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Taumata Rau
**University
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INNOVATIONS



Te Huinga
Hāpai Tāngata
COMPASS
Research Centre



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Using administrative data to explore the health and service use of people with diagnosed rare disorders in Aotearoa

These results are not official statistics. They have been created for research purposes from the Integrated Data Infrastructure (IDI) which is carefully managed by Statistics New Zealand. For more information about the IDI please visit <https://www.stats.govt.nz/integrated-data>

Acknowledgements



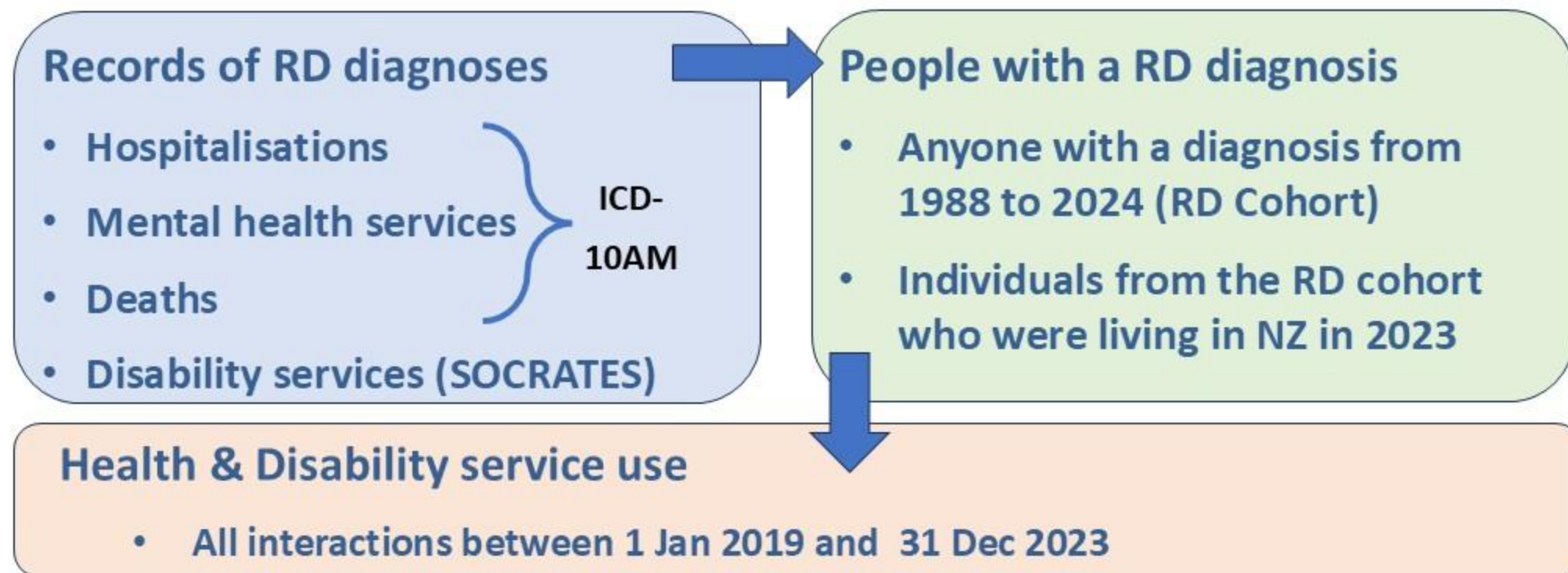
Today's presentation

The (perceived & real) challenges of using the Integrated Infrastructure (IDI) to obtain information on people with Rare Disorders (RDs)

- Reliance on ICD-10 coding
 - Do a sufficient number of RDs have an ICD-10 code for meaningful data collection?
 - Are ICD-10 codes specific enough to identify RDs?
- RDs are rare!
 - Will IDI searches yield sufficient numbers of people with RDs for meaningful data analysis?
 - How do IDI confidentiality rules restrict output on RDs?

Rare Disorders in Aotearoa NZ Project

- Socio-demographic & clinical characteristics of people with a diagnosed RD
- Diagnostic & care pathways experienced by people with a diagnosed RD



Individual linkage to these data: population, Census (2023), health, disability, aged care

Integrated Data Infrastructure (IDI)



The Integrated Data Infrastructure (IDI) is a research database that holds de-identified (anonymous) information about people and households in Aotearoa New Zealand.

