24 February 2015

NZORD - the New Zealand Organisation for Rare Disorders
Press release: Rare Disease Day highlights the orphans in our health system

February 28th is Rare Disease Day and an opportunity for those who often slip below the radar in our health system, to have some focus on their needs and interests. Getting a timely and accurate diagnosis can be one of the major hurdles they face, and is often a significant frustration.

That's not to say it is all bad for those with rare diseases. There is a small army of dedicated health professionals out there working on a daily basis to improve the health and quality of life of rare disease patients, as for all health patients. Their commitment makes a very significant difference for many of us.

But to achieve good outcomes, there first needs to be an accurate diagnosis to act upon. And there's the problem. Getting a diagnosis is a major challenge for many of the 7000 or more rare diseases and a cause of significant problems for many rare disease patients. Surveys in the UK and parts of Europe suggest 6 or 7 years is the likely time delay in diagnosis for many rare diseases, and anecdotal evidence in New Zealand suggests similar problems, especially for those whose problems emerge in their teens or adult years.

Diagnosis time for children with rare diseases tends to be faster than for adult rare disease patients, mainly because many rare diseases that present in childhood, do so with significant or even dramatic symptoms. This improves the chances of the child being seen early by the appropriate specialists, including the genetic health service, and the right treatment or management plan being put in place. The primary role of genetic health services is to accurately identify the disease and provide guidance on the appropriate course of action, and it is a service highly valued by families and other health professionals too.

Though rare disease in children take an awful toll, with many of the diseases having no effective treatment, and many of the children not living past age 5, the knowledge of what the condition is can be such an important thing for families. Knowledge can mean better management of symptoms, less anxiety or guilt for parents, improved chances of ensuring the condition is not repeated in another pregnancy if it is an inherited condition, and opportunities to connect with other families with the same condition to share experiences and practical management strategies.

New diagnostic tools make the process of diagnosis faster and more reliable now. Databases with clever algorithms, faster and cheaper sequencing technology, and new insights into interpreting DNA sequences, all dramatically improve the likelihood of a more timely and accurate diagnosis, provided the right specialist is seen and the test is funded.

This Rare Disease Day it is worth reflecting on the challenge there has been to get good quality genetic services set up in New Zealand. It has taken literally decades for New Zealand to get near the standard of services provided in most comparable countries, and we owe a great debt to the

dedicated few who have provided and promoted this largely neglected and under-resourced, but vital service. Bringing these services under one national service five years ago, certainly helped improve access and coordination.

In 2014 there was another big step forward when District Health Boards provided funding for genetic health services to clear the most serious backlogs in the waiting lists for genetic assessments. Now we need this improvement to be sustained so that those with complex and obscure conditions always get to see the genetic specialists within the six month timeframe which is the standard for all specialist assessments in New Zealand, as well as ensuring those with urgent referral needs get those attended to quickly too.

This will require the employment of additional genetic consultants and associated health professionals and of course that takes money. But for rare disease patients that will mean getting a fairer share of the health budget, less time on waiting lists, and less time untreated. This would be a big step forward in gaining equitable access to healthcare, just as those needing cancer treatment and hip and knee surgery, rightly expect from our health system.

So while many support groups are running awareness and fundraising events in celebration of Rare Disease Day, Saturday 28 February, behind the bit of fundraising fun is a serious message to our health system; that it is time for the relative neglect of rare diseases to stop, and for some serious work to ensure equity in healthcare provision for these orphans of the health system. Last year most political parties agreed with our position that New Zealand needs a rare diseases policy and action plan, and we sense this is now becoming mainstream thinking among politicians as well as among many health officials and health professionals. Now we need the political and health sector leadership to make this happen.

More information about Rare Disease Day can be found on the website www.rarediseaseday.org.nz