections*

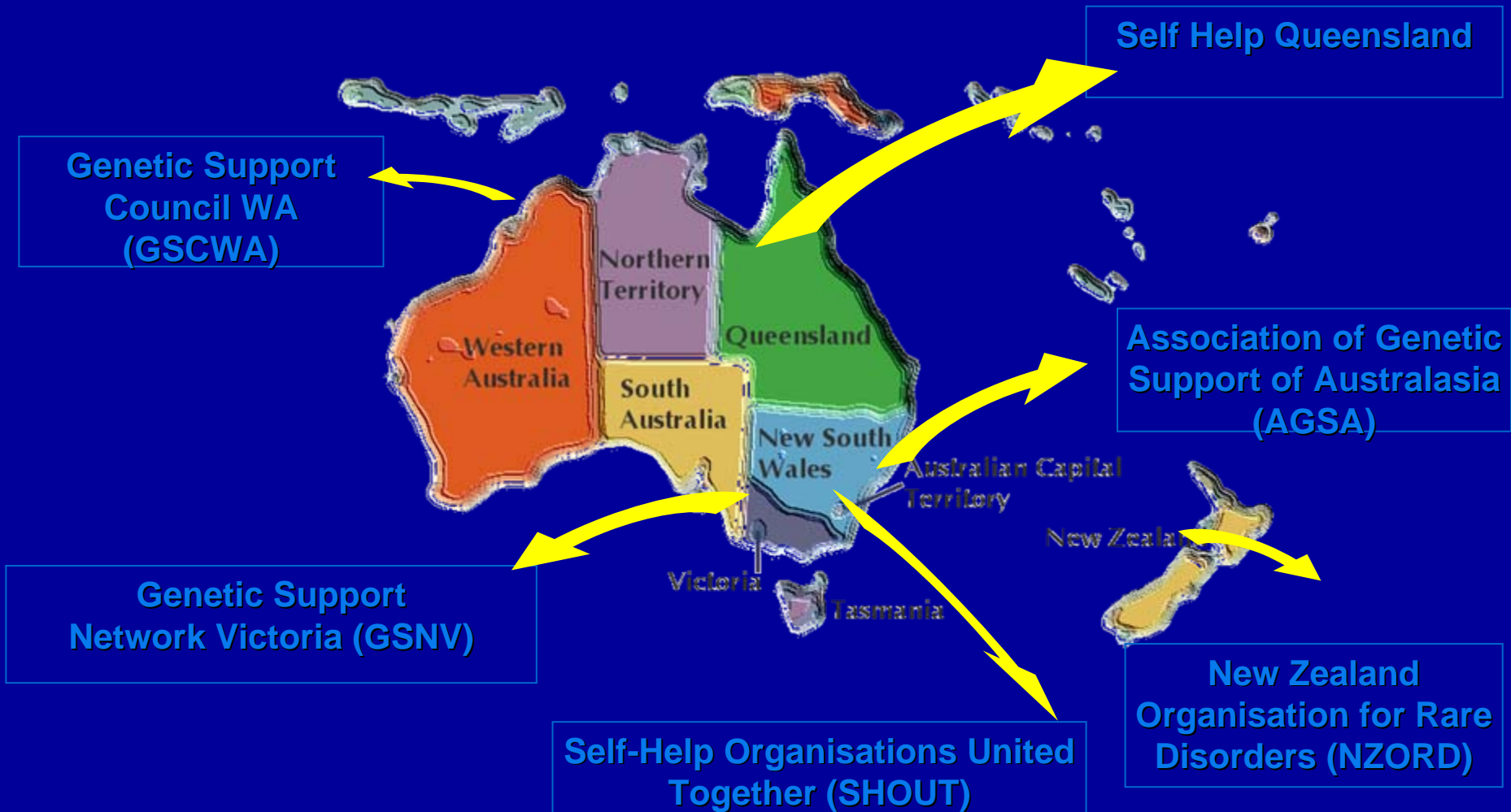
- AGSA is a one stop referral centre.
- AGSA facilitates interaction between support groups, professionals, community organisations and scientific researchers.

