

Coding for rare disorders in the health record

Rare Disorders Research Forum, 5–6 August 2026

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Aotearoa New Zealand Rare Disorders Strategy – Te Rautaki o Aotearoa e Pā ana ki ngā Mate Mokorea

Collecting information on rare disorders

National health data sets will be able to routinely collect rare disorder information.

The level of diagnostic information that is available to readily search and report on will become more extensive and detailed over time. Increasingly, people and their whānau living with rare disorders will be able to access their information as well as more general information on rare disorders in Aotearoa New Zealand.

Clinical classifications will become increasingly detailed as national data collections adopt more advanced and specific classification systems. Classification systems will need to connect easily to supplementary data sets on rare disorders in particular.

Using information to support provision of care

In the future, services will use rare disorder information to guide, monitor and improve care for patients.

As rare disorder information becomes more visible in the system, it will have many uses. These include to plan care, follow care provision and results, fill gaps and make improvements. These activities will build on work already done for some rare disorders, such as metabolic disorders.

Where clinical coding is not yet available, methods will be developed to connect high-quality rare disorder information with national health data.

Over time, planners and researchers will use non-identifiable information to improve care.

Rare diseases are a **proving ground for medical innovation**, shaping breakthroughs in genomics, gene therapy, AI and precision medicine

Stronger, **interoperable data foundations** can accelerate diagnosis, unlock new treatments, reduce costs and improve outcomes

By investing in **better data, smarter regulation** and cross-border collaboration, rare disease innovation can benefit entire health systems

World Economic Forum

Aotearoa New Zealand Rare Disorders Strategy 2024 – 2034

Implementation Plan 2026-27

Learning and sustaining progress

Actions begin the development of a health system that has robust, usable information on rare disorders to support planning, care delivery, monitoring and investment decisions.

Actions	Lead	Outcomes
- Health New Zealand continues with primary care, hospital and specialist care coding improvements by: - publishing to the sector a SNOMED CT code set of all 7,000 approx. rare disorders covered in the Orphacodes. The code set would be distributed in the July 2026 release of the SNOMED CT NZ edition. Due date 20 July 2026. - providing guidance to software suppliers on how to use the code set (including how to map between SNOMED CT and Read Codes) September 2026 (ongoing).	DONE HNZ	Responsive system
Health New Zealand identifies and plans steps towards improving the continuity and coordination of care for people with rare disorders	HNZ	Responsive system

hiso

Health Information Standards Organisation

PAEREWĀ PĀRONGO HAUORA



Our standards products

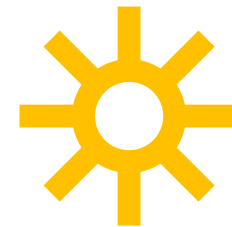
HISO Standards

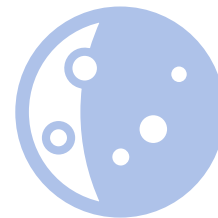
SNOMED NZ Edition

NZ Health Terminology Service

NZ Core Data for Interoperability

NZ Patient Summary





Leading healthcare terminology, worldwide



World Health Organization



GDHP

Global Digital Health Partnership



Spark



orphanet



openEHR



350,000,000
are affected by a
rare disorder globally



50%
of those affected
by a rare disorder
are children



7,000+
known rare disorders



80%
are genetic in
origin



500+
types of
rare cancer



All childhood
cancers
are rare



Making sense of the numbers

Rare Disorders Insights Report

Pathways towards better health outcomes

Here-turi-kōkā 2022

Advocate for the Government health agencies to develop a rare disorder patient registry, which uses the **Orphanet classification**

This would:

- Improve the way data is collected and classified
- Make international research and collaboration easier
- Increase diagnosis speed and accuracy

How do we record a diagnosis of
aHUS – atypical haemolytic uraemic syndrome
in the person's health record?

