

Title

A systematic approach to diagnosis, screening, referrals, information, clinical care, treatment, rehabilitation, disability support, registries and clinical research for rare disorders.

Type of Referral

- New model of care/programme

Description

The establishment of a rare diseases action plan that recognises the significant public health burden of rare disorders, estimated to affect approximately 8% of the whole population. That is approximately 350,000 New Zealanders whose health needs are often poorly understood and inadequately met.

The rare diseases action plan would ensure co-ordinated effort to improve systems and models of care to accurately identify these conditions early, minimise their impact, improve health outcomes and quality of life for affected patients and families, and promote registries as the interface between clinical care and research, to assist improvements in clinical care and therapy development for them.

Area(s)

Category: *Rare Disorders.* The European Union definition of prevalence less than 1 in 2,000 in the population, is a well established measure of rarity in health conditions, and is a recommended definition here in New Zealand.

Condition, disease and/or disability type: Any disease affecting less than 1 in 2000. Most are inherited genetic conditions, but the total of an estimated 9,000 rare disorders may also result from sporadic, cancerous, congenital malformation, infectious, or gene-environment interaction causes.

Service specialty: Covers all services and disease types.

Target Population

Target Group/Clinical Indication(s): Rare diseases defined as a prevalence of less than 1 in 2,000 in the population.

Estimated Current Demand or Size of Group: 8% of the whole population, or approximately 350,000 New Zealanders.

Demography: As defined above, 8% of the whole population, or approx 350,000 New Zealanders. In context for New Zealand, these figures are about the equivalent size of the Pacific population of New Zealand, or the equivalent of 50% of the entire Maori population of New Zealand.

Future Demand: The proportion of the population is expected to remain at 8% over time, but of course absolute numbers will grow in direct proportion to population size.

Current Availability

Diagnosis, information, clinical services and social supports for rare diseases currently range from extremes like:

- conditions such as PKU, Congenital Hypothyroidism and Cystic Fibrosis with a high level of early detection through newborn metabolic screening, with well established and effective treatment from experienced clinical teams, leading to very significant improvements to health and quality of life
- the vast majority of rare conditions where there is very mixed success with diagnosis, limited expertise to treat, frequent structural problems with access across DHB boundaries or getting adequate priority within a DHB, considerable difficulty getting access to specialist care, let alone access to certain innovative therapies, and overall poorer health and quality of life outcomes for the patient and their family.

Historically there had been little that could be done for many rare diseases. The technical ability to even diagnose, let alone to intervene, was very limited. Therefore the condition was often something that just had to be borne by the patient and their family, with little more to be done by our health system other than provide symptomatic management, disability support and palliative care.

Things are changing and are continuing to change rapidly. The ability to accurately diagnose has improved significantly. Medical interventions to ameliorate or treat rare conditions are rapidly evolving. Knowledge sharing is empowering both patient and clinician. Clinical care is being rapidly informed by new genetic knowledge and by the shared efforts of patients, support groups, clinicians and research groups to gather natural history studies and develop registries for the benefit of clinical care and research.

The particular dilemma faced by rare disease patients in New Zealand is that our health system has not yet taken steps to adapt to the new paradigm. Even in the first set of examples for PKU, CH and CF, the early detections and interventions may be partly frustrated by regional or structural impediments to quality care.

We think this work needs to start now, and it needs to come at a high level such as the National Health Committee. All other initiatives that are relevant to the global needs of patients with rare

disorders tend to be taken in isolation from a high-level perspective, resulting in many piece-meal changes that rarely bring a cohesive and positively joined-up impact.

Proposed Service/Patient/Programme Pathway

In keeping with models of rare disease action plans established elsewhere, but especially in Europe, there would be comprehensive systems put in place to ensure diagnosis and access to the most appropriate clinical care for all rare disease patients, with appropriate multi-disciplinary input when needed, plus provision of information, along with the development of registries to assist with knowledge gain about best clinical care and understanding of the disease.

The services and patient pathways could be modelled on those presently in place nationally for Paediatric oncology, or regionally for Paediatric neuromuscular conditions in the Auckland region, as examples, but with adaptation of those models to ensure best fit for national delivery across all ages and with specific attention to the barriers that those models, however advanced, still face.

The model and pathway cannot realistically be described in greater detail at this stage as work would be required to identify all its component parts and best design. However the intention is to provide seamless care that avoids the many access problems and blockages that occur now.

Current or Alternative Intervention

Currently there are a number of discrete services or programmes that are relevant for most rare diseases. These range from maternity care, ante-natal and newborn screening, genetic services, metabolic, endocrinology, oncology, neurology and other specialty services, specialised medicines, equipment services, disability support services, community support groups, palliative care, and more. Many of these services act in isolation from each other most of the time. Generating suitable multi-disciplinary team approaches is successful for some conditions but a major challenge for most.

