Step by step towards Global Digital Child Care

Liesbeth Siderius, Rare Care World, The Netherlands
Sahan Damsiri Perera, University of Colombo, Colombo, Sri Lanka

Digital Modelling of Primary Child Health

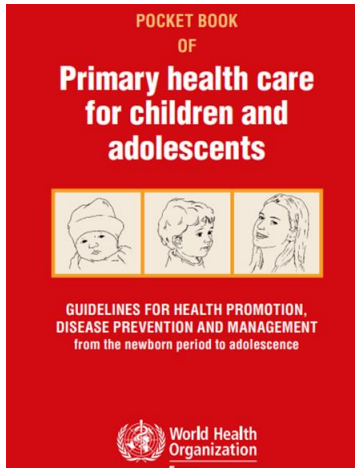
- Illustrative cases from The WHO Pocket Book on Primary health care for Children and Adolescents (WHO Europe, 2022)

- Proof of concept for a comprehensive implementation guide that harnesses the digital terminologies and HL7/FHIR standards

- Facilitating the seamless integration of WHO's quality healthcare standards into diverse primary care environments for children and adolescents.

Digital Modelling of Primary Child Health

World Health Organization



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

Introduction

Dissemination and generation of knowledge on complex diseases in children depends on the availability and interoperability of primary child health data.

Method

Identify structured set of interoperable international data terminologies such as the International Classification of Diseases (ICD) and International Classification of Function (ICF) as well as the numerical Logical Observation Identifiers Names and Codes (LOINC).

International terminologies as a tool for interoperability in child health

Towards a Global Integrated Digital Preventive Child Health Model

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

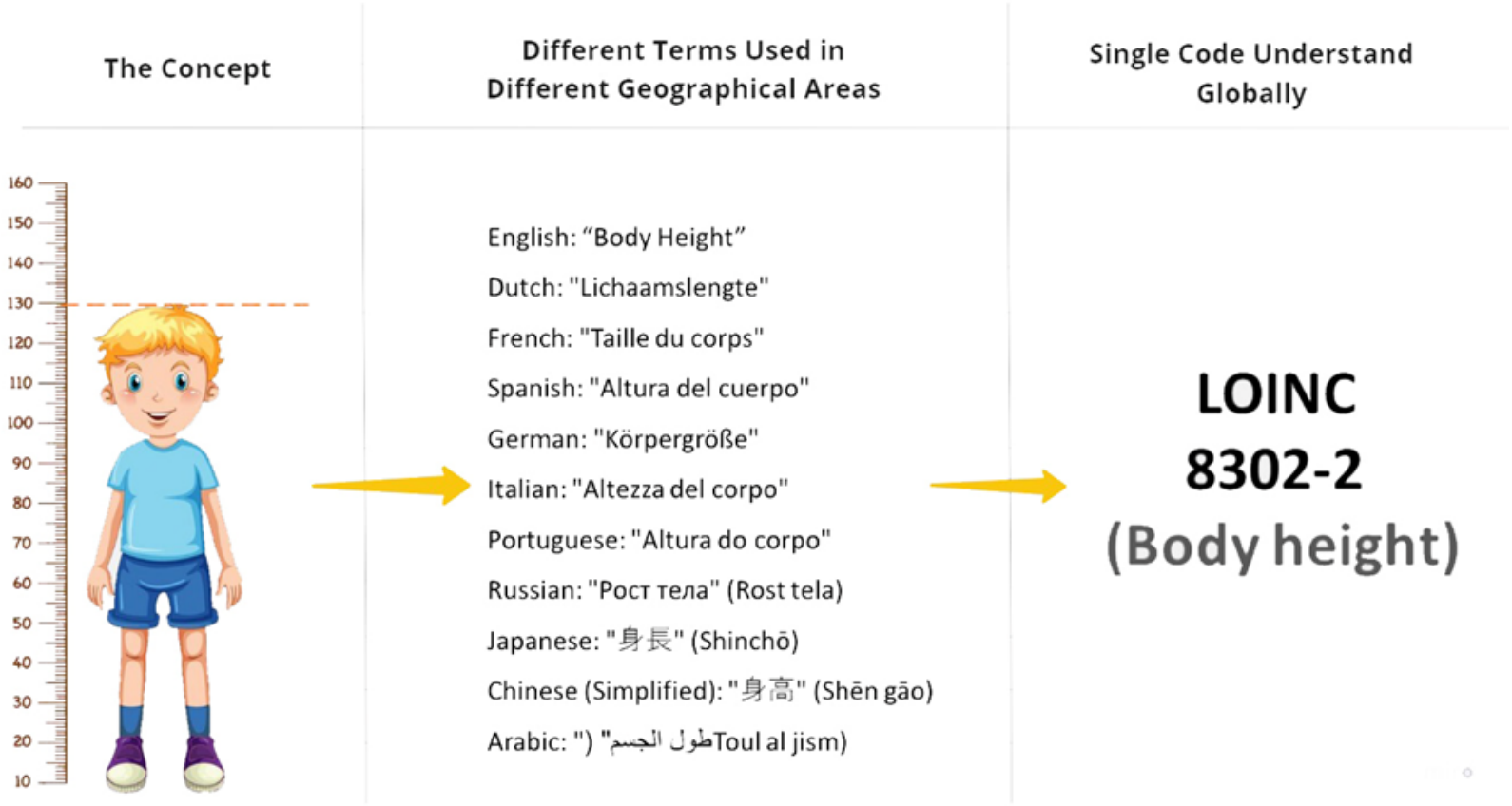
ATC: Anatomical Therapeutic Chemical Classification System

