Organisation: Rare Disorders New Zealand (RDNZ)

Submission to: Pae Ora Legislation Committee (Pae Ora Legislation - New Zealand Parliament (www.parliament.nz))

Subject: Pae Ora (Healthy Futures) Bill

Charlotte's story[1]

Charlotte is a four year old kiwi kid with type 2 spinal muscular atrophy (SMA), a genetic condition that affects the control of muscle movement and causes a person's muscles to waste away. Although a treatment, Spinraza, is available in New Zealand it's not publicly funded, so her family moved to Australia where the drug is funded. At that time Charlotte was unable to do basic things for herself like roll over in bed, or walk. After the second dose she could roll over, by the eighth dose she was able to start walking and today she can take about 250 steps a day unassisted. If her family had stayed in New Zealand Charlotte would have been unable to do any of these things, would have remained severely disabled, and most likely would have had a significantly shortened lifespan

James's story[2]

"My cardioversion procedure for the atrial fibrillation I'm experiencing has been postponed, yet again. Though I've experienced it before, this abnormal heart rhythm is deeply disconcerting, adding to the anxiety of an already difficult time. It's a symptom of the damage being done to my heart by Fabry disease, a rare genetic disorder that runs through my family and for which there is no available treatment in New Zealand."

[1] https://www.1news.co.nz/2021/05/04/pharmac-asked-to-fund-drug-for-children-with-spinal-muscular-atrophy-after-it-changes-four-year-olds-life/

[2] https://themedicinegap.co.nz/voices/james-mcgoram/

“In my own case, there are treatments that would slow or halt the progress of Fabry disease. They will not cure me, but they will improve the length and quality of my life. I know this because members of my family who live in Australia and the UK, and who have the identical gene that causes this disorder, receive these treatments”.

“We get to compare notes, and I have the privilege of being their unwitting control group. To my friends I’ve sometimes described my quest for treatment as a game of chicken – at what point do I flinch, and leave the country I love to seek treatment in Australia?”

“Such a solution sits uneasily with me, however, as even though it offers a possible treatment path for me – and my children – it does little to address the systemic inequity of care faced by others with a rare disorder. The fact that I have the privilege to be able to leave is the luck of the draw. These deep-seated challenges are not solved by a single drug, a single breakthrough, or a single Givealittle campaign. They require instead a system-wide change of mindset”.

Lizzie’s story[3]

Lizzie, 17, was born with 22q11.2 deletion syndrome, a complex condition with more than 180 potential anomalies. She has learning difficulties, a serious heart condition and multiple other physical and mental health problems.

Her mum, Sue, says that Lizzie's care has lacked any kind of shape or co-ordination. Because of her rare and complex condition the Haldanes tap into three or four different ministries at one time, but even within one ministry Lizzie's details are not shared.

"In theory with your NHI (National Health Index number) all your medical information is available to every clinician that you see but that appears not to be the practice."

Haldane says as a result, she has told Lizzie's story “hundreds of times to different clinicians”.

“There is no support per se, we basically attack each speciality one at a time. So you work through a speciality, through the health system at the moment, and you continue to be treated in that particular silo, or you are signed off out of that silo and then when you need to address another issue you start again, and you get a referral, and you go through the same process all over again.”

Sue says that under a national rare disorders framework a care plan for Lizzie would have been put in place very early in her life which would co-ordinate the different services she required.

[3] https://www.stuff.co.nz/national/the-detail/300237596/the-detail--urgent-call-for-framework-to-treat-kiwis-with-rare-disorders

It's not fair for rare

Anyone, anytime, could find themselves or a loved one having to cope with the symptoms of a rare disorder. The reality of living with a rare condition can mean waiting years for a diagnosis, juggling specialist appointments and no clear pathway to care or social supports including mental health and education support. The community of people living with rare disorders face the same challenges and systemic barriers despite the type of condition they happen to have. This has been acknowledged in other OECD countries and needs recognition in New Zealand so we can take action to leave no-one behind in our health system reform.