ORPHA:2134

SNOMED CT 789660001

ICD-11 3A10.Y

aHUS is a rare, genetic thrombotic microangiopathy due to dysregulation of the alternative complement pathway and characterized by the triad of haemolytic anaemia, thrombocytopenia, and acute renal dysfunction



Recognised international code system specifically for rare disorders

SNOMED CT

Clinical terminology standard for health records of all kinds

ICD-11

Classification system for statistics and reporting

**ORPHA****C****O****D****E****S**

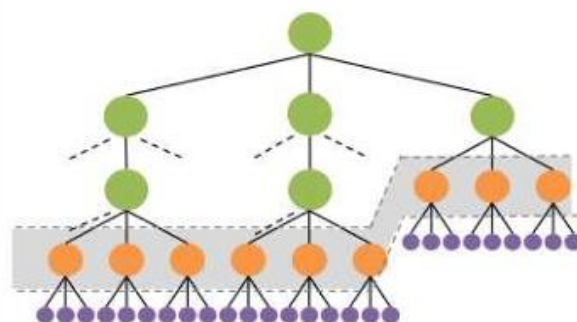
41	ORPHA:2130	Non-syndromic longitudinal meromelia	Synonym	Group of disorders	
42	ORPHA:2131	Alternating hemiplegia of childhood	Main name	Disorder	
43	ORPHA:2131	AHC	Synonym	Disorder	
44	ORPHA:2132	Hemoglobin C disease	Main name	Disorder	
45	ORPHA:2132	HbCC disease	Synonym	Disorder	
46	ORPHA:2133	Hemoglobin E disease	Main name	Disorder	
47	ORPHA:2134	Atypical hemolytic uremic syndrome	Main name	Disorder	
48	ORPHA:2134	aHUS	Synonym	Disorder	
49	ORPHA:2134	Atypical HUS	Synonym	Disorder	
50	ORPHA:2135	Cutaneous mastocytosis-deafness-microtia syndrome	Main name	Disorder	
51	ORPHA:2135	Cutaneous mastocytosis-hearing loss-microtia syndrome	Synonym	Disorder	
52	ORPHA:2135	Mastocytosis-short stature-deafness syndrome	Synonym	Disorder	
53	ORPHA:2135	Mastocytosis-short stature-hearing loss syndrome	Synonym	Disorder	
54	ORPHA:213500	Rare ovarian cancer	Main name	Group of disorders	
55	ORPHA:213500	Rare ovarian malignant tumor	Synonym	Group of disorders	

What is an ORPHAcode?

A RD-specific classification system for secondary use



ORPHA code	Preferred term	Synonyms	Classification level
ORPHA:2134	Atypical hemolytic uremic syndrome	Atypical HUS aHUS	Disorder
Definition	a rare thrombotic microangiopathy disorder characterised by mechanical hemolytic anemia, thrombocytopenia, and renal dysfunction.		



Group
Disorder
Subtype

"Classification level"



The Orphanet Nomenclature is comprehensive, standardized, evidence-based, interoperable, versioned, computable and free (CC-BY 4.0). It includes more than 6500 rare diseases, as of the July 2025 release.



Rare diseases

Search

[Clinical Signs and Symptoms](#)

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Search for a rare disease

AHUS

Search

(*) mandatory field

- ☒ Disease name
- ☐ OMIM disease
- ☐ Gene name or symbol
- ☐ ORPHAcode
- ☐ ICD-10
- ☐ ICD-11

Other search option(s) ▼

Atypical hemolytic uremic syndrome

[Suggest an update](#)

Disease definition

A rare, genetic thrombotic microangiopathy due to dysregulation of the alternative complement pathway and characterized by the triad of hemolytic anemia, thrombocytopenia, and acute renal dysfunction.

ORPHA:2134

[Classification level: Disorder](#)



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REVIEW ARTICLE

Rare disease nomenclature and coding: challenges, systems, and policies in the global context

Ashray Vohora,^a Sylvie Maiella,^a  Caterina Lucano,^a Kurt Kirch,^b Carina Thomas,^b Stefanie Weber,^b and Ana Rath^a

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standard had limitations due to its potential inaccuracies and the lack of granularity in its code categories (14).

The 2024 New Zealand Rare Disorders Strategy (90) highlights the collection of information on RD as a priority and includes a pledge that diagnostic and clinical information in national health datasets will become more detailed and extensive over time. The plan, however, does not specify the coding systems intended to be utilised to achieve this objective.

