Digitalisation of primary health care

Leave no child behind

Liesbeth Siderius & Sahan Damsiri Perera

The Voice of 12,000 Patients , EURORDIS 2009

Rare Disease Working Group, EAP 2010

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

2013

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

ICF
ISO9999

HL7/ FHIR

Registry
Data collection with systematically organised computer processable collection medial terms

Guideline
Social services and rehabilitation

REGULATION (EU) 2016/679 OF THE EUROPEAN PARLIAMENT AND OF THE COUNCIL

of 27 April 2016

on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation)

General Data Protection Regulation 2016

Art. 20 GDPR

Right to data portability

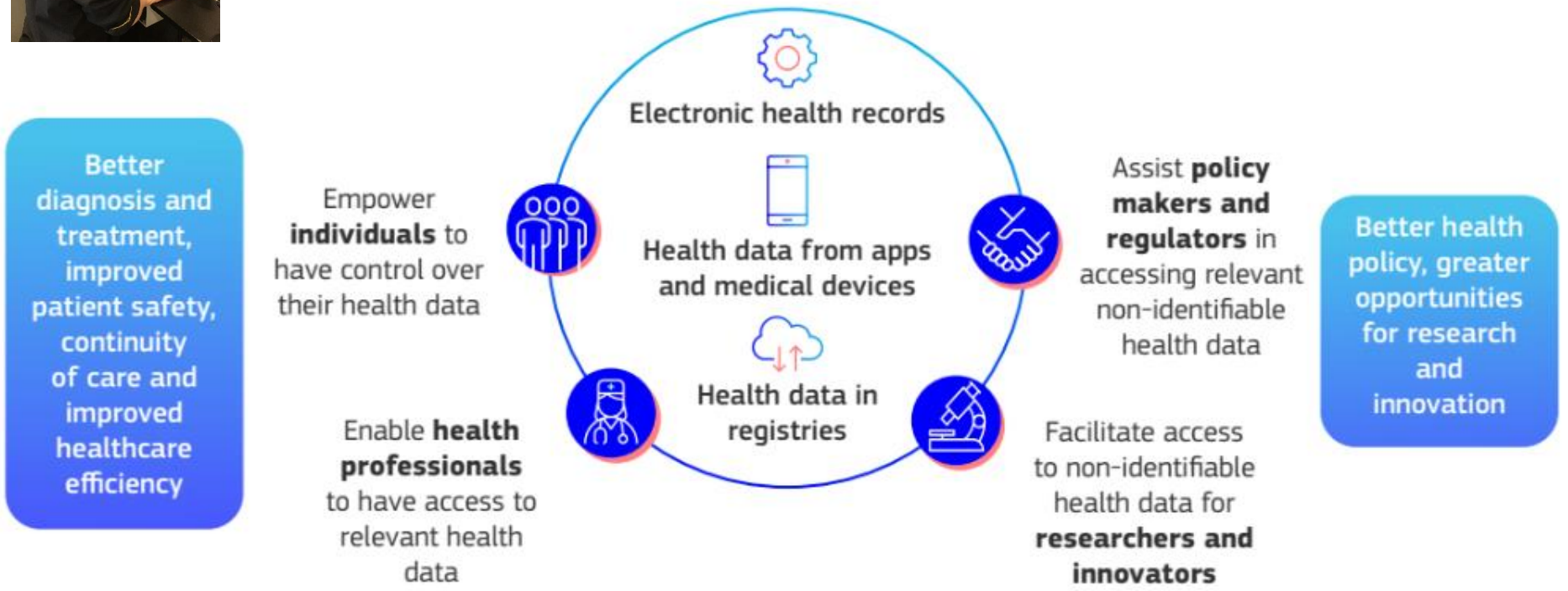
The data subject shall have the right to receive the personal data concerning him or her, which he or she has been provided to a controller, in a **structured, commonly used and machine-readable format** and have the right to transmit those data to another controller without hindrance from the controller to which personal data have been provide….

Digital Health Europe

ABOUT NEWS & EVENTS TWINNINGS RESULTS & PUBLICATIONS SUPPORT SERVICES RESOURCES Participate

DigitalHealthEurope recommendations on the European Health Data Space

3 May 2022



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**.