ORPHA: Classification of rare diseases

OMIM: Catalog of Human Genes and Genetic Disorders

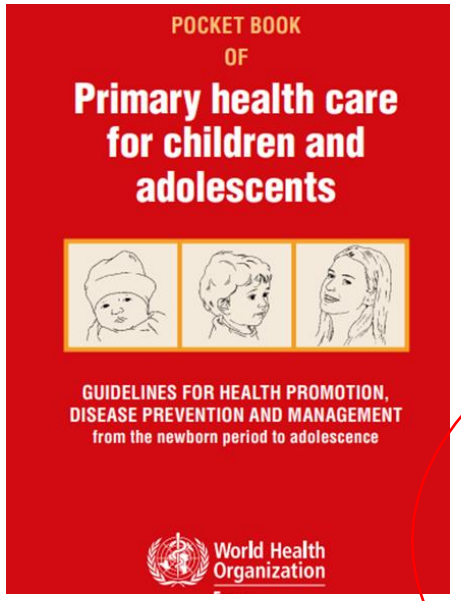
Use of terminologies enables semantic interoperability between systems using HL7 CDA and FHIR

Title "Digital child health: opportunities and obstacles", by Liesbeth Siderius*, Sahan Damsiri Perera, Lars Gelander, Lina Jankauskaite, Manuel Katz, Arunas Valiulis, Adamos A. Hadjipanayis, Laura Reali and Zachi Grossman, published in "Frontiers in Pediatrics-Children and Health".



Author Figure S.D. Perera, University Colombo, Sri Lanka

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.

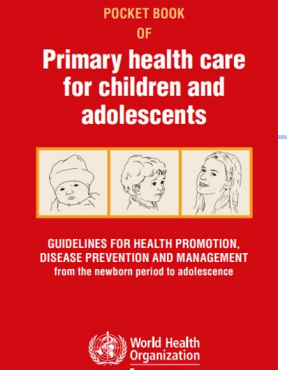
History

- 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

Results

data terminologies

- achondroplasia
- thalassemia
- Shwachman Diamond syndrome



Maternal Achondroplasia

Vitamin K

— 1 mg vitamin K IM within the first hour of birth (during initial breast-feeding while the infant is in skin-to-skin contact with the mother) **or**

— 3 doses of 2 mg vitamin K orally: at birth, at 4 to 6 days, and at 4 to 6 weeks.

— Preterm newborns should receive a lower dose 0.4 mg/kg IM.

Vitamin D

— Daily dose of 400 IU vitamin D starting within days after birth for at least the first 12 months of life.

History

Take a thorough medical history including:

- **Baby's progress since birth:** any parental concerns, feeding, problems in passing urine (usually within 24 hours of birth) and meconium (usually within 48 hours of birth) (p. 150).
- **Maternal history:** age, social background, chronic maternal diseases, medical treatments and drugs, recreational drugs including alcohol and smoking.
- **Family history:** father's age, genetic conditions, consanguinity of parents, previous pregnancies and health of siblings.
- **Present pregnancy:** medical conditions that may have influenced the pregnancy (e.g. gestational diabetes), complications, screening tests and special diagnostic procedures, exposure to maternal infectious diseases such as hepatitis B (p. 168), HIV (p. 167), cytomegalovirus (p. 163), syphilis (p. 164) or toxoplasmosis (p. 165) during pregnancy or delivery.
- **Labour and delivery:** mode of delivery, length of labour, signs of fetal distress, drugs and/or anaesthesia given, APGAR score (p. 24).
- **Risk factors for neonatal infections:**
  - — Premature rupture of membranes (> 18 h before delivery)
  - — Maternal fever > 38 °C before delivery or during labour
  - — Foul-smelling or purulent (chorioamnionitis) amniotic fluid
  - — Maternal colonization with Group B streptococcus
  - — Preterm delivery.

