Leave no child behind

Liesbeth Siderius

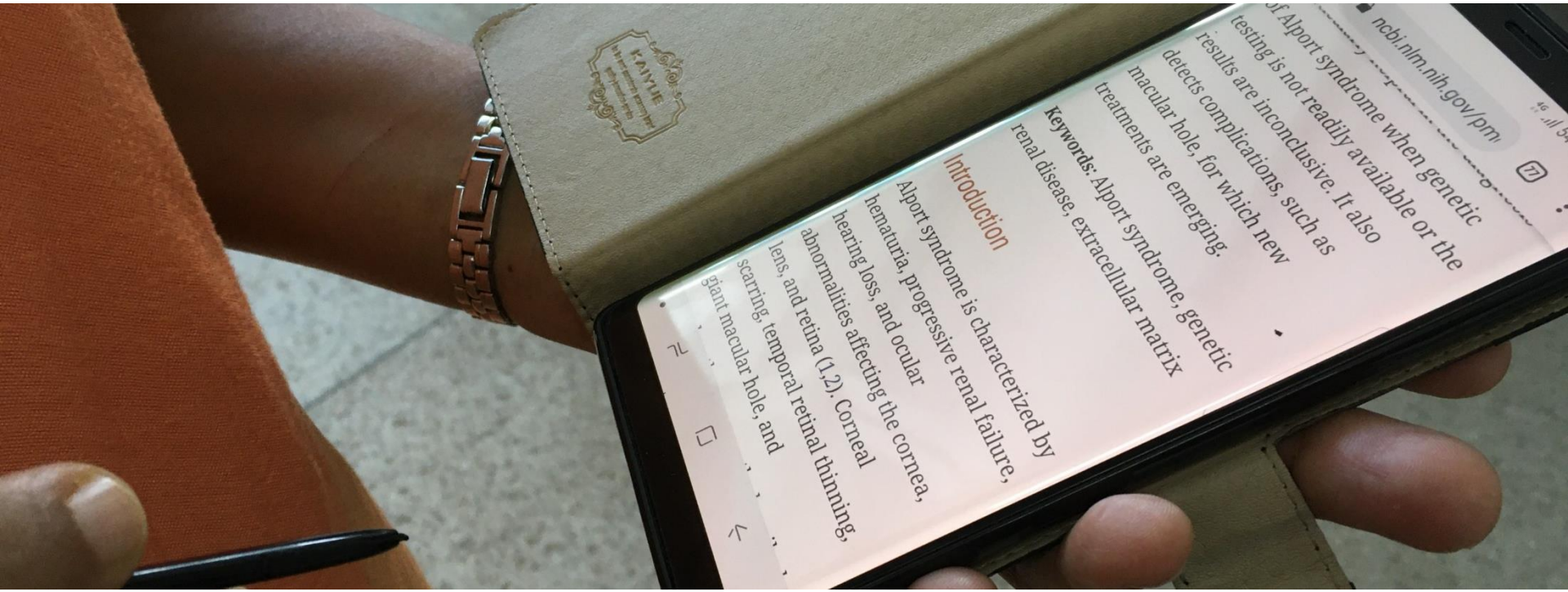
Rare Care Word & Shwachman diamond Syndrome Support Holland

UNICEF, January 2022
Millions of children with disabilities around the globe continue to be left behind,

despite

- **Convention on the Rights** of the Child, embedded in the
- **Convention on the Rights of Persons with Disabilities** and a mandate set by the
- **Sustainable Development Goals**.

Often this neglect is the result of limited data



Universal Health Coverage

What can we do, together?

From first feature to rare disease co-management with LOINC

Universal Health Coverage

23/09/2019

MOVING TOGETHER
TO BUILD A HEALTHIER WORLD

UN High-Level Meeting on Universal Health Coverage, 23 September 2019, New York

Patient Information	Primary Care	Diagnosis Collaborative care	Social Services
www.shwachman.nl https://rarecare.world	Growth retardation Recurrent infections (LOINC)	Guideline SDS (Orphanetcode; SNOMED, ATC e.a.)	Recurrent illness Fatigue, Short (ICF-CY; ISO 9999)

Stichting Shwachman syndroom Support Holland

New Diagnostics

Diagnosis

Hurler syndrome

PKU, Duchenne MD, FOP

Shwachman Diamond Syndrome

ICD - 10

Orphacode

OMIM

SNOMED -CT

DCOM

HPO

LOINC

ICPC

Sign primary care

Heelstick screening

Hearing screening

Growth; Development

Guideline

Collaborative Health Care

Interoperable data model

ISO 3166-1

HL7/ FHIR

ATC

New Therapeutics

ICF

ISO9999

Registry

Data collection with systematically organised computer processable collection medial terms