Abandoned in hospital

Often, this neglect is the result of limited data

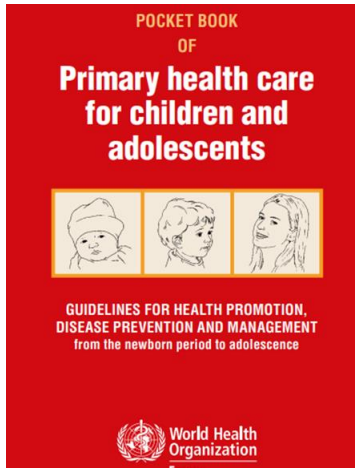

EQUITY
FOR PEOPLE LIVING WITH A RARE DISEASE
IS EQUITABLE ACCESS TO DIAGNOSIS, TREATMENT, HEALTH, SOCIAL CARE AND OPPORTUNITY.
#RAREDISEASEDAY
RAREDISEASEDAY.ORG

2024


70%
OF GENETIC RARE DISEASES START IN CHILDHOOD!
ARTICLE 'ESTIMATING CUMULATIVE POINT PREVALENCE OF RARE DISEASES: ANALYSIS OF THE ORPHANET DATABASE, EUROPEAN JOURNAL OF HUMAN GENETICS (2019)
#RAREDISEASEDAY
RAREDISEASEDAY.ORG
29 FEB 2024

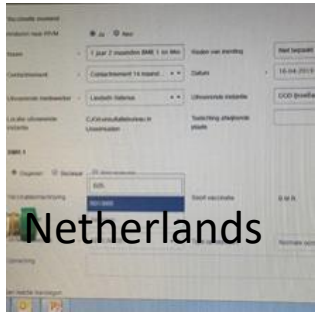
Digital Modelling of Primary Child Health