Pregnant woman visit PCH at 22 weeks pregnancy

Mother : Diagnosed with achondroplasia (data academic hospital)

Pregancy & **Fetus :** Short femur by ultra sound observations at 22 weeks of pregnancy

PCH officer considers child has achondroplasia & Refer to academic hospital

Child at birth : Macrocephaly and short stature at birth

After birth:

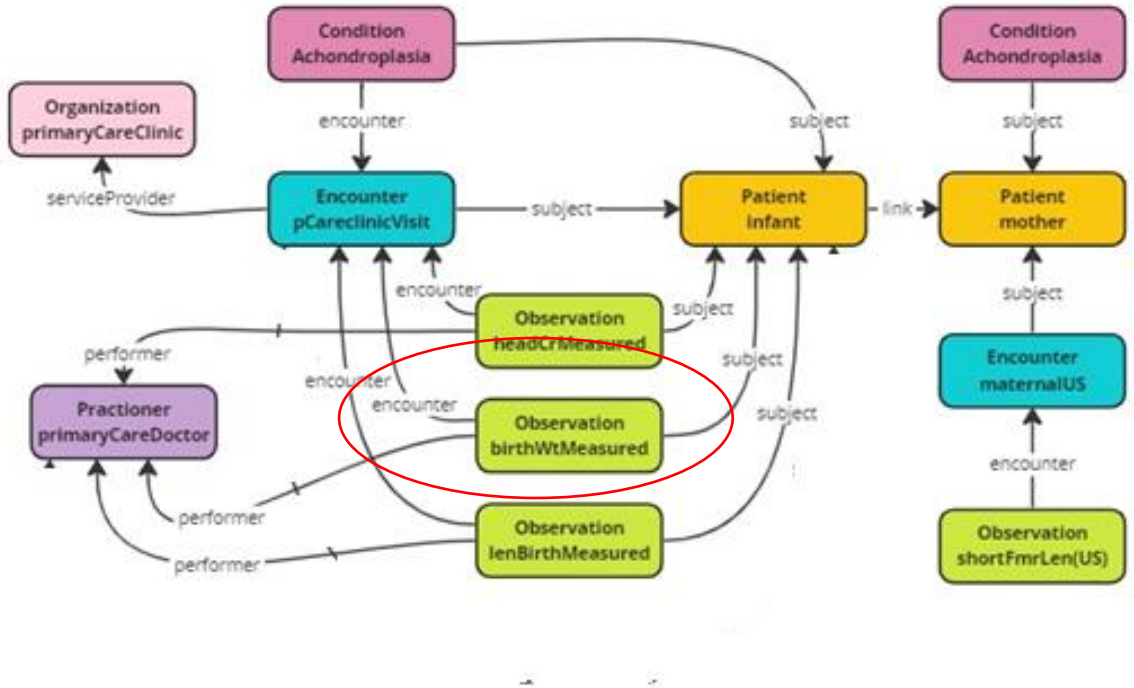
Child head circumference, body length and weight are followed according to achondroplasia growth curves

Maternal Achondroplasia

Integration in electronic health records

Date Flow HL7

Maternal Record → Home Based Record



Terminologies

Femur Length US	LOINC	11963-6
Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
Birth weight Measured	LOINC	8339-4
Body height Measured --at birth	LOINC	89269-5
Achondroplasia	ICD 10	Q77.4

Digital language JSON

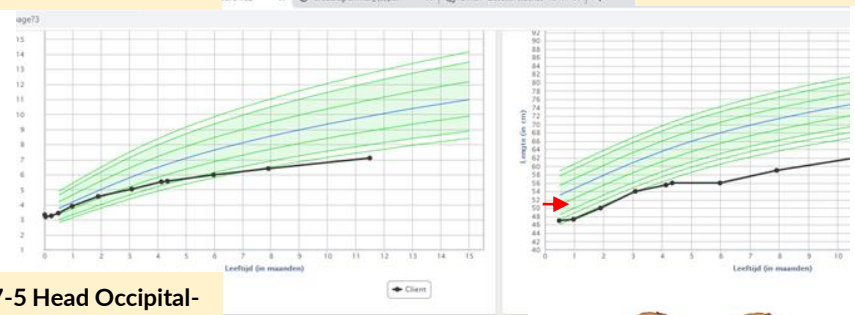
{"resourceType":"Observation","id":"cfsb1704636128883","status":"final","code":{"coding":[{"code":"3141-9","system":"http://loinc.org","display":"Body weight Measured"}]},"valueQuantity":{"value":10,"unit":"kg"},"interpretation":[{"coding":[{"code":"L","system":"http://terminology.hl7.org/CodeSystem/v3-ObservationInterpretation","display":"Low"}]}],"subject":{"reference":"cfsb1704507726008"},"effectiveDateTime":"2024-01-01T00:00:00.000Z"}

