

Briefing

Rare disorders strategy: update on development and emerging themes

Date due to MO:	N/A	Action required by:	N/A
Security level:	IN CONFIDENCE	Health Report number:	H2023023661
To:	Hon Dr Ayesha Verrall, Minister of Health		
Consulted:	Health New Zealand: <input type="checkbox"/> Māori Health Authority: <input type="checkbox"/>		

Contact for telephone discussion

Name	Position	Telephone
Maree Roberts	Deputy Director-General Strategy Policy and Legislation	s 9(2)(a)
Allison Bennett	Acting Group Manager Health System Settings	s 9(2)(a)

Minister's office to complete:

- | | | |
|---|------------------------------------|--|
| <input type="checkbox"/> Approved | <input type="checkbox"/> Decline | <input type="checkbox"/> Noted |
| <input type="checkbox"/> Needs change | <input type="checkbox"/> Seen | <input type="checkbox"/> Overtaken by events |
| <input type="checkbox"/> See Minister's Notes | <input type="checkbox"/> Withdrawn | |

Comment:

Rare disorders strategy: update on development and emerging themes

Security level: IN CONFIDENCE **Date:** 09 May 2023

To: Hon Dr Ayesha Verrall, Minister of Health

Purpose of report

1. This report outlines development processes for a rare disorders strategy and discusses themes that are emerging.
2. This report discloses all relevant information and implications.

Summary

3. In its June 2022 response to the independent review of Pharmac, the Government announced the development of a strategy to improve the lives of people and whānau living with rare disorders. The strategy would allow the health sector as a whole to provide much better support for people with rare disorders, make it easier for people, practitioners and organisations to get the information and support that would help.
4. Manatū Hauora | the Ministry of Health is working to deliver a strategy in December 2023. The strategy will seek to seize opportunities created through:
 - a. a more unified and consistent health system
 - b. a heightened focus on equity of outcomes
 - c. advances in science
 - d. progress being made internationally.
5. Work to date has focussed on engagement with rare disorders communities, planning to highlight Māori and Pacific perspectives and outcomes, reviewing international approaches and definitions, and exploring potential collaborations with other countries.
6. We have partnered with Rare Disorders New Zealand (RDNZ) to ensure rare disorders voices are informing the wider strategies and policy statement required by the Pae Ora (Healthy Futures) Act 2022. Themes identified include:
 - a. themes in common with more general health service, consumer and whānau groups including access to support, practitioners and services
 - b. themes particular to rare disorders communities including awareness and access to diagnosis, treatments and coordinated care.
7. The next stages of work are being planned around a series of topics and around sector and industry groups and interests. We aim to provide sufficient opportunities for and actively seek out input from diverse groups, and consider that public confidence is likely to be served without an added formal public consultation process.

8. Our partnerships with RDNZ and with Te Whatu Ora, Te Aka Whai Ora, Pharmac, the Health Quality and Safety Commission and Whaikaha will steer the work to most effectively and efficiently draw on expertise, both lived and professional. It will be important to take minimal time away from people, whānau and practitioners who are already stretched.
9. You may wish to consider copying this report to the Associate Ministers of Health with delegations for Māori health, Pacific health and rural health and/or the Minister for Disability Issues.

Recommendations

We recommend you:

- a) **Note** that we are working with the sector and in partnership with Rare Disorders New Zealand (RDNZ) to provide a rare disorders strategy to you in December 2023;
- b) **Note** that the development work will involve sector expert and working groups and engagement with rare disorders communities and Māori, Pacific, practitioner and provider groups through the coming months;
- c) **Note** that we will keep you informed about progress in strategy development

Maree Roberts
Deputy Director-General
Te Pou Rautaki | Strategy Policy and Legislation

Date: 10/05/23.

Hon Dr Ayesha Verrall

Minister of Health

Date:

Rare disorders strategy: update on development and emerging themes

Background / context

10. The Government announced the development of a strategy to improve the lives of people with rare disorders in its June 2022 response to the independent review of Pharmac¹. Many of the independent review panel's recommendations for rare disorders were aligned with changes being brought in through the Pae Ora (Healthy Futures) Act 2022. The first focus for Manatū Hauora was on the strategies and policy statement required by that Act.
11. The Minister of Health asked Manatū Hauora to lead development of a rare disorders strategy, once the Pae Ora strategies were well progressed. The strategy would allow the health sector as a whole to provide much better support for people with rare disorders, make it easier for people, practitioners and organisations to get the information and support that would help, and improve the lives of people with rare disorders. It would lead to better, more timely services and more equitable support and outcomes for people and whānau with rare disorders².
12. Rare disorders are a significant burden on people, communities and the health system. For example, a 2010 study in Western Australia found that a cohort of people with rare disorders, representing 2% of the Western Australian population, accounted for 9.9% of hospital discharges and 10.5% of state inpatient hospital costs³. Many estimates of rare disorder prevalence are around 6% to 8% in populations. Rare Disorders New Zealand (RDNZ), the peak body for the large number of people and organisations concerned with rare disorders, have been advocating for a strategy for some years.
13. A December 2022 Health Report (2022017711 refers) explained how the rare disorders strategy was being developed alongside and integrated with the Pae Ora and Health Workforce strategies.