Guideline

Social services and rehabilitation

1. Recognize

- **Preventive Child Health**
  LOINC, HPO, ICPC

2. Multidisciplinary care

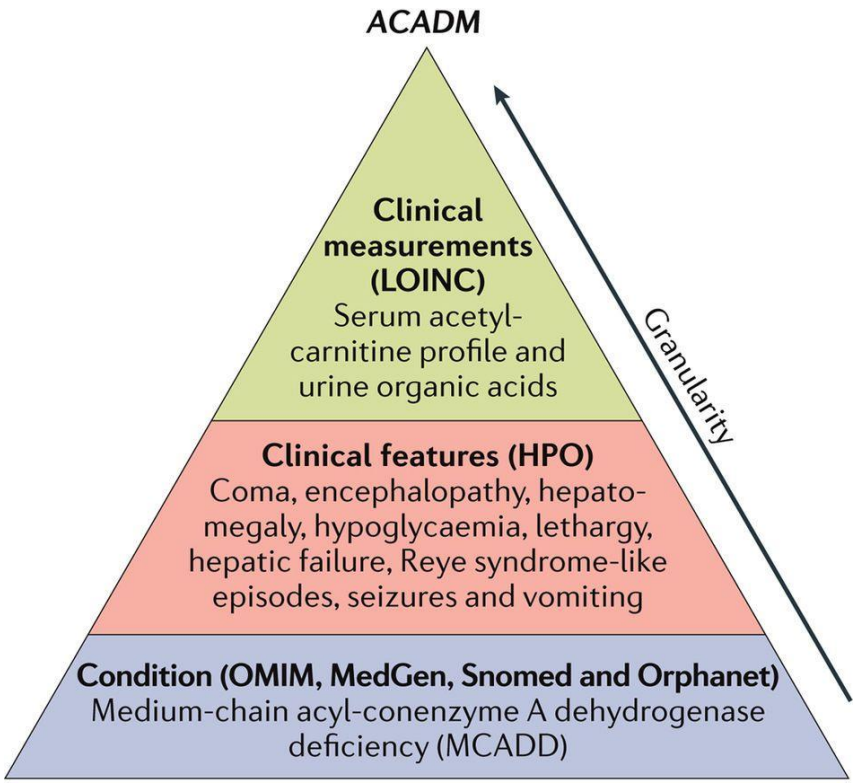
- **Digital medical guidelines**

ICD, LOINC, ATC, Snomed, ORPHA, OMIM

3. Social support

- **Community**

International Classificatie Function (ICF) for everyone



Nature Reviews | Genetics

New variants found in Mendelian disease, what next?
Review #bioinformatics scoring to prioritise 2017
https://www.nature.com/nrg/articles

One code = One meaning

LOINC Standard for identifying health measurements, observations, and documents

HPO: Human Phenotype Ontology

ICPC: International Classification of Primary Care

ICD: International Classification of Diseases

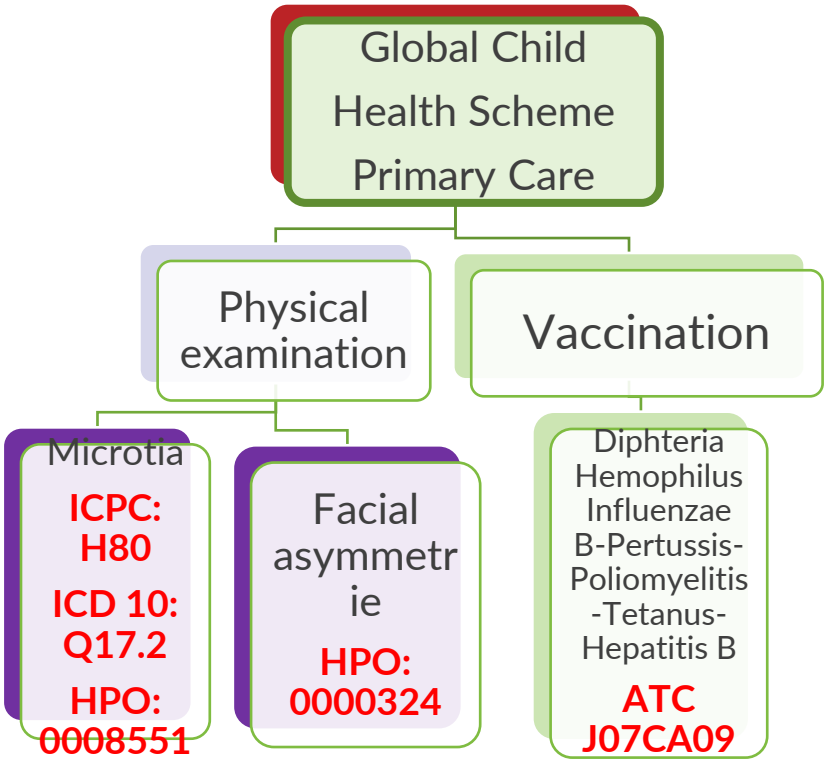
ATC: Anatomical Therapeutic Chemical Classification System

ORPHA: Classification of rare diseases

OMIM: Catalog of Human Genes and Genetic Disorders

Use of terminologies enables semantic interoperability between systems using HL7 CDA and FHIR

International classifications
the tool for interoperability in child health



Oculo-Auriculo-Vertebral Spectrum/Goldenhar Syndrome
ORPHA:141132 Oculo-auriculo-vertebral spectrum
OMIM # 164210 HEMIFACIAL MICROSOMIA; HFM

International Conference on Birth Defects and Disabilities in the Developing World
23 - 26 Feb 2020, Colombo, Srilanka

