Unravelling the Jargon.

By Lesley Murphy
Co-Director
Adjunct Research Associate
Centre For Comparative Genomics
Murdoch University

rare voices
www.rarevoices.org.au

The unified voice for all Australians living with a rare disease
Brief History

• Australian Rare Diseases Symposium 2011. A unanimous call for a national organisation.

• Outcomes included;

• National Organisation who can do the following:
  ➢ Networks & Collaboration
  ➢ Information
  ➢ Services
  ➢ National Plan
RVA Logo

- RVA formed March 2012
- V formation of logo
- Australian birds
- Fingerprints
- Australian colours
RVA Office - Sydney
Vision

‘Australians with a rare disease will have extended and improved lives.’
Access to, and advocate for, world class, best practice in:

- Diagnosis
- Treatment
- Care
- Services
- Therapies
RVA Objectives

- United voice for ALL Australians living rare disease
- Identify Common issues/Unmet needs
- Policy/position papers
- Raise awareness/Advocacy
- National Rare Disease Registry
- National Rare Diseases Plan
- Education
Unraveling the Jargon

c.402_405CCAA p(Ans 135Val*fs)

- How do we unravel this?
- What is the Patient Organisation perspective?
- Can POs influence decisions made in Australia?
- The role of RVA in patient advocacy
- Education
Diagnosis

- Shock wears off
- News settles in
- What can I do?
- Light bulb moment!!!
- Use Dr Google!!!
- Start educating yourself
- Join a support group
- Get on a registry
Oh No, there is no registry!!
Now what?

• Start lobbying
• Get your act together!
• Patient advocacy
• National Duchenne Registry.
• FSGA (Fabry Support Group Australia Inc.)
• REG4ALL
• Real impetus for registries from patient perspective
• Chance of access trials and treatments
Phew! We can access a registry.

- Patients are better positioned to support research
- Lobbying, advocacy begins again
- We can influence research priorities?
- Standards of care?
- Patients are becoming experts in their own rare disease
- Time for ALL stakeholders to embrace and welcome this
Education for Rare Disease Community

- Australia doesn’t have a national rare disease plan, or a national rare disease registry.
- Community needs education
- Patients don’t understand
  - clinical trials
  - importance of data
  - their role in all of this
Things are looking up

- Robust research data has been collected
- Some promising progress
- Keep raising funds and keep the pressure on
- ANRI, MDWA and exon skipping
- POs can influence regulatory process
- Thus resulting in adoption of new therapies and drugs
Fabry Support Group Australia

Example

• E.g. ‘Patient Empowerment at work!’
• FSGA formed - 1994
• First Fabry Clinic – Royal Melbourne Hospital
• FSGA Funded Medical Research Student
• Fundraising efforts
• Prof Desnik U.S. visited talked of new treatments
• 2000 – Enzyme Replacement Therapy trials began
• Fabry Clinics in 5 states for adults and paediatrics

Source: Fabry Support Group Australia Director; M Fookes
Drug A developed for a Rare Disease

Pharmaceutical company considers producing Drug A

Profitable

Pharmaceutical company applies to TGA for assessment of safety and clinical efficacy for licensing of Drug A

An application is made to the Pharmaceutical Benefits Scheme (PBS) to include Drug A in the drugs partly paid for by the government according to cost-effectiveness.

PBS finds Drug A is cost effective

Yes

Drug A approved by the PBS

Drug A is available to patient’s with a rare disease at no cost through LSDP

Not Approved

Drug A not produced

Not Approved

Drug A produced and available to patient’s with a rare disease

Approved

Application made to the Pharmaceutical Benefits Advisory Committee to include Drug A in the Life Saving Drugs Program (LSDP) if shown to significantly extend the patient’s lifespan

Source: Prof J Goldblatt
Challenges Ahead in Oz

- **10 drugs for 7 rare conditions** currently funded
- Life Saving Drugs Program (LSDP)
- Federally Government funded Program
- How will this regulatory process cope if IRDiRC meets its goal of 200 new treatments by 2020?
- Patient advocacy vs increasing calls for rationing of health budgets?
Australian Experience cont’d

- Demands for accurate robust data!
- Accurate genetic information = increasing effectiveness of patient advocacy
- ‘fundamental knowledge to the application of fundamental knowledge’. (Francis Collins. TED talks)
- The three R’s;
  - Registries, Research and the Regulatory Process
Final Comments

- This is not a linear process.
- Engagement with patient groups at every stage, is crucial to the success and funding of the 3R’s.
- The political is personal & the personal becomes political.
RVA Parliamentary Event
RVA Parliamentary Event

The unified voice for all Australians living with a rare disease

www.rarevoices.org.au
Acknowledgements

- Rare Voices Australia Ltd. Executive Director; Megan Fookes
- RVA Board; Dr Tracy Dudding, John Forman (present)
- Prof Hugh Dawkins; WA Health Dept and OPHG
- Dr G Baynam, Prof J Goldblatt, Prof S Wilton, Prof S Fletcher, Prof M Bellgard
- CCG Murdoch University
- Everyone involved in IRDiRC and RD Connect
- Fabry Support Group Australia Inc.
- Muscular Dystrophy WA