Summary of the Rare Disorders NZ: Implementation of the Rare Disorders Strategy Round Table 14 May 2025

A focus on rare disorder diagnosis and medicine access.

A collection of officials from the Ministry of Health, Health New Zealand and Pharmac joined Rare Disorders NZ, Consumer Representatives, Clinicians, Pharma and industry representatives to discuss where New Zealand wants to be with rare disorder diagnosis and medicine access in ten years' time and stepping stones to getting there. The event ended with presentations to the Minister of Health and Associate Minister of Health (Pharmac) and their responses.

Presentation on the draft Health New Zealand | Te Whatu Ora Genomics Operational Strategy

The Ministry of Health and Health New Zealand, on the back of the work undertaken in Precision Health identified that genomics is a key piece of any implementation. They have undertaken work and developed a draft operational strategy document.

The vision is that by 2030, Health New Zealand | Te Whatu Ora has the infrastructure, mechanisms and workforce required to capitalise on advances in genomic medicine to improve the lives of all New Zealanders.

In terms of genomics, NZ is currently 10 years behind other OECD countries.

The Operational Strategy is made up of key building blocks:

- Lab
- Data- that is available and useful
- Workforce

Without these building blocks, diagnosis for rare disorders cannot be implemented.

A governance structure is also needed, and it is important there is a focus on addressing inequity both in terms of private vs public options and geographical availability.

The overarching aim is to have genomic services available to patients and informed clinicians and workforce who know how use genomic services appropriately/ a pathway that is appropriately accessed.

NZ needs a working national clinical genetic service and a database of NZ patients. The exome analysis being undertaken in Christchurch is a step to bringing testing back to NZ- work that is otherwise being undertaken in Finland.

There is a want to see health dollars invested in NZ. The Christchurch pilot has demonstrated that we can do it in NZ – though can't do sequencing as we don't have a sequencer the size needed- but otherwise can do it in a reasonable timeframe and at a reasonable cost.

Discussion on diagnosis and where would we like to be in ten years

A clinician shared that there has been movement made in the past 5 years that is enormous, and it is having an huge impact on patients- patients who now aren't dying without a diagnosis. They also noted that we know it is cheaper to keep testing in NZ and that it is vital to keep data in NZ to understand our information. They emphasised that we must move forward with this. They identified a ten-year goal of rapid genomics at the bedside that is properly funded, enabling following evidence-based medicine with diagnosis achievable in 1 to 2 weeks.

A Pharma representative noted that the potential of whole genome sequencing as part of newborn screening has extraordinary potential, but to achieve this the community needs to be taken on the journey. Then asked if whole genome sequencing as part of newborn screening is on the ten-year horizon.

A clinician responded that internationally this is being investigated and that a genomic approach may achieve diagnosis of treatable conditions, but that NZ needs to walk before we run. They also emphasised that there are legal and ethical considerations as well as that NZ does not have the infrastructure for that scale at present. The clinician shared they see the next step as introducing whole genome sequencing in a pilot fashion in NZ (Christchurch) for a period to assess feasibility, costs, and benefits. They noted there are some isolated disorders where we should be thinking about whole genome sequencing.

Another clinician shared the importance of there being pilot programs and research alongside an established framework when it is set up. They also noted the need for looking at the indigenous voice and highlighted that consumers are concerned about genetic testing. They emphasised that we must be mindful that the success of newborn screening (and its excellent uptake rates) could be affected by genetic screening being introduced).

A Health NZ official shared agreement with the prior points that introduction of genomics needs to be taken slowly and with discussion with the general public. They raised the question of how we go from a metabolic program for newborn screening to the next stratosphere. They noted that the genomics operational strategy is a good start.

A clinician raised that preconception screening is a conversation that needs to be had as well.

An industry representative suggested that NZ takes the opportunity to learn from Australia including their mistakes. They noted that NZ has the opportunity to have a nationalised approach. They mentioned a preconception carrier screening program. They cautioned not to do what Australia did and encouraged the room to not just catch up but to be leaders.

A consumer shared that their children are carriers and when starting their own families their partners had the opportunity to undergo carrier screening. They noted they see this as a must.