World Health Organization

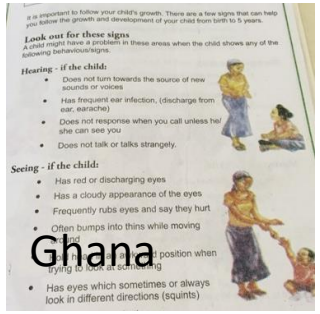


https://www.who.int/europe/publications/i/item/9789289057622

Preventive Child Health Records



Netherlands



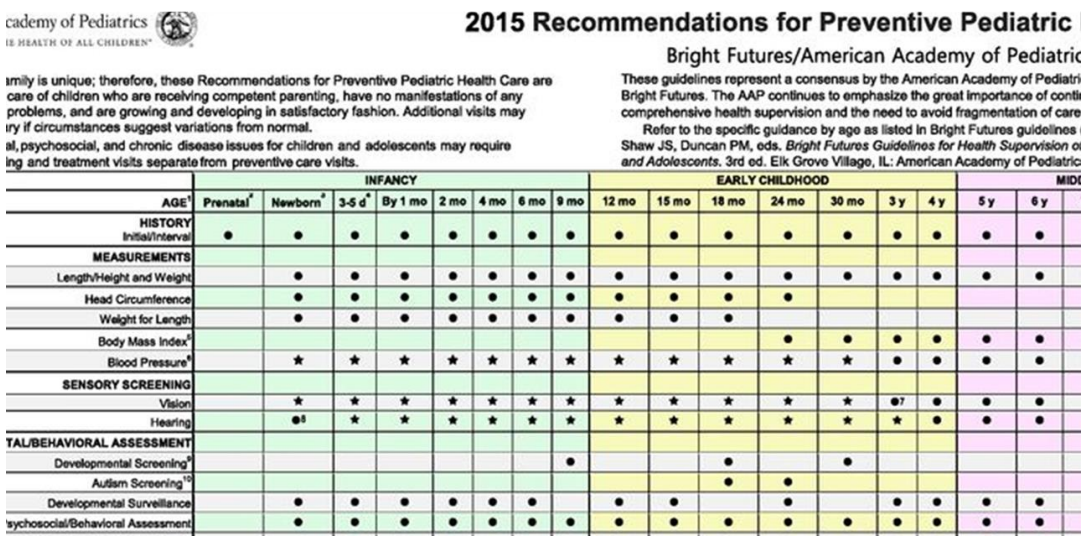
Ghana

Spain

Peru

Sri Lanka

Poland



cademy of Pediatrics
IE HEALTH OF ALL CHILDREN®

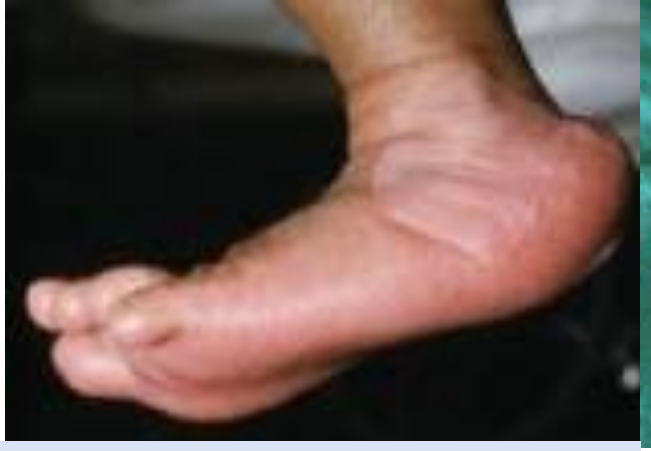
2015 Recommendations for Preventive Pediatric

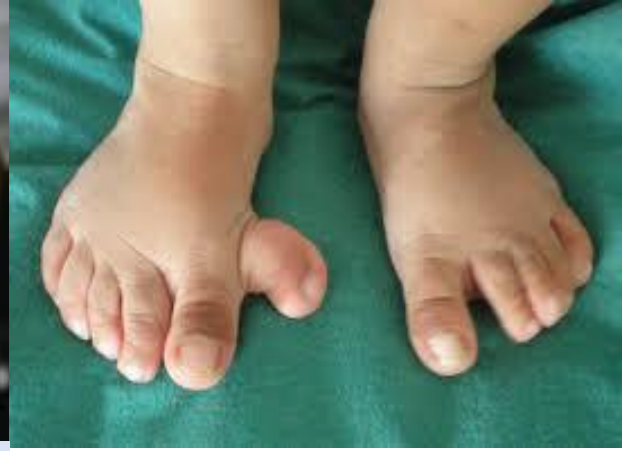
Bright Futures/American Academy of Pediatric

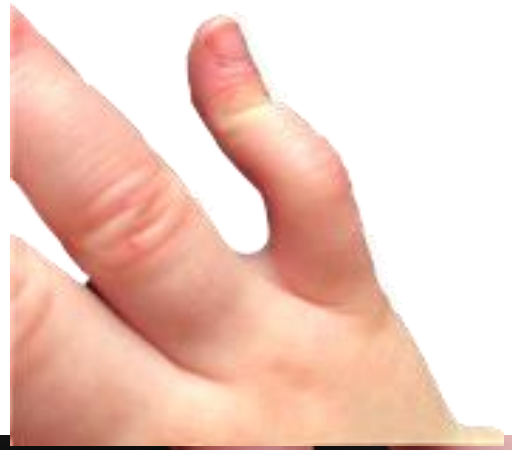
mily is unique; therefore, these Recommendations for Preventive Pediatric Health Care are care of children who are receiving competent parenting, have no manifestations of any problems, and are growing and developing in satisfactory fashion. Additional visits may ry if circumstances suggest variations from normal.
al, psychosocial, and chronic disease issues for children and adolescents may require ng and treatment visits separate from preventive care visits.

These guidelines represent a consensus by the American Academy of Pediatri Bright Futures. The AAP continues to emphasize the great importance of conti comprehensive health supervision and the need to avoid fragmentation of care
Refer to the specific guidance by age as listed in Bright Futures guidelines Shaw JS, Duncan PM, eds. *Bright Futures Guidelines for Health Supervision o and Adolescents*. 3rd ed. Elk Grove Village, IL: American Academy of Pediatric

