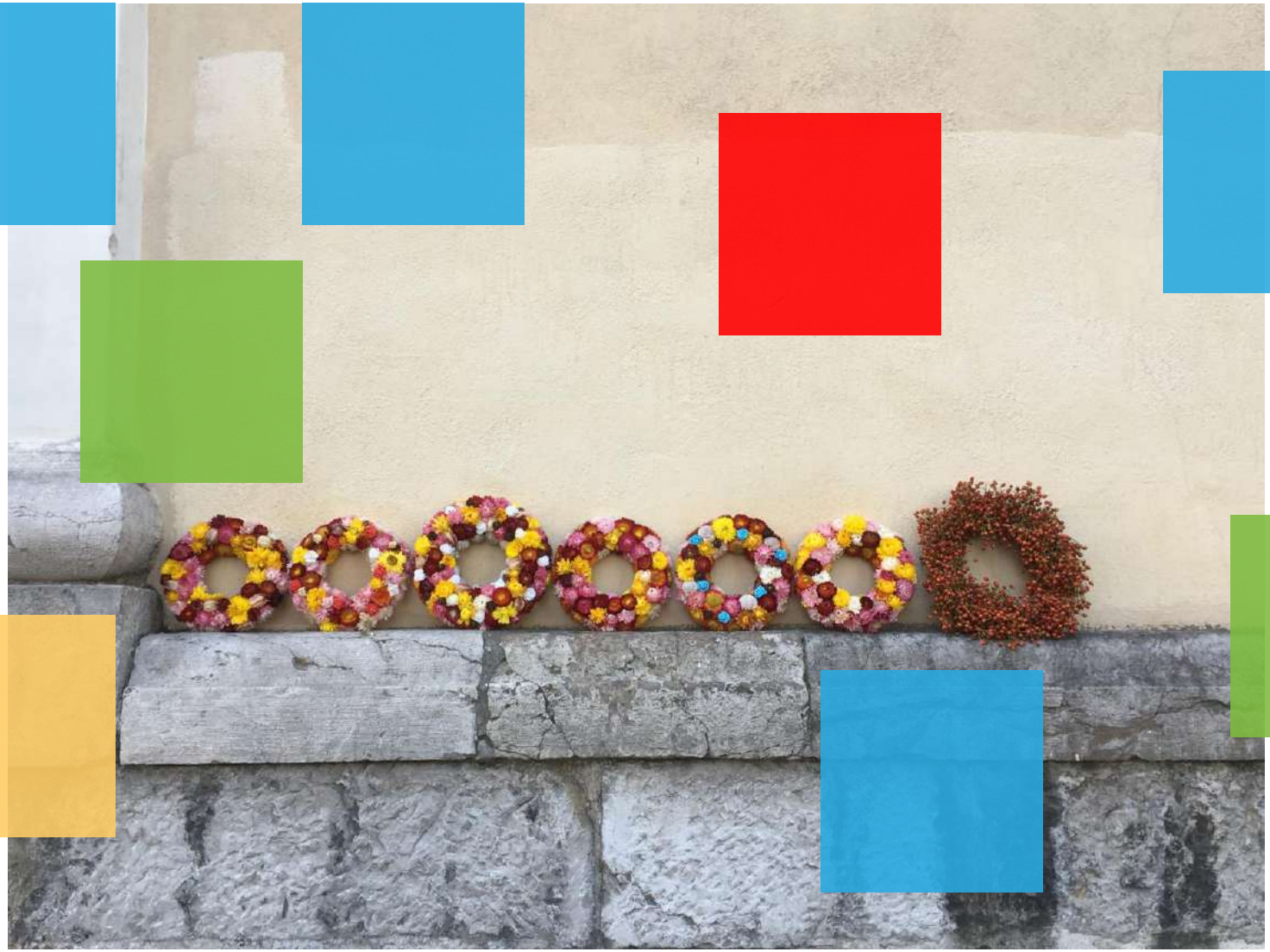


GLOBAL MOTHER – CHILD DATA SETS TO UNLOCK DIGITAL RECORDS

Louisa Kyerematenge , Youth Health Nurse , Almere, Netherlands

Liesbeth Siderius, Youth Health MD , Almere, Netherlands

Rob Braamburg , Rare Care Word, Netherlands

LEAVE NO CHILD BEHIND

Faculty Disclosure

x	No, nothing to disclose
	Yes, please specify:

	Honoraria/ Expenses	*Consulting/ Advisory Board*	*Funded Research*	*Royalties/ Patent*	*Stock Options*	*Ownership/ Equity Position*	*Employee*	*Other (please specify)*

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

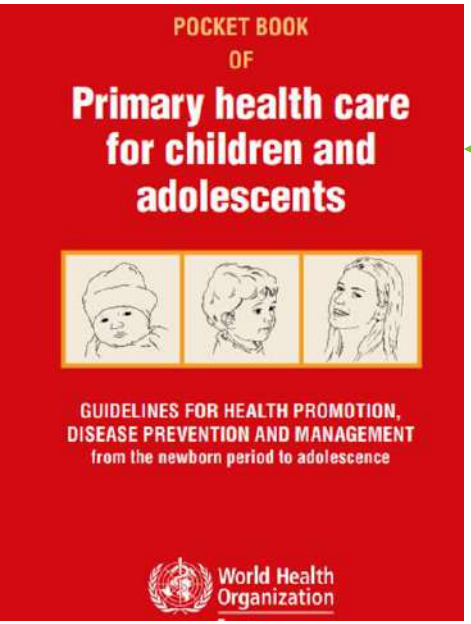
despite the near-universal ratification of the

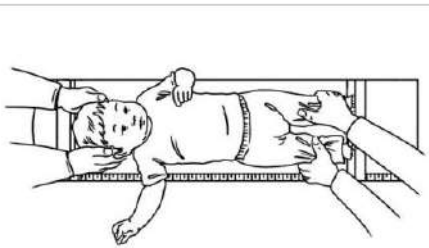
- **Convention on the Rights** of the Child, the call for action embedded in the
- **Convention on the Rights of Persons with Disabilities** and the clear mandate set by the
- **Sustainable Development Goals**.

Often, this neglect is the result of limited data

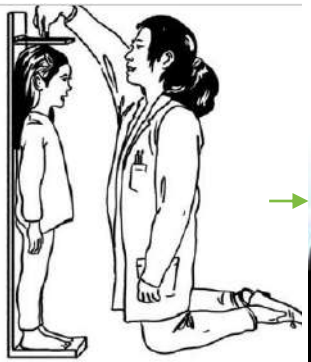
Universal Health Coverage, leave no child behind