The data comes from surveys, government agencies and non-government organisations (NGOs).

Data are linked at the individual (person) level. We can connect information about a person across different sectors. E.g., education records can be linked to hospital admissions.

Updated with new information a few times a year.

Rare Disorders in Aotearoa NZ Project

Identifying people with a diagnosed RD

RDs defined as conditions that:

- Are classified as a disorder in the Orphanet database of rare diseases, and
- Affect not more than 1 person per 2000 in the population

RDs were considered for inclusion in the project if they:

- Have a corresponding ICD-10 code in the Orphanet rare disease knowledge base (ORPHAcode)
- Were not among groups of disorders excluded from the project (see Slide 8)
- Have a corresponding ICD-10 code that only identifies RDs

Phase 1: Orphanet to ICD-10AM coding

Sources: Orphanet nomenclature pack (2024).

<https://www.orphacode.org/pack-nomenclature/>

Australian Independent Health and Aged Care Pricing Authority (IHACPA). ICD-10 to ICD-10-AM Eleventh Edition mapping tables

Orphacodes at disorder level

- N=7,195

Orphacodes with ICD-10 code

- N=6,370

Unique ICD-10/Orphacodes

- N=2,110



Exclusion criteria:

Infectious disorders (A00-B99)

Injury, poisoning & other external causes (S00-T88)

Malignant Cancers

Mental health disorders F00 to F99 (except Rett Syndrome & Landau-Kleffner)

Illness, unspecified (R69)



1,307 unique ICD-10 codes
relating to 5,547 unique Orphacodes



ICD-10 codes mapped to ICD-10AM & ICD-9
ICD-9 codes harmonised to one ICD-10AM code

Relationships between ICD-10 & Orphacodes

1. Exact match
2. Orphacode narrower than ICD-10 code
3. Orphacode broader than ICD-10 code

! Diagnoses not always coded using
narrowest ICD-10 code !

Phase 2: ICD review & classification

1. ICD code relates to one condition

- ✓ Condition considered to be rare
- ✗ Condition is not considered to be rare

2. ICD code relates to multiple conditions

- ✓ Overarching ICD code considered to be rare
- ✗ None or only some of the conditions are considered to be rare
- ✓ Individual conditions all considered to be rare

3. Only some forms of a condition are rare

- ✗ ICD code includes specifiers for rare forms of a condition but overall that condition is not considered rare even though it is included in Orphanet (e.g. Down Syndrome)

Phase 3: Additional codes and pilot searches

SOCRATES Diagnosis codes: N=140

- ✓ Label for code corresponds to a rare condition



23 SOCRATES Diagnosis codes
harmonised to one ICD-10AM code

Conditions covered by support groups under the RDNZ (as at June 2024)

- ✓ Condition has an ICD code that meets the project inclusion criteria
- ✓ Condition has an ICD code that was not in the 2024 ORPHA Nomenclature pack but is considered rare
- ✗ Condition does not have a corresponding ICD code
- ✗ Condition has an ICD code or is coded under a broader ICD code that was excluded in Phase 1 or 2