A truly integrated health system must address the needs of this vulnerable population and that cannot happen without awareness of what barriers they face and authentic partnership for solutions. International evidence clarifies that rare disorders need specific pathways and policies, and taking action makes economic, social and moral sense. New Zealanders are shocked to learn how far New Zealand is falling behind, and being a small country is not a good enough reason to dismiss this community of scale with its clear vulnerabilities. Most other countries including Brazil, China, the Philippines, Sweden, the UK and Australia have taken action to ensure equitable diagnosis, treatment and care.

Rare disorders are life-threatening or chronically debilitating conditions associated with high levels of complexity and uncertainty, with an estimated prevalence of less than 1 in 2,000. Examples include well-known conditions such as cystic fibrosis, Batten's disease, Ehlers-Danlos syndrome, muscular dystrophy, and also rare cancers - plus other conditions with no formal diagnosis. Approximately 50% of rare disorder patients are children, 30% of whom will not live to celebrate their 5th birthday[4] thus being denied the opportunity to experience the healthy futures promised in the Pae Ora Bill, and in contrast to Prime Minister Ardern's (paraphrased) sentiment that "*our success as leaders depends on no lesser standard than the well-being of children to whom as the most vulnerable we owe a huge duty of care*".

Many of the 300,000 New Zealanders with a rare disorder fall into one of about 150 different types. A high proportion of these will have been mis-diagnosed and a small number of others have a collection of undiagnosed symptoms, leaving both groups at an elevated risk of missing out on appropriate therapies for their conditions. Those living with a rare disorder in New Zealand experience inequitable, ineffective, and/or no access to the diagnostic and treatment services they require for them to have healthy futures. This is due to a combination of lack of clinician awareness and systemic barriers to appropriate services which are embedded within the current health system. We have the opportunity to improve this picture.

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

These sentiments are reinforced by the following key findings from RDNZ's 2019 NZ Voice of Rare Disorders Survey[5] [6]:

- 60-75% of people and their families reported that their rare disorder makes a number of everyday activities difficult.
- The time burden is substantial for people living with a rare disorder and their carers; the majority requiring over two hours per day for care and coordination.
- There is high utilisation of healthcare services including specialist and GP visits, and diagnostic tests. One in three people were in hospital for an average of 16 days per year. One in twenty people were in ICU for an average of eight days per year.
- There were almost no effective treatments accessible for the majority of people other than for reducing inflammation.
- Most people living with a rare disorder and their carers consider that professionals are poorly prepared to support them and that there is a clear lack of communication between service providers.
- Full employment and education are a challenge for people living with a rare disorder and their carers: this may require modifying work arrangements through part-time contracting; or continuing absence from school for children.
- The disorder has serious effects on social and family life, increasing tension with family members and triggering isolation and feelings of being neglected.
- Mental health of people living with a rare disorder is worse in comparison with the general population with one in three often unhappy and depressed and feeling they cannot overcome their problems.

Rare is common (and cutting edge)…

Collectively rare disorders are not rare, with a potential 10,000 types worldwide. This figure is ever increasing as our understanding of the genetic bases for many diseases develops, and disorders hitherto thought to be common become disaggregated by their much rarer and sometimes unique underlying genotypes. Diagnosis and treatment of rare disorders is therefore at the cutting edge of innovative therapies for all New Zealanders who might become unwell, challenging the "one size fits all" approach in favour of potentially more effective precision medicine techniques and therapies. Far from being "orphan diseases" treatment of rare disorders represents the future of health care and as such they deserve special recognition within New Zealand's health system - in contrast to the marginalisation and indifference that has typically been the experience of both people living with rare disorders and the support organisations which advocate on their behalf.

[5] HealthiNZ. February 2020. Impact of Living with a Rare Disorder in NZ: Why a different approach is needed to improve outcomes for people living with rare disorders, their family and whānau.