World Health Organization

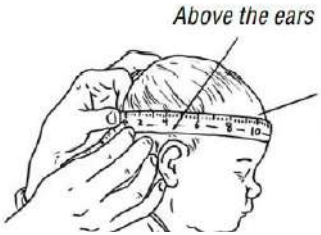




Length measurement from birth to 2 years of age

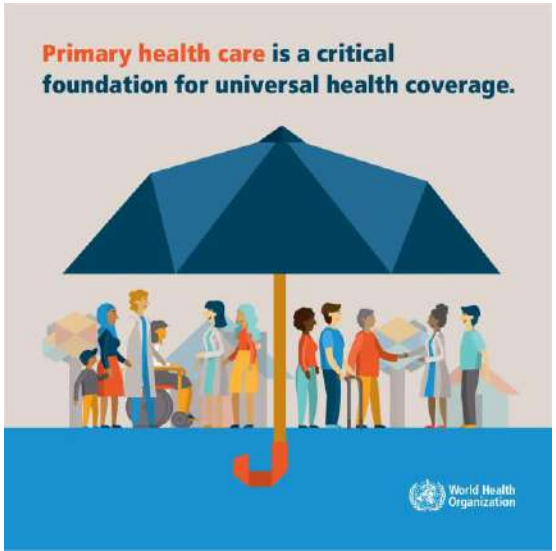


Height measurement in children from 2 years of age



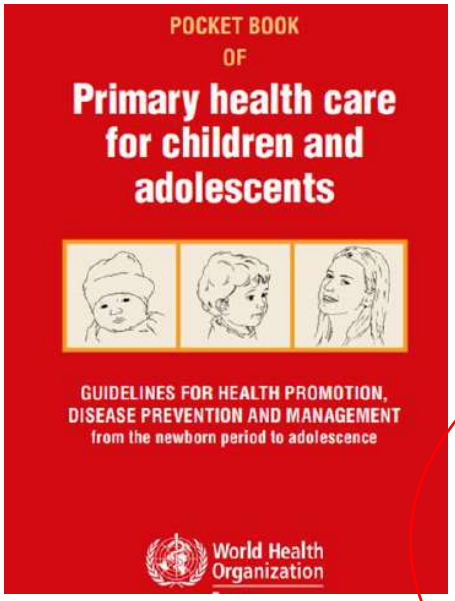
Above the ears

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

Patient Informatiom	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

HPO
LOINC
ICPC

Diagnosis
Hurler syndrome
PKU, Duchenne MD, FOP
Shwachman Diamond Syndrome

ICD - 10
Orphacode
OMIM

SNOMED -CT
DCOM

Sign primary care
Heelstick screening
Hearing screening
Growth; Development

Guideline
Collaborative Health Care

Paracetamol

Stofnaam	Paracetamol
Merknaam	Perfalgan, Apotel
ATC code	N02BE01

ATC
New Therapeutics

Real world data

ISO 3166-1

HL7/ FHIR

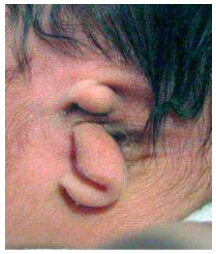
ICF
ISO9999

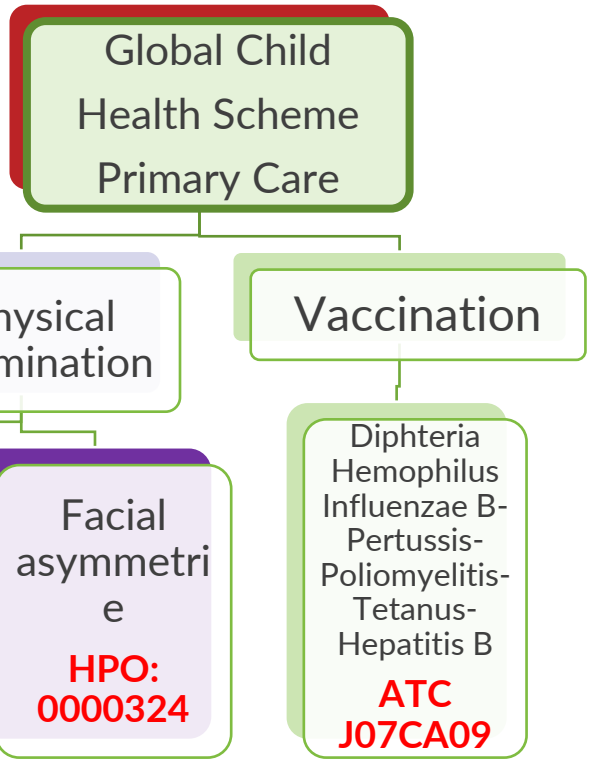
Registry
Data collection with systematically organised computer processable collection medial terms