924 ICD-10AM codes and associated ICD-9 and SOCRATES Diagnosis codes

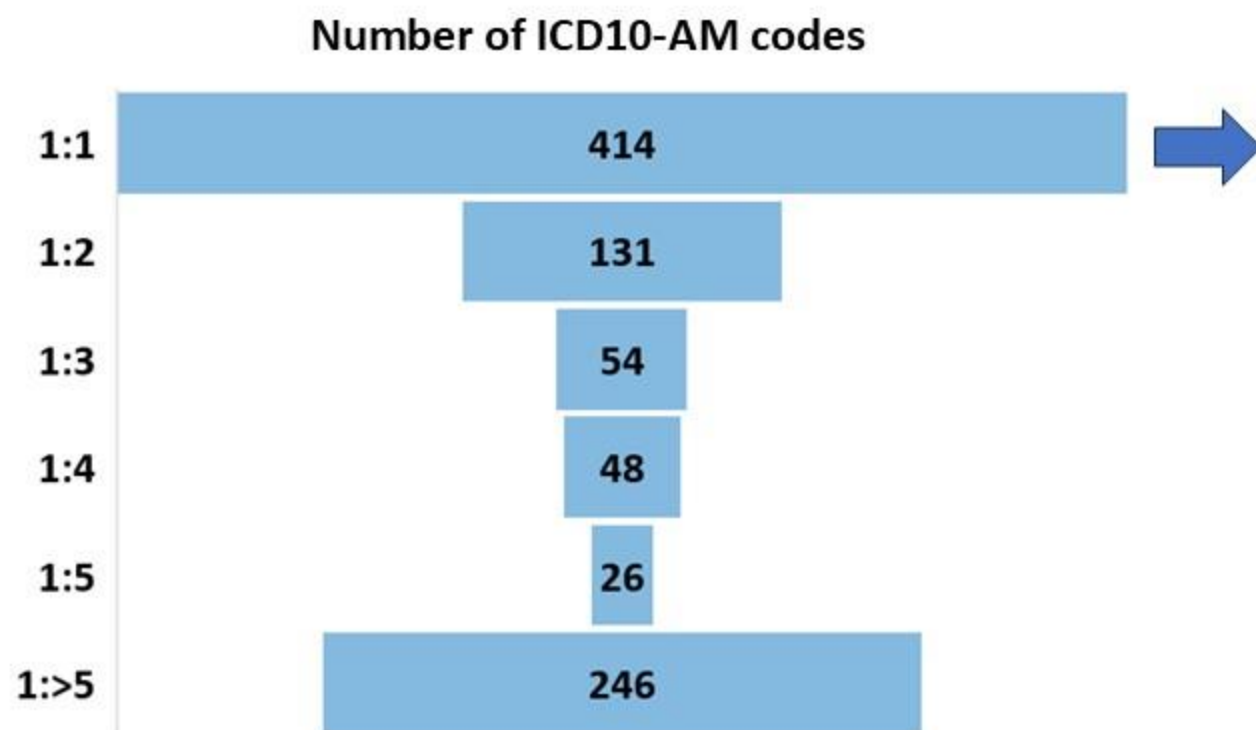
IDI search of public hospital records (1988 to 2024) & review of codes with >5,000 individuals

919 ICD-10AM codes with associated ICD-9 & SOCRATES Diagnosis codes

Final ICD-10AM selection

The 919 ICD-10AM codes included in the project correspond to 3,431 Orphacodes

- 45% of the ICD-10AM codes correspond to a single Orphacode disorder



	N	%
ICD-10 groupcodes		
Congenital, Chromosomal & Genetic	182	44
Blood & Immune	45	11
Nervous System	44	11
Skin	40	10
Musculoskeletal	39	9
Endocrine, Nutritional & Metabolic	26	6
Circulatory	16	4

Number of people with a diagnosis of these 414 conditions ranges from <100 to >5,000

Number of people with a diagnosis of the 919 ICD-10AM codes ranges from <10 to >5,000

Outputting IDI data for small cohorts or subgroups

Tables of aggregated data (unweighted counts)

Age	European	Māori	Pacific	Asian
Under 1 Yr	3,675	1,422	828	600
1 to 4	522	255	162	69
5 to 11	432	171	96	39
11 to 17	507	246	111	51

Any counts <6 are suppressed
All counts are rounded to base 3

	Died aged under 65
Neuronal ceroid lipofuscinosis	21
Conjoined twins, unspecified	15
Zellweger syndrome	12
Hydranencephaly	12
Smith-Lemli-Opitz syndrome	12



Safe output

Source: Stats
NZ. (2022).
Microdata
output guide.
5th edition,
version 2.

Summary statistics (e.g. means, medians)

	N	Mean	SD
20-24 year olds	48	13.22	1.54
25-29 year olds	75	9.81	1.65
30-39 year olds	123	7.22	0.98
40-49 year olds	45	5.00	1.07
50 and over	S	S	S

Data for underlying
counts <20 are
suppressed

All counts are
rounded to base 3

Regression models

Outcome	N	Estimate	Lower CI	Upper CI
Any benefit receipt	229743	0.0728	0.0075	0.1381
Any mental health problem	229743	0.1616	0.0969	0.2263
Any non-cancerous pain	229743	0.1516	0.0868	0.2163

Data for underlying counts <5 are suppressed

People with diagnosed RDs in Aotearoa New Zealand

The most common type of RD is “Congenital malformations, deformations, chromosomal abnormalities, and genetic disorders” (ICD-10 Q codes)

Many conditions in other categories are also congenital, hereditary or have a strong genetic component