Currently work is occurring in the development of clinical networks and national services that are relevant to some rare diseases. However these positive initiatives of the National Health Board to develop clinical networks with the Paediatric Society are restricted in their scope primarily to Paediatric services, seem hampered by their inability to operate at a higher level than the current DHB silos and power blocs, and by dealing with such discrete service initiatives are unlikely to generate widespread change to service delivery problems for the bulk of patients affected by rare conditions, in the foreseeable future.

A higher level plan of action is required, in much the same way as the unmet health needs of Maori and Pacific populations, and the significant relative disadvantage they experience, could begin to be addressed effectively only when high level strategies and action plans were put in place by government and Ministry initiatives to deal more effectively with them.

Safety Information

A rare diseases action plan would be directed to improvement of outcomes and filling gaps in an otherwise frequently unsafe system for many people with rare disorders.

Estimated Benefits

The most significant benefit of a rare disease action plan would be the delivery of equitable access to health care and improvements to health and quality of life, for a significant but frequently neglected portion of our population.

These benefits of a well-designed action plan could result in measurable benefits such as:

- Faster and more accurate diagnosis of rare disorders – less failure to diagnose, fewer misdiagnoses, reduction in the stresses of diagnostic odysseys for families
- More timely intervention with appropriate clinical care and social supports – fewer inappropriate treatment or missed opportunities, reduced mortality and morbidity
- Improved access to specialist services – avoiding DHB silo barriers which can be a significant impediment to best care
- Better knowledge generation through development of disease registries and natural history studies – avoiding the lost opportunities to gather this information and act upon it
- Improved knowledge for individual patients and support groups to assist the development of expert patients as partners with clinical advisors in the management of their disease – less isolation and ignorance means reduced risk and improved self-management
- Improved partnerships between patient group, clinicians and officials in the design and delivery of rare disease information, clinical care, research and social supports – less tension and contention about service aims, objectives and capacity

Estimated Costs

Unknown at this stage.

Cost Effectiveness

Unknown at this stage. However an important consideration in the investigation of this topic will be the extent to which the ethical considerations inherent in this proposal may challenge standard approaches to cost effectiveness.

For example, the arguments in favour of proceeding with this proposal will include emphasis on equity and social dimensions of Health Technology Assessment or any other tool used to evaluate the proposal.

Service Configuration and Implementation Issues

The development of this proposal would require significant commitment from officials, clinicians and patient groups.

Ethical, Equity and Acceptability Issues

The rationale for this proposal is essentially one of equity in access to health care and health outcomes. It also involves a human rights dimension. 8% of the population is a sizeable minority

population. Leaving their health needs in a state of relative neglect, especially as we see increased capacity to understand and intervene, is not an ethically justifiable situation.

Just as our society now realises that the relatively poorer health status of Maori, Pacific and other groups is not acceptable and must be addressed, our capacity to do better puts a duty on us to do so for other significant identifiable populations.

Relevant Stakeholders

Patients, support groups, health professionals, DHBs, Ministry of Health, professional bodies, government.

Is there any other relevant information?

Note that support from the LAM Trust is presented as indicative of wider support within the rare disease community. We have a network of 150 rare disease support groups and have conducted a number of conferences and seminars on these topics since we formed in 2000. There has been substantial support for the development of a rare disease strategy/policy/action plan in New Zealand for many years.

NZORD has represented its wider network in taking this issue up with Ministry officials and with Ministers in the past. In 2008 we received support from then Health Minister David Cunliffe for improvements to Genetic Services as the first step towards the improvement of services for those with rare disorders, and the National Health Board has made some progress on this by designating Genetic Services as a national service. However the Minister's support for a group or section within the Ministry to have specific responsibility for policy and services for rare disorders has become lost in the changing bureaucracy and changing health priorities.

In addition to the well developed rare disease action plans in many European countries, there are efforts under way in Australia, Canada, Latin America, and various other countries to make progress on rare diseases as a significant public health problem. New Zealand is at risk of being significantly behind in the development of plans which would seem an essential part of catching the wave of innovative approaches to medicine such as applied genome knowledge in personalised medicine.

Additional material supplied by email re Lysosomal Diseases Group and HD Commissioner; and ethics issues

Lysosomal Diseases

Early in 2007 Lysosomal Diseases New Zealand (which I chair) made a complaint to the H&D Commissioner regarding health care and disability support problems for 14 of the 64 families in the LDNZ network at that time. The problems related to difficulties getting appropriate transition from Paediatric services to Adult services, failure of referral systems across DHB silos, inadequate needs assessment and disability support provision, and failure to accept the knowledge and expertise of families in managing these complex rare conditions.