Progress to date

14. As part of initial work, Manatū Hauora partnered with RDNZ to ensure rare disorders voices inform the Pae Ora strategies and the long-term insights briefing on precision health. This has been received very positively, though with a degree of frustration among some grass roots members of RDNZ that improvements will take time – there have been calls for improvements, along with access to treatments not available in New Zealand, for some years.
15. A number of comments on the needs of whānau with rare disorders were also made in hui to discuss the interim Hauora Māori strategy in development.

¹ [Government response to the independent review of Pharmac](#)

² [Cabinet paper Government response to the Pharmac Review Panel's report](#)

³ [The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort](#)

16. We have reviewed international approaches to rare disorders and to defining rare disorders, and started to plan how to ensure Māori and Pacific perspectives can underpin the work.

Engagement with rare disorders communities

17. Manatū Hauora has been working in partnership with Rare Disorders New Zealand (RDNZ) to facilitate a series of engagements with the rare disorders community.
 - a. A hui of experts (both lived and professional expertise) gave valuable insights on precision health for the long term insights briefing in development.
 - b. Two webinars attended by some 70 people helped inform the six Pae Ora strategies near completion. The webinars gave voice to rare disorders issues and direct insights into people's lived experiences, adding richness to other input including submissions made by RDNZ.
 - c. Updates through RDNZ newsletters and quick surveys are keeping rare disorders communities of interest abreast of health system reform and direction and the coming rare disorders strategy development and informing our future engagement.
18. Prominent themes have included themes common across wider communities of health service users, such as access to support, practitioners and services. Current pressures the sector is facing have amplified concerns that needs are not being addressed promptly.
19. Other prominent themes are more particular to the rare disorders community. Awareness of rare disorders is considered by people with a rare disorder to be low among health practitioners as well as the wider community. Many people say they simply want to be believed when they tell practitioners about their concerns. Patients frequently face long delays before diagnosis that may mean the disease progresses before treatment can start; treatments may not be available in New Zealand; supportive or symptomatic relief is often limited; and coordinating care across specialties and carers is a very large barrier, especially for whānau who live far from specialist centres.

Māori and Pacific perspectives and outcomes

20. We have started work on the following important inputs to the strategy:
 - a. capturing Māori perspectives on life for whānau with rare disorders, how whānau ora and pae ora can be promoted for whānau with rare disorders, what is most important and what the health system can do better
 - b. ensuring Māori partnership in development and oversight of the strategy
 - c. examining quantitative evidence on impacts of rare disorders, and how this may differ for Māori, such as through the Global Burden of Disease
 - d. similarly capturing Pacific peoples' perspectives and those of other diverse communities
 - e. picking up on related themes and trends that will be highlighted in the long-term insights briefing on precision health.

Review of international approaches to rare disorders

21. Over the last 15 years, many countries have developed rare disorders strategies, registries and centres of excellence. Most have struggled to define rare disorders and

definitions vary. Definitions commonly include a prevalence threshold (such as disorders that occur in less than 1 person in 2000) or thresholds (such as for rare and ultra-rare disorders) and many countries have adopted a specific definition for orphan drug regulation purposes.

22. Countries have faced similar barriers when addressing issues faced by those living with rare disorders, in particular:
 - a. timely diagnosis
 - b. access to treatment and support services
 - c. workforce capabilities
 - d. research.

These areas are common themes in many of the rare disorders strategies, along with infrastructure such as registries; however, it appears that no country has yet addressed all of these areas well through their policy initiatives.

23. The European Union (EU) has played an important role in promoting cross-country support and knowledge exchange to improve care and support across members and neighbouring countries. An EU platform coordinates data registries and facilitates cross-country multidisciplinary collaboration for diagnosis and clinical expertise sharing. The EU promotes rare disorders research across members and translation into clinical services.
24. European Reference Networks (ERNs)⁴ are virtual networks of health professional specialists in the diagnosis and treatment of rare/very rare illnesses in almost all fields of medicine including rare cancers. Their objective is to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources. The ERNs involve 1600 experts embedded in more than 300 EU hospitals, generally distributed in most or all EU countries as well as Ukraine and Norway. They facilitate the exchange of knowledge between health care professionals across borders, giving people access to the expert knowledge they need even if it's not available in their own country or region. The EU supports the ERNs with shared methodology for development of tools like clinical practice guidelines, patient information booklets or quality measures. An evaluation of the ERNs is currently underway.
25. Other countries have a range of strengths in their rare disorders programmes. For example, Canada has strong new-born screening and well-established diagnostic centres that exceed what is available in many other countries. Australia, which published its plan in 2020, has a partnership approach with its consumer organisation Rare Voices Australia. Evidence of effectiveness of the various approaches remains limited.