	Number of people	Number of ICD-10 codes in search
Congenital, Chromosomal & Genetic Disorders (Q)	99,555	489
Blood & Immune Diseases (D)	73,701	80
Nervous System Diseases (G)	62,709	87
Musculoskeletal Diseases (M)	43,041	71
Endocrine, Nutritional & Metabolic Diseases (E)	42,651	86
Circulatory Diseases (I)	31,509	19
Skin Diseases (L)	31,350	49
Eye & Ear Diseases (H)	10,791	10
Genitourinary Diseases (N)	6,114	7
Digestion Diseases (K)	5,712	9
Mental, Behavioural & Neurodevelopmental Disorders	942	2
Neoplasms	3,075	1
Respiratory Diseases	1,914	9

People with diagnosed RDs in Aotearoa New Zealand

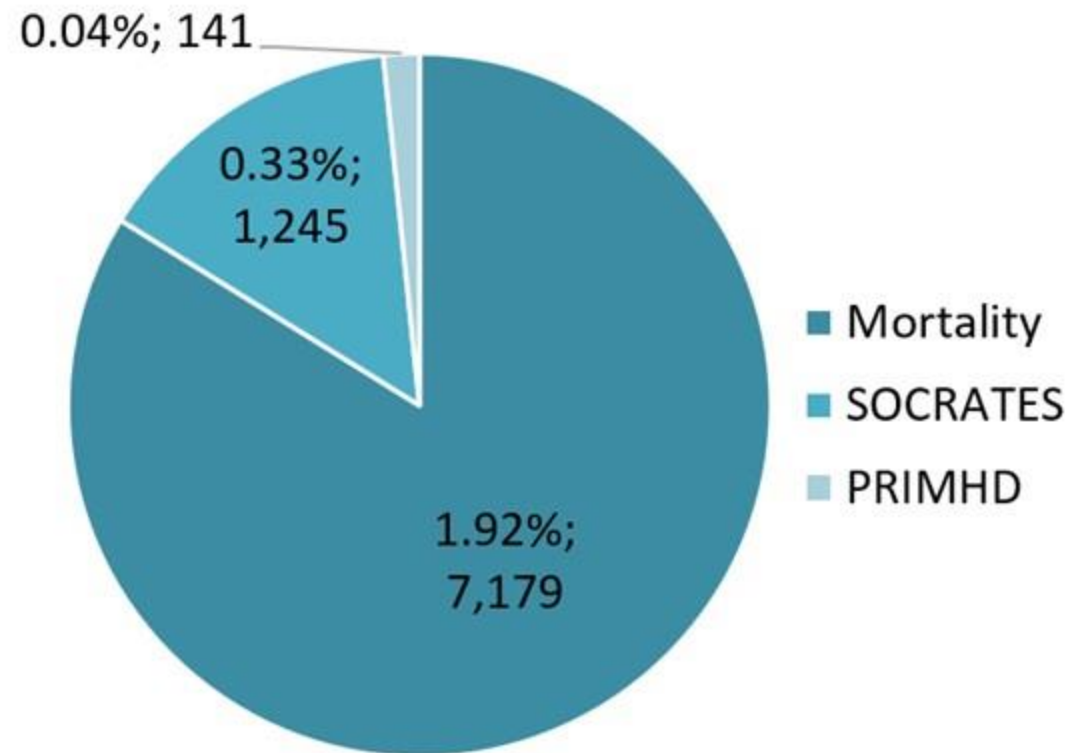
374,000 people with ≥ 1 of 870 ICD-10 disorders (records from 1988 to 2024)

- 63,200 people (17%) had more than one RD diagnosis recorded

**97.7% of the cohort were
identified from NMDS
(public & private hospitalisations)**

**The number of people with a first
recorded RD diagnosed in a given
year increased from around 10,000
in 1988 to 15,000 in 2023**