A clinician pointed out that carrier screening is ultimately cost saving and while we need to be cautious not to belittle the lives of people with rare disorders, often there is no funded treatment available.

Another clinician noted that what was being discussed is vital but requires an investment in genetic health services and reproductive services, as with existing services, there is already limited capacity. They highlighted that there are massive implications for the health workforce.

RDNZ's CE noted that there are significant health and societal costs of delayed diagnosis also.

The clinician agreed that these costs of delayed diagnosis go much wider than health services.

A consumer asked what framework the pre-implementation screening process is currently under and if it is inequitable.

A clinician responded that it is functional and funded for limited patients and limited cycles. They reflected that it is working but that there are waitlists. They noted the need will increases as we progress down the path and as has been seen in Mackenzie's Mission [$20 million study of reproductive genetic carrier screening, called Mackenzie's Mission, undertaken in Australia][i]. They raised the question: are we resourced?

A Pharmac representative asked about lessons learnt following SMA being added to the newborn screening program and the gap between funded medicine access and newborn screening. A clinician responded that it was two separate but parallel processes that could have been better aligned but in the scheme of things the timeframe between the two was short.

An industry representative noted that there is a need for a national strategy and that infrastructure is needed which industry is actively involved in conversations about. They highlighted concerns with the current position of not screening for conditions in newborns where there is no budget for treatment. They noted that diagnosis allows decisions and that parents want to know what is wrong with their child, regardless of access to treatment.

A Pharmac representative signalled that Pharmac wants to know more and understand the challenges- both ethical and in terms of social license.

A MoH representative shared that equitable access resonates with them. They acknowledged that the Ministry has been slow in taking a leadership role but that it was good to see the progress that is happening. They noted the societal, financial and ethical considerations associated with increased access to genomic testing.

Discussion on rare disorder medicine access and where would we like to be in ten years

A consumer representative acknowledged that Pharmac has opened the door for discussion between Pharmac and Consumers. They cautioned not to discount the voice of the consumer, and that without the voice of the people, discussion can't move forward.

A clinician shared that internationally NZ is falling behind and that this is increasingly obvious to clinicians. A consumer seconded this view and noted it is obvious to patients too.

A consumer noted there have been years of assessment conversations and recommendations from, for instance, the Valuing Life Summit and Consumer Engagement Workshops. They noted that the conversations have been had, and reports written. They asked: What are we going to do? And who will lead this work.

A consumer raised the key issue of equity noting that rare disorders will never stack up in Pharmac's processes. They compared a child with cancer with access to medicines under rule 8..b vs a child with a rare disorder who has no way to access their medicine. They emphasised where we want to be is not here and asked how we are going to move forward collaboratively.

A Clinician shared they returned to NZ from London, and they just want access to drugs they were prescribing to patients there back in 2008.

A Clinician shared that through working closely with Pharmac they have made huge progress, but these are cost neutral medicines that increase quality of life and processes that decrease paperwork. They shared that they are just absolutely stuck when it comes to the high-cost therapies. They emphasised that we must look at it differently. They agreed with the consumer that there have been so many reports and conversations, and we need to get this across the line quicker as patients are dying.

A Pharma company representative shared that 5 to 10 years ago their company would automatically register products in Australia and then NZ. Now the discussion is which wave would NZ fall into for just registration, let alone Pharmac submission. They shared they see the change happening at Medsafe but that this is not where is needs to happen. They noted their company has products on waitlists that are lifesaving with a low recommendation and not moving. They said that NZ is not in the position of other OECD countries.

A RDNZ representative shared that they are aware many companies are frustrated with the Pharmac processes.

A consumer representative noted that there is a medicine that is funded for infants but not for adults, but they can't get a diagnosis as infants as it isn't screened for.

A consumer noted that clinical trials are being pulled away due to there being no forward pathway for the medicines being trailed and that as a result the patient suffers. They shared a hope that NZ will be a better place for clinical trials.

A Pharma representative agreed noting that they must think about what happens when the trial stops, and that they don't start as they worry about the end.