# Brief outline of AGSA

- Formed in 1987
- AGSA is an umbrella group for less common genetic conditions providing peer support and information.
- 380 members (individuals and organisations)
- Funded by NSW Health
- Contact Register is the largest in Australia and has over 850 rare genetic conditions represented
- And 250+ chromosomal abnormalities - Rare Treasures Support Group



## Members of The Australasian Genetic Alliance



# AGSA's Position

AGSA is a consumer representative on

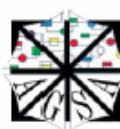
- The Newborn Screening Advisory Committee
- NSW Genetics Advisory Committee
- Sydney/Sydney Eye /Prince of Wales Hospitals (CAC) Committee
- Acting President International Genetic Alliance
- Board Member of the World Alliance of Organisations

# What will I/We gain from the project?

- Meeting others in the same or similar position.
- Sharing stories and strategies.
- Learning about services currently available in the area.
- Accessing specialists in a non clinical setting.
- Opportunities to discuss future options.

## 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 particular genetic disorders.
- 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.



**AGSA**

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Funded by

**NSW HEALTH**

Under the NSW Carers Program

Printed July 2005

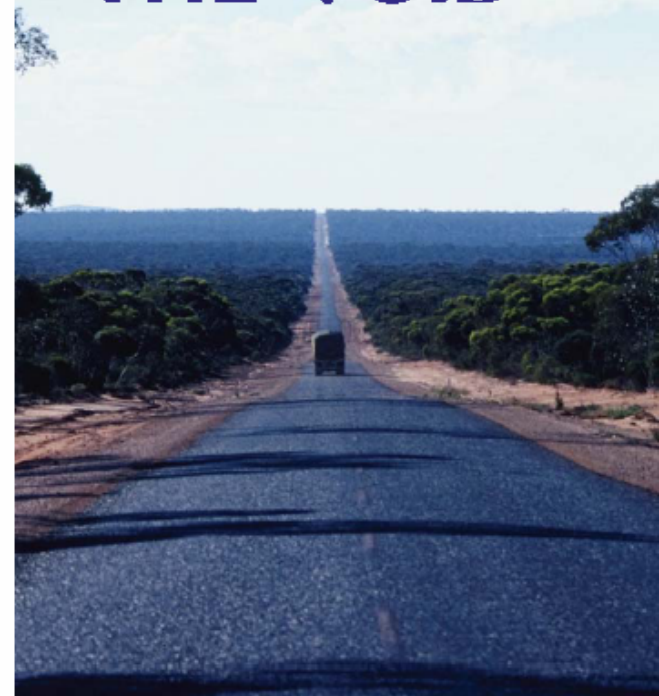
Ready Set Design (02) 9922 4924

Printed by PR Print (07) 3205 1235

Frag illustration by Annabel Craighead

Supporting people living in isolation who are caring for someone with a genetic condition.

# Filling THE VOID



**AGSA**

The Association of Genetic Support of Australasia Inc.