Maternal Achondroplasia

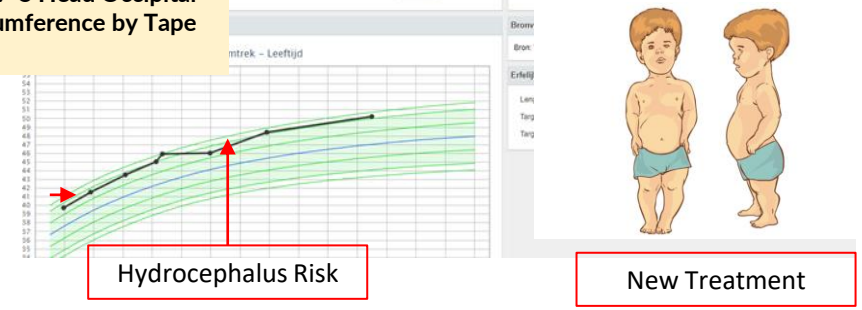
Real life data

LOINC 29463-7 Body weight

LOINC 8302-2 Body height



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



Hydrocephalus Risk

New Treatment

Achondroplasia-growth curve at each primary care visit

The company will price the treatment at roughly $300,000 per year

Beta Thalassemia

Case History

A four Month Old Child at PHC Visit In Sri Lanka

7.14 Thalassaemia

Thalassaemias are a group of autosomal-recessive hereditary blood disorders, which are characterized by defective haemoglobin chains. Based on the defective globin chain, they are classified as either α- or β-thalassaemia. They are more common in Mediterranean countries but immigration has led to wider distribution.

History

Assess for risk factors:

- Family history of α- or β-thalassaemia
- History of recurrent need for transfusions in patient or family member
- Prenatal diagnosis declined by the pregnant woman or couple at risk of thalassaemia carrier status
- Ethnic background from sub-Saharan Africa, Mediterranean and Arabian peninsula, Southeast Asia, Indian subcontinent.

Symptoms

Symptoms and timing of clinical manifestation depend on the type of thalassaemia. Severity of symptoms ranges from asymptomatic minor forms or silent carrier status to death in utero in severe forms (alpha-thalassaemia major).

Symptoms include:

- Pallor
- Abdominal distension
- Failure to thrive, poor feeding, decreased activity, lethargy
- Enlarged liver and spleen
- Jaundice
- Symptoms of gallstones: sudden intense pain in upper right abdomen
- Skeletal deformities: large head with frontal and parietal bossing, "chipmunk" facies, misaligned teeth.

Investigations

- Full blood count: microcytic hypochromic anaemia
- Ferritin
- Further investigations: peripheral smear, DNA analysis, X-ray for skeletal deformities.

PCH

Vaccination: DTP

Physical exam: **Pale | Large spleen and liver**

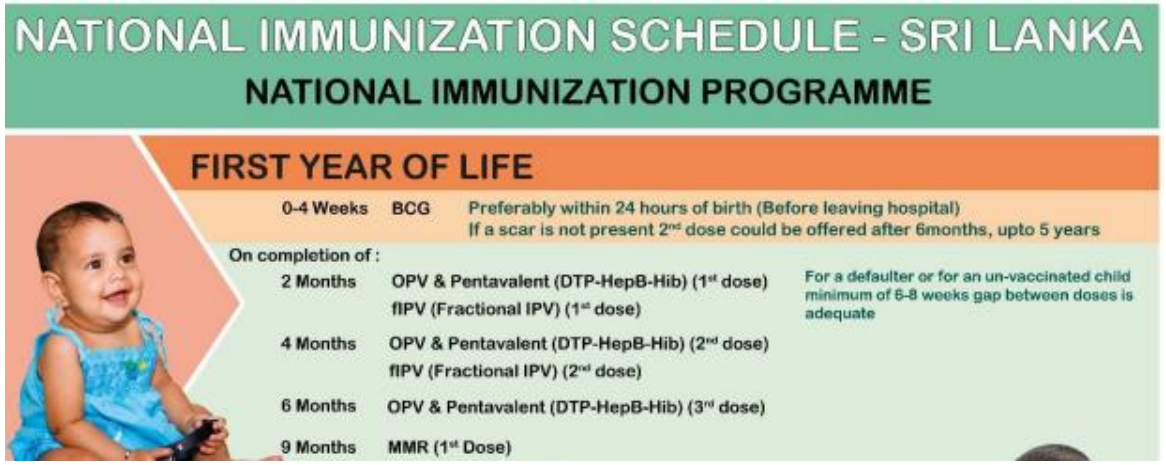
Laboratory test : Hemoglobine | Microcosis red blood cells