A Medicines NZ representative pointed out that clinical trials are about finding answers, not a substitute for a proper funded pathway. They are there to give a chance when there is no registered treatment, and for clinicians to become familiar with new treatments. They endorsed earlier comments about good recommendations in the Valuing Life Summit White Paper and noted that the same wheels are being turned but we know where we need to go. They emphasised that not funding treatment is not an excuse to not screen for disorders.

A MoH representative noted that to a degree prioritization will always be needed. They asked what unique element can NZ offer to Pharma? How can we construct our environment in a unique way that sets us apart from other countries with higher GDPs to ensure people here have sustainable access.

A Pharma representative responded that one option is second order benefits through Pharmac, and that NZ could be a world leader in incorporating these. They noted that the structure of NZ's health budget puts less proportionately towards medications than for instance Australia. They noted that unless Pharmac has more money it will always be a struggle. They queried rare disorders specific funding mechanisms and quarantined funding. They asked about savings to hospitals if people are treated upfront.

A MoH representative shared that they had had discussion with their Pharmac counterpart in the past in relation to the optimisation of medicine use and how savings in the health system are transferred to Pharmac. They recognised a need to work as a whole system, not a bunch of silos.

A Pharma representative noted the OECD average of people living with rare disorders in the workforce is 80% compared to only 50% in NZ.

A clinician shared that from 2012 conversations have changed significantly with Pharmac and their thinking about rare disorders has changed. They noted they have never seen rare disorders in the budget. They highlighted the need to think about genomics and Pharmac. They said that cancer drugs got their time and now it's time for rare disorders. They asked whether people living with rare disorders should be seen as equivalent to people with a common disease, noting they didn't ask for this disorder.

An industry representative noted that industry have tech that can start personalised medicine and this is proven overseas, especially around adverse effects to, for example, oncology medications. They asked how we can collaborate better.

A Pharmac rep responded that industry could reach out and talk to them, that they are open to discussion and recommended reaching out to acting CE Brendan Boyle. They shared that Brendan has been meeting with Pharma companies and wants more engagement.

There was discussion about how companies can collectively talk to Pharmac and Health NZ and other parties, and it was noted that the Rare Disorders NZ Round Table of Companies had previously met as a group with Paula Bennett (Pharmac Chair).

A consumer shared that Trikafta has been a great case study and shows the bigger picture needs to be considered and that Pharmac shouldn't be siloed. They shared data that two years on from the funding of Trikafta hospitalisations are down 50% for the patient population and primary lung transplants are nearly non-existent. They noted these savings aren't going back to Pharmac, that there is no linkage for these savings or reinvestment to get more medicines funded. They emphasised that Trikafta shows what can be done when we work together and asked how we do more of this.

A Pharmac representative shared that they have been meeting with the Social Investment Agency about similar points.

A Pharma representative highlighted that NZ should learn from other countries and consider how to harness information, including planning around a registry or centralised data collection to see the benefits of medicines and care. They also noted that it is important to make sure those that don't have the voice of a patient association also have a voice in the system.

Session with Ministers Brown and Seymour

The main corporate sponsor Alexion shared two messages.

1. A rare disorders strategy has internationally been a foundational step in improving access to healthcare and medicines for those living with rare disorders. However, they cautioned that it does require government commitment and leadership from the top. Supported with funded resource and an implementation plan and framework.
2. New Zealanders are struggling to access new innovative medicines. The Pharmac Review identified a high and urgent unmet need, and medicines NZ has identified that approximately 11% of rare disorder medicines available overseas in OECD countries have made it to NZ. They noted that they welcomed the recent additional investment in Pharmac but that it hasn't moved the dial for rare disorders.

They closed by encouraging and supporting a continued pursuit of increased funding and consideration of a ring-fenced rare disorders fund.