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

# Filling the Void

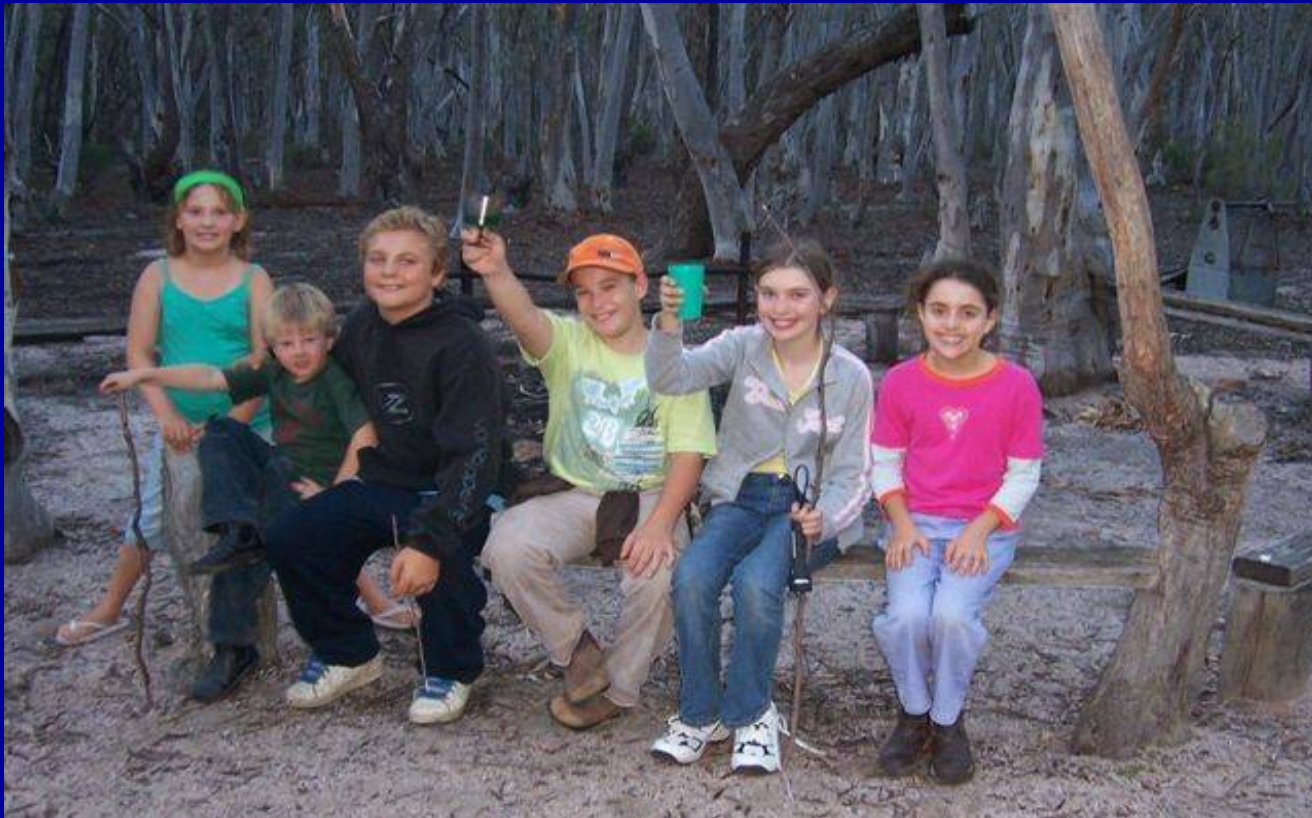
- Seminars held in rural towns, e.g. Lismore, Orange, Dubbo, Newcastle, Tamworth, Albury
- Sibling workshops
- Face to face counselling
- Telegroup counselling 3 times a year
- Metro seminars



# Filling the Void Project

## Working with Carers in Remote and Rural Areas

Funded by NSW Health under the Carers Program





# AGSA's Activities

- Annual genetic awareness week
- Annual BRAC1 /2 Information Day
- Counselling and referral
- Genetic seminars
- Presentations and posters
- Support group leaders meetings

# Mauritius

## Sydney Children's Hospital

- AGSA worked closely with this family from Mauritius whose son had a very rare genetic condition. AGSA liaised with the Sydney Children's Hospital resulting in the family travelling to Australia for a medical consultation which resulted in a very successful operation.

# AGSA'S Seminars

- Turner syndrome
- Klinefelters syndrome x 3
- Lissencephaly
- Double Y syndrome x 3
- Triple X syndrome x 2
- Rare Chromosome x 4
- MEN2
- Leighs disease
- BRCA1/2 x 8
- HNPCC
- Caring for the carer
- TMAU
- Rural/Sibling seminars
- Sotos syndrome
- Noonans syndrome
- Ehlers Danlos syndrome x 3
- HHT x 2
- Beckwith-Wiedemann syndrome x 2
- Genetic Awareness Week x 14
- How to run a support group seminar
- Williams syndrome conference
- Cornelia de Lange
- Marfan syndrome
- Fragile X conference x 3
- Lysosomal storage disorders
- Haemochromatosis
- Living Grief seminar
- Androgen Insensitivity Syndrome

# Support groups

750 support groups Australia wide

New Zealand Organisation for Rare Diseases  
(NZORD)

Taiwan Foundation

Malaysian Rare Disorder Society (MRDS)  
formed 2004

Williams Syndrome Group Singapore

# Resources

- The Australasian Genetics Resource Book  
[www.genetics.edu.au](http://www.genetics.edu.au)
- Genetics in Family Medicine  
The Australian Handbook for General Practitioners  
[www.gpgenetics.edu.au](http://www.gpgenetics.edu.au)

The School and the Telethon Institute for  
Child Health Research.