One of these cases was selected for urgent attention which resulted in action to address the problem and the publication of a patient safety statement by the H&D Commissioner. The rest were referred to the Director General via Ron Paterson's letter with the expectation that a systemic solution would be found. Four years later we have reached the point where the first tentative steps have been taken towards a solution with the decisions to implement national services for genetics, metabolics and some other services.

These national services and other clinical networks being set up are positive steps but at the current rate of progress, issues about appropriate care for rare conditions that have been identified for many years, and increasingly since NZORD was established in 2000, are unlikely to be resolved in any systematic way before 2020.

Patients with rare conditions like Rett syndrome, Fragile X syndrome and Dystrophic Epidermolysis Bullosa could benefit enormously from improved coordination of services across the country and an organised multi-disciplinary approach in appropriate cases. The practical

solutions that are well understood and supported by patients and families and many of the clinicians dealing with these conditions, do not receive the attention of planners and funders at most levels in our health system. Even in the case of EB, where there was a very welcome interim solution from the National Health Board to fund specialist EB nurses salaries, problems with DHBs have meant the contract for these nurses salaries had to be given to the small patient support group, and the opportunity to exploit expected saving from central purchasing were lost. Having the contract managed by the patient group, in my view, is unlikely to provide a sustainable long-term solution and may carry some service risks with it.

I support the work being done by the Paediatric Society with the support of the National Health Board to develop clinical networks and service improvement programmes. I am a member of the advisory group for that exercise. But NZORD remains very concerned that the approach is incremental. Limited to Paediatrics, and unlikely to generate major improvements across all rare conditions in a hurry. That is why we have submitted the referral to the National Health Committee seeking a high level endorsement of a systematic approach to the health and disability needs, and clinical research, for rare disorders.

Additional comments on the issue of ethics, rights and duties.

NZORD believes that the issues identified in the complaints to the H&D Commissioner, and the general arguments set out in this referral, touch on important issues relating to how such matters are addressed by our health system. We appreciate that economic restraints and the need to prioritise services do actually mean a significant restraint on our “rights” to the highest attainable standard of care, and to services of an appropriate standard, as set out in a variety of legislation, international instruments, national codes and common law.

However we also believe that it is inappropriate and unacceptable for the needs of rare disease patients and their families to suffer such obvious ongoing systemic neglect. If this applied to other segments of our population it would be easily identified as unacceptable discrimination. We ask the National Health Committee to carefully evaluate the moral dimension of this case, as well as the concepts of rights and duties (however strong or weak they might be) alongside the need for consideration of cost effective interventions in health.

Ranking (if submitting multiple referrals)

N/A

Referrer Contact Details

Title	Mr
First Name	John
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Position	Executive Director
Organisation	New Zealand Organisation for Rare Disorders

Endorsing/Sponsoring Organisation Contact Details

Title	Ms
First Name	Bronwyn
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Position	Director
Organisation	NZ LAM Trust

Available Evidence and Assessment of Quality (including references to information stated above)

References and notes

[Position statement - on transition from paediatric to adult health services, for those with rare and complex disorders](#) Nov 2002 - NZORD worked with the Ministry of Health and the Paediatric Society to develop this statement calling for better care management of transition to adult services. Some modest gains have been made, especially with service specifications for the new national congenital cardiology and metabolic services dealing effectively with this problem by providing continuity of care regardless of age, but this positive step for these few services will not reach quickly across many services which need similar specification changes to ensure equitable access.

[Minister helps to rescue genetic services report](#) This link summarises the briefing given to Health Minister David Cunliffe in March 2008 when he agreed to support a central point of responsibility in the Ministry for rare disorders and to support implementation of the Genetic Services Report. There has been action on Genetic Services from the National Health Board but the point of responsibility in the Ministry has drifted into a “place to have our questions answered”, rather than a place where responsibility for policy initiatives is housed.

Forman, John. Health inequalities and need associated with rare diseases. Book chapter in Understanding Health Inequalities and Need in Aotearoa New Zealand. 2008. Editors K Dew and A Matheson. Otago University Press.

European Commission policy on rare diseases. Various papers available from this link http://ec.europa.eu/health/rare_diseases/policy/index_en.htm

The need for worldwide policy and action plans for rare diseases - Position Statement from the International Conference of Rare Diseases and Orphan Drugs (ICORD) Unpublished paper (submitted for publication) copy attached. This paper includes many relevant references also.

ATTACHMENTS

ICORD Position Statement
Letter from H&D Commissioner