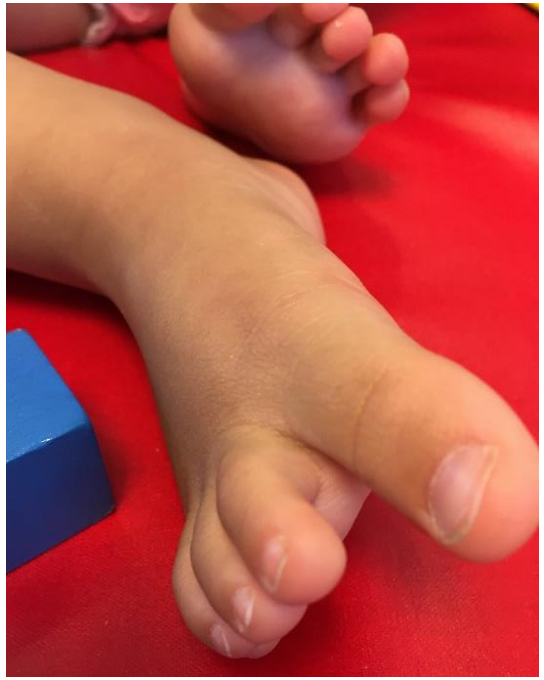
	INFANCY								EARLY CHILDHOOD							MID		
AGE[1]	Prenatal[2]	Newborn[3]	3-5 d[4]	By 1 mo	2 mo	4 mo	6 mo	9 mo	12 mo	15 mo	18 mo	24 mo	30 mo	3 y	4 y	5 y	6 y	
HISTORY Initial/Interval	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	
MEASUREMENTS																		
Length/Height and Weight		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	
Head Circumference		●	●	●	●	●	●	●	●	●	●	●						
Weight for Length		●	●	●	●	●	●	●	●	●	●							
Body Mass Index[5]												●	●	●	●	●	●	
Blood Pressure[6]		★	★	★	★	★	★	★	★	★	★	★	★	●	●	●	●	
SENSORY SCREENING																		
Vision		★	★	★	★	★	★	★	★	★	★	★	★	●[7]	●	●	●	
Hearing		●[8]	★	★	★	★	★	★	★	★	★	★	★	★	●	●	●	
TAL/BEHAVIORAL ASSESSMENT																		
Developmental Screening[9]								●			●		●					
Autism Screening[10]											●	●						
Developmental Surveillance		●	●	●	●	●	●		●	●		●		●	●	●	●	
sychosocial/Behavioral Assessment		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	

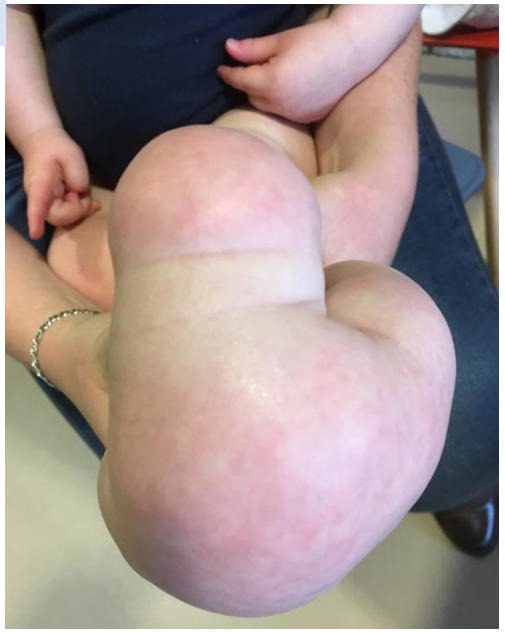






Access to Diagnostics





International classifications 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

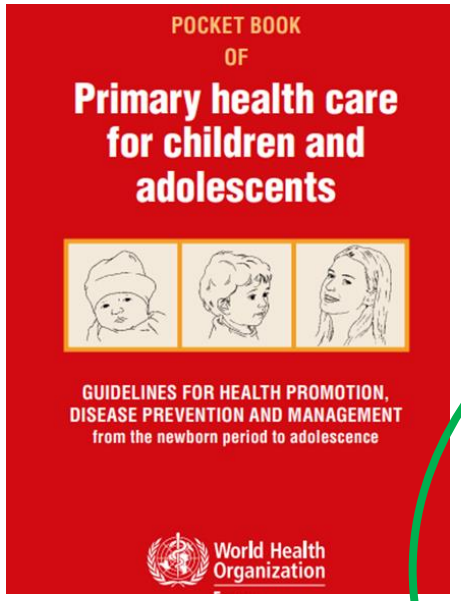
Use of terminologies enables semantic interoperability between systems using HL7 CDA and FHIR

Global Child Health

Cases

1. Fetal Alcohol syndrome
2. **Maternal Achondroplasia**
3. Breech-Cleft Palate-Microtia
4. Neonatal Jaundice & Hyperbilirubinaemia
5. **Juvenile Cataract**
6. Beta Thalassemia
7. Down Syndrome

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.