*Involving People in Research Symposium*  
*Communiqué*

March 5th & 6th 2008, Perth, WA

- National Health and Medical Research Council (NHMRC) provided funds to bring consumers and researchers together at this inaugural symposium.
- The symposium attracted some 240 researchers, consumers, community members and students from Australia, New Zealand and the UK.



# 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*

# Partnership –

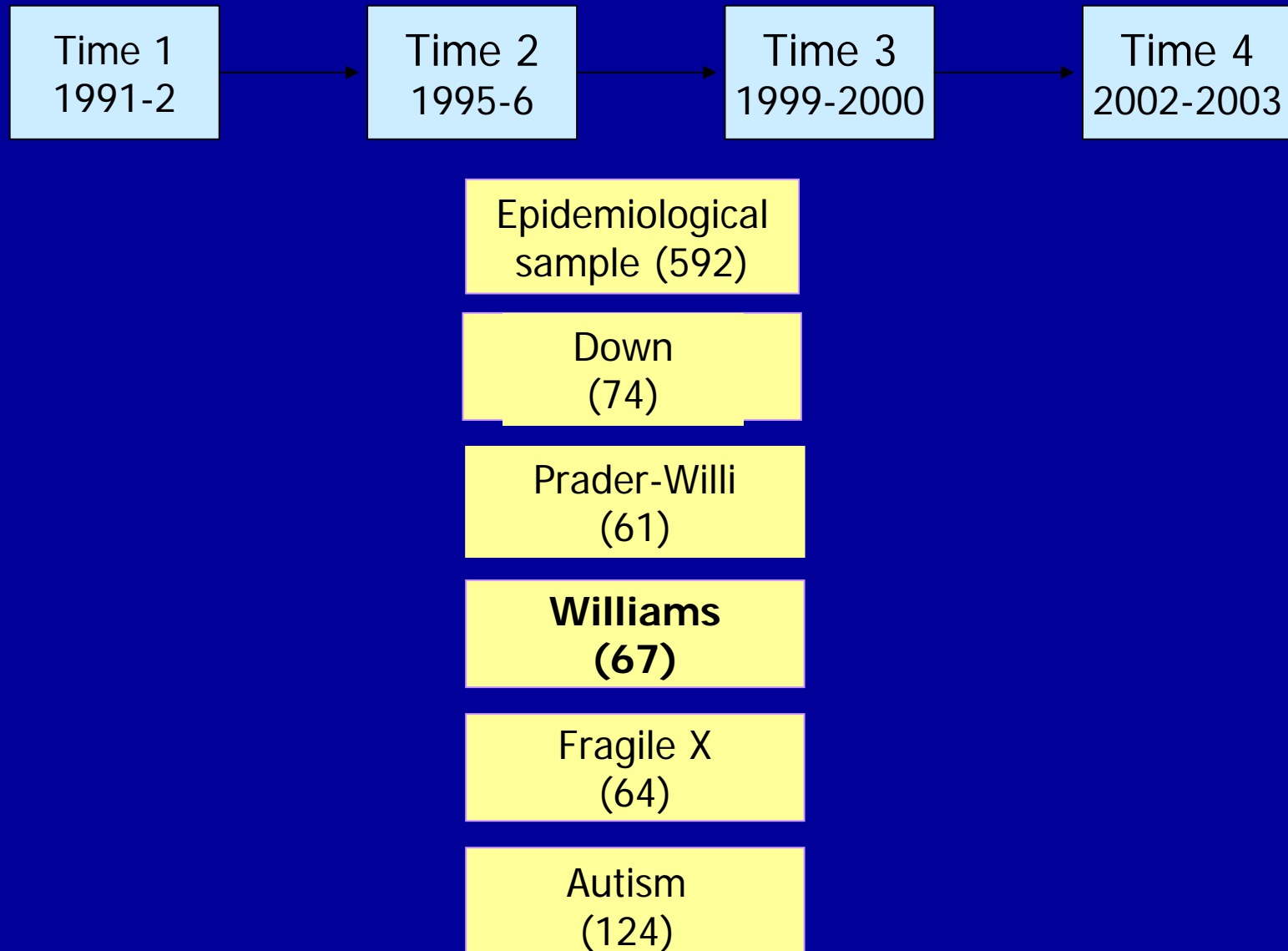
Prof Stewart Einfeld, Faculty of Health Sciences & Brain  
Mind Research Institute, University of Sydney and Bruce  
Tonge, Monash University

The Australian Child to Adult Development Study (ACAD) - a  
longitudinal cohort study of young people with intellectual  
disabilities.