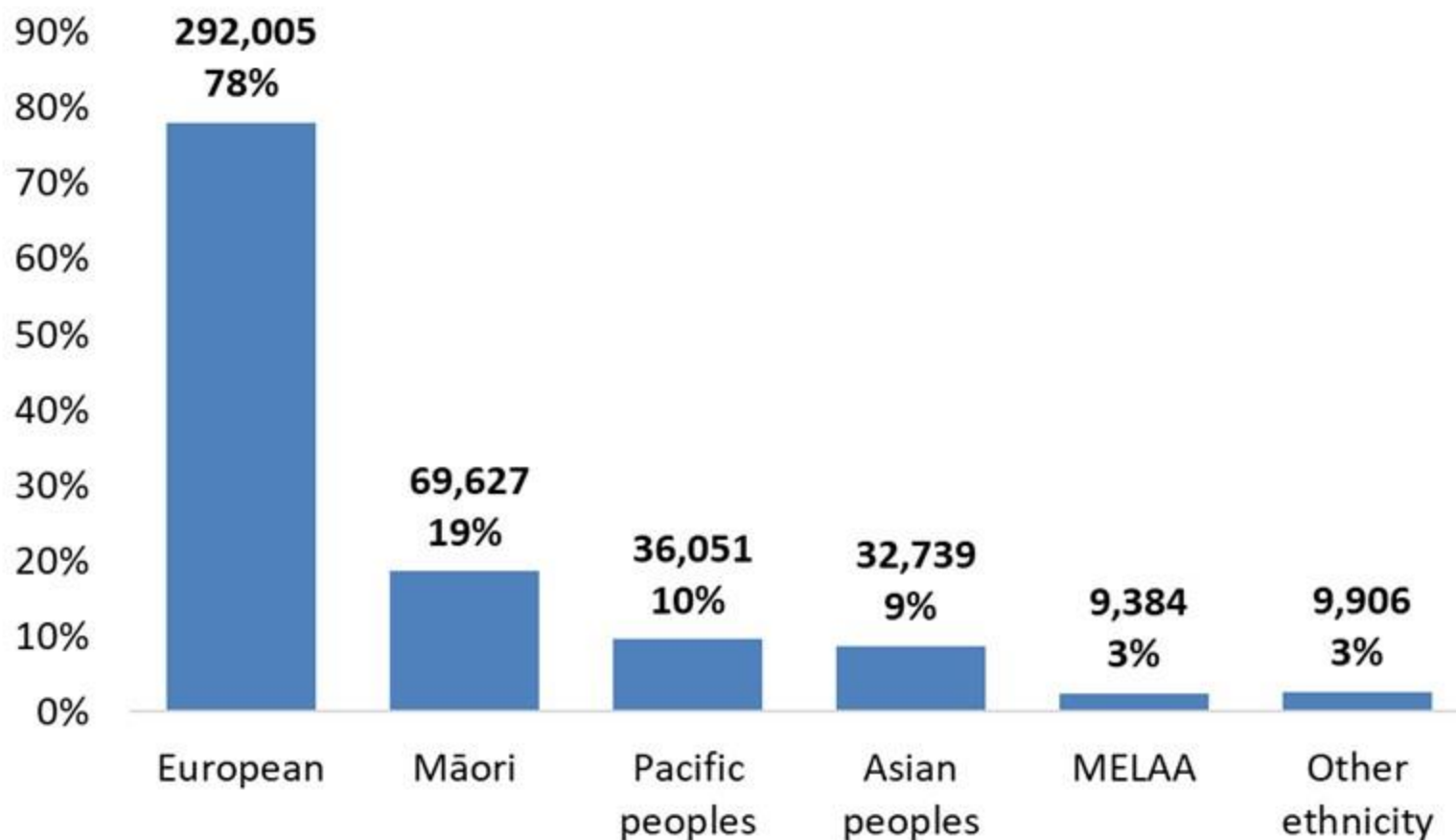
8,565 individuals identified by other sources



Characteristics of people with diagnosed RDs (1988 to 2024)

- No gender imbalance within the RD cohort

Number and % of RD cohort that identified with each ethnicity group



Gender, birth date and total response ethnicity data were sourced from the Personal details table and Censuses.

MELAA = Middle Eastern, Latin American or African

First record of RD diagnosis – age of individuals (1988 to 2024)

27% of RD cohort had their diagnosis first recorded when they were under 18 years of age