Guideline
Social services and rehabilitation

International classifications as a tool for interoperability in child health

Towards a Global Integrated Digital Preventive Child Health Model





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

One code = One meaning

ICPC: International Classification of Primary Care

HPO: Human Phenotype Ontology

LOINC Standard for identifying health measurements, observations, and documents

ICD: International Classification of Diseases

ICF: International Classification of function

ATC: Anatomical Therapeutic Chemical Classification System

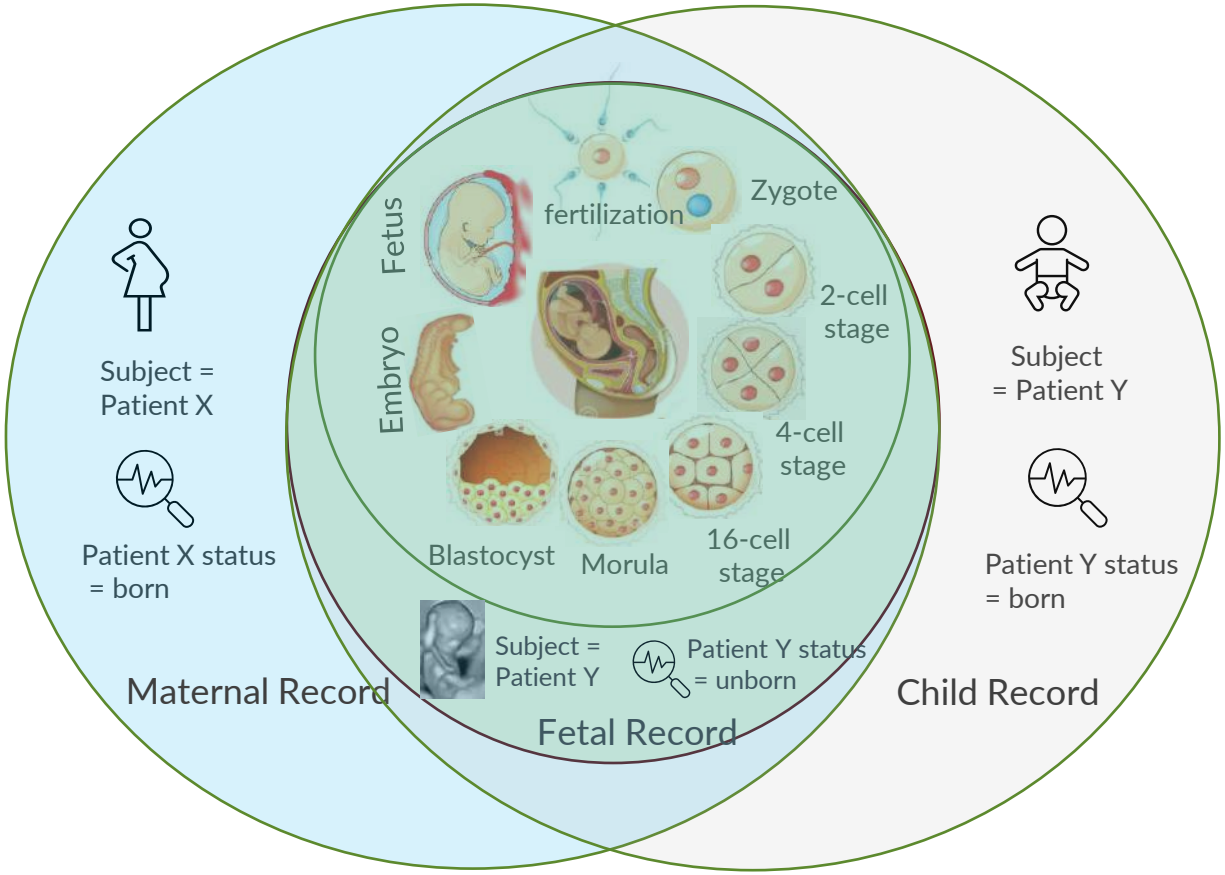
ORPHA: Classification of rare diseases

OMIM: Catalog of Human Genes and Genetic Disorders

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

Child Health & Obstetrics International Collaboration and Exploration

Conceptual diagram: Mother-Fetus-Child concepts, 2022



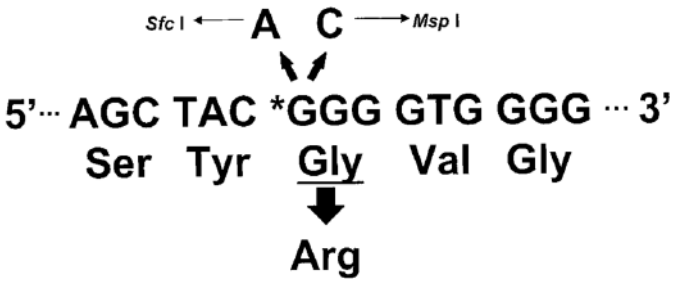
HL7 International

Key modelling question:
At what stage/when does a fetus stop being a body part of the pregnant woman and becomes an entity

Cases Achondroplasia *Primary Care 2021-2022*

4 weeks old girl presented: normal weight, reduced length , large head
What was observed during pregnancy and after birth?