Funded by NHMRC

Australia National Institutes of Health USA

# Timeframe and Subject Groups

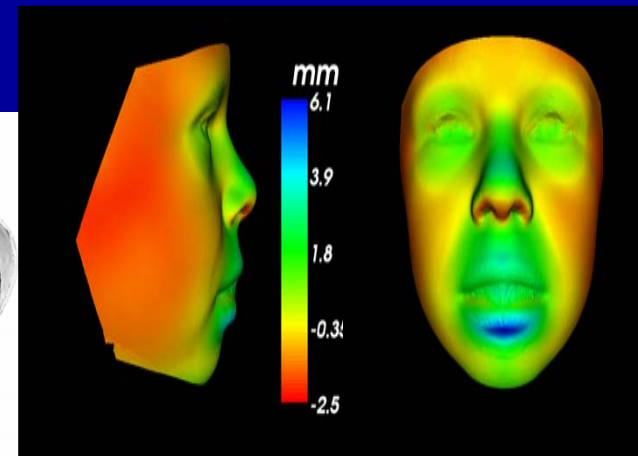
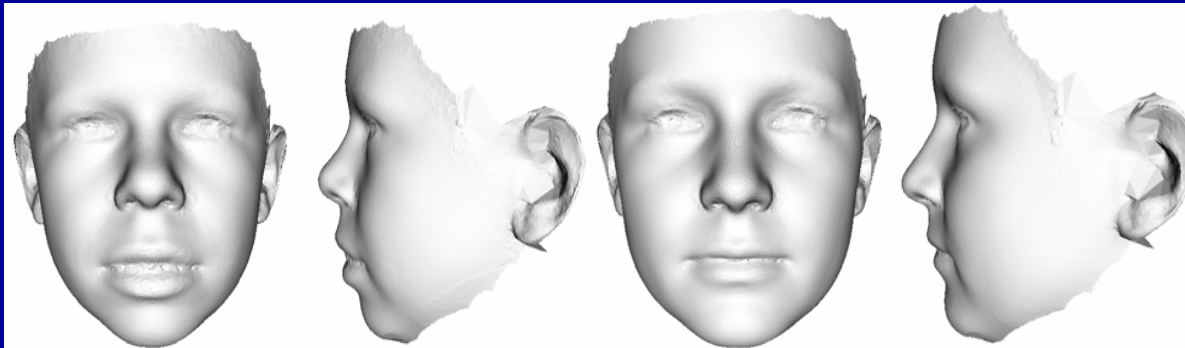


# National Williams Syndrome Conference,

Sydney, September 2007

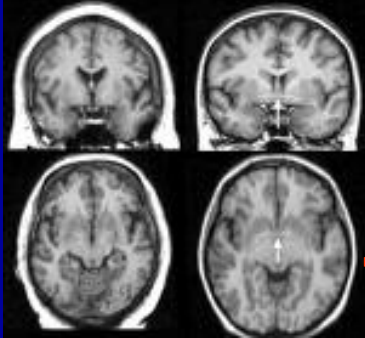
Participants provided the following

- 25 bloods
- 38 Eye movement study
- 51 3D images of faces

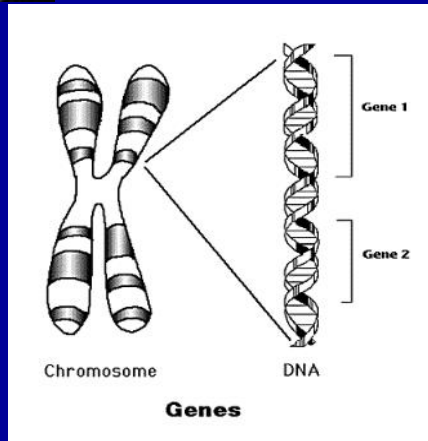


# Correlating Genetic, Cognitive and Clinical Variability in Williams syndrome

- Melanie Porter, Dr May Tassabehji (Manchester University, UK) and Prof. Peter Hammond (University College London, UK)
- research exploring the role of individual genes and groups of genes within the WS critical region in partnership with Williams Syndrome Association and AGSA



