Improving outcomes for rare diseases in Australia:

A National Plan for Rare Diseases

Equal rights to information, services and quality of life for people with rare diseases

A "rare" disease has been variously described as one:
1. with a prevalence of 1 in 2000 people or less (European Organisation for Rare Diseases)
2. affecting less than 200,000 people in the population (National Organisation for Rare Diseases, USA)
3. with prevalence of 1 in 10,000 people or less (Therapeutic Goods Authority, Australia)

One difficulty of accurately estimating prevalence or incidence rates for rare diseases is the lack of population based epidemiological research on rare diseases. This is compounded by a general perception that rare diseases have minimal impact and are therefore considered less important and a low priority for research. Paradoxically, however, rare diseases are common. There are approximately 8000 known rare diseases, collectively affecting up to 10% of the population or over 2 million Australians including ~400,000 Australian children. Rare diseases have the following common features:

- Most begin in childhood and continue throughout life
- Obtaining a definitive diagnosis is often difficult and delayed
- Many rare diseases have no cure but some can be prevented
- Neurological and intellectual disabilities occur in about half of all cases regardless of disease type and lead to loss of independence and opportunities
- Families experience isolation, psychological and financial stress
- Health professionals have inadequate access to information, education and resources
- Of deaths in children aged < 1year 35% are due to rare diseases
- Of deaths in children aged between 1 and 15 years, ~10% are due to rare diseases

Equity in access to health and community services is lacking for people affected by rare diseases.

The impacts of rare diseases go beyond affected individuals. They have wide-reaching impacts on families and carers (mostly women), health professionals, health resources and the wider community. The significance of this has been recognised by many countries including the US, Canada, UK, and at least 16 countries in the EU who have responded to the burden of rare diseases by developing co-ordinated national plans outlining strategies in favour of rare diseases. On 9th June 2009 an EU Council Recommendation on a European Action in the Filed of Rare Diseases called on all EU member states to implement national plans and strategies for rare diseases ensuring that patients with rare diseases have access to appropriate high quality health care – this is a recognised ***human right***.

In Australia there is a general lack of awareness of the impacts of rare diseases, and lack of strong and coordinated advocacy. However, Australia is in an excellent position to learn from extensive international initiatives. The Australian Paediatric Surveillance Unit convened the National Rare Diseases Task Force to develop, seek wide endorsement and to advocate for the implementation of a national plan to support research, education, service development, to promote awareness of rare diseases and advocacy for affected individuals and their carers.

The Task Force met on the 9th of February 2009 and agreed on the following **eight principles** for the Australian National Plan for Rare Diseases:

1. **Raise awareness** of the burden of rare diseases on patients, families, health professionals and the community
2. Provide **educational resources and networking opportunities for health professionals** to allow them to better identify and manage rare diseases
3. **Improve health care for people with rare diseases** through access to diagnostic tests, new drugs and other treatments, improved primary care and specialised services
4. **Promote research** on rare diseases through advocacy for targeted research funds and development of national and international multidisciplinary research partnerships.
5. **Increase knowledge** of the epidemiology and impacts of rare diseases in Australia through research
6. Develop and disseminate information to **educate patients, parents, carers and the general public**, about rare diseases that is relevant in the Australian context
7. **Develop an umbrella organisation** to support people affected by any rare disease by linking existing organisations to facilitate the co-ordinated development of integrated peer support networks, contact among families and contact among rare diseases interest groups.

DRAFT

8. **Advocate to government** in partnership with families, for people affected by rare diseases

The Need for a National Plan

A national plan provides a platform and framework for the development of strategies

for the implementation of the eight principles identified

In Australia, people affected by rare disease do not have a coherent strategy for access to appropriate healthcare and support. Rare diseases are often chronic and complex, requiring many different specialised health services and community support. Unfortunately, people affected by rare diseases, have difficulty accessing health services and the poor coordination among existing health services results in sub-optimal care. Furthermore, healthcare professionals are often ill-equipped to manage patients with rare diseases due to lack of educational opportunities and lack of easily accessible information, expert advice, and optimum care pathways. Better coordination, and innovative models of access to expertise that is often dispersed geographically and across disciplines and sectors, would optimize the benefits offered to people affected by rare diseases.