LOINC
RESULT in VALUE and UNITS

LocalCode^LocalName^CodeSystem | LOINCcode^LOINCname^CodeSystem

OBX|2|NM|123^WBC^HSP_A^26464-8^Leukocytes [#/volume] in Blood^LN||10.8|K/MM3|||||F|
OBX|3|NM|234^RBC^HSP_A^26453-1^Erythrocytes [#/volume] in Blood^LN||4.82|MIL/MM3|||||F|
OBX|4|NM|345^HGB^HSP_A^718-7^Hemoglobin [Mass/volume] in Blood^LN||15.2|GM/DL|||||F|
OBX|5|NM|456^HCT^HSP_A^20570-8^Hematocrit [Volume Fraction] of Blood^LN||45|%|||||||F|

Notice how the result value and units have their own places in the message

Data type of result (OBX-5) is a coded element

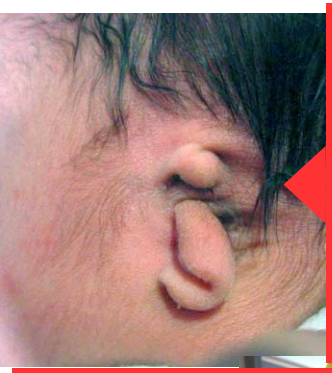
This code is from LOINC

This code is from SNOMED

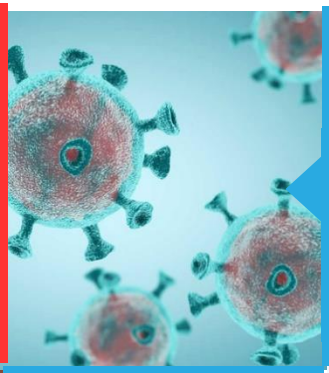
OBX||CE|57131-5^Newborn conditions with pos markers^LN|7573000^PKU^SCT

Code identifying this observation (what are these results? Conditions identified by newborn screening)

Code identifying the result (Phenylketonuria)

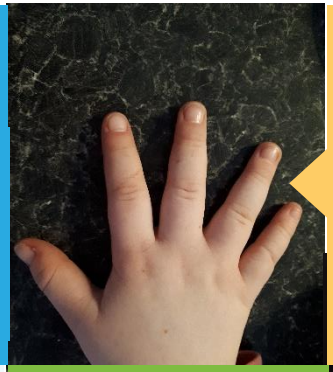


Goldenhar s
Abnormal ear
.
HP:0008551



Immunodeficiency

LOINC
94500-6
SARS

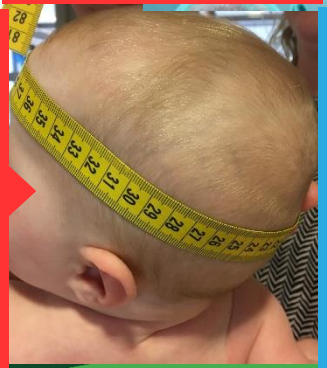


Coffin Lofry s
Tapered fingers

HP:0001182

Measuring Head Circumference

LOINC
8287-5



Neutropenia

LOINC
751-8
Neutrophils

Shwachman DS

ATC

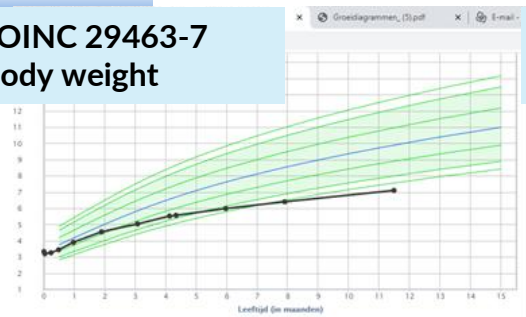
. A09AA02
Pancreatine

Achondroplasia

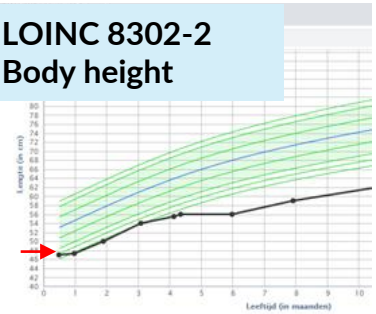
- Bijzonderheden groei: 234 0..1 (W0082, AN, Alfanumeriek 4000)
- Lengte: 235 0..1 (W0252, PQ, Lengte in millimeters)
- Methode lengtemeting: 236 0..1 (W0253, KL_AN, Methode lengtemeting)
- Groeicurve lengte naar leeftijd: 237 0..1 (W0167, BER, Berekend veld)
- Target height: 809 0..1 (W0167, BER, Berekend veld)
- Target Height Range: 810 0..1 (W0167, BER, Berekend veld)
- Gewicht: 245 0..1 (W0260, PQ, Gewicht in grammen)
- Methode gewichtsmeting: 246 0..1 (W0261, KL_AN, Methode gewichtsmeting)
- ...ht/lengte)
- BMI-curve: 813 0..1 (W0167, BER, Berekend veld)
- Gewichtsklasse op basis van BMI: 1492 0..1 (W0668, KL_AN, Gewichtsklasse op basis van BMI)
- Middelomtrek in millimeters: 1485 0..1 (W0252, PQ, Lengte in millimeters)
- Hoofdomtrek: 252 0..1 (W0267, PQ, Hoofdomtrek in millimeters)
- Groeicurve hoofdomtrek naar leeftijd: 253 0..1 (W0167, BER, Berekend veld)