# + Update on WS Research



+



Jobs



=



Dr Melanie Porter

Ms Helen Dodd



# International Genetic Alliance (*IGA*)

Founded: Lyon, France: April 7-10, 2003

- 7 Representatives of Genetic Alliances
- 16 representatives of Genetic Patient Organizations from 6 Continents
- Founding Member countries
- Australia, Brazil, Germany, India, Iran, New Zealand, South Africa, Switzerland, Ukraine, USA



"Voicing the interests of people affected by genetic diseases"

"Representing Patient and Parent Organisations from Australia, Eastern Europe,  
Western Europe, Middle East, India, New Zealand, South Africa, South America and  
North America"

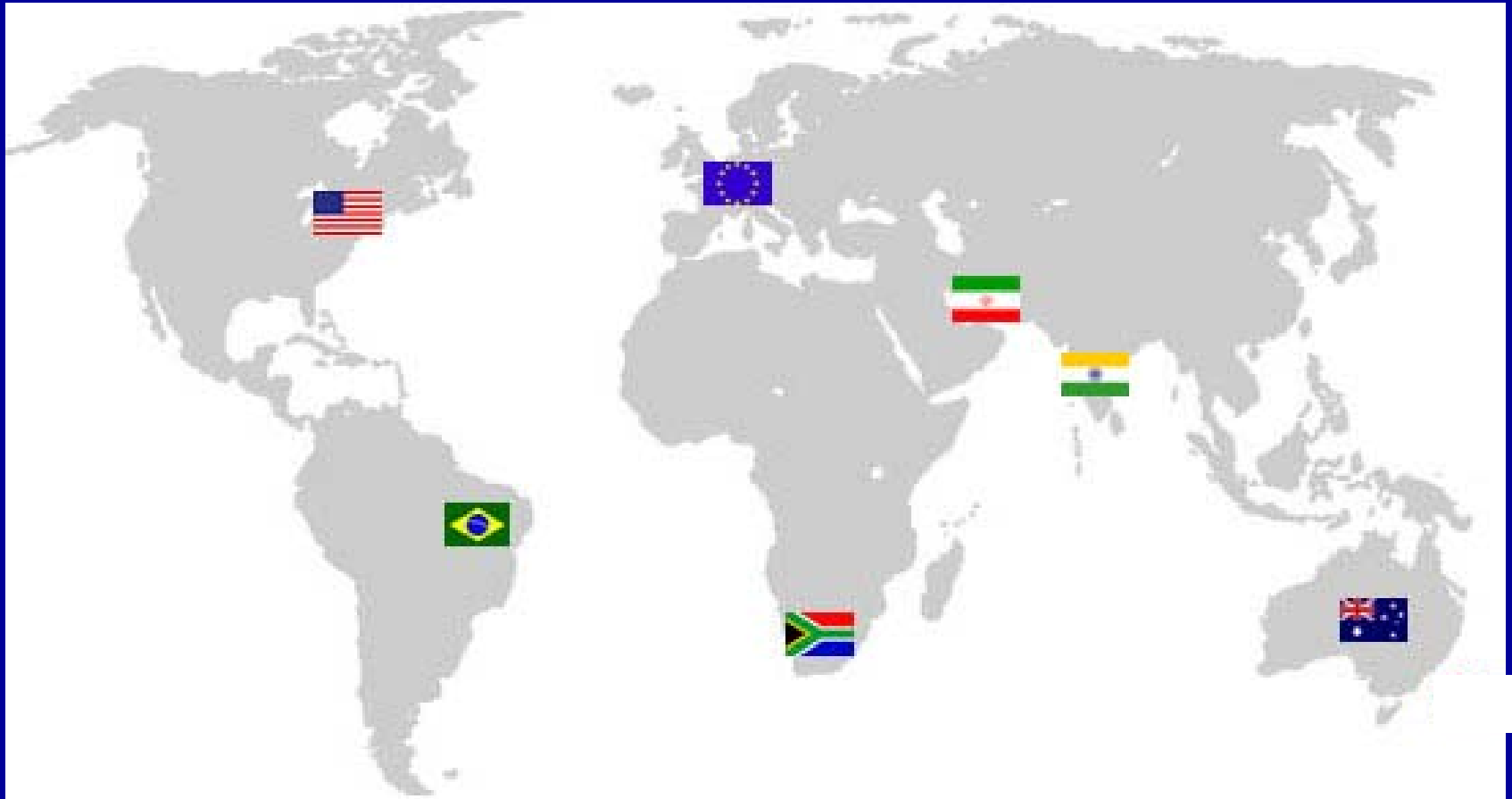
[www.intga.org](http://www.intga.org).