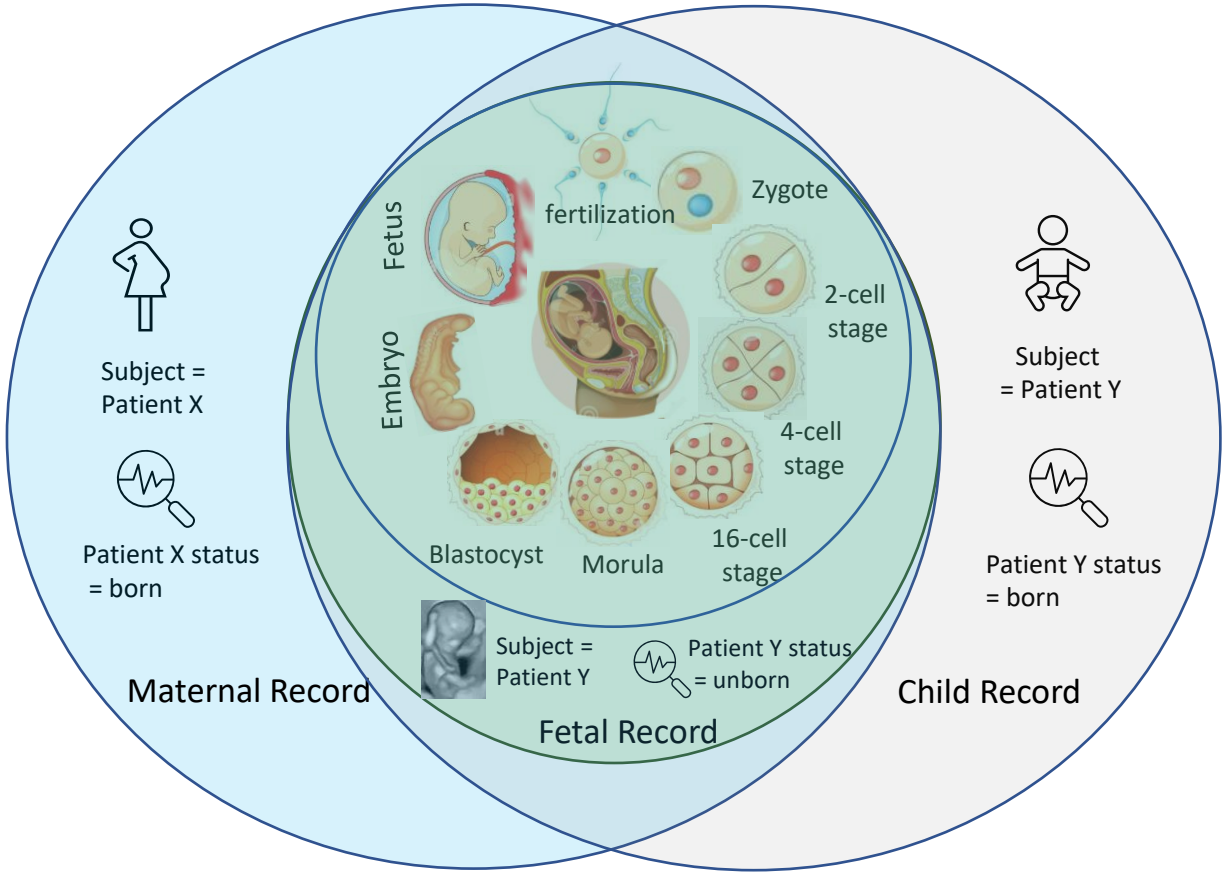
- Look for congenital diseases and jaundice
- Support caregivers.

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

Child Health & Obstetrics International Collaboration and Exploration

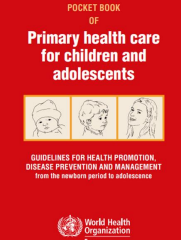
Conceptual diagram: Mother-Fetus-Child concepts, 2022



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

[Child Health & Obstetrics International Collaboration and Exploration - Patient Care - Confluence (hl7.org)]



Case 2 - Maternal Achondroplasia

Case History

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.

115

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 and body length are followed according to achondroplasia growth curves

