BRIEFING TO MINISTER OF HEALTH

NEW ZEALAND ORGANISATION FOR RARE DISORDERS - *To support, assist and promote better health care and well-being for patients and their families/whānau living with a rare disorder in New Zealand.*

Rare Diseases:

There are over 7,000 rare or orphan disorders that collectively affect around 8% of the New Zealand population. Approximately 50% of rare disease patients are children, 30% of whom will not live to celebrate their 5th birthday [1].

While the individual diseases are rare, the total number of people who have a rare disease is high. In New Zealand rare disease patients exceed the total number of people who are affected by diabetes.

Unlike the US or EU, New Zealand has no official definition of what constitutes a rare disease, however they are considered to be life-threatening or chronically debilitating diseases which are statistically rare – and have an estimated prevalence of less than 5 in 10,000, and have a high level of complexity.

Examples of rare diseases include rare cancers such as childhood cancers and some other well-known conditions such as cystic fibrosis, Batten's disease, Ehlers-Danlos syndrome, muscular dystrophy, Epidermolysis Bullosa - and other conditions that haven't even been named yet.

While research in the field of rare diseases and the technological advancements accompanying this are constantly evolving, rare disease patients and health systems globally face common challenges, including:

- Difficulty in obtaining accurate diagnosis
- Lack of experience amongst health professionals to treat
- Good information is harder to find, for professionals and families
- Research is less likely to occur than for common diseases
- Developing medicines may not be economic for the pharmaceutical industry
- Treatments are often very expensive

NZ Organisation for Rare Disorders (NZORD):

NZORD is the umbrella group that provides patients and their families with information, support and advocacy to achieve the best possible health outcomes for the patient.

NZORD's role also involves working with health care professionals, researchers and health planners to improve knowledge about rare diseases, improve clinical care and assist in accessing treatments for rare diseases.

Our network includes more than 100 rare disease support groups and partners, and we work closely with these groups in a range of health and disability sector improvement initiatives.

[1] https://globalgenes.org/rare-diseases-facts-statistics/

Opportunities for Rare Disease Patients in New Zealand

Patient Registry:

A critical issue for New Zealand patients is the lack of information we have about those living with a rare disorder. Understanding who the patients are, where they live and what type of rare disease they have are essential elements in ensuring treatment and care are allocated appropriately and in the most cost-effective manner.

Properly designed and executed, patient registries can provide a real-world view of the rare disease community in New Zealand.

NZORD's goal is to work with the Ministry of Health to explore ways in which we can establish a national database that drives improvements in the treatment and care of our patients and ensures resources are allocated to where they are needed. The database would capture and hold information on diagnosis, disease type, treatment and health outcomes for patients throughout New Zealand and would provide data that assists health care providers and planners with their decision making.

Orphan Drugs Fund:

The issue of providing access for suitable treatments to patients living with a rare disorder is a challenge for many health systems around the world. Rare diseases, by their nature, do not naturally fit within public health benefit equations because patient numbers are low and typically, treatment costs per patient are high.

NZORD wants to ensure that the structures put in place by the Government get the best outcomes for patients with rare disorders and we believe decisions relating to treatments for these diseases need to be considered differently.

PHARMAC have recently undertaken a pilot scheme that enabled them to look at new funding solutions for rare disorder medicines and NZORD has been advised that as a result of this pilot, 10 medicines were funded and PHARMAC are introducing a set of dedicated features for considering rare disease medicines. These include:

- Establishing a standing Pharmacology and Therapeutics Advisory Committee (PTAC) expert subcommittee for rare disorders which will provide objective clinical advice on rare disorder medicines.
- Regularly calling for rare disorder funding applications.
- Undertaking dedicated pre-engagement with new, as well as existing, suppliers prior to each call for funding applications.
- Formally adopting adjusted policy settings for rare disorders treatments; notably the set of prerequisites that define 'treatments for rare disorders', and that Medsafe approval would no longer be required for rare disorder treatments to participate in the funding application process (but would be required prior to listing).

While we recognise the efforts being made to ensure rare disease patients receive timely access to treatments, there are many challenges for the New Zealand health system that remain with this approach, including:

1. The lack of a timetable for PTAC;
2. The current backlog of 20 funding applications that are still to be considered;

3. Two out of the ten medicines funded by PHARMAC have no patients currently in New Zealand which will mean that patients with rare diseases will continue to miss out on potentially lifesaving or life extending medicines that are freely available to patients in other countries;
4. One of the ten approved medicines is used to treat tuberculosis, which may occur rarely in New Zealand but is not a rare disease.

We recommend the Government establish an Orphan Drugs Fund which sits inside the Ministry of Health but outside of PHARMAC's current legislative requirements.

National plans for rare diseases are evolving in countries around the world, with several countries implementing policies to ensure timely patient access to treatments following regulatory approval. Scotland, a country with a comparable population of 5.4 million people, established an Orphan Drugs Fund in 2013 and have since dedicated £140 million to funding medicines for people with rare disorders.

NZORD would like to work with the Minister of Health to explore ways to establish an Orphan Drugs Fund which would allow for drug provision for rare disorders that can be treated in a way that benefits everyone.

The Scottish Medicine Consortium has developed a new approach for reviewing rare disease medicines designed to increase the influence of patients and clinicians in the appraisal process. This "Patient and Clinician Engagement group" can address additional benefits that may not be represented in the conventional clinical and economic assessment process. Scotland also has a New Medicine Fund that was recently expanded to £80 million year allocated to orphan drugs to ensure patient access to the most advanced therapies for diseases with unmet needs.[2]

NZORD recommends establishing an Orphan Drugs Fund that would take notice of factors other than cost benefit. This would need a substantial budget that recognises the cost of orphan drugs, and is entirely separate from other medicines budget.

Neural Tube Defects:

Neural tube defects, including spina bifida, are rare disorders, and fall under NZORD's umbrella. Unlike other conditions that we cover, about two thirds of them are preventable, and yet successive governments have not taken the necessary steps to address this significant public health issue. We estimate the incidence of preventable neural tube defects in live births to be about 24 cases per year, half of which are associated with a diagnosis of spina bifida, and half with anencephaly (which becomes fatal soon after birth). This means that there are about twelve New Zealanders per year who are unnecessarily destined for a lifetime of disability (with annual childhood rehabilitation costs of about $400,000 per year, and a significant burden of care which is borne by their families), and a further 12 who are denied the right to life altogether.

NZORD's recommends the Government adopt a regime of mandatory fortification of flour with folic acid. The scientific and medical literature supports this approach and many other countries have already done this.

NZORD's recommendation is to implement an effective fortification regime, with likely little or no opposition from the Bakers' Assocation, by fortifying flour at the mill rather than bread. This is the regime followed by Australia and many other countries. Fortification of flour at the mill would be the easiest implementation method as there are only four or five millers to work with, rather than 2,000

[2] Fund for new medicines doubles, 2015; https://news.gov.scot/news/fund-for-new-medicines-doubles

separate bakers. Note that plain flour for organic or other specialty foods could still be sourced separately, like in Australia, so niche brands would not be impacted by this change.

Dr Collette Bromhead
CEO, NZ Organisation for Rare Disorders