[6] HSC-submission-from-Rare-Disorders-NZ_-Sue-Haldane-Petition-v2.pdf (raredisorders.org.nz)

…yet neglected by health planners…

Unlike in other jurisdictions, such as the UK or Australia, New Zealand's health officials have declined RDNZ's repeated offers to co-create a New Zealand Rare Disorder Framework. This framework, the development of which should be consumer led, would offer a cohesive plan and address seven key elements comprising screening and diagnosis; establishment of a national rare disorder registry; planned pathways for critical care; access to appropriate disability and social support; rare disorder medicines; research; and workforce development. Among other things the framework would help to ensure vital support is available for carers of people with rare disorders – who are disproportionately women who may also be suffering from rare disorders themselves (e.g. genetic conditions). It will also assist the New Zealand government in meeting international obligations (eg UN Declaration of Human Rights, CEDAW, UNCROC, UNCRPD, and SDGs).

…and neglected by Pharmac

New Zealanders have the worst access to publicly funded modern medicines out of 20 OECD countries[7], and of the 36 modern medicines launched in the OECD between 2011-2018 to treat rare diseases only two were publicly funded in New Zealand.

This indicates, again in stark contrast to other jurisdictions, that access to life saving and life changing medicines for New Zealanders with rare disorders is unreasonably curtailed, a concern which is extensively and sympathetically considered in the Pharmac Review Interim Report[8] released on 2nd December 2021.

Key points indicated in the report which came through the submitters were that a strategy for patients with rare disorders was needed and that submitters found it unfair that Pharmac has no alternative assessment pathway. The message from the Chair of the report indicates appreciation of the input from the rare disorder community who voiced their collective concerns:

"This interim report assembles much of what the review has heard from patient advocates, clinicians, industry lobbyists, pharmaceutical companies, consumers, Māori and Pasifika health providers and Pharmac itself. Our most grateful thanks go to those who shared deeply personal stories about the struggle – either their own or those of whānau – with rare disorders or disabilities and attempts to get public funding for their medication. No review member was left unmoved".

Although the report confirms that its terms of reference include a requirement to make recommendations on whether Pharmac's *"objectives maximise its potential to improve health outcomes for all New Zealanders (in particular, equitable outcomes for*

[7] IQVIA. November 2021. A Decade of Modern Medicines: An International Comparison 2011 – 2020.

[8] Pharmac Review Panel. 2021. Pharmac Review: Interim report. Wellington: Ministry of Health. Interim Report | Pharmac Review / Te Arotake i Te Pātaka Whaioranga (health.govt.nz)

Māori and Pacific people) as part of the wider health system, and whether and how these should be changed" it is yet to indicate any recommendations. However the preface on the report's webpage states that in the next phase the panel will look at Pharmac's legislative requirements in terms of the new health and disability system and the Pae Ora (Healthy Futures) Bill and also look further at funding for people with rare disorders.

Rather than wait for the review panel's final report RDNZ is taking the opportunity now to submit to the Pae Ora Committee on these matters, and where appropriate we will also convey these submissions to the Pharmac review panel in response to their invitation to contact them regarding new information or feedback on the interim report. In any case it would be seriously remiss if the Pae Ora legislation were to be progressed without taking into account the review panel's recommendations, and RDNZ strongly recommends that it does so.

RDNZ maintains that New Zealand would serve its citizens better if it were to emulate other jurisdictions. We especially commend the approach adopted in England where pharmaceutical funding decisions are made by the National Institute for Health and Care Excellence (NICE).

This agency is established under sections 232 and 233 of the UK's Health and Social Care Act 2012[9], which requires NICE in exercising its functions (including, among other things, deciding on which medicines are to publicly funded) to have regard to

(a) the broad balance between the benefits and costs of the provision of health services or of social care

(b) the degree of need of persons for health services or social care, and

(c) the desirability of promoting innovation in the provision of health services or of social care

Listen to the people

RDNZ is deeply frustrated, albeit not surprised, that despite having engaged with the health system for over twenty years since our inception in 2000, and having specifically engaged with the Health Transition Unit prior to the introduction of the Pae Ora Bill, there is little in the legislation which will make a significant difference to the lives of people living with a rare disorder. We are not surprised because we know that government officials, including Ministry of Health and Pharmac officials, advise elected government members against distinct arrangements for rare disorders[10] in favour of continuing with the existing ineffective generic arrangements. RDNZ therefore urges the Pae Ora Legislation Committee to make decisions and recommendations which first and

[9] Health and Social Care Act 2012. United Kingdom. https://www.legislation.gov.uk/ukpga/2012/7/pdfs/ukpga_20120007_en.pdf

[10] Ministry of Health. Undated. Letter to Health Committee Chair Liz Craig. Petition-MOH-response-to-HSC.pdf (raredisorders.org.nz)

foremost serve the interests of the New Zealand public - to whom they are accountable - without undue influence from officials with an interest in the status quo.