ACHONDROPLASIA
OMIM #100800 Orpha:15

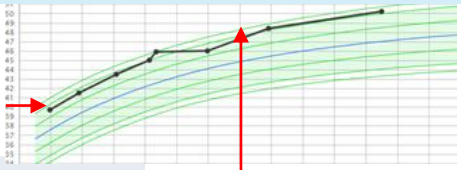
LOINC 29463-7
Body weight

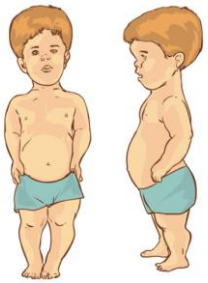


LOINC 8302-2
Body height



LOINC 8287-5 Head Occipital-frontal circumference by Tape measure





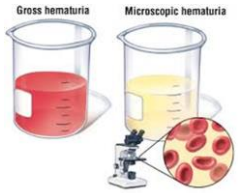
Hydrocephalus Risk

New Treatment

VOXZOGO™
(vosoritide) for injection

Achondroplasia-growth curve at each primary care visit

Alport syndrome



33051-4 Erythrocytes [Presence] in Urine

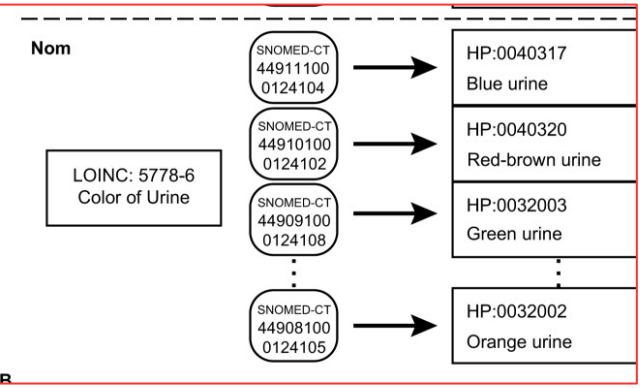
2888-6 Protein [Mass/volume] in Urine

ICD-10-CM Diagnosis Code N04.9 Steroid-resistant nephrotic syndrome

53853-8 COL4A5 gene targeted mutation analysis

 # 301050
ALPORT SYNDROME 1, X-LINKED
ORPHA:63 Alport syndrome

98422-9 Hearing loss degree Ear - left



Zhang XA, Yates A, Vasilevsky N, et al. Semantic integration of clinical laboratory tests from electronic health records for deep phenotyping and biomarker discovery. *NPJ Digit Med*. 2019;2:32. doi:10.1038/s41746-019-0110-4

Fish-eye disease

is extremely rare

Partial deficiency of lecithin–cholesterol acyl-transferase (LCAT)

very high serum free cholesterol

corneal opacities, beginning in adolescence or early adulthood

Haemolytic anaemia and renal involvement

persistent proteinuria (LOINC 2888-6 Protein [Mass/volume] in Urine)

steroid resistant nephrotic syndrome ICD-10-CM N04.9.)

high total cholesterol (491 mg/dl) (LOINC 2093-3 Cholesterol [Mass/volume] in Serum or Plasma)

high LDL (331 mg/dl) (LOINC 2089-1 Cholesterol in LDL [Mass/volume] in Serum or Plasma).

136120 FISH-EYE DISEASE; FED

Genetic studies revealed a mutation in the lecithin: cholesterol acyl-transferase gene

Duchenne Muscular Dystrophy

Muscular power function
ICF b 730

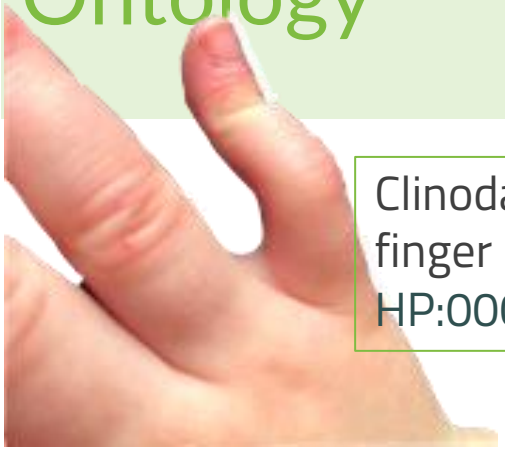
49551-5
Creatine kinase.MB [Mass/volume] in Blood

22075-6 DMD gene mutations tested for in Blood or Tissue by Molecular genetics method Nominal

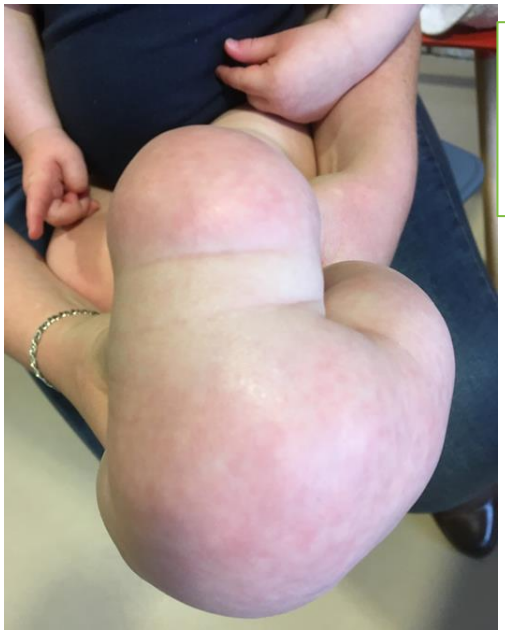
OMIM # 310200
MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD
ORPHA:98896 Severe dystrophinopathy, Duchenne type

Tests for **creatine kinase**.
Children with DMD always have a very high level of creatine kinase (about **10-100 times normal**).
Creatine kinase level is normal, then DMD is ruled out
Creatine kinase level is high, further tests are needed to see whether this is due to DMD or to some other condition.

