

International conferences on rare diseases: initiatives in commitment, patient care and connections

Andrew W Knight and Domenica Taruscio

An Australian GP's pilgrimage to Rome to sound the voice of primary care

Rather than finishing paperwork after a busy Monday in a Katoomba general practice, I (AWK) found myself sitting (jet-lagged) in a marble auditorium in Rome within the Istituto Superiore di Sanità (Institute of Public Health; Box 1) with about 200 other conference participants from the United States, Australia, and more than 22 different European Union (EU) and non-EU member states. We were waiting for an announcement by the Honourable Livia Turco, the Italian Minister of Health. An article I co-authored, which was published in the *Journal* in July 2006,¹ had led to an invitation to participate in the 2-day International Rare Disease Conference (IRDC) and the subsequent 3-and-a-half-day NEPHIRD (Network of Public Health Institutions on Rare Disease) conference organised by Dr Domenica Taruscio, Director of the Centro Nazionale Malattie Rare (National Centre for Rare Diseases) in Rome, together held from 18–23 September 2006.

The Ministry of Health announcement confirmed the Italian Government's commitment to rare diseases and to orphan drugs research and development. The IRDC proceeded with an overview of initiatives in rare diseases in Italy and some of the more than 22 countries represented. The NEPHIRD involved morning presentations and afternoon small-group work, with the first day devoted to prevention and epidemiology, and the second to diagnosis and treatment. The third day dealt with the social aspects of rare diseases and, on the final morning, we heard plenary sessions on specific rare diseases such as neurofibromatosis, Prader–Willi syndrome, myasthenia gravis, Cornelia de Lange syndrome and Rett syndrome.

Plenary presenters were significant people in the field of rare diseases from Europe and the US, including Dr Kerstin Westermark, Chair of the Committee for Orphan Medicinal Products of the European Medicines Agency, Dr Ségolène Aymé, the Chair of the European Rare Disease Task Force, and Dr Marlene Haffner, Director of the Office of Orphan Products Development, which is part of the US Food and Drug Administration.

The patient voice

As the week unfolded, a striking theme was the presence of patients. Patients and carers dealing with Ehlers–Danlos syndrome, cystic fibrosis, narcolepsy, multiple endocrine cancers, Sjögren syndrome, chronic fatigue syndrome, fibromyalgia, fibrodysplasia ossificans, muscular dystrophy, cyclical vomiting, neurofibromatosis, and many others disorders, participated in and presented sessions. Specific sessions were devoted to patient groups to allow them to present the problems they face. In one moving contribution, Claudio Buttarelli, President of the neurofibromatosis support group Ananas (Italian for pineapple — rough on the outside but sweet on the inside), described the impact of this rare and misunderstood disease on every aspect of his life from socialisation in his teenage years through to the limitations imposed by nerve palsies on playing soccer with

1 The Istituto Superiore di Sanità (Institute of Public Health), conference venue, Rome



his children. Such sessions kept presentations on genetic research, new compounds, and public health initiatives grounded in the everyday experience of patients.

However, the presence of patients should have been no surprise, as patient voices were instrumental in bringing the problem of rare diseases to the attention of governments. In the 1980s in the US, a peak patient group, the National Organization for Rare Disorders (NORD),² was established to lobby for funding and research that no single rare disease could attract (Box 2). Eurordis (the European Organisation for Rare Diseases) fulfils a similar function. Notably, there is no peak patient body for rare diseases in Australia.

Different stories

The US

Dr Stephen Groft, Director of the Office of Rare Diseases (ORD) of the US National Institutes of Health,⁴ spoke at both conferences, describing ORD and some of its activities. ORD was set up in 1993 to stimulate and coordinate research on rare diseases and to support research to respond to the needs of patients with rare diseases. ORD supports a grants program to establish a network for research on rare diseases; grants are provided for such activities as the training of rare diseases researchers and programs to stimulate clinical research on rare diseases. Of particular interest to Australians because of its online accessibility is an information centre aimed at the public, researchers, and health care providers (http://rarediseases.info.nih.gov/asp/resources/rardis_info.asp). ORD also supports a national scientific conferences program to stimulate research and regional workshops to help patient support groups obtain assistance through the National Institutes of Health.