In Europe, excellent models of coordinated multidisciplinary clinics that accept patients with many different rare diseases have been shown to provide significant savings when compared with the conventional often fragmented services. In Australia, there are excellent models of multidisciplinary clinics for single rare diseases (eg. Rett Syndrome) providing a template for the development of other clinics. The utility of such clinics and other centres of clinical excellence in rare diseases could be optimised by offering virtual access to health professionals around Australia via videoconferencing, secure internet access, or telephone. Similar, virtual clinic models are being developed in Europe.

As well as highly specialised services, patients with rare diseases require local support, general medical care and skilled coordination of services. The role of primary care in rare diseases has been neglected till now. Australia has an opportunity to model comprehensive joined up care across all sectors of the health system for patients with rare diseases.

Developing strategic planning for rare diseases will improve the current fragmentation of services and enable patients and health professionals to provide and use best practice care. Much progress has been made in the field in this past decade in Europe and the USA, with the establishment of the European Union Task Force for Rare Diseases; the implementation and evaluation of the French National Plan for Rare Diseases; adoption of national plans by approximately 16 European nations; establishment of the Office for Rare Diseases and the Undiagnosed Diseases Program in the National Institutes for Health in the USA. All of these initiatives strive to improve healthcare and wellbeing of people with rare diseases by developing innovative services and evidence-based policy.

In 2002 the USA passed the Rare Diseases Act and established the Office for Rare Diseases[20] within the National Institutes of Health to promote research into rare diseases, stating:
"Because of the small number of affected patients in any one location, rare disease research requires collaboration of scientists from multiple disciplines and the capacity to share access to geographically distributed national research resources and patient populations. Knowledge about rare diseases may offer leads for scientific advancement in other rare diseases and in more common diseases."

Furthermore, in June 2009 the European Council's Recommendation on an Action in the Field of Rare Diseases, was adopted unanimously by EU Health Ministers, stressing the importance of strengthening research programmes into rare disease, encouraging the development of national rare disease policies, and developing and sharing common policy guidelines such as access to good quality information.

There has been no coordinated approach for the development of health services and policy in favour of Australians affected by Rare Diseases. The Rare Diseases Task Force aims to work in partnership with government, both federal and state, to develop a National Plan for Rare Diseases in Australia and to determine strategies for its implementation.

A coordinated National Plan for Rare Diseases will bring together patients, support groups, clinicians and researchers, supporting best practice and clinical policy for the treatment of rare diseases. Furthermore, the establishment of a comprehensive, website devoted specifically to rare diseases: Australian Rare Diseases Network (ARDNet), will improve ease of access to the most up-to-date information and educational resources on rare diseases to the public, patients and health professionals in Australia.

DRAFT

Expected key benefits from the adoption of a National Plan for Rare Diseases in Australia

1. A coordinated research strategy and targeted research funding will:

a. Generate new knowledge about rare diseases and their impacts
b. Enable the development of an evidence base to support the development of appropriate health and community services
c. Identify new research priorities
d. Enable sharing of knowledge and expertise among researchers and institutions

In 2002The Office for Rare Diseases in the National Institutes for Health in the USA was established because…..
"…of the small number of affected patients in any one location, rare disease research requires collaboration of scientists from multiple disciplines and the capacity to share access to geographically distributed national research resources and patient populations. Knowledge about rare diseases may offer leads for scientific advancement in other rare diseases and in more common diseases."

3. Coordinated provision of healthcare for people with rare diseases will result in:

- **Patient centred care which is streamlined across the health system from general practice to tertiary care offering better outcomes and cost savings**
- **Earlier diagnosis, intervention and secondary prevention**
- **Reduced stress for individuals and families**

Parent of a child with a rare disease, as yet undiagnosed: "I had to find the right doctors and to be insistent but not obstinate. On the one hand, I had to save Mathilda from unnecessary diagnostic tests, often saying 'no' and 'why?' On the other hand, I had to ensure that she received everything she needed." [7]

3. An umbrella organisation that brings together single-disease support groups will enable for more effective advocacy, giving a stronger and more united voice to people with rare disease in Australia.

"Rare diseases patients and their representative organisations have already played an active role in shaping rare disease policies…." Yann Le Cam, CEO European Orgnisation for Rare Diseases, in The Voice of 12,000 patients. Experiences and expectations of Rare diseases patients on diagnosis and Care in Europe. Eurordis 2009 (www.eurordis.org)

4. Opportunities to meet other affected individuals and families and easy access to information for patients, families, carers and the community will reduce isolation, fear and stress for families affected by rare diseases.