41% had their diagnosis recorded when they were 18-64 years of age

Around 1/3 had their diagnosis recorded when they were 65 years of age or older

Age at first record of RD

Number of people

% of people with RD

Under 1 Yr

45,222

12%

1 to 4

23,298

6%

5 to 11

19,887

5%

12 to 17

14,142

4%

18 to 29

30,144

8%

30 to 44

45,012

12%

45 to 64

77,340

21%

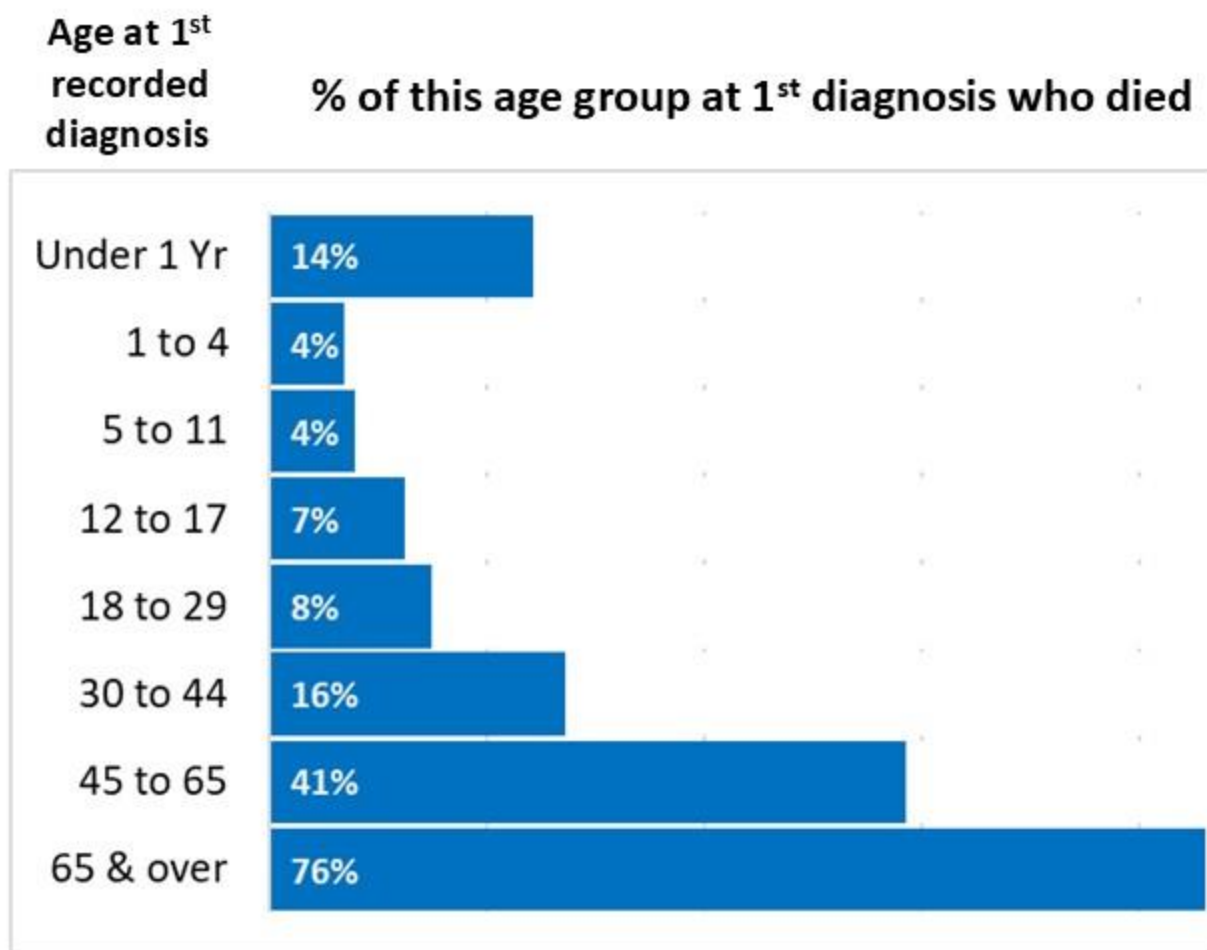
65 & over

119,037

32%

People with diagnosed RDs in Aotearoa New Zealand (1988 to 2024)

- 140,200 people in the RD cohort died (37%) = 13% of all deaths
- 7,780 people were <18 years of age when they died = 27% of deaths of under 18s
- 30,100 died at 18-64 years of age = 14% of deaths at 18-64 years
- 1 in 10 infants who were <1 year old when diagnosed died within 2 years of their diagnosis



People with diagnosed RDs in Aotearoa New Zealand (1988 to 2024)