Consumer presentations

A consumer shared their journey to diagnosis and the vast difference obtaining a diagnosis made for them and their family. They emphasised that an earlier diagnosis would have been central to many of their life's big decisions - family planning, financial decisions, house purchasing. They noted an earlier diagnosis would have helped them seek ways to slow the deterioration of their condition, potentially keeping them out of a wheelchair. They shared their eventual diagnosis has enabled access to treatment, stating "If I hadn't fought the health system and been a strong advocate for myself, I

would have died long ago, or at the very least, been stuck in a long-term care facility on a tracheostomy". They noted that earlier diagnosis and access to treatment may have

Another consumer shared that it's all been said before. There are clear sets of recommendation and responsibilities set out for instance in the valuing life white paper and a year in it is gathering dust. The consumer highlighted that we are ready, willing, able and smart. That we understand and want to help. They emphasised the need for a clear plan, actions and accountability. They highlighted the need for equity and compared children with cancer accessing medicines through rule 8.1.b vs children with rare disorders with no access. They noted that it has been proven with Trikafta that medicines can decrease the pressure and burden on the health system. They said they want to see this reinvested into medicines and finished with the statement we just need action.

Clinician presentation

A Clinician presented on behalf of the group of clinicians present on why understanding, diagnosing and caring for rare disorders so important. They highlighted that early diagnosis and treatment of rare disorders can reduce morbidity, mortality, and recurrence.

They outlined the current state:

- 3 Genetics laboratories working independently Labplus in Auckland, Wellington Regional Genetics Laboratory (WRGL), Canterbury Health Laboratories (CHL).
- Variation in access to testing and in test performed (gene content) by geographical location
- >4000 gene panel and exome tests being sent overseas at a cost of $3.9M p.a. and rising
- Commercial laboratories control and retain the data
- Re-analysis incurs additional charges
- Rapid genomic testing (Liggins Institute Auckland only) with Oxford Nanopore

They outlined the future state:

- Establishment of pathways operating to agreed national standards
- Coordinated and comprehensive genomic testing for inherited and acquired disease performed in NZ
- Genetic laboratories work as a network
- Education and training for clinical and laboratory workforce
- Strong links to research and industry, including cross-Tasman partnerships

They identified next steps:

- 2 year Pilot (based at CHL with support from Illumina)

i. Whole genome sequencing (WGS)

ii. Cancer comprehensive genetic profile (CGP) – aligns with GPS Health Target - faster cancer treatment

- Real world experience
- Build capability (and capacity)
- Model for retention of send-away testing
- Benefits of WGS workflow and analysis capability with potential for additional diagnoses
- Aligns with current international practices

They identified what they believe NZ needs:

- Nationalised and equitable genomic care
- Nationalised and equitable access to specialist services
- Improved education of all medical staff on rare disorders and diagnostics
- Change of PHARMAC model for high-cost therapies for rare disorders
    - An alternative that works within the economic system that is transparent
- Orphan drugs will always have high cost, low evidence and low numbers of patients – but they are no less in need than others, and they did not cause their condition

Ministers' responses

David Seymour

Minister Seymour identified that within his influence is how rapidly NZ absorbs medical technologies into our health system through Pharmac and Medsafe. He noted that the Government is doing what they can to make the barrier as permeable as possible. He noted that Pharmac is being more proactive in listening to patients. He shared a hope for more personalised care, support and dignity for people with rare disorders.

Simeon Brown

Minister Brown said that in relation to the Rare Disorders Strategy he said "I think it has a huge amount of opportunity and it is now about focusing on delivery. I will be making that clear to Health NZ".

He noted that how we focus our resources in our health system is important. He observed as a fraction of GDP per head we spend the same as comparable countries on health, but that proportionately we spend far less on medicines and a lot more on hospitals.

He outlined the need to focus on medicines, diagnosis, treatment and keeping people well in communities. He noted that we need to look over time as we consider how we

keep people well at home and well in communities. He shared he sees the answer as better diagnosis, medication, technology and AI. He identified a key challenge is the need to ensure that Health New Zealand is focused on delivery. He shared that patients should be at the centre of decisions and that understanding the patient journey is key to this.

He highlighted the importance of nationwide consistency and mentioned the Clinical Senate established by Health New Zealand. This is a clinically led group responsible for decisions around clinical pathways and national consistency. The aim is to make the patient journey more simple.

He closed noting "I am ready to work with you and committed to investing more in health".

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