Considering a definition of rare disorders

26. The Pharmac review panel recommended a definition be developed and agreed; RDNZ has been calling for a definition for some years.
27. Internationally, there is no standard definition of rare disorders or diseases internationally, nor standard criteria for describing the population affected, prevalence, severity of disorders or whether unrecognised or undiagnosed disorders are included.

⁴ [European Reference Networks](#)

Approaches are affected by many factors, including the level of social security, social and economic development, and human cognition of disease in a country.

28. There are a number of advantages in using a descriptive rather than quantitative definition. A description can:
- a. raise awareness of and alertness to the very diverse range of rare disorders
 - b. validate the experiences of people and whānau living with rare disorders or unusual circumstances
 - c. include as yet unrecognised and undiagnosed disorders
 - d. allow flexibility to include low-prevalence conditions affected by changes in social or environmental conditions, such as communicable (eg, rheumatic fever, meningococcal disease) or work-related (eg, asbestosis) conditions
 - e. incorporate prevalence thresholds within the description, both to illustrate people's life circumstances and to indicate boundaries for particular access issues.

Clear boundaries can, on the other hand, improve ease and consistency in clinical decisions and in access to certain services.

29. Pharmac uses 'a clinically defined disorder that affects an identifiable and measurable patient population of less than 1 in 50,000 people' to define a rare disorder for the purpose of its rare disorders medicines funding consideration process (other criteria are also involved). This accords with other countries' prevalence boundaries for orphan drug, high-cost medicine or ultra-rare disorder processes. Most pharmaceuticals for rare disorders are considered through Pharmac's standard funding consideration processes, often because they are also used for less rare conditions.

Access to novel high-cost medicines

30. Major steps in biopharmaceutical research and advances in prevention, screening, diagnosis and treatment have been made in recent decades and the global market for highly specialised medicines is likely to expand significantly over the next decade. This will offer new opportunities for patients. However, high prices for specialised new medicines restrict access, can increase inequities and financial hardship when patients seek to meet costs themselves, and challenge health system sustainability.
31. The World Health Organization Europe region (WHO/Europe) is now establishing a new Access to Novel Medicines Platform (NMP) a formal collaboration mechanism to promote collective action with dialogue and knowledge exchange between Member States, non-state actors, partners and other stakeholders to improve access to novel, high-cost medicines.
32. The strategic aims of the NMP are to:
- a. agree on actions to improve the transparency of the markets to build trust, promote collaboration, enable horizon scanning, facilitate accountability and allow corrective actions to be implemented
 - b. support solidarity-based voluntary collaborations that focus on patient access, including horizon scanning, demand aggregation and life-cycle management
 - c. develop methodology, indicators and systems that recognize the need for sustainability of health-care systems and industry, and enable risk sharing and good governance of the market.

33. New Zealand has accepted an invitation to be an observer on this project.

Key opportunities to be realised by a strategy

34. The strategy will seek to seize opportunities created through:
- a. a more unified and consistent health system, such as through improving access to highly specialist and culturally informed services, and virtual liaison, consultation and care coordination
 - b. a heightened focus on equity of outcomes, especially for Māori, Pacific peoples and disabled people, and on gathering evidence to inform equity improvement
 - c. advances in science, such as through new genetic and pathological testing capability
 - d. progress being made internationally, such as through clinical and research collaboration and virtual specialist networks.
35. Work with the sector will explore these and other opportunities. It will focus on what is most likely to improve equity and health outcomes for people and whānau with rare disorders. It will draw on themes and trends that will be highlighted in the long-term insights briefing on precision health.
36. Cross-jurisdictional approaches, as the EU has championed, appear desirable. Discussions with international colleagues will explore potential for collaboration and how issues that could pose barriers, such as information governance or system interoperability, can be harmonised.