"Obtaining the correct diagnosis has had significant implications for us ……We were able to understand and accept Rett syndrome…We are finally able to explain to others, when they ask, what is wrong and to foster more realistic expectations of our child's future including planning ahead for equipment and schooling and also to further extend our family." Mother of a child diagnosed with Rett Syndrome.

Key references:

Zurynski Y, Frith K, Leonard H, Elliott E. Rare childhood diseases: how should we respond? *Arch Dis Child*. 2008 Dec;93(12):1071-4.

French National Plan for rare Diseases 2005-2008. Ensuring equity in the access to diagnosis, treatment and provision of care. 2004. Available from: http://www.orpha.net/actor/EuropaNews/2006/doc/French_National_Plan.pdf

Zurynski Y, Reeve K, Elliott E. International conferences on rare diseases: initiatives in commitment, patient care and connections. *MJA* 2007, 187(10):597 (Letter)

Schiepatti A, Henter JI, Darina E, Aperia A. Why rare diseases are an important medical and social issue. *Lancet* 2008; 371:2039

Zurynski YA, Peadon E, Bower C, Elliott EJ. Impacts of national surveillance for uncommon conditions in childhood. *J Paed Child* Health 2007; 43:724-731

National Plan for Rare Diseases

1. Raise awareness of the burden of rare diseases on families, health professionals and the community

Rationale and Context

Individual rare diseases are perceived as having limited impact and limited importance. However, when taken collectively, the ~8000 known rare diseases impact on a significant proportion of our population (~400,000 children)[1-3]. Small, often poorly resourced support groups for individual rare diseases have limited impact for raising awareness. This limitation has been recognised overseas by the formation of umbrella organisations such as the National Organisation for Rare Diseases (NORD)[4], European Organisation for Rare Diseases (EURORDIS)[3], Canadian Organisation for Rare Diseases (CORD)[4] and New Zealand Organisation for Rare Diseases (NZORD)[5], which coordinate education, services, support and advocacy for people with rare diseases.

In Australia we need a strong united voice to raise awareness of rare diseases. To achieve this we need to implement a national rare diseases plan and develop an organisation similar to those developed overseas.

Organisations representing sufferers and their families, health professionals, and researchers are excellent advocates for rare diseases. Bringing people from all of these organisations together under one umbrella will ensure a united voice and dissemination of consistent messages that can be effectively delivered to many stakeholders to raise awareness of rare diseases among health professionals, government, and community. More importantly, such umbrella organisations have proven that they can be effective agents of change for rare diseases policy and clinical practice.

Objectives

To raise awareness of the importance of rare diseases by:

- Implementing a National Rare Diseases Plan
- Establishing a national umbrella organisation for rare diseases in Australia
- Developing media awareness campaigns
- Joining international awareness campaigns
- Engaging with health professional groups, the wider community, parent support groups, philanthropic groups, and government

Strategies

- Develop the **Australian Rare Diseases Network (ARDNet)** an umbrella organisation that aims to raise awareness of rare diseases in Australia. APSU is currently seeking seeding funding to develop the structure and content for ARDNet.
- To recruit wide membership of users to ARDNet, including parent and carer organisations, individuals, specialised services (clinical and social), health professionals, and peak professional organisations.
- Join the global effort to raise awareness through participation in Global Rare Diseases Day 28th February every year.
- Engage relevant groups and individuals in the development of ARDNet and in efforts to raise awareness for rare diseases eg. Rare Diseases Day

DRAFT

2. Provide educational resources and networking opportunities for health professionals to allow them to better identify and manage rare diseases

Rationale and Context

Rare diseases pose a challenge to clinicians working in primary, secondary and tertiary health settings.[1,7,10,28] For many rare diseases standardised diagnostic criteria do not exist, resulting in multiple, often avoidable investigations, and diagnostic delays of a year or more are common.[1,7,10,14, 28]

For example, in Australia, a diagnosis of Rett syndrome is frequently delayed, sometimes for years.[14] In a recent survey only 19% of Australian paediatricians knew the diagnostic criteria for fetal alcohol syndrome. Furthermore 70% were concerned that making a diagnosis would stigmatise the child.[29]

Once a diagnosis is reached, clinicians are challenged by lack of management guidelines, referral pathways and peer support services for their patients.[26-29] Difficulties with diagnosis and management often stem from lack of knowledge, lack of evidence-based information, or difficulty accessing information.[1,7,10,28] Because there are approximately 8000 rare diseases, it is unrealistic to provide training to health professionals about every disease. Nevertheless, primary care clinicians and paediatricians believe there is insufficient knowledge about rare diseases and want access to evidence-based information for themselves and their patients about diagnosis, management and specialised referral clinics.[1,6,7,28-31]

In France, a module on rare diseases is offered in the undergraduate medical program to raise awareness of rare diseases among future clinicians and to provide training on accessing evidence-based rare diseases databases such as Orphanet.