RDNZ's submission

The Pae Ora (Healthy Futures) legislation as currently drafted does little to improve the situation or health outcomes for Charlotte, James or Lizzie, or to ensure that they and thousands like them and their whānau and families will experience the healthy futures promised by the legislation.

RDNZ's submission recommends amendments to the Bill to address the current systemic deficits and to ensure that Charlotte, James and Lizzie and others experience significant and positive outcomes from the Pae Ora (Healthy Futures) legislation. The specific issues which we have consistently raised with government policy and decision makers and which we believe could effectively be addressed through the Pae Ora legislation are:

1. Consistent access, irrespective of geographical location, to effective services which are provided in accordance with contemporary evidence based standards of care
2. Access to life saving and life changing medicines
3. A government rare disorders strategy which both acknowledges that people living with a rare disorder comprise a vulnerable population and mandates how services must be delivered to people living with a rare disorder

NZHR's recommended amendments to the Bill in respect of these issues are set out in the following tables[11] (pages 8 – 14):

[11] Links to the relevant sections in the legislation are underlined and blue

RDNZ's recommendations

Healthy futures for New Zealanders with rare disorders through consistent access to effective services which are provided in accordance with contemporary evidence-based standards of care, irrespective of geographical location.

Issues	RDNZ recommends that:
General Policy Statement Successive reviews of the publicly-funded health system in New Zealand, including the 2020 Health and Disability System Review, the subsequent 2021 White Paper and indeed the Pae Ora (Healthy Futures) Bill itself have repeatedly overlooked the consistently poor outcomes experienced by those diagnosed and living with rare disorders. In contrast however the Interim Report of the Pharmac Review does highlight this in respect of access to medicines. RDNZ holds that the second paragraph of the General Policy Statement (GPS), which states that "*one of the root causes of… inequity and variation was the structure of [a] health system…that had become fragmented and complex, leading to unclear roles, duplication, misalignment, and a lack of a common whole-system ethos*", applies particularly to those living with rare disorders.	In light of the content of the Interim Report of the Pharmac Review the opening sentence of the General Policy Statement be amended to: *Successive reviews of the publicly-funded health system in New Zealand…have found consistently poor outcomes for some groups, in particular Māori, Pacific peoples, people living with rare disorders, and people with disabilities, and significant unwarranted variation in service availability, access, and quality between population groups and areas of New Zealand.*
Regional Arrangements RDNZ notes that the Health and Disability Review report contemplated 6-8 DHBs, the subsequent health reforms White Paper proposed four Health New Zealand subregions, and the current Bill leaves the actual number to be determined by the Minister. Irrespective of the details Charlotte, James and Lizzie and people like them are not served well when services are	Section 97(1)(a) be deleted and replaced by *National service arrangements* *The Governor-General may, by Order in Council, on the recommendation of the Minister, make regulations…* *(a) specifying national service arrangements—* *(i) through which Health New Zealand and the Māori Health Authority must provide and arrange services; and*

Issues	RDNZ recommends that:
organised regionally. People with rare disorders deserve to receive evidence-based services which are consistently available, delivered with consistently high quality in accordance with internationally agreed standards of care for their particular condition, as part of a nationally managed service stream. This is the opposite of the current postcode lottery arrangements which would risk being perpetuated under any future regional arrangements. We believe that the same arguments could be made for other conditions (such as mental health and cancer services for example), and that rather than have a patchwork quilt where some services are provided regionally and some nationally, it makes best sense for them all to be provided along national service lines. People with rare disorders could come under the umbrella of a dedicated national rare disorders and precision medicine service (RDNZ's preference), or be a discreet sub-service of a broader national service.	*(ii) which must be maintained by the Health New Zealand and the Māori Health Authority*
Evidence based services As a corollary to the above RDNZ believes that the current postcode lottery approach to delivering services to people with rare disorders is indicative of their not receiving services which are consistently evidence-based (ie supported by results of best available, credible contemporary research). Were service provision to be consistently evidence based there would be little to no variation in clinical practice, in marked contrast with the current experiences of those who live with rare disorders.	Section 7 be amended to include an additional health system principle: *(f) all health system strategies, policies, practices, services, programmes and interventions are evidence based* Section 13 (a) be amended to: *The objectives of Health New Zealand are—* *(a) to design, arrange, and deliver evidenced based services to achieve the purpose of this Act in accordance with the health system principles*