At present, New Zealand, like Australia, utilises the ICD-10-AM for coding. In a 2023 insights report on Rare



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accurate and specific code	
ORPHAcodes	Reference: 100% coverage
ICD-10	6.4% exact matches
ICD-11	14.7% exact matches
SNOMED-CT	93.8% exact matches
Online Mendelian Inheritance in Man (OMIM) phenotypes	50.9% of Exact matches
Mondo Disease Ontology	88.8% exact matches
Unified Medical Language System (UMLS)	97.1% exact matches

Figures are recent as of July 2025. Also shown in the online form



SNOMED CT



Taxonomy

Search

Favorites

Refset

Search

Options

Search: Prefix any order

Status: Active and inactive concepts

Description type: All

Language Refsets

☒ Group by concept

Filter results by Language

english4

Filter results by Semantic Tag

☐ disorder4

Filter results by Module

SNOMED CT core4

Filter results by Refset

CTV3 to SNOMED CT simple map9

SNOMED CT to ICD-10 extended map4

Type at least 3 characters ✓ Example: shou fra

atypical hemolytic

4 matches found in 0.49 seconds.

Atypical hemolytic uremic syndrome	Atypical hemolytic uremic syndrome (disorder)
Atypical hemolytic uremic syndrome with anti-factor H antibodies	Atypical hemolytic uremic syndrome with anti-factor H antibodies (disorder)
Atypical hemolytic uremic syndrome with complement gene abnormality	Atypical hemolytic uremic syndrome with complement gene abnormality (disorder)
A rare, genetic, primary immunodeficiency disease characterized by increased susceptibility to recurrent, usually severe, infections (particularly by Neisseria meningitidis, Haemophilus influenzae and Streptococcus pneumoniae), typically manifesting as otitis, sinusitis, bronchitis, pneumonia, and/or meningitis. Autoimmune disease (e.g. systemic lupus erythematosus, glomerulonephritis) and atypical hemolytic uremic syndrome may be associated. Laboratory serum analysis reveals, in addition to diminished or undetectable complement factor I, variably decreased complement C3, complement factor B and complement factor H.	Immunodeficiency with factor I anomaly (disorder)

All results are displayed

Concept Details

Expression Constraint Queries

Concept Details

Summary

Details

Diagram

Expression

Refsets

Members

History

Parents

Atypical hemolytic uremic syndrome (disorder)

SCTID: 789660001

789660001 | Atypical hemolytic uremic syndrome (disorder) |

en Atypical hemolytic uremic syndrome (disorder)