Referal to Thalassemia clinic

Parents are advised about routine vaccinations

Cascade Screening of Family

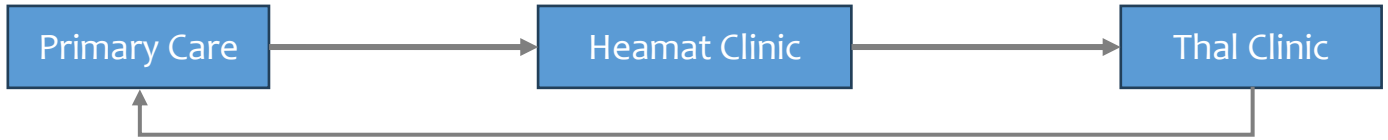
Diagnosis : Beta Thalassemia



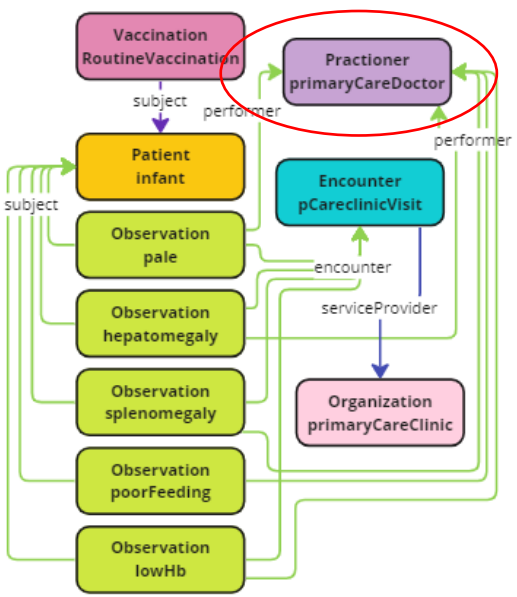
Beta Thalassemia

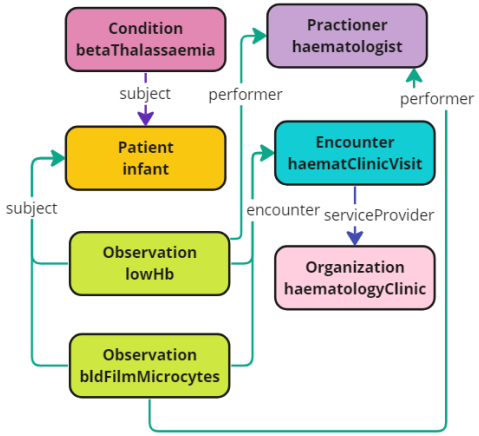
Integration in electronic health records

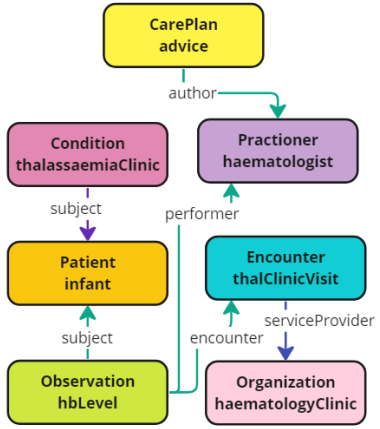
Date Flow HL7



FHIR







Terminologies

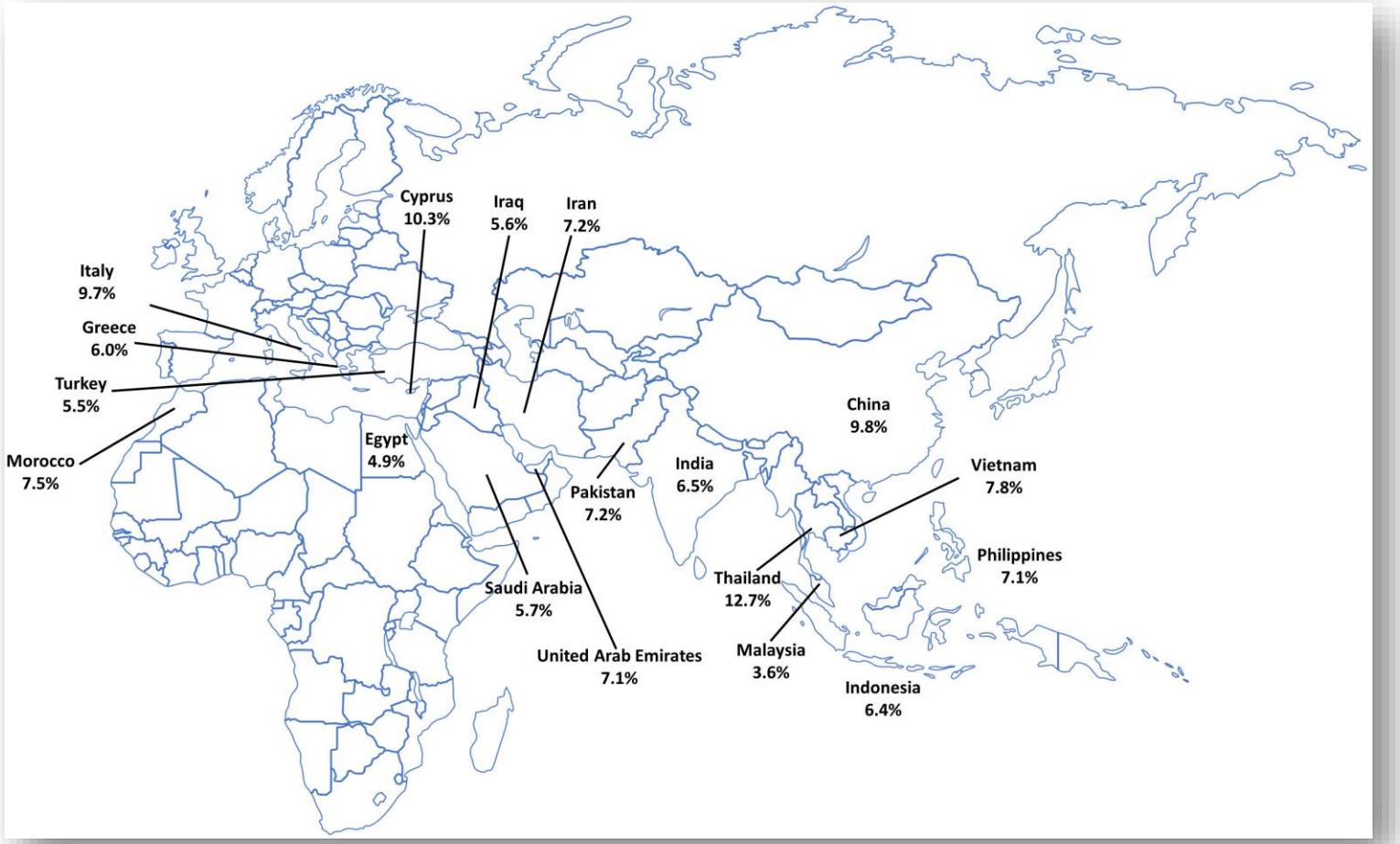
diphtheria-poliomyelitis-tetanus	ATC	J07CA01
Pallor	SNOMED CT	1237486008
Hepatomegaly	SNOMED CT	80515008
Splenomegaly	SNOMED CT	16294009
Haemoglobin concentration in blood	LOINC	718-7
Microcytes in blood film	LOINC	741-9
Feeding disorder of infancy and childhood	IDC 10	F98.2
Beta Thalassaemia	ICD 10	D56.1