Objective

- Improve awareness of and knowledge about rare diseases among health professionals
- Provide networking opportunities for health professionals and access to existing expertise

Strategies

Support Australian health professionals who look after patients with rare diseases by:

- Disseminating information and providing links to existing clinical treatment guidelines and fact sheets for rare diseases via a comprehensive national information portal
- Facilitating the development of new diagnostic and clinical treatment guidelines
- Developing new easily accessible, evidence based educational resources for clinicians including fact sheets on specific diseases, diagnostic tests, specialised services and referral pathways
- Identifying rare diseases experts and specialised clinics who can provide information and advice to health professionals
- Provide opportunities for networking among health professionals looking after people with rare diseases.
- Providing training for medical undergraduates on rare diseases and the use of existing educational resources and databases

DRAFT

3. Improve health care for people with rare diseases through access to diagnostic tests, new drugs and other treatments and specialised services

Rationale and Context

Access to appropriate health services has been described as a "post code lottery"[10] where 6% of people in the USA with rare diseases had to move home, and almost half had to travel more than 50 miles to access appropriate medical services.[2] In Europe, a quarter of patients with rare diseases had to travel to a different region and 2% to a different country for diagnosis,[11] and 26% of patients found it difficult or impossible to access medical services.[10,11] In Australia, a population-based longitudinal study of Rett syndrome highlighted difficulties in accessing services, including lack of services and appropriately trained therapists.[12] Transition from paediatric health services to the adult health setting is also recognised as a significant problem including for girls affected by Rett syndrome.[12]

Diagnostic delays for rare diseases are common due to under-recognition by health professionals and limited access to diagnostic services. Although many rare diseases are incurable, parents value a definitive diagnosis because this allows them to understand what is wrong with their child, to plan for ongoing care, and for future pregnancies.[13] Having a diagnosis alleviates some of the stress and powerlessness associated with not knowing.[13-16] Multidisciplinary, integrated, clinics that treat children with many different rare diseases have operated in Europe since 1989. These clinics are more cost effective for treating children with rare diseases than conventional health services.[17]

The Undiagnosed Diseases Program, an innovation of the NIH (USA) brings together experts from many institutions and is accessible to patients with longstanding undiagnosed diseases and their health professionals.[18] Although diagnostic tests for many rare diseases are becoming increasingly available, access to these is difficult due to lack of availability in Australia and prohibitive costs. Genetic counselling and screening services are also important for prevention of rare diseases. However, information about such services and ways of accessing them is not readily available to Australian health professionals.[1,7] There is a need for clear pathways for patients between general medical services and expert resources, and for the sharing of expertise and information across all health sectors.

The development of new drugs for rare diseases is supported by legislation both overseas and in Australia. The Life Saving Drugs Program, which is essentially the Orphan Drug Program[19] fast tracks the approval of drugs for release to the Australian market, however, few drugs have been approved under this scheme since its introduction in 1998, and costs of many drugs remain extremely high, preventing access for may people with rare diseases.

Objectives

- Improve availability and visibility of information about health services for rare diseases in Australia.
- Improve the quality of and access to health services for patients with rare diseases.
- Improve access to new treatments and diagnostic tests to people with rare diseases or those who may not yet have a definitive diagnosis
- Capitalise on existing models of healthcare delivery eg. centres of excellence for rare diseases

Strategies

- Provide e.g. through ARDNet, current evidence-based information about diagnosis, screening, genetic counselling, health services and orphan drugs for rare diseases and links to reputable websites.
- Identify specialised clinics and clinicians with expertise in rare diseases and link these experts to provide a "virtual centre of expertise" that could be accessed by health professionals working in primary, secondary and tertiary health services.
- Develop strategies for improved linkage of primary care with specialised services
- Advocate for the development of integrated health services for rare diseases to streamline and coordinate patient care, including the transition from child services to adult services
- Advocate for the extension of the Life Savig Drugs Program

DRAFT

4. Promote research on rare diseases through advocacy for targeted research funds and development of national and international multidisciplinary research partnerships.