en Atypical hemolytic uremic syndrome

en aHUS - atypical hemolytic uremic syndrome

en Atypical haemolytic uraemic syndrome

en aHUS - atypical haemolytic uraemic syndrome

Interprets → Renal function

Has interpretation → Impaired

Interprets → Platelet count

Has interpretation → Below reference range

Clinical course → Sudden onset

AND/OR short duration

Interprets → Measurement of total hemoglobin concentration

Has interpretation → Below reference range

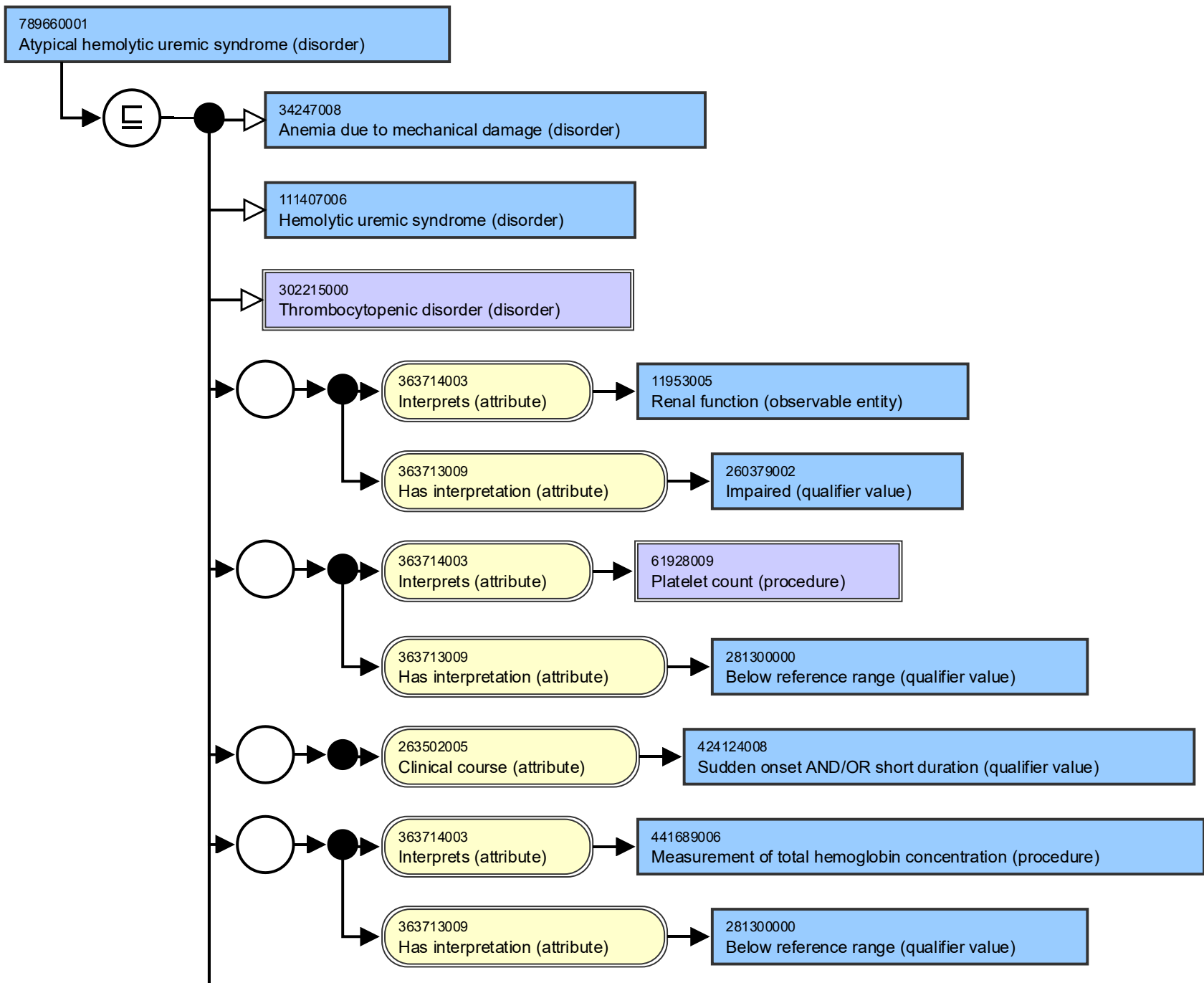
Finding site → Erythrocyte

Interprets → Hemolysis

Has interpretation → Present

Interprets → Red blood cell count

Has interpretation → Below reference range



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< **Health professionals**

Guidance and standards

• **By topic**

• Antenatal and newborn screening

• Antimicrobial resistance (AMR)

• Border health

• Bowel screening

• Breast screening

• Cervical screening

FOR HEALTH PROFESSIONALS

SNOMED CT New Zealand edition

We work with health agencies and the health software industry to publish a standard set of SNOMED CT content for digital health solutions in Aotearoa New Zealand.

On this page

[About the New Zealand edition](#)

[April 2026 release](#)

About the New Zealand edition

The SNOMED CT NZ Edition is a comprehensive source of concepts and terms for recording health information at point of care. All local content is governed by recognised national clinical networks.

The SNOMED CT New Zealand edition is released 3 times each year — in April, July and October.



No need to use ORPHAcodes directly



Instead use the SNOMED content that mirrors the ORPHAcodes and is supported by all our digital public infrastructure



ICD-11

And map SNOMED CT to ICD for national collections and reporting

Rare disorders count!

CAN YOU RECORD A RARE DISORDER WITHIN YOUR PATIENT MANAGEMENT SYSTEM?

With the collaboration between **SNOMED International** and **INSERM**, it is now possible to easily code rare disorders, **ensuring visibility for those who have been diagnosed with a rare disorder**, or have a suspected rare disorder.

SNOMED CT
The global language of healthcare

Inserm

SNOMED is a global language for data-driven healthcare that promises to improve outcomes for all New Zealanders, including people with rare disorders. SNOMED is used with other standards such as the International Patient Summary to enable the exchange of critical health information across boundaries near and far.

Help move those with rare disorders towards a brighter future and **advocate for getting SNOMED into your patient management system!**

Contact snomed@tewhatuora.govt.nz and we'd love to help you get started on your next step



Health New Zealand
Te Whatu Ora

Ngā mihi nui