Achondroplasia curves are available in PCH and home-based record

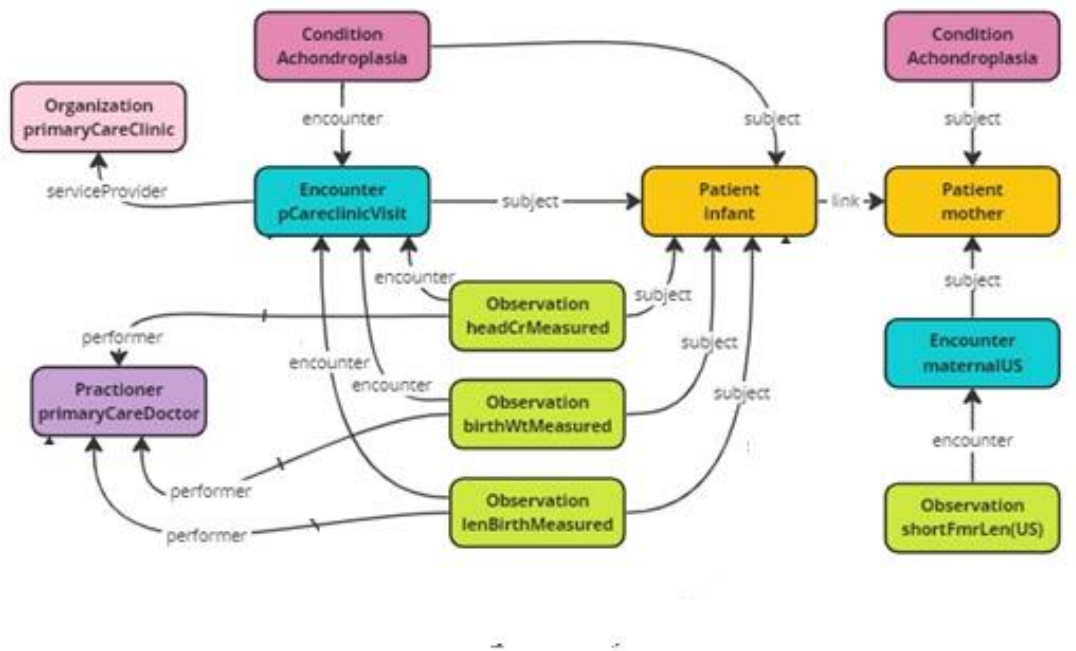
Case 2 - Maternal Achondroplasia

Management in Home

Date Flow

Maternal Record → Home Based Record

FHIR



Terminologies

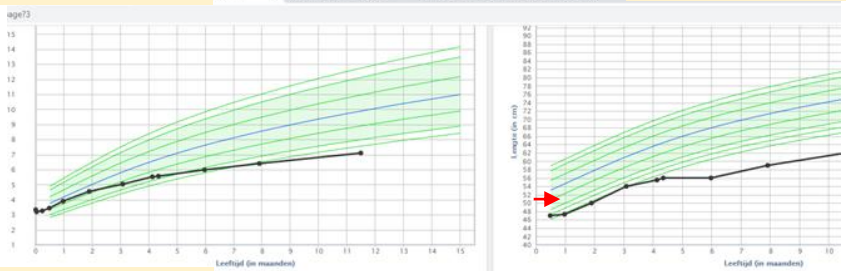
Femur Length US	LOINC	11963-6
Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
Birth weight Measured	LOINC	8339-4
Body height Measured --at birth	LOINC	89269-5
Achondroplasia	ICD 10	Q77.4

Maternal Achondroplasia

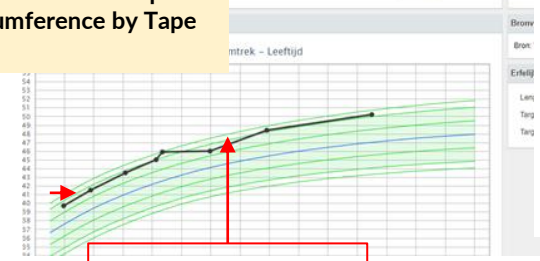
VOXZOGO™
(vosoritide) for injection

LOINC 29463-7 Body weight

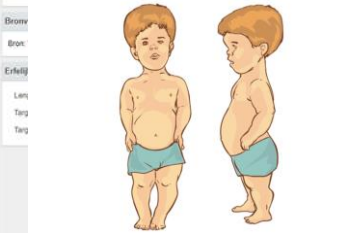
LOINC 8302-2 Body height



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



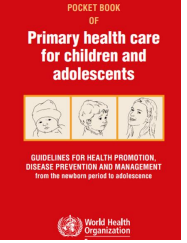
Hydrocephalus Risk



New Treatment

Achondroplasia-growth curve at each primary care visit

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



Case 5- Juvenile Cataract

Case History

3.3 Well-child visit: 1 week

- Look for congenital diseases and jaundice
- Follow up weight gain and vaccinations
- Support caregivers and counsel on feeding, activity and safety

History

- Care situation and exceptional burdens in the family
- Feeding difficulties
- Abnormal crying
- Congenital disorders in the family, e.g. hip problems, eye conditions