# IGA

International Genetic Alliance

Europe - Africa - Middle East - Asia - Australia & New Zealand - North America - South America





# IGA

International Genetic Alliance

## Members: *15 groups*

<b>AGA</b>	<b>American Genetic Alliance</b>
<b>AGSA</b>	<b>Association of Genetic Support of Australasia</b>
<b>ASRIM</b>	<b>Swiss Alliance for Neuromuscular Diseases</b>
<b>FB</b>	<b>Fighting Blindness</b>
<b>GIG</b>	<b>Genetic Interest Group</b>
<b>HEART</b>	<b>HEART EU Patient Network</b>
<b>IBIS</b>	<b>Ukrainian Alliance for the Prevention of Birth Defects</b>
<b>IGAO</b>	<b>Indian Genetic Alliance of Organisations for Prevention of Genetic Disorders and Care of the Families</b>
<b>IGAIP</b>	<b>Iranian Genetic Alliance of Parent/Patient Organisations</b>
<b>NZORD</b>	<b>New Zealand Organisation for Rare Disorders</b>
<b>PXE</b>	<b>PXE International</b>
<b>RDB</b>	<b>Belgian National Alliance for Rare Disorders and National Alliance Committee</b>
<b>SAGAP</b>	<b>South American Genetic Alliance of Patient Organisations</b>
<b>SAIDA</b>	<b>Southern African Inherited Disorders Association</b>
<b>VSOP</b>	<b>Dutch Genetic Alliance of Parent/Patient Organisations</b>



# Indian Genetic Alliance

*of Organizations for Prevention of Genetic Disorders and  
Care of the Families*

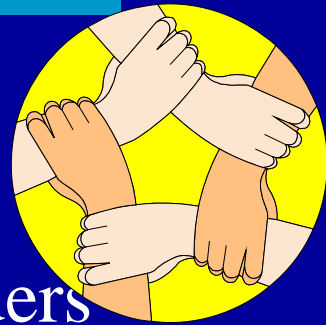
- Founded: Amritsar, 2nd December, 1998
- Represents 13 Genetic Support Groups
- Aims & Mission
- Information availability & sharing
  - Improve genetic services
  - Emphasize needs of women
  - Networking of IGA components & those providing genetic services in India
  - Promote pre- & post- natal screening & pre-conception counselling in high risk cases in India





# Genetic Interest Group

Working to benefit all people affected by genetic disorders



## Alliance of over 130 patient organisations

- Support children & families with genetic disorders
- Educate & promote awareness of genetic disorders among public, opinion formers, and media
- High quality services for people affected by genetic conditions

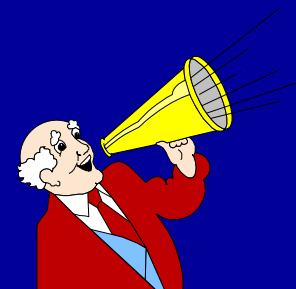




# The Major US Alliance

1984, 85: Alliance of Genetic Support Groups

1986: Genetic Alliance



- > 600 members organizations involving millions of individuals with genetic conditions

# Take home messages

- Diagnosis of a genetic disorder brings about a complex reaction that is often a crisis. The coping process is often similar to what occurs in the experience of loss in the death of a loved one which is compounded by the rarity of the disorders – “being the only one in the world”
- Support groups supplement professional services not a replacement of professional services.
- Provide accurate information and psycho-emotional support

# AGSA's Vision

- Genetics to be a major part of GP's, paediatrics training
- Increase genetic services to shorten diagnosis time
- Raise awareness of the inequity in treatments

# Partnerships with a smile.





AGSA

“Making the right connections since 1988



# AGSA

**“Making the right connections  
since 1988”**

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**Thank you**