Digital Language JSON

{"resourceType":"Practitioner","id":"cfsb1704509045558","name":[{"text":"Doctor Practioner","given":["Doctor"],"family":"Practioner"}],"telecom":[{"system":"email","value":"xxx@xxx.com","use":"work"},{"system":"phone","value":"0771111111","use":"work"}],"address":[{"text":"No x, Stree x, City X","use":"work","type":"both","line":["No X"],"city":"CIty X","country":"XXX"}]}

Beta Thalassemia

Carrier rate of β-thalassemia in endemic countries. Data taken from the global burden of disease collaborative network.



Front. Hematol., 20 June 2023
Sec. Red Cells, Iron and Erythropoiesis Volume 2 - 2023 | https://doi.org/10.3389/frhem.2023.1187681

From Feature to Medical Guideline

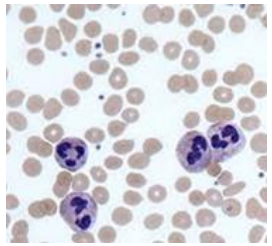
Feature

- **Fatty Stool**
- **Failure to thrive**
- **Common infections**

Shwachman Diamond

Syndrome- *Management*

- Pancreas insufficiency
- Neutropenia
- Skeletal Dysplasia
- Autisme like



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

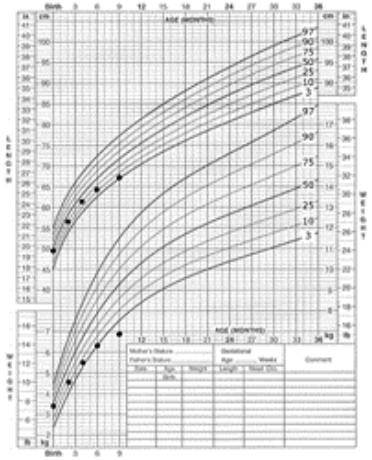
Yigal Dror,[1] Jean Donadieu,[2] Jutta Koglmeier,[3] John Dodge,[4] Sanna Toiviainen-Salo,[5] Outi Makitie,[5] Elizabeth Kerr,[1] Cornelia Zeidler,[6] Akiko Shimamura,[7] Neil Shah,[3] Marco Cipolli,[8] Taco Kuijpers,[9] Peter Durie,[1] Johanna Rommens,[1] Liesbeth Siderius,[10] and Johnson M. Liu[11]