Rare diseases are under-researched in Australia, resulting in a lack of evidence for translation into clinical practice and health and social policy. In 2002 the USA passed the Rare Diseases Act and established the Office for Rare Diseases[20] within the National Institutes of Health to promote research into rare diseases, stating:

"Because of the small number of affected patients in any one location, rare disease research requires collaboration of scientists from multiple disciplines and the capacity to share access to geographically distributed national research resources and patient populations. Knowledge about rare diseases may offer leads for scientific advancement in other rare diseases and in more common diseases."

Stimulating research effort into rare diseases is a priority in the USA and Europe, where a significant amount of targeted funding has been allocated. For example in 2007 Italy allocated €8.3 million p.a. for basic and clinical research and clinical trials.[21,22] There has also been considerable investment in the establishment of *reference centres* throughout Europe to bring together multidisciplinary expertise on rare diseases. This has provided the critical mass required to conduct research on complex, rare conditions. Rare diseases are not considered a health or research priority by governments and funding agencies in Australia, yet the economic impacts of rare diseases are enormous. For Example in the USA the medical costs alone of Fetal Alcohol Syndrome amount to US$3.6 billion annually, and the lifetime cost per individual is estimated at US$2.9 million.[24]

Early and accurate diagnosis and appropriate management will reduce the costs and morbidity associated with rare diseases and will enable prevention.

For diseases of low frequency national and international research collaborations are essential to provide adequate case number for meaningful interpretation and to bring together specialised multidisciplinary expertise.[1,20]

Objectives

- Identify knowledge gaps in Australia and set priorities for research
- Identify potential national and international collaborators
- Promote scientific and social research on rare diseases

Strategies

- Review Australian data and publications relating to rare diseases identify gaps and set priorities for research
- Identify groups and key individuals currently involved in rare diseases research in Australia and facilitate communication, networking and collaboration among these groups
- Identify potential international research collaborators
- Advocate to the NHMRC, ARC and other funding bodies for allocation of targeted funding for rare diseases research
- Through ARDNet, provide infrastructure to support research on the social and economic impacts of rare diseases on families and health professionals
- Establish infrastructure to support research into rare diseases eg. enabling establishment of disease-specific registries and a single repository for registry data that is accessible to researchers. This would save scarce resources and provide efficiencies of scale in terms of registry administration and access to data by researchers

DRAFT

5. Increase knowledge of the epidemiology and impacts of rare diseases in Australia through research

Rationale and Context

In Australia there has been limited support for the systematic collection and provision of accurate data to inform the development of effective policy, health and community services for rare diseases.[1,7,8] In recognition of the importance of high quality data to inform policy development, governments overseas have prioritised support for research and surveillance to provide the following information about rare diseases:

- Incidence, prevalence, geographical distribution, morbidity and mortality
- Psychological impacts on affected individuals and families
- Social impacts: including wellbeing of patients, families and siblings, and impact on the community
- Financial costs to: hospitals, health services, schools, families, siblings, caregivers, and community
- Burden of care faced by health professionals and health services

In Australia there has been little investment in support of existing surveillance systems and registries or in development of new ones, resulting in a piece-meal approach. Although the following national organisations are devoted to the collection of data on rare diseases in Australia, none is adequately resourced:

- Australian Paediatric Surveillance Unit (APSU) – established in 1993 to collect prospective national epidemiological data simultaneously on a range of childhood diseases simultaneously.[9]
- Paediatric Active Enhanced Disease Surveillance (PAEDS) is a hospital based active surveillance system that collects data on children admitted to hospital with selected rare conditions, currently being piloted in four tertiary paediatric hospitals in four different states.
- The National Perinatal Statistics Unit collates data from state birth defects registries to provide a national dataset.
- Numerous rare diseases registries, eg the Australasian Registry for Orphan Lung Diseases (www.ARNOLD.org.au), the Autism Register, Primary Immunodeficiency Register, etc
- Australian Maternal Outcomes Surveillance System (AMOSS) will begin data collection in the second half of 2009

Objectives

- To increase knowledge and promote access to rare disease data collections
- To enable translation of knowledge into policy and clinical practice
- To improve data linkage among existing sources
- To support existing surveillance systems and registries and develop new mechanisms for data collections as required
- To support research to evaluate the impact of rare diseases on families, individuals, health professionals, health services, and communities