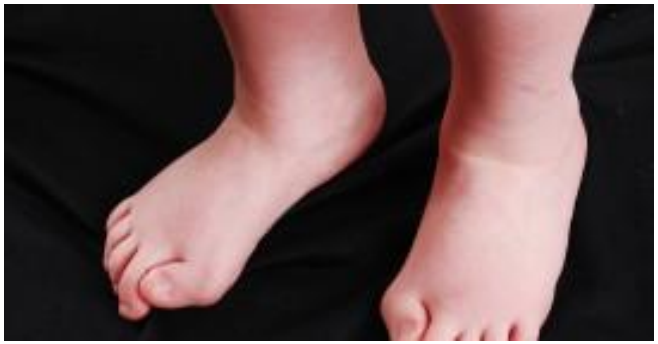
HPO: Human Phenotype Ontology



Clinodactyly of the 5th finger
HP:0004209



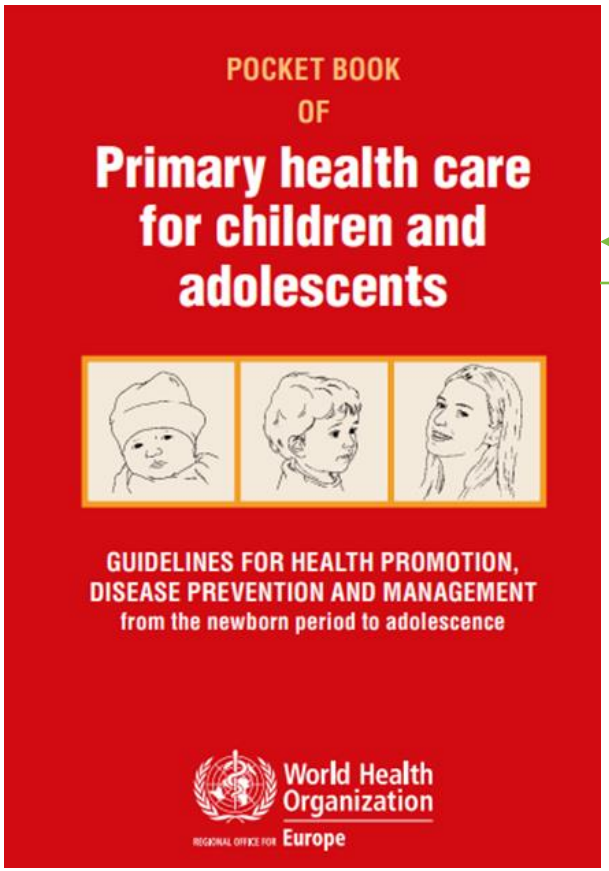
Foot oligodactyly
HP:0001849



Short hallux HP:0010109

Hemihypertrophy HP: 0001528

Primary health care, the pillar of universal health coverage



LOINC

LOINC CODE
39294-4

LONG COMMON NAME
Children's preventive health services attachment Set

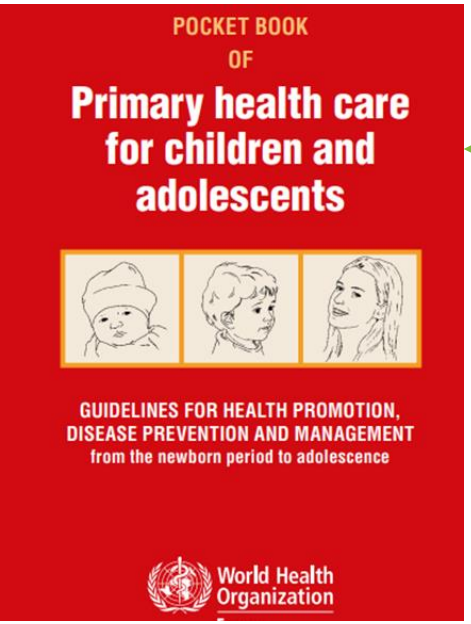
Panel Hierarchy

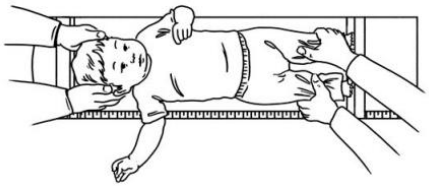
Details for each LOINC in Panel

LOINC	Name	R/O/C	Cardina
39294-4	Children's preventive health services attachment Set		
39157-3	Screen type indicator CPHS	1..1	
39158-1	Screening on schedule to patient age CPHS	1..1	
39159-9	Screening extent CPHS	0..1	
39160-7	Visit was for recheck CPHS	0..1	
39255-5	Date previous screen visit CPHS	0..1	
39161-5	Date next screen visit CPHS	0..1	
39155-7	Family history or condition or disease and action Family CPHS	1..1	
39162-3	Chronic illness indicator CPHS	0..1	