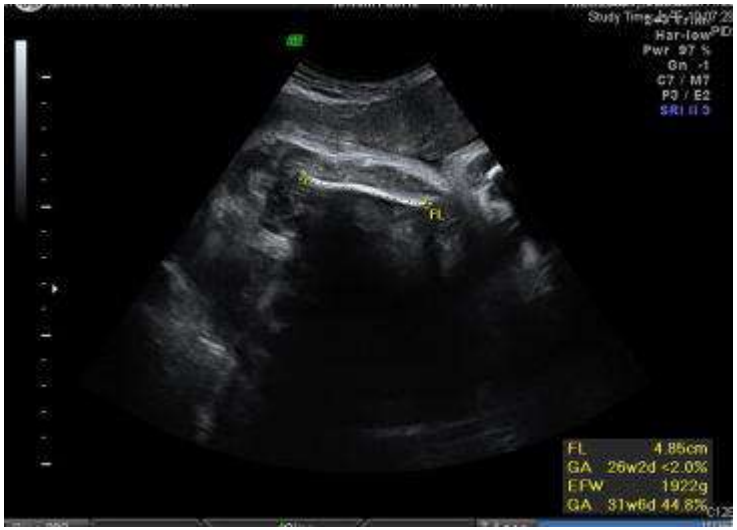
2,5 year old boy with achondroplasia: sign of autism
Does Autism occur more often in achondroplasia?

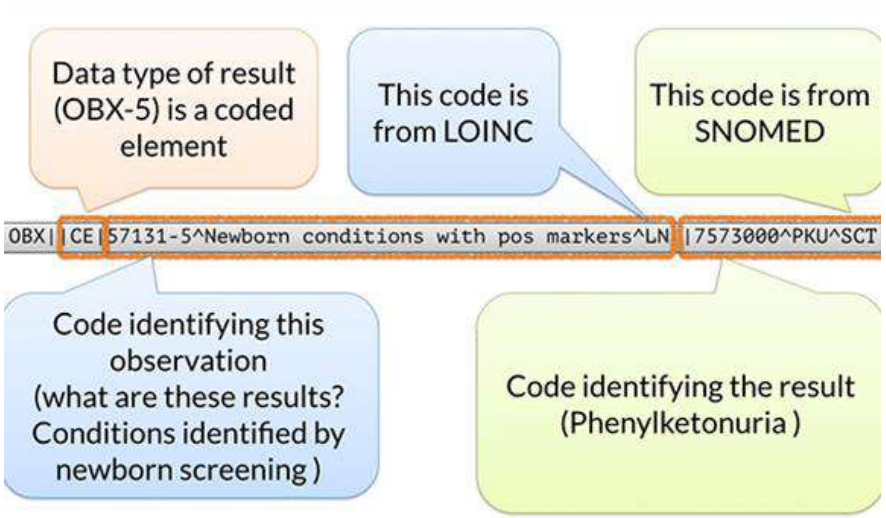


Common cause achondroplasia is
FGFR3 gene p.Gly380Arg mutation

Achondroplasia-growth curve at each primary care visit

Achondroplasia, prenatal ultrasound





SHORT FEMUR – Ultra sound
A 29-year-old female was transferred to our clinic because of short femurs (<2 percentile) on ultrasound scan.
https://doi.org/10.1016/j.jmu.2012.07.010.

LOINC 11963-6
Fetal Femur diaphysis [Length] US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LOINC License: https://loinc.org/license/\n\nThis CodeSystem resource descr
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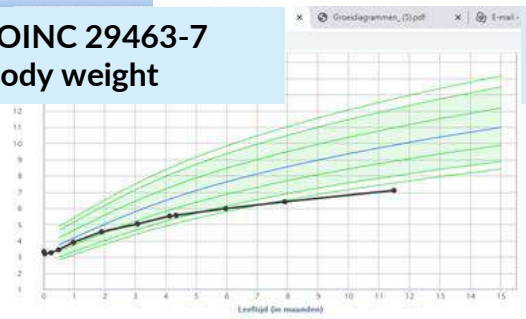
LOINC® from Regenstrief HL7® FHIR®