Strategies

1. Identify Australian organisations collecting data on rare diseases; describe the type of data collected, its accuracy, timeliness, and usefulness; determine similarities and synergies among data collections, and develop priorities for improving data quality and filling knowledge gaps.
2. Improve access to, and dissemination of, currently collected data
3. Determine the impacts of rare diseases on patients, families and their specific service and support needs through research in collaboration with family support groups.
4. Determine the impacts and needs of clinicians managing rare disease with regard to education and support (eg. APSU will survey the 1300 paediatricians who regularly report cases of rare diseases to the APSU)
5. Audit hospital admissions for rare diseases to demonstrate their impact on tertiary services. (APSU has begun this process.)
6. Seek adequate funding from state and federal government to support existing surveillance systems and registries for rare diseases and develop new data collections if required.
7. Enable data linkage among different sources

DRAFT

6. Develop and disseminate information to educate parents, carers and the general public, about rare diseases that is relevant in the Australian context

Rationale and Context

Australian families affected by rare diseases, need accurate, accessible and locally relevant information, about rare diseases including:

- lay descriptions of signs, symptoms and prognosis;
- opportunities to access educational courses and other educational resources;
- lists of available health and community services and peer support groups.

The internet is increasingly used as a source of information and support by families affected by rare diseases.[25-27] In Europe, the UK, USA, Canada and New Zealand, national web portals provide information on rare diseases, health and social services for rare diseases, educational opportunities for families, access to family support groups and opportunity to contact similarly affected individuals. Examples include "Contact a Family" in the UK,[26] The European Organisation for Rare Diseases (Eurordis) and The Rare Disease Patient Solidarity Project, OrphaNet (www.orpha.net),the National Organisation of Rare Diseases (NORD) in the US and the Canadian Organisation for rare diseases (CORD) and New Zealand Organisation for Rare Diseases (NZORD). A number of groups in Australia provide information on rare diseases, especially organisations focussed on genetic diseases, many of which are classified as rare. Association of Genetic Support Australasia (AGSA) is an umbrella organisation providing support and information for people with genetic conditions and their families (www.agsa-geneticsupport.org.au). The Centre for Genetic Education (NSWHealth) provides a directory of ~750 support groups for genetic diseases, many of which are classified as rare as well as providing lay language fact sheets. The SMILE Foundation provides small relief grants for families affected by rare diseases (www.smilefoundation.com.au), and the Steve Waugh Foundation supports children with very rare diseases by providing grants for equipment and services that cannot be accessed though other means (www.stevewaughfoundation.org.au). Association for the Wellbeing of Children in Healthcare (AWCH) provides support for children affected by serious diseases and their families (www.awch.org.au).

Many of the organisations providing resources on rare diseases are small and under-resourced and often unable to maintain the currency of the information provided. Overseas, national centralised rare diseases organisations linking multitudes of small, single disease support groups are common. In Australia however, there is little linkage and communication or coordination among organisations. Furthermore, there is a general lack of locally relevant resources for parents, teachers, childcare and community workers. There is no centralised organisation for rare diseases and no coordinated effort to provide information or educational opportunities.

Objective

- Improve access to information about rare diseases and educational opportunities for Australian communities

Strategies

Bring available resources eg. Genetic Education Services, disease support groups, educational organisations and research groups providing education on rare diseases under an umbrella organisation for rare diseases and disseminate information via a centralised portal eg. ARDNet to provide:

- Evidence-based and regularly updated information about rare diseases including summaries of latest research in lay language
- Information about health, respite and community services for people with rare diseases
- Educational opportunities for affected families, educators, childcare workers and others in the community
- Opportunities to contribute to the research effort for rare diseases
- Enable organisations already conducting research into rare diseases (eg. APSU) to provide patient focused summaries of rare diseases

DRAFT

7. Develop an umbrella organisation to support families affected by rare diseases by facilitating the development of integrated peer support networks, contact among families and contact among rare diseases interest groups.

Rationale and Context

Families affected by a rare disease are under-supported, overburdened, and experience financial hardship[1,2,11,-14, 32] These problems are common for most families affected by rare diseases, regardless of the specific disease. Peer support has been shown to be beneficial and to improve coping. However, the low frequency of individual rare diseases and the dispersed geography of the Australian population pose difficulties when seeking peer support for rare diseases.

Web portals developed overseas and described under Aim 6, are a useful way of facilitating contact among affected families via email, blogs, and on-line chat rooms. Furthermore, these web portals facilitate the development of new support groups leading to regular face to face meetings to discuss common issues as well as providing opportunity for outings and other social occasions where families can interact.

