

**Patient support services for people with rare diseases and for
health professionals:**

A Creswick Fellowship Project Summary Report

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January 2011

Travel undertaken July 2009

Project Summary Report and Recommendations

Background and Rationale

There are between 5000 and 8000 rare diseases that affect approximately 6-10% of the population,¹⁻³ or an estimated 1.2 million Australians³, including ~400,000 Australian children. Rare diseases often begin in childhood, continue throughout life, are disabling, difficult to diagnose, and have a significant burden on children, their carers, health professionals and health resources, teachers and the community. Health professionals need access to current information to help diagnose manage, and prevent rare diseases and to improve the quality of life of affected children and the wellbeing of families. Overseas organizations such as Orphanet have provided integrated information services on rare diseases for many years.

For Australian families affected by rare diseases it is difficult to find and to access information, appropriate health services and support groups. Many small single-disease support groups exist but are under-resourced and struggle to provide support and up-to-date information for families. There are limited opportunities to interact with other families affected by rare diseases, and limited information on educational activities for families, teachers, and the community. These opportunities are even more limited for families who have a child who hasn't been diagnosed yet. In Australia there is a lack of coordination and integration, and no central point of access to information on diagnostic services, treatment clinics, family support groups or up-to-date research.

Families affected by rare diseases are often isolated, stigmatized, and stressed emotionally and financially. The value of linking families affected by rare diseases has been recognized overseas. Organizations such as Contact-a-Family provide one-on-one e-mail linking, telephone help line and a web-based forum. Eurordis provides similar facilities throughout Europe and links national rare diseases alliances. Some of these services have been operating for over 30 years. Gathering information from these long-established and experienced organizations will inform the development of similar services in Australia.

The Australian Paediatric Surveillance Unit (APSU, www.apsu.org.au) conducts surveillance and research into rare childhood conditions including genetic disorders, infectious diseases, rare injuries and mental health problems (45 conditions studied so far). This provides an excellent starting point from which to develop integrated information service for parents, patients, clinicians, teachers and others interested in rare diseases.

Organisations visited in July 2009

- British Paediatric Surveillance Unit
- Royal College of Paediatrics and Child Health
- Rare Diseases UK
- Contact-a-Family Head Office in London and the Wandsworth Project
- INVOLVE (DHS)
- Steps
- Orphanet
- European Organisation for Rare Diseases (Eurordis)

Key findings

- The European Commission has recently released a Communique to urge all EU countries to develop and implement integrated national plans to address the public health priority of rare diseases.
- The first French National Plan for rare diseases has been evaluated and the second National Plan for rare diseases is currently under development.
- National organisations devoted to supporting families with rare diseases, each with budgets of approximately €2,000,000 per annum and financially supported by governments, industry and charitable trusts have been operating in Europe and the United Kingdom for up to 30 years.
- Eurordis and Rare Diseases UK have specific transparent policies on accepting and using industry funding.
- National alliances of rare diseases support groups provide an efficient, integrated infrastructure for the dissemination of high quality information on many different rare diseases with a central access point for parents, patients, clinicians and the community. To improve accessibility information is provided via a number of methods: telephone help lines, websites, email alerts, podcasts, printed brochures and reports.
- In recognition of the relatively small impact that small organisations serving single rare diseases are likely to have in advocacy, national alliances of rare diseases support groups provide a united and strong platform from which to advocate to the community, health professionals and policy makers on behalf of all people affected by rare diseases.
- Inclusion of parents and patients within health professional bodies and in research enables these bodies to share information and to acknowledge the view of the communities they serve.
- Many research organisations and professional bodies have benefited from systematic inclusion of parents and patients in their organisational structures eg. The BPSU's Executive Committee and the RCPCH's Parent and Carer Advisory Committee.
- Families value the ability to access information on rare diseases via a central point
- Families value the opportunity to interact with other families via one-on-one email contact and via moderated on-line forums. Approximately 100 Australian families are registered with the UK based Contact-a-Family organisation, demonstrating a lack of a similar integrated support service in Australia for these families.
- National alliances provide an opportunity to conduct research on the impacts of rare diseases on families, to identify needs and to feed this information back to policymakers, health services and clinicians as demonstrated by the Euro Care surveys summarised in the "Voices of 12000 patients" published by Eurordis.
- Clinicians access professional information services such as Orphanet to learn about rare diseases, particularly when faced with diagnosing a rare disease.
- Clinicians also access information provided by parent/patient organisations to learn about rare diseases and the impacts they have on families.
- Inclusion of rare diseases as a topic within the medical curriculum in France raises awareness among future clinicians about rare diseases provides training on evidence-based resources where information on rare diseases can be accessed.