% of people in each ethnicity group

	1st Dx at <18 years of age	1st Dx at 18-64 years of age	1st Dx at ≥65 years of age	Died	% of deaths that were ≤2 years after 1st Dx	% of deaths at <18 years of age	% of deaths at 18-64 years of age	% of deaths at ≥65 years of age
European	25%	38%	36%	40%	36%	4%	18%	78%
Māori	44%	44%	13%	26%	39%	11%	42%	47%
Pacific	46%	41%	13%	23%	43%	15%	39%	47%
Asian	39%	45%	16%	17%	47%	14%	28%	58%

People with diagnosed RDs living in NZ as at 30th June 2023

213,590 people living in NZ in 2023 with a recorded RD diagnosis

- = 1 in 24 people living in NZ
- 14,930 people with a first recorded diagnosis of a RD in 2023

Children (under 18s) comprised
19% of those living with a
diagnosed RD in NZ in 2023
(n>41,000)

50% of the 2023 RD cohort were
aged 45 or over (n>106,000)

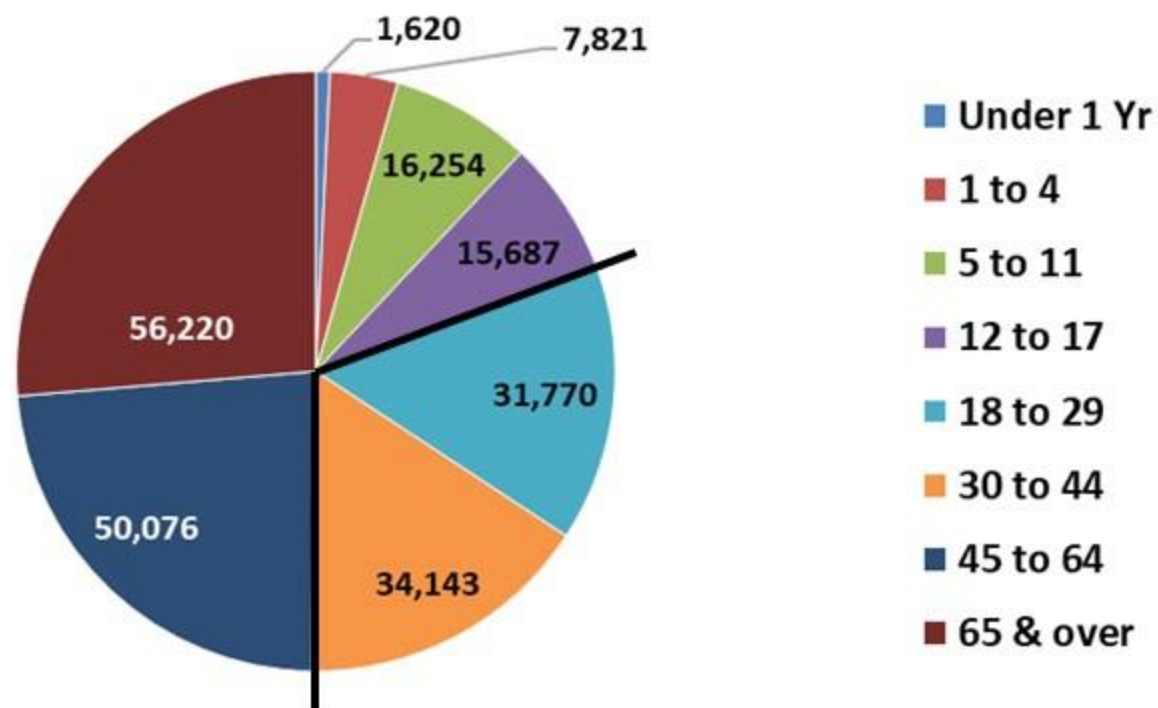


Chart shows the number of people in each age group

Challenges of using IDI to obtain information on people with RDs

These are likely to be undercounts of the true number of people with RDs in NZ

- Many RDs do not have an ICD-10 code or share an ICD-10 code with common conditions
- Disability services data are based on a limited range of RD diagnoses (23 diagnosis codes)
- Many people with RDs will not have been hospitalised or used disability services

Those included in the entire RD cohort for this project are likely to be those who:

- Have a RD or comorbid health condition that has severe impacts, resulting in hospitalisation, death and/or disability service use
- Had a first recorded RD diagnosis at a time when they were seriously ill or died. These people may have been living with a RD before this first record

Classifying RDs and types of RD using ICD-10AM diagnostic codes is a significant challenge when there is not an exact match between an ICD-10AM code and an Orphacode disorder

Thank you!

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