In Australia there is no overarching patient support organisation to facilitate contact among affected families or the development of peer support networks.

Objective

- Improve opportunities for peer support among families affected by rare diseases and improve opportunities for support groups to share information and resources

Strategies

- Use existing support groups for rare diseases (eg. AGSA, Rett Syndrome Associations etc.) as a basis for the development of an umbrella organisation that provides opportunities for networking and resource sharing
- Under the umbrella organisation (ARDNet) develop a family linking service similar to that developed by Contact-a-Family in the UK to bring individuals and families together for peer support
- Develop and provide access to an on-line e-mail service or teleconferencing service for families affected by rare diseases to facilitate peer support, especially for geographically dispersed families
- Enable ARDNet to facilitate the development of new peer support groups for rare diseases

DRAFT

8. Advocate to government in partnership with families, for people affected by rare diseases

Rational and Context

Overseas, the introduction of rare diseases policy resulted largely from lobbying by patient advocacy groups such as NORD and Eurordis. Similar umbrella organisations representing rare diseases sufferers and their families have been established in Canada (CORD) and New Zealand (NZORD) and both organisations are advocating for national government plans in their countries.

On February 29th 2008, Eurordis, NORD and many other rare diseases organisations around the world held the first Rare Disease Day to raise awareness of rare diseases and to lobby governments to respond to the significant impacts of rare diseases. In Europe, the 2008 Rare Diseases Day culminated in a Public Hearing on rare Diseases at the European Parliament in Brussels.[33]

In response to lobbying by individuals with rare diseases, parents and health professionals, the USA passed the Rare Diseases Act in 2002 and established the Office for Rare Diseases within the National Institutes of Health to promote research and peer support for rare diseases.

These examples illustrate the need for a coordinated approach to advocacy in Australia.

Objective

- Advocate for the implementation of the national plan for rare diseases

Strategies

- Develop an umbrella organisation for rare diseases in Australia (ARDNet) whose remit is to advocate for people affected by rare diseases
- Join the global efforts to raise awareness of rare diseases and to advocate for people with rare diseases during Rare Disease Day, February 28th or 29th each year
- Use evidence generated by research to advocate to government for a coordinated response to rare diseases
- Develop clear and consistent messages to be delivered via the media to advocate for people with rare diseases

DRAFT

APPENDIX

French National Rare Diseases Plan 2005-2008

Ten strategies to ensure 'equity in the access to diagnosis, to treatment and to provision of care' for people with rare diseases:

•Increase knowledge of the epidemiology of rare diseases

•Recognise the specificity of rare diseases

•Develop information for patients, health professionals and the general public concerning rare diseases

•Train health professionals to better identify rare diseases

•Organise screening and access to diagnostic tests

•Improve access to treatment and the quality of patient care

•Continue efforts in favour of orphan drugs

•Respond to the specific accompaniment needs of people suffering from rare diseases