2 Rare disease facts

- Rare diseases are life-threatening or chronically debilitating diseases that have such a low prevalence (not more than 5 per 10000) that specially combined efforts are needed to prevent morbidity and perinatal or early mortality, and to address quality-of-life and equity issues.
- There are approximately 6000 defined rare diseases.
- It is estimated that up to 6%–10% of the community have a rare disease.²
- Patients with rare diseases have common experiences — including delayed diagnosis, wrong diagnosis, inappropriate surgery, lack of access to evidence-based care, and social consequences — because their diseases are rare.³
- Orphan drugs are pharmaceuticals developed to treat diseases that affect relatively few people. ♦

Europe

In April 1999, the EU Parliament set forth Decision No. 1295/1999/EC, adopting a program of community action on rare diseases within the framework for action in the field of public health (1999–2003). Many projects were funded under this program, and important initiatives that continued in the 2003–2008 European public health program⁵ include ORPHANET, a database for the general public on rare diseases (<http://www.orpha.net/consor/cgi-bin/home.php?Lng=GB>), and EURO-CAT (<http://www.eurocat.ulster.ac.uk/>), which surveys more than one million births per year in 19 countries to provide epidemiological information on congenital abnormalities.

We heard about innovative Italian initiatives, including the national network for the prevention, surveillance, diagnosis and therapy of rare diseases made up of certified centres expressly identified by the regions (decentralised administrative units) and the National Registry of Rare Diseases. This registry is established at the Centro Nazionale Malattie Rare,^{6,7} led by Dr Domenica Taruscio. The centre carries out a wide range of activities including genetic research into rare diseases, quality assurance of genetic testing, primary prevention projects, maintenance of the rare disease registry, dissemination of information, development of guidelines, involvement and coordination of EU projects such as NEPHIRD, qualitative research on patients' quality of life, narrative medicine, and training of health professionals.

Lessons for an Australian GP

A need for coordinated activity

The relative lack of coordinated activity in rare disease in Australia compared with the US and Europe is striking — we believe there is a need for a peak patient group (such as NORD or Eurordis) in Australia to lobby for patients with rare diseases.

Australia's small population (with consequent small numbers of patients with any given rare disease) and geographic dispersal presents particular challenges in connecting patients with rare diseases with each other and with expert care. Presentations at the rare diseases conference covered a number of potentially useful strategies for connecting and empowering patients. Of particular note is Ågrenska in Sweden,⁸ which organises week-long camps at which families and patients with similar problems receive intensive education and establish connections with each other, and hear

about specialist services. Ågrenska has been able to demonstrate better outcomes and cost savings through its strategy.⁸

Europe is establishing networks of excellence in which researchers and institutions with expertise in particular diseases are linked. We wonder whether Australian patients and clinicians with interests in particular diseases could join these networks, perhaps even participating in e-medicine consultations.

The role of primary care

The specialists and scientist researchers at the conferences expressed frustration about the supposedly “low” level of skills of their primary care colleagues in identifying the rare disease in which they were expert. One specialist exclaimed that some GPs had never even heard of neurofibromatosis type 1. Some sessions at the conference presented this as an equity issue — surely, a patient with Prader–Willi syndrome has as much right to prompt diagnosis and evidence-based treatment as a patient with type 2 diabetes? The natural reaction is to call for more education of primary care clinicians in individual rare diseases. However, we do not think it would be realistic or even wise for GPs to use their time learning all the details of the 6000 identified rare diseases.⁴

We do believe that primary care has a neglected but important role in rare diseases. To date, progress in rare diseases has been driven by patients through their specialist clinicians and through public health institutions. Primary care clinicians provide a key link between patients in our community and the very specialised services those with rare diseases require. The Australian proposal of a generic model of general practice care¹ was presented and discussed at the IRDC. One important and simple strategy identified during the week was careful monitoring of infant development as a generic strategy to screen for many rare congenital diseases.

A voice from the perspective of primary care seemed to be quite strange to this very specialised community. In the main, they welcomed the general practice contribution and were excited by the possibilities of adding a primary care perspective to the rare disease agenda. Patients at the conference in particular confirmed the need to continue to develop the role of primary care clinicians in rare disease.

Future connections

The pursuit of an idea (the common problem of rare disease in general practice) through to publication in the *Medical Journal of Australia* has led to a number of connections which have been professionally stimulating, satisfying and helpful. One of them was the IRDC in Rome 2006, which highlighted the international agenda on rare diseases. This conference has stimulated one Australian GP (AWK) to think further about and conduct more research into the role of GPs in rare disease. Policymakers, other clinicians, and patient groups in Australia also have the opportunity to reflect on the way forward. In particular, a funded initiative to establish a peak patient body for rare diseases seems long overdue.

Competing interests

None identified.

Author details

Andrew W Knight, FRACGP, MMedSci(Clin Epid), Medical Educator¹

CONFERENCE REPORT

Domenica Taruscio, MD, Director of Research²

1 WentWest Ltd, Sydney, NSW.

2 Centro Nazionale Malattie Rare, Istituto Superiore di Sanità, Rome, Italy.

Correspondence:

andrew.knight@wentwest.com; aknight@pnc.com.au

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