Achondroplasia

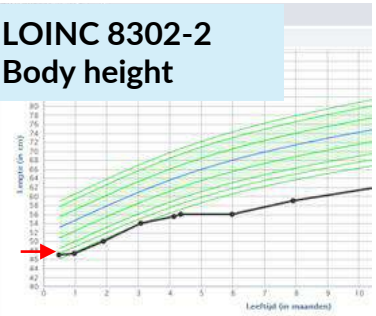
- Bijzonderheden groei: 234 0..1 (W0082, AN, Alfanumeriek 4000)
- Lengte: 235 0..1 (W0252, PQ, Lengte in millimeters)
- Methode lengtemeting: 236 0..1 (W0253, KL_AN, Methode lengtemeting)
- Groeicurve lengte naar leeftijd: 237 0..1 (W0167, BER, Berekend veld)
- Target height: 809 0..1 (W0167, BER, Berekend 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

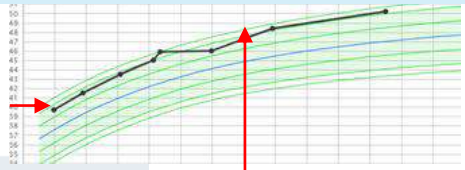
LOINC 29463-7
Body weight

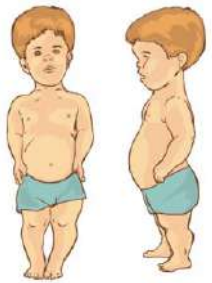


LOINC 8302-2
Body height



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





Hydrocephalus Risk

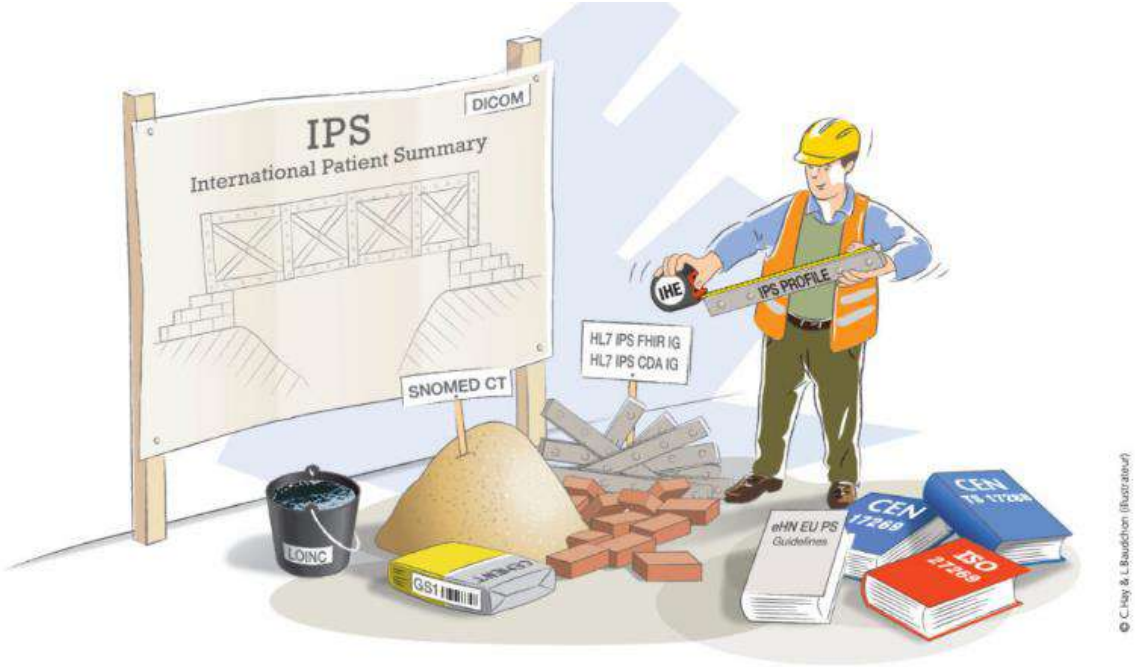
New Treatment

VOXZOGO™
(vosoritide) for injection

GLOBAL MOTHER – CHILD DATA SET
Achondroplasia

Case Achondroplasia			
age	feature	standard	Type of care
29 weeks Prenatal	Short Femur	LOINC 11963-6 Fetal Femur diaphysis [Length] US	Prenatal
4 weeks	Large head	LOINC 8287-5 Head Occipital-frontal circumference by Tape measure	Primary Care
4 weeks	Short	LOINC 3137-7 Body height Measured	Primary Care
6 weeks	Achondroplasia (clinical)	2022 ICD-10-CM Diagnosis	Primary Care
8 weeks	vaccination	ATC J07CA09	Primary Care
10 weeks	Achondroplasia (Genetic)	LOINC 21678-8FGFR3 gene p.Gly380Arg [Presence] in Blood or Tissue by Molecular genetics method	Multi-disciplinair
3 years	Communication problem Sign of Autism	ICF b 160 Thought functions	Primary Care

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 o **standard** components and bespoke **specifications** to make it all work together.