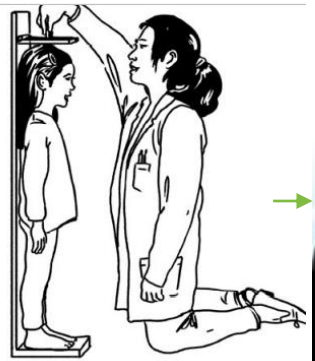
Universal Health Coverage, leave no child behind

World Health Organization

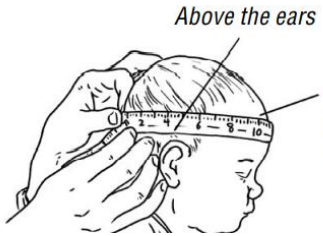




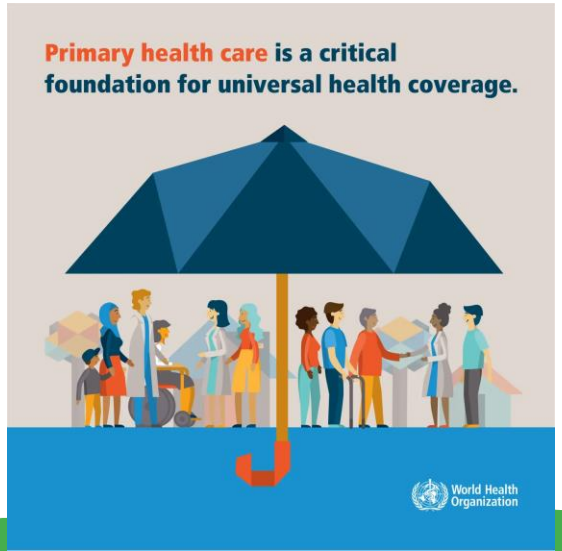
Length measurement from birth to 2 years of age



Height measurement in children from 2 years of age

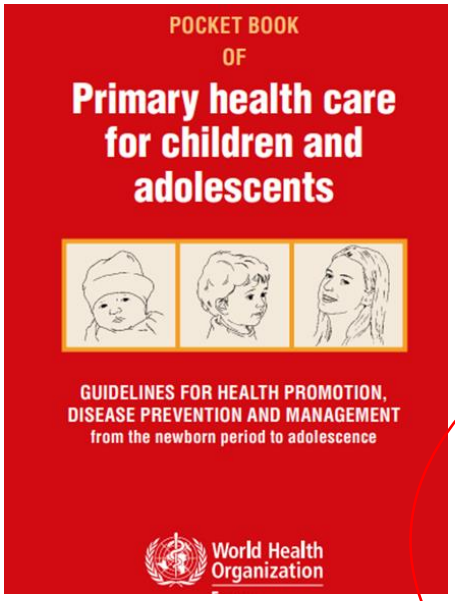


Broadest part of the forehead, midway between the eyebrows and hairline



https://www.who.int/europe/publications/i/item/9789289057622

World Health Organization



The health information system ensures the collection, analysis and use of data to ensure early, appropriate action **to improve the care of every child**

3.2 Well-child visit: birth – 72 hours

Most children will be seen in hospital for these visits; if not, they ought to be seen by the primary care provider within 24 hours of birth and again at 48–72 hours.

- Look for congenital diseases and jaundice
- Support caregivers.

History

- Problems during pregnancy, e.g. diabetes, medications, substance abuse, acute or chronic infections, mental or social stress, abnormal test results, e.g. positive group B Streptococcus, HIV, hepatitis B
- Mode of delivery and problems during or after birth
- Congenital disorders in the family, e.g. hip problems
- Hip dysplasia risk factors, e.g. twin pregnancy, breech position
- Problems passing meconium and urine

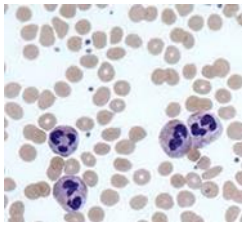
From Feature to Medical Guideline

Feature

- Fatty Stool
- Growth Retardation
- Common infections

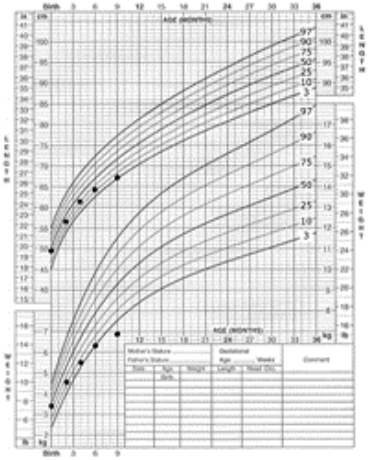
Shwachman Diamond Syndrome- Management

- Pancreas insufficiency
- Neutropenia
- Skeletal Dysplasia
- Autisme like



Failure to thrive
- HP:0001531
- LOINC:42819-3 Failure to thrive [CCC]
- ICPC: T10