•Promote research on rare diseases

•Develop national and European partnerships

DRAFT

References

1. Zurynski Y, Frith K, Leonard H, Elliott E. Rare childhood diseases: how should we respond? *Arch Dis Child*. 2008 Dec;93(12):1071-4. Epub 2008 Aug 6.
2. Kramer MG. The National Organisation for Rare Diseases and the experiences of the rare disease community. National Organisation for Rare Diseases (NORD), January 2003. Available from: http://www.rarediseases.org/pdf/Eval_Final_Paper.pdf (accessed 30 May 2008).
3. European Organisation for Rare Diseases. Rare Diseases: understanding this Public Health Priority. November 2005. Available from: http://www.eurodis.org/IMG?pdf/princeps_document-EN.pdf (accessed 30 May 2008).
4. Canadian Organisation for rare Diseases (CORD) http://www.cord.ca/
5. New Zealand Organisation for Rare Diseases (NZORD) http://www.nzord.org.nz/
6. Dan B. The paediatrician's role in support groups for rare diseases. *Acta Paed* 2008; 97:1510-1511
7. Knight AW, Senior TP. The common problem of rare disease in general practice. *Med J Aust* 2006;185:82-3.
8. Zurynski Y, Reeve K, Elliott E. International conferences on rare diseases: initiatives in commitment, patient care and connections. (Letter), *MJA* 2007, 187(10):597
9. Zurynski YA, Peadon E, Bower C, Elliott EJ. Impacts of national surveillance for uncommon conditions in childhood. *J Paed Child* Health 2007; 43:724-731
10. Le Cam Y. Patients' needs and expectations concerning access to health services – Eurodiscare 3 study. In: 4th European Conference on Rare Diseases, Conference Report, 2007. Available from: http://www.rare-diseases.eu/2007/IMG/File/ECRD%202007%20report.pdf (accessed 30 May 2008)
11. European Organisation for Rare Diseases (Eurodis). EurodisCare 2 Survey 2004. Available from: http://www.eurordis.org (accessed 30 May 2008).
12. Moore H, Leonard H, de Klerk N, et al. Health service use in Rett syndrome. J Clin Neurol 2005; 20:42-50.
13. Knott, Marisa. Diagnosing Rett Syndrome – My experience. Available from: www.apsu.org.au/.../**Rett**%20workshop/M**KnottRett**%20**Syndrome**%20clinician's%20workshop%20talk.pdf (accessed 30 May 2008).
14. Dunkelberg S. A patient's journey: our special girl. *BMJ* 2006; 333:430-431.
15. Tooth reveals cause of child's death. *Sydney Morning Herald*, 15 Nov 2005.
16. Russell E. *Alcohol and Pregnancy: A mother's responsible disturbance*. Zeus Publications, 2005.
17. Olauson A. The Agrenska Centre. A Socioeconomic Case Study of Rare Diseases. *Pharmacoeconomics* 2002;**20**(Suppl 3):73-75.
18. Office of Rare Diseases. Undiagnosed Diseases Program. Available from:

http://rarediseases.info.nih.gov/Resources.aspx?PageID=31&PHPSESSID=46125b6f7f3d86c1c9f50d4236d22ca2 (accessed 30 May 2008)

19. Australian Therapeutic Goods Administration. Orphan Drug Program. http://www.tga.gov.au/docs/html/tganews/news25/dseb.htm
20. Office of Rare Diseases, National Institutes of Health (NIH). Available from: http://rarediseases.info.nih.gov/ (accessed 30 May 2008).
21. Ayme S. Overview of member states public health policies for rare diseases: 2007 update. The Rare Diseases Task Force. Available from: http://www.eurordis.org/IMG/pdf/ECRD2007-Ayme.pdf (accessed 30 May 2008).
22. Rare Diseases Task Force. Centres of reference for rare diseases in Europe: State-of-the-art in 2006 and recommendations of the rare Diseases Task Force. December 2006. Available from: http://ec.europa.eu/health/ph_threats/non_com/docs/contribution_policy.pdf (accessed 30 August 2007)
23. SMILE Foundation. http://www.smilefoundation.com.au
24. Lupton C, Burd L, Harwood R. Cost of Fetal Alcohol Spectrum Disorders. *Am J Med Genet C Semin Med Genet* 2004; 127:42-50.
25. Lasker JN, Sogolow ED, Sharim RR. The role of an online community for people with a rare disease: Content analysis of messages posted on a primary biliary cirrhosis mailinglist. *J Med Internet Res* 2005; **7**: e10.
26. Contact a Family. Available from: http://www.cafamily.org.uk/ (accessed 19 November, 2007)
27. RAPSODY, the Rare Diseases Patient Solidarity Project. Available from: http://eurordis.org/article.php3?id_article=1066 (accessed 19 November, 2007)
28. Phillips WR. Zebras on the commons: rare conditions in family practice. *J Am Board Fam Pract* 2004;17:283-6.

DRAFT

29. Elliott EJ, Payne J, Haan E, Bower C. Diagnosis of foetal alcohol syndrome and alcohol use in pregnancy: a survey of paediatricians' knowledge, attitudes and practice. *J Paediatr Child Health* 2006;42:698-703.

30. Cullen RJ. In search of evidence: Family practitioners' use of the internet for clinical information. *J Med Libr Assoc* 2002; 90:370-379.

31. Williamson S and Christodoulou J. Rett Syndrome: new clinical and molecular insights. *Eur J Human Genet* 2006; 14:896-903

32. Laurvick CL, Msall ME, Silburn S, *et al.* Physical and Mental Health of Mothers caring for a Child with Rett Syndrome. *Pediatrics* 2006;**118**:1152-1164.

33. Eurordis. Rare Disease Day 2008 was a huge success. Available from http://www.eurodis.org/article.php3?id_article=1707 (accessed 30 May 2008).

DRAFT