IPS Datablocks for Rare Disease

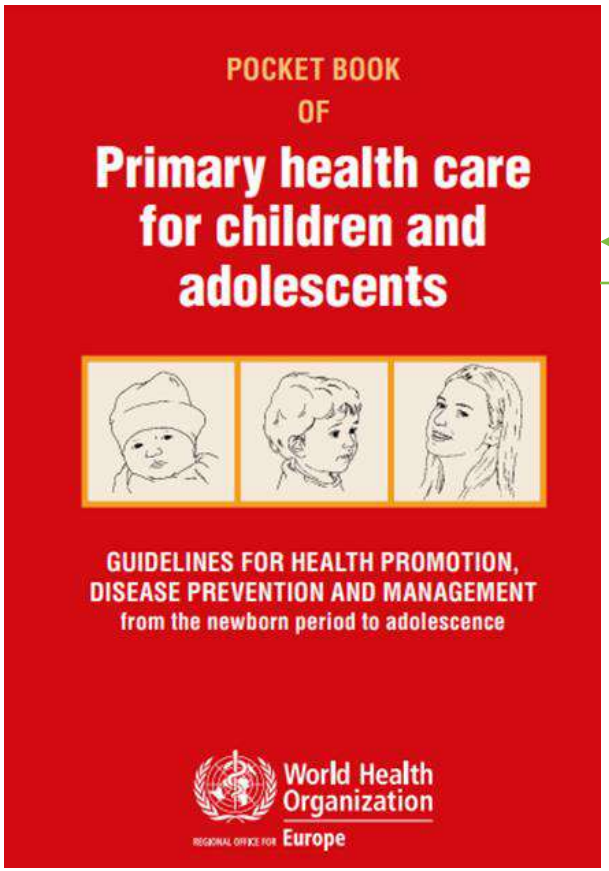
(SK's suggestions, breadth)

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

X-eHealth

Global Pediatric Digital Health



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

DigitalHealthEurope recommendations on the European Health Data Space

3 May 2022

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

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

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

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

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

Better health policy, greater opportunities for research and innovation

Acknowledgement:
European Paediatric Rare Disease Network
Consensus in Pediatrics and Child Health
Forum Rare Diseases, Sri Lankan Pediatric Society
Anjan Bhattacharya, ICF expert, India

Marc de Graauw, IT Expert, Netherlands
Martin Postma, IT Expert, Netherlands

Yvonne Heerkens, ICF Expert, Netherlands
Gonda Stallinga, ICF Expert, Netherlands

People with a rare condition and their families.

Siderius, L., Neubauer, D., Bhattacharya, A., Altorjai, P., Margvelashvili, L., Lamabadusuriya, S., Wierzba, J., Mazur, A., Albrecht, P., and Tasic, V. (2021). **Universal Health Coverage "Leave No Child Behind".** Pediatria Polska - Polish Journal of Paediatrics, 96(1), pp.1-6. https://doi.org/10.5114/polp.2021.104822

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Stichting Shwachman syndroom Support Holland

Health and Well-being for all