Issues	RDNZ recommends that:
RDNZ therefore recommends that legislation be amended to require that service provision together with the principles, plans and strategies referred to in the Bill be evidenced based.	Section 41(1) be amended to include *When preparing a health strategy, the Minister must—* *(c) have regard to all available evidence including results of best available, credible contemporary research*

Fair access to life saving and life changing medicines

Issues	RDNZ recommends that:
Health system principles Clauses 7(1) (b) and (c) of the health system principles require engagement with…other population groups…to develop and deliver services and programmes that reflect their needs and aspirations…and provision of opportunities for Māori to exercise decision-making authority…having regard to…the interests of other health consumers. Clause 7 (4) states that these two health system principles do not apply to Pharmac and the performance of its functions. Pharmac's inadequate engagement with people with rare disorders contributes to poor health outcomes by creating undue barriers to equitable access for small populations to innovative modern medicines. Without treatment New Zealanders with rare disorders impact hospital and ICU beds, require family carers and create a high societal economic burden RDNZ maintains that Pharmac should be bound by clauses 7(1) (b) and (c) in the same way as all other components of the health system. Inclusion cannot exclude a community of scale and still remain equitable.	The health system principles section of the Bill be amended by deleting clause 7 (4) in its entirety.

Issues	RDNZ recommends that:
Functions of Health New Zealand Clause 14 (3) states that "in performing any of its functions in relation to the supply of pharmaceuticals, Health New Zealand must not act inconsistently with the pharmaceutical schedule". The problem with this clause is that it will result in people such as Charlotte, James and Lizzie being denied life-saving and life improving therapies which will enable them to contribute to the economy and society generally through valued social and occupational roles, simply because Pharmac claims it does not have the resources or mandate to fund such treatments. RDNZ maintains that there has to be a provision to override Pharmac where a good efficacy and funding case can be made. This could be by way of an alternative assessment pathway such as those in place in other OECD countries. Health New Zealand should not have to automatically have its mandate to achieve best health outcomes for all held hostage by Pharmac's heavily proscribed decision making processes.	Clause 14 (3) be amended to: *"in performing any of its functions in relation to the supply of pharmaceuticals, Health New Zealand may with the consent of the Minister of Health, act inconsistently with the pharmaceutical schedule where this is warranted by:* *(a) the broad balance between the benefits and costs of the provision of pharmaceuticals, medical devices and other therapies it is required or authorised to purchase* *(b) the degree of need of persons for pharmaceuticals, medical devices and other therapies, and* *(c) the desirability of promoting innovation in the provision of pharmaceuticals, medical devices and other therapies*
Pharmac As above, the problem with Pharmac's current objectives is that people such as Charlotte, James and Lizzie are being denied life-saving and life improving therapies which will give them a healthy future and enable them to contribute to the economy and society generally through valued social and occupational roles, simply because Pharmac claims it does not have the resources or mandate to fund such treatments. Compared to other OECD countries New Zealanders' access to medicines is the worst[12] and RDNZ maintains that New Zealand	Section 61 (1) and (2) be deleted and replaced with *(1) The objectives of Pharmac are—* *(a) to secure for eligible people in need of pharmaceuticals, the best health outcomes that are reasonably achievable from pharmaceutical treatment; and* (b) any other objectives it is given by or under any enactment, or authorised to perform by the Minister by written notice to the board of Pharmac after consultation with it.

[12] IQVIA. November 2021. A Decade of Modern Medicines: An International Comparison 2011 – 2020.