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

Shwachman Diamond S
ORPHA:811
OMIM# 260400

Cystic Fybrosis
ORPHA:586
OMIM # 21970

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

Interoperable data model

ISO 3166-1

ATC

New Therapeutics

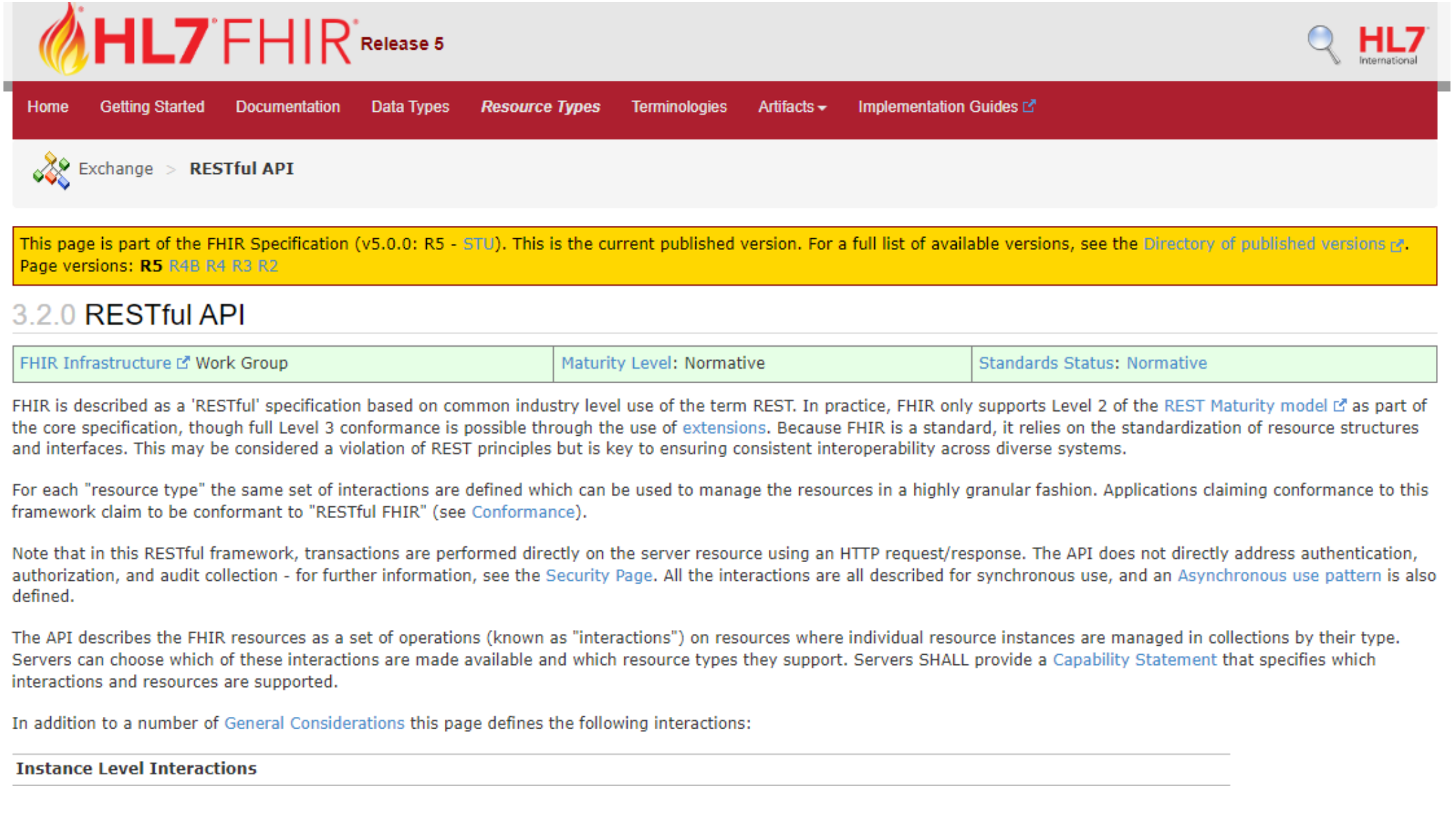
HL7/ FHIR

ICF
ISO9999

Registry
Data collection with systematically organised computer processable collection medial terms

Guideline
Social services and rehabilitation

Terminologies enable semantic interoperability in health information exchange standards systems using HL7 CDA and FHIR



HL7 FHIR Release 5

HL7 International

Home | Getting Started | Documentation | Data Types | *Resource Types* | Terminologies | Artifacts | Implementation Guides

Exchange > **RESTful API**

This page is part of the FHIR Specification (v5.0.0: R5 - STU). This is the current published version. For a full list of available versions, see the Directory of published versions.
Page versions: **R5** R4B R4 R3 R2

3.2.0 RESTful API

FHIR Infrastructure Work Group	Maturity Level: Normative	Standards Status: Normative

FHIR is described as a 'RESTful' specification based on common industry level use of the term REST. In practice, FHIR only supports Level 2 of the REST Maturity model as part of the core specification, though full Level 3 conformance is possible through the use of extensions. Because FHIR is a standard, it relies on the standardization of resource structures and interfaces. This may be considered a violation of REST principles but is key to ensuring consistent interoperability across diverse systems.

For each "resource type" the same set of interactions are defined which can be used to manage the resources in a highly granular fashion. Applications claiming conformance to this framework claim to be conformant to "RESTful FHIR" (see Conformance).

Note that in this RESTful framework, transactions are performed directly on the server resource using an HTTP request/response. The API does not directly address authentication, authorization, and audit collection - for further information, see the Security Page. All the interactions are all described for synchronous use, and an Asynchronous use pattern is also defined.