Recommendations

- Advocate for a stock-take of Australian facilities and supports for rare diseases including specialist clinics and parent/patient support groups
- Advocate for the establishment of an Australian Rare Diseases Network (ARDsNet) to provide a network of parent support groups, clinicians, clinical services and researchers. This overarching organisation would enable strong, united advocacy to government by representing many different small organisations which individually tend to have a weaker voice
- Advocate for the establishment of an ARDsNet website to link network members and to disseminate information and resources
- Use existing platforms such as Orphanet to disseminate information about Australian clinical trials, specialised clinics and educational opportunities.
- Advocate for the development of parent support services that link families affected by rare diseases, while building on existing services already provided by Australian groups such as AGSA, Genetic Alliance Groups, SMILE Foundation
- Develop information summaries for parents on the diseases already studied by the APSU and make them available on the APSU website with links to other organisations such as AWCH, Children's Hospitals Australasia, Medical Faculties in Universities and RACP Division of Paediatrics and Child Health
- Amend APSU procedures to include an information sheet for the public for every new surveillance study undertaken
- Include a consumer representative on the Scientific Review Panel of the APSU
- Liaise with the Royal Australasian College of Physicians Division of Paediatrics and Child Health regarding the introduction of patient/parent liaison at College level
- Seek funding to support research which aims to determine the impacts on and needs of families, clinicians and health services.

Table 1. Description of organisations visited in England and France (add website and e-mail)

Organisation and address	When established	Contact person interviewed	Primary purpose or mission	Governance	Funding Model
<p>Contact-a-Family 209-211 City Road LONDON EC1V1JN United Kingdom</p> <p>www.cafamily.org.uk</p>	1979	Louise Derbyshire and Sasha Henriques +44 020 76088715	<p>To provide information, linking and support services to children and families affected by rare diseases and those with disability</p> <p>Main Services Provided:</p> <ul style="list-style-type: none"> • E-mail linking of families and chat room (moderated) • Facilitate families meeting face to face • Free-call information service, pod-casts, on-line videos • ~70 information brochures on services and benefits • A large (compendium of brief fact-sheets on specific diseases (diagnosis, prognosis and treatment) including links to specific support groups and specialist clinics 	Registered UK Charity Board of trustees Medical Advisory Panel	<p>Registered Charity Reliant on donations Volunteers Funding via the Lady Hoare Trust</p> <p>Budget ~£4mill</p>
<p>Contact-a-Family Wandsworth Project 1 Siward Road Wandsworth LONDON SW17OLA</p>	1979	Rosie Noble +44 020 89475260	As above and to work through the local council area to provide information and support services including family visiting by family workers	Contact-a-Family structures and Wandsworth Borough Council structures.	<p>Local council support and funding from Contact-a-Family</p> <p>Funding for coordinator and family worker (part-time)</p>