Forward plan for strategy development

37. We understand many of the issues for people and whānau with rare disorders and for the sector, and are starting to identify improvement options. Sector input is critical from this stage to identify all options and test feasibility, acceptability and priorities. This work will require effective management to ensure scarce clinical and other expertise is drawn on without adding pressure.
38. The work will include:
- a. continued partnership with RDNZ to facilitate broad rare disorders community input on a series of themes – RDNZ have since 2020 been promoting seven strategic priorities to improve health and wellbeing for people living with a rare disorder⁵, and have suggested in depth discussions on these themes as a next step
 - b. agreeing oversight with a health sector joint leadership group including the health system entities and Whaikaha
 - c. appointing a reference panel/s in discussion with Te Whatu Ora, Te Aka Whai Ora, Whaikaha and RDNZ, to facilitate effective and efficient expert input from both professional and lived experience, across the broad range of relevant topics
 - d. developing an agreed forward programme of workshops and engagements together with the reference and oversight groups, so that people can plan their input accordingly.

⁵ [Rare Disorders Strategy progress](#)

Should you wish, there could be opportunities for Ministerial announcements of working groups or reference panels prior to the pre-election period.

39. Table 1 summarises our plan to complete and agree the strategy. The plan is subject to changes such as to sequencing that may be needed after further discussion with sector experts.
40. If required, and to meet Budget 24 timeframes for the Health package, we will work with relevant teams to ensure any potential initiatives over and above current and planned services and infrastructure are known, costed and escalated to you as early as practical.

Table 1. Work plan, subject to refining in discussion with oversight and reference groups

Month	Work plan for further refinement
May 23	Form working groups, invite experts for reference panel and agree oversight
June - August 23	Focused engagement and analysis on: <ul style="list-style-type: none"> • definition of rare disorders • key services such as paediatric, genetic, metabolic and pathology • equity improvement for Māori and Pacific peoples • potential international policy, regulatory and research collaborations
September / October 23	Drafting of strategy directions and improvement proposals and testing with reference panel Industry-focused discussions in late October
November 23	Report to Minister on: <ul style="list-style-type: none"> • outcomes of work programme • proposed directions for strategy development • alignment with the government policy statement (GPS) being developed • improvement proposals to progress and any resource implications.
December 23	Provide strategy to Minister (Cabinet consideration to follow)

Equity

41. Many people with rare disorders and undiagnosed health issues also have disabilities. Equity issues for the rare disorders community are likely, on the whole, to align with those prevalent in disabled communities. Though there may be wide individual variation, in the rare disorders community health needs are very high. Access to health services may be higher than average but low to very low in proportion to need.
42. Also similar to disabled communities, disproportionality high rates of unhappiness, depression and anxiety are reported; and income, education and other socioeconomic indicators are lower and/or become lower over time, compounding access issues.
43. The rare disorders population have significantly poorer health outcomes than the whole population. This is further exaggerated among Māori and Pacific peoples for whom culturally informed services may be lacking, and for people including Māori and Pacific peoples who live in remote and high-deprivation communities.

44. A rare disorders strategy is expected to improve equity and health outcomes for disabled people, and to have a high focus on improving equity of outcomes within the rare disorders community, especially for Māori and Pacific people with rare disorders.

Risk management

45. Achieving better outcomes for people and whānau with rare disorders will inevitably raise expectations and demand for health and other services. Achieving gains affordably and equitably will require changes to practices and service provision, potentially for all New Zealanders. It will be important to continue to emphasise the gains in effectiveness and efficiency being made by a more unified and consistent health system, and the consequent ability to direct more healthcare to those needing it most.
46. There is a risk that continuing necessary changes to the broader health system, especially to hospital-based services, may limit the speed with which agreement can be reached on the strategy and on resources that may be required. It will be important to integrate work on this strategy with the system and clinical improvements being made through other work.
47. Our planned sector-facing development process will allow the fastest progress to be made. This will address expressed concerns from the rare disorders community that they have been asking for a strategy for some years; further public consultation could be seen as too much time spent talking about this rather than getting on with making progress. Efficient sector input will also manage risks of adding further pressure to health practitioners and services already struggling to meet healthcare demand. A risk remains that winter illness surges may affect input from health sector personnel.
48. We will endeavour to promote all voices being heard and involve all parts of the sector including those providing high-demand services in the face of workforce shortages and those dominated by non-government providers (such as laboratory services). If this promotion is not sufficient, public consultation on a draft strategy could be considered to provide additional opportunities for input.

Next steps

49. We will continue the work as described in Table 1. Engagement with RDNZ has been positive and fruitful and we are planning ongoing joint hosting of engagements. Communication will stress the potential benefits of a strategy created at this stage of the newly reformed health system.
50. We will continue to engage with RDNZ and with Te Whatu Ora, Te Aka Whai Ora, Pharmac, the Health Quality and Safety Commission and Whaikaha as further work develops. This will help in ensuring reference groups and other inputs inform and steer the work in ways that take minimal time away from people, whānau and practitioners who are already stretched.
51. We will keep you updated with regular progress reports, and can tailor reports for Ministers and Associate Ministers with related responsibilities, should you wish.

ENDS.

Minister's Notes

PROACTIVELY RELEASED