Fatty Stool
- LOINC 16142-2Fat [Mass/time] in 24 hour Stool



Pancreas insufficientie
- ICD -10 K86.81

Ann. N.Y. Acad. Sci. ISSN 0077-8923

ANNALS OF THE NEW YORK ACADEMY OF SCIENCES
Issue: Annals *Meeting Reports*

Draft consensus guidelines for diagnosis and treatment of Shwachman-Diamond syndrome

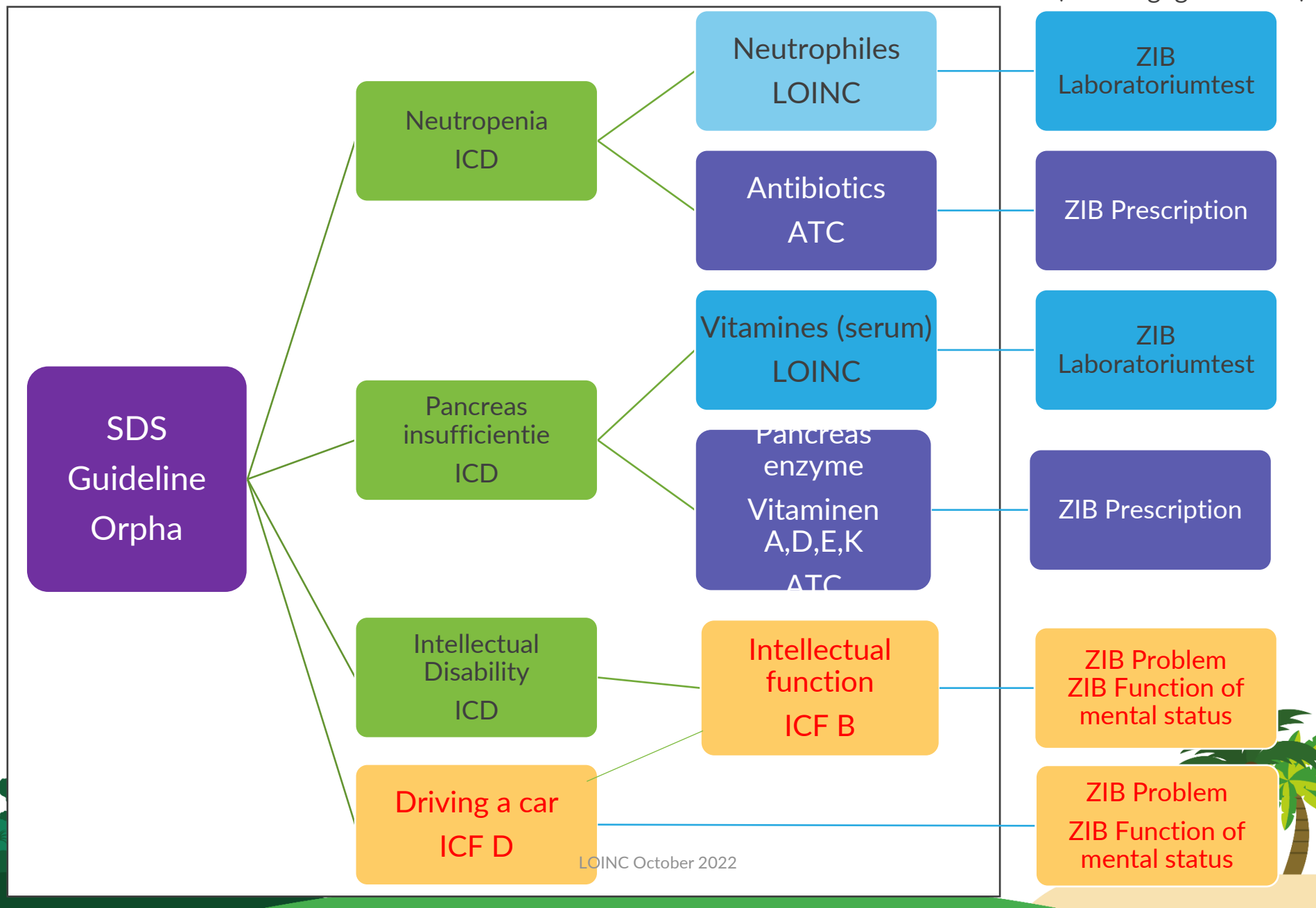
Yigal Dror,[1] Jean Donadieu,[2] Jutta Koglmeier,[3] John Dodge,[4] Sanna Toiviainen-Salo,[5] Outi Makitie,[5] Elizabeth Kerr,[1] Cornelia Zeidler,[6] Akiko Shimamura,[7] Neil Shah,[3] Marco Cipolli,[8] Taco Kuijpers,[9] Peter Durie,[1] Johanna Rommens,[1] Liesbeth Siderius,[10] and Johnson M. Liu[11]

Shwachman Diamond S
ORPHA:811
OMIM# 260400

Cystic Fybrosis
ORPHA:586
OMIM # 21970

Rare Care World

MedMij ZIB's
(binnen gegevenssets)



LOINC October 2022

FHIR Profile chronic condition

Home Artifacts

Table of Contents > **Home**

RarecareFHIRIG - Local Development build (v0.1.0). See the Directory of published versions

1 Home

This repository contains the FHIR resources for the "Een PGO voor iedereen" ("A personal healthcare environment for everyone") project.