Issues	RDNZ recommends that:
would serve its citizens better if it were to emulate other jurisdictions. We especially commend the approach adopted in England where pharmaceutical funding decisions are made by the National Institute for Health and Care Excellence (NICE), as outlined above. RDNZ recommends in the strongest possible terms that the Pae Ora legislation be amended to require Pharmac to operate with the same parameters, and that the references in the Bill to having to operate within a fixed budgetary allocation be removed. This measure taken together with RDNZ's other recommendations would be a hugely significant step towards healthy futures for people living with rare disorders. If the Labour government will not see itself clear to do the right thing and adopt this recommendation, then at the very least the Bill should equip Pharmac with an alternative assessment pathway for provision of innovative life-changing treatments, allowing Pharmac to spend beyond its funding envelope where the costs of investing in a treatment can be justified by both the overall benefits to society and notions of human decency and fairness. RDNZ further believes that to minimise the need for the proposed "alternative assessment pathway" provision there should be a requirement for Pharmac to be sufficiently funded so that New Zealanders receive pharmaceutical treatments in a way that is consistent with what is available to citizens of comparable advanced economies.	*(2) in carrying out its objectives Pharmac shall be bound by:* *(a) the broad balance between the benefits and costs of the provision of pharmaceuticals, medical devices and other therapies it is required or authorised to purchase* *(b) the degree of need of persons for pharmaceuticals, medical devices and other therapies, and* *(c) the desirability of promoting innovation in the provision of pharmaceuticals, medical devices and other therapies* *(3)* In this section, eligible people means people belonging to a class specified in regulations made under section 97 as being eligible to receive services funded under this Act. By way of a less satisfactory alternative RDNZ's fallback position is that: Section 61(1)(a) be amended to the objectives of Pharmac are to *"secure for eligible people in need of pharmaceuticals, the best health outcomes that are reasonably achievable from pharmaceutical treatment, either from within the amount of funding provided, or with the consent of the Minister of Health, as otherwise warranted by societal costs, benefits and notions of human decency and fairness,* and section 62 be amended to include a new subclause 62(3): *Pharmac shall be allocated sufficient funds to enable it to carry out its objectives in a way that is consistent with comparable international norms*

A government rare disorders strategy which both acknowledges that people living with a rare disorder comprise a vulnerable population and mandates how services must be delivered to people living with a rare disorder

Issues	RDNZ recommends that:
Interpretation RDNZ believes that the legislation should make explicit reference to rare disorders, which should be defined as per international norms best exemplified by European Union Orphan Drug Regulation 141/2000, which defines a disease or disorder as rare when it affects less than 1 in 2000. Additionally the Bill makes frequent references to the "term population groups" without providing a corresponding interpretation of what this means.	The Interpretation section of the Bill be amended to include: *"rare disorder means a disease or disorder which affects less than 1 in 2000 people in the New Zealand population"* *"population groups means Māori, Pacific, people with a disability, and people with a rare disorder"*
Key roles and health documents RDNZ maintains that people with rare disorders have long been neglected by the health system, as illustrated by the stories of Charlotte, James and Lizzie above. We believe that people with rare disorders have unique issues and challenges which should be recognised through a specific health strategy.	Sections 10 (1)(a) 29 (1)(b) "key roles and health documents" and "overview of important health documents" be amended by adding *(v) Rare Disorders Health Strategy* The Bill be amended by including a new Section 41 as follows: ***Rare Disorders Health Strategy*** *(1) The Minister must prepare and determine a Rare Disorders Health Strategy.* *(2) The purpose of the Rare Disorders Health Strategy is to provide a framework to guide the health system in improving health outcomes for people with rare disorders.* *(3) The Rare Disorders Health Strategy must—* *(a) contain an assessment of the current state of health outcomes for people with rare disorders and*

Issues	RDNZ recommends that:
	the performance of the health system in relation to people with rare disorders; and *(b) contain an assessment of the medium and long-term trends that will affect the health of people with rare disorders and health system performance; and* *(c) set out priorities for services and health system improvements relating to the health of people with rare disorders, including workforce development.* *(4) **Subsection (3)** does not limit what may be included in the Rare Disorders Health Strategy.*