The API describes the FHIR resources as a set of operations (known as "interactions") on resources where individual resource instances are managed in collections by their type. Servers can choose which of these interactions are made available and which resource types they support. Servers SHALL provide a Capability Statement that specifies which interactions and resources are supported.

In addition to a number of General Considerations this page defines the following interactions:

Instance Level Interactions

Open Access FHIR RESTfull API Library

Mother and Child Health

- Growth & Development
- Conditions

Computable clinical guidelines

- Thalassemia
- Shwachman Diamond Syndrome

Immunizations

- Vaccination schemes

Social Support

- ICF
- ISO 9999

Conclusion

- **Evidence based guidelines** are the basis of global child health care
- **Clinical data** points derived from guidelines by clinicians serve as interoperable **terminologies**
- Care delivery is ‘translated’ into **digital language** as JSON
- FHIR enables semantic interoperability in health information exchange standards systems for **clinical and administrative content**
- A defined FHIR profile can be offered as **application programming interface** (API) to enable two or more computer programs to communicate with each other

Title "Digital child health: opportunities and obstacles", by Liesbeth Siderius, Sahan Damsiri Perera, Lars Gelander, Lina Jankauskaite, Manuel Katz, Arunas Valiulis, Adamos A. Hadjipanayis, Laura Reali and Zachi Grossman, published in "Frontiers in Pediatrics-Children and Health".*

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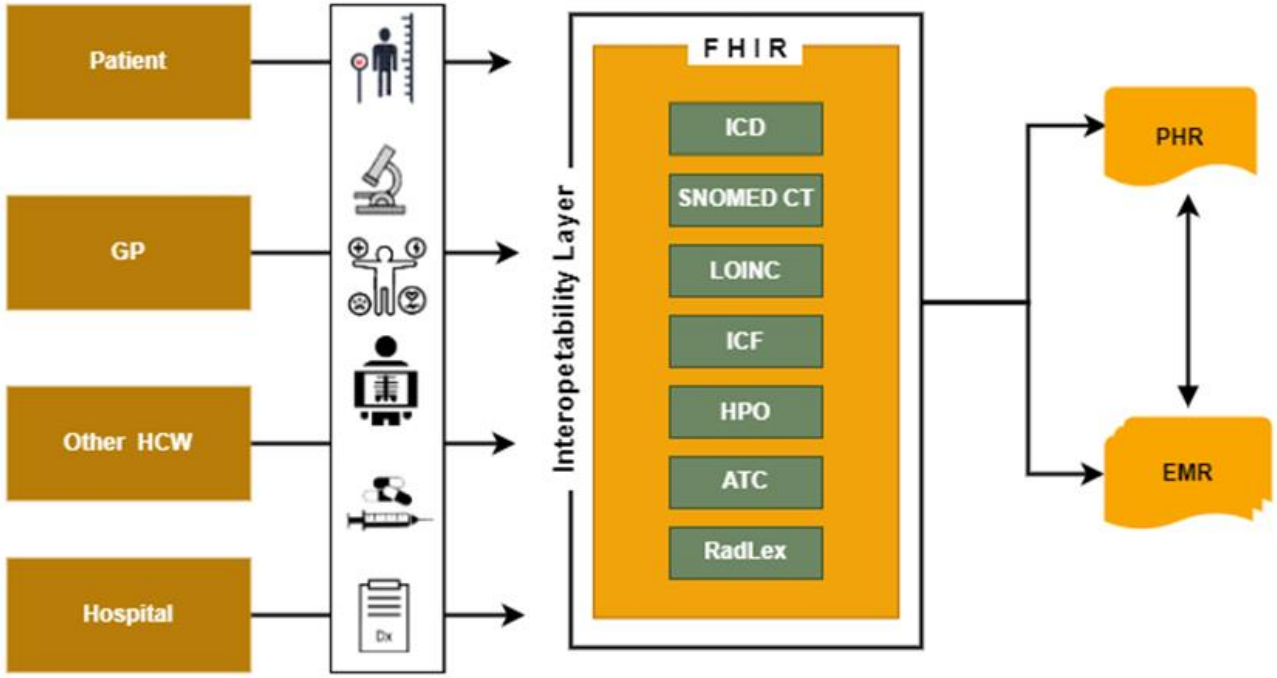
Sec. Children and Health
Volume 11 - 2023 | https://doi.org/10.3389/fped.2023.1264829



Author Figure S.D. Perera, University Colombo, Srilanka

Thank

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John Dodge, U.K.
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Jola Wierzba, Poland
Jernej Zavrsnik, Slovenia

- Consensus in Pediatrics and Child Health

Manual Katz, Israel

- Forum Rare Diseases, Sri Lankan Pediatric Society

Anjan Bhattacharya, ICF expert India
Sahan Damsiri Perera, IT Expert, Sri Lanka
Marc de Graauw, IT Expert, Netherlands
Martin Postma, IT 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

e.siderius@kpnplanet.nl

Stichting Shwachman syndroom Support Holland