Note: *All example content is example only!* It is based on Shwachman Diamond Syndrome (SDS) data from rarecare.world, but for brevity it is much shorter than the actual data would be.

1.1 MedMij and the "PGO for everyone" project

The Netherlands has a national effort, MedMij, to provide all Dutch citizens with a personal healthcare environment, web or mobile. However, persons with rare diseases have trouble seeing their condition properly represented. Due to the rare nature of their condition, vendors are hesitant to invest in small populations. Having a machine-readable Rare Condition profile would enable vendors to simply read the necessary data to provide those persons with customized dashboards, graphs and questionnaires to address their conditions properly. Moreover, healthcare professionals, patient organizations and researchers could all benefit from the structured collection of data.

The "PGO for everyone" project aims to provide such a machine-readable API for PGO's. The definitions for specific rare conditions are published as FHIR resources. PGO's can pull those in with an API, and use the definitions to provide disease-specific dashboards and questionnaires for those rare conditions.

1.2 The RareCare Data Model

The Rare Care models are maintained at https://rarecare.world

and (only partially complete yet) https://decor.nictiz.nl/art-decor/decor-datasets-zaz-

From those resources FHIR profiles are generated. The basis is a Data Model of Rare Conditions:

Building the Rare Disease

knowledge and information ecosystem

- B87 Splenomegaly (1)

LOINC

- 718-7 Hemoglobin [Mass/volume] in Blood (4)
- 24325-3 Hepatic function 2000 panel - Serum or Plasma (2)
- 2243-4 Estradiol (E2) [Mass/volume] in Serum or Plasma (1)
- 2276-4 Ferritin [Mass/volume] in Serum or Plasma (1)
- 10501-5 Lutropin [Units/volume] in Serum or Plasma (1)

Show more

OMIM

- # 613985 BETA-THALASSEMIA (1)

ORPHA

- ORPHA:231214 Beta-thalassemia major (1)

SNOMED

B78.01 Thalassemia

LOINC

46740-7 Hemoglobin disorders newborn screen interpretation

Thalassemia

ORPHA231214

Splenomegaly in thalassemia

Symptom

... Splenomegaly in **thalassemia** Splenomegaly (enlarged spleen) is common in **thalassemia** major and in of ... by an enlarged liver. B87 Splenomegaly Large spleen **Thalassemia** major or Beta **Thalassemia** ...

Rare Condition

Thalassemia major or Beta Thalassemia

Feature

Large spleen

ICPC

B87 Splenomegaly

Carrier screening programs

Feature

... member Examples of conditions screened in populations: **Thalassemia** , an autosomal recessive condition. When both ... measures, such as diet and medications Carrier screening **thalassemia** **Thalassemia** major or Beta **Thalassemia** ...

Rare Condition

Thalassemia major or Beta Thalassemia

Symptom

Carrier screening thalassemia

Carrier screening thalassemia

Pretty Raw Preview Visualize JSON


```
{
  "id": {
    "S": "231214"
  },
  "value": {
    "S": "{\"resourceType\":\"PlanDefinition\",\"id\":\"plan-thalassemia-major-or-beta-thalassemia\",\"meta\":
      {\"profile\":[\"http://rarecare.world/fhir/StructureDefinition/rare-care-plan\"]},\"text\":
      {\"status\":\"generated\",\"div\":\"<div xmlns=\\\"http://www.w3.org/1999/xhtml\\\"><p><b>Generated
      Narrative</b></p><p><b>url</b>: <code>https://rarecare.world/fhir/PlanDefinition/
```


API endpoint ORPHA231214

International Patient Summary

ISO 27269:2021
Health informatics — International patient summary

The International Patient Summary (IPS) is building the bridge between the "home" health and care environment of the patient and any other place where the patient needs to visit a clinical professional, whether within or across borders. The construction of the IPS involves a number of **standard** components and bespoke **specifications** to make it all work together.

IPS Datablocks for Rare Disease

(SK's suggestions, breadth)

Patient attributes	Allergies & intolerances	Problems incl. diagnosis	Medication summary	Immunization (incl. Vaccinations)	Results	Vital signs
Healthcare provider	History of procedures	History of past illness/ problems	History of Pregnancy	Medical Devices (incl. implants)	Functional status	Social history (incl. life style factors)
Address-book	Advance directives (i.e., living wills)	Care plan				
Provenance			Alerts (incl. Risks)	Child-health	Family history	Genetic details
Cross-border (conditional)				Recent Encounters	Computable Clinical Guidelines	Patient Story

From Presentation X-eHealth project
Stephen Kay, december 2021

DigitalHealthEurope recommendations on the European Health Data Space

3 may 2022

Better diagnosis and treatment, improved patient safety, continuity of care and improved healthcare efficiency

Empower **individuals** to have control over their health data

Enable **health professionals** to have access to relevant health data

Electronic health records

Health data from apps and medical devices

Health data in registries

Assist **policy makers and regulators** in accessing relevant non-identifiable health data

Facilitate access to non-identifiable health data for **researchers and innovators**

Better health policy, greater opportunities for research and innovation

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People with a rare condition and their families.

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e.siderius@kpnplanet.nl

Stichting Shwachman syndroom Support Holland