<p>Rare Diseases UK Unit 40 Leroy House 436 Essex Road LONDON N13QP</p> <p>http://www.raredisease.org.uk</p>	2008	Alastair Kent + 44 020 77043141	<ul style="list-style-type: none"> To provide a means of communication about rare diseases and their impacts among patient organisations, professionals, industry and government; To influence policy and to develop a National Plan for Rare Diseases To advocate and raise awareness 	<p>Unincorporated Association Board of directors Five Working Parties:</p> <ul style="list-style-type: none"> Information Access Diagnosis and prevention Coordinated care Research Commissioning <p>Offshoot of the Genetic Interest Group (GIG)</p>	<p>Membership fees form organisations and individuals Mainly pharmaceutical industry companies</p> <p>Budget~ £90K</p>
<p>INVOLVE Involve, 33 Corsham Street, London, N1 6DR</p> <p>http://www.involve.org.uk</p>	1999	Helen Hayes +4402 380651088	<p>To support and facilitate the involvement of public and health users in health research Provide structure and resources needed by research and other organisations when involving consumers Undertake commissioned work for organisations</p>	NHS Programme initiative Small group	Funded via the Wellcome Trust and NHS
<p>STEPS Wright House, Crouchley Lane, Lymm, Cheshire WA13 0AS</p> <p>http://www.steps-charity.org.uk</p>	1987	Sue Banton	<p>To support and advocate on behalf of families who have children affected by a mobility problem related to feet legs or hips. Specifically: Supporting & guiding Training & educating Empowering & enabling Providing & connecting Seeking & campaigning</p> <ul style="list-style-type: none"> Information booklets, videos Blogs/on-line forums Helpline 	<p>Limited Company and registered charity Board of Trustees Parent Panel CEO</p> <p>Links with health professionals and other support organisations</p>	<p>Donations Volunteers</p> <p>Budget ~£180K</p>

<p>Orphanet INSERM Plateforme Maladies Rares 102 rue Didot 75014 Paris France</p> <p>http://www.orpha.net</p>	1997	Segolene Ayme and Charlotte Rodwell +33 1 56538141	<p>To provide an easily accessible information database on rare diseases that is available for health professionals and lay people. To advocate for clinician education and health service development</p> <p>Information on: 5781 Diseases; 4291 Clinics; 4486 Laboratories; 13440 Professionals; ~10,000 daily visitors to the site</p> <p>Developed a teaching module for undergraduates medical course</p> <p>Orphanet Journal of Rare Diseases</p> <p>Clinical guidelines</p>	<p>Programme within INSERM Rare Diseases Platform International Consortium led by a coordinating team in France</p> <p>Steering Committee (representatives from funding bodies)</p> <p>Management Board (~36 country coordinators)</p> <p>Each country has a Scientific Advisory Board</p>	<p>Funded by French Government Directorate for Health INSERM European Union</p> <p>Grants</p> <p>Financial and in-kind contributions from member organisations</p> <p>Industry</p> <p>Budget: €3mill</p>
<p>Eurordis INSERM Plateforme Maladies Rares 102 rue Didot 75014 Paris France</p> <p>http://www.eurordis.org</p>	1997	Anja Helm Anna Cole	<p>To provide an information service for patients and families affected by rare diseases, To facilitate interaction among patients and families, To identify gaps in service provision; To advocate on behalf of rare diseases community To conduct research on issues relevant to patients</p> <p>Provides information, advocacy activities, linking of families – online chat/communities, blogs, podcasts, educational resources</p>	<p>Europe-wide organisation Coalition of National Rare Diseases organisations from ~30 countries</p>	<p>European Commission Member organisations Grants Pharmaceutical companies Volunteers</p> <p>Budget: €2mill</p>

RCPCH Theobald's Road London WC1X 8SH http://www.rcpch.ac.uk British Paediatric Surveillance Unit	1986	Richard Lynn	To conduct research on rare childhood diseases; Provide information to the public about rare diseases being studied Information sheets for the public on conditions being studied Media after publication of results to reach wider community	Royal College of Paediatrics and Child Health BPSU Executive Board (inc consumers) Scientific Coordinator	DHS grant RCPCH support
RCPCH Children and Young People Participation Program	2008	Bharti Mehan Children and Young People Participation Manager	To develop strategies to include patients' interests and views in the College	Processes and structures currently under development Feeds into existing College structures eg. Advocacy Committee	RCPCH funds manager
RCPCH Parent and Carer Advisory Committee	2007	Ann Sanson	To represent the interests of parents and carers to the RCPCH and to provide input into college policy, paediatrician training, and service provision in paediatrics	Parent and Carer Advisory Committee consisting of 5 paediatricians and 5 parents/carers	RCPCH provides secretarial support for the committee and covers expenses