Examination

- Perform a complete physical examination (p. 116). Look for signs of acute illness or congenital conditions:
  - **Growth:** measure body weight, length and head circumference (p. 21) and confirm the z-score according to the WHO growth charts (Annex 3). Newborn typically lose up to 10% of their birth weight during the first days of life and regain it within 10–14 days. If weight loss exceeds 10% of birth weight, see p. 119.
  - **Skin:** pallor, cyanosis, jaundice (p. 148), rashes (p. 143), hydration
  - **Head and neck:** bulging fontanelle (p. 128), crepitations, cleft palate (p. 129), caput succedaneum (p. 126), ptosis (p. 134), absent red eye reflex (p. 133), coloboma (p. 133), nystagmus, ear deformities (p. 131)

Cloudy lens or absent red reflex

A lens opacity (grey-white clouding of the lens) or absence of the red reflex, during the red reflex examination (p. 119), can be a sign of both congenital cataract (p. 459) and early retinoblastoma (p. 459).

- Refer newborns with an absent red reflex or a cloudy lens immediately to an eye specialist. Early detection and treatment are essential.

A Two Month old at PCH

PCH

Child comes for a regular screening at PCH

Physical exam

Red eye reflex

Referal to ophtalmologist

Observation:

Study observation Left optic lens Slit lamp biomicroscopy Ophtalmol >

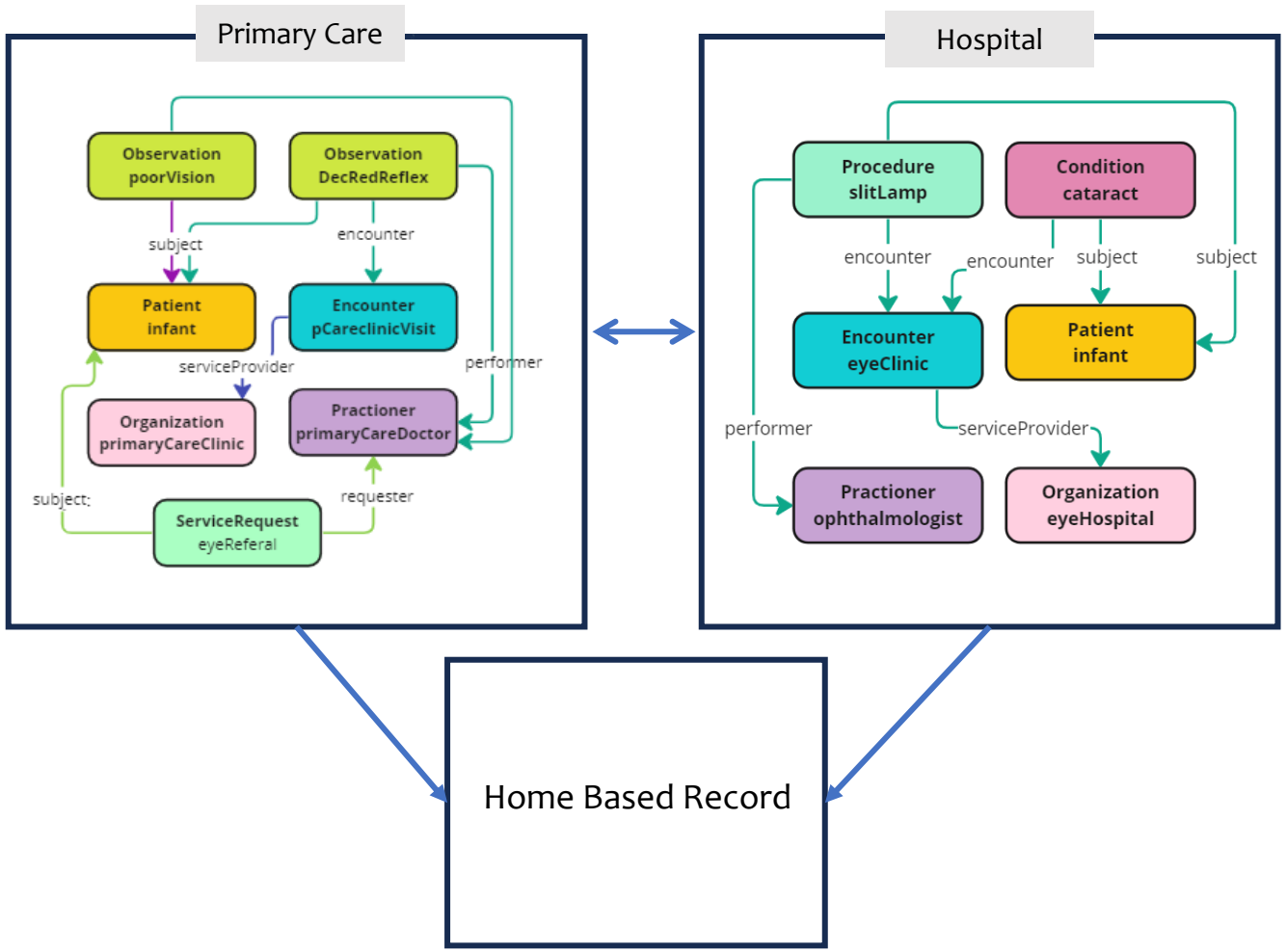
Diagnosis : Infantile cataract

Case 5- Juvenile Cataract

Diagnosis

Date Flow

Primary Care → Hospital Record → Home Based Record



Terminologies

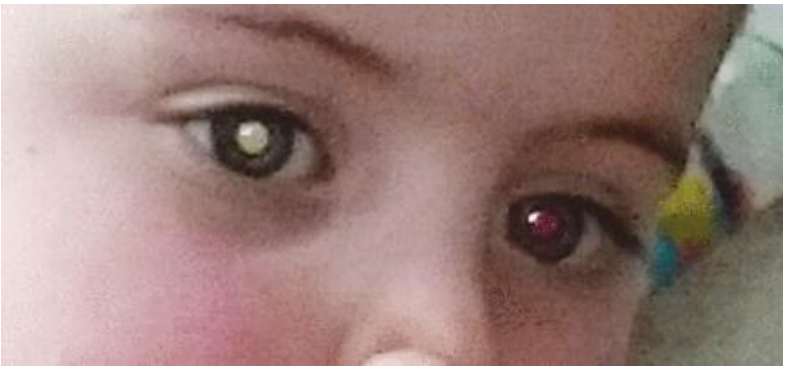
Red reflex absent	SNOMED CT	247079003
Abnormal vision	SNOMED CT	7973008
Study observation Left optic lens Slit lamp biomicroscopy	LOINC	79866-0
Infantile, juvenile and presenile cataract	ICD 10	H26.0

Visual impairment

Infantile cataracts remain one of the **most treatable causes of lifelong visual impairment**.

While the chance of improving vision for children with infantile cataracts has never been better,

Significant global and socioeconomic disparities still exist in their early management.



Lenhart PD, Lambert SR. Current management of infantile cataracts. Surv Ophthalmol. 2022 Sep-Oct;67(5):1476-1505. doi: 10.1016/j.survophthal.2022.03.005. Epub 2022 Mar 17. PMID: 35307324; PMCID: PMC10199332.

From Feature to Medical Guideline

Feature

- Fatty Stool
- Growth Retardation
- Common infections

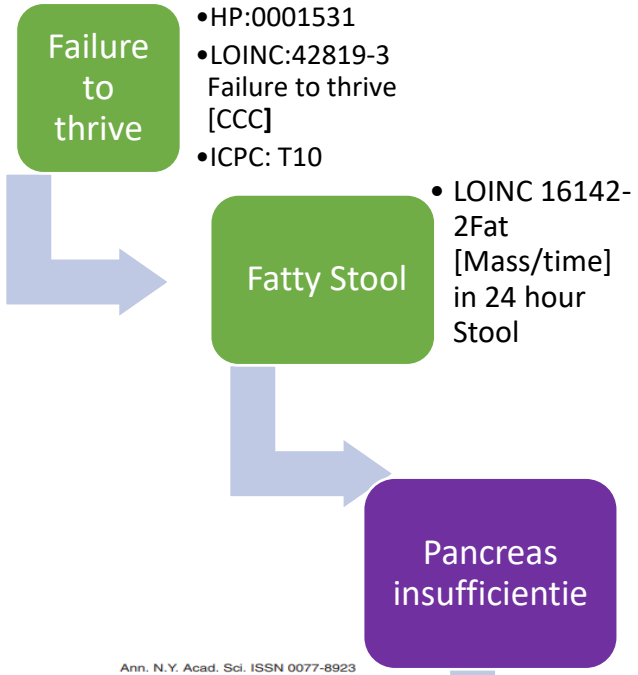
Shwachman

Diamond

Syndrome-

Management

- Pancreas insufficiency
- Neutropenia
- Skeletal Dysplasia
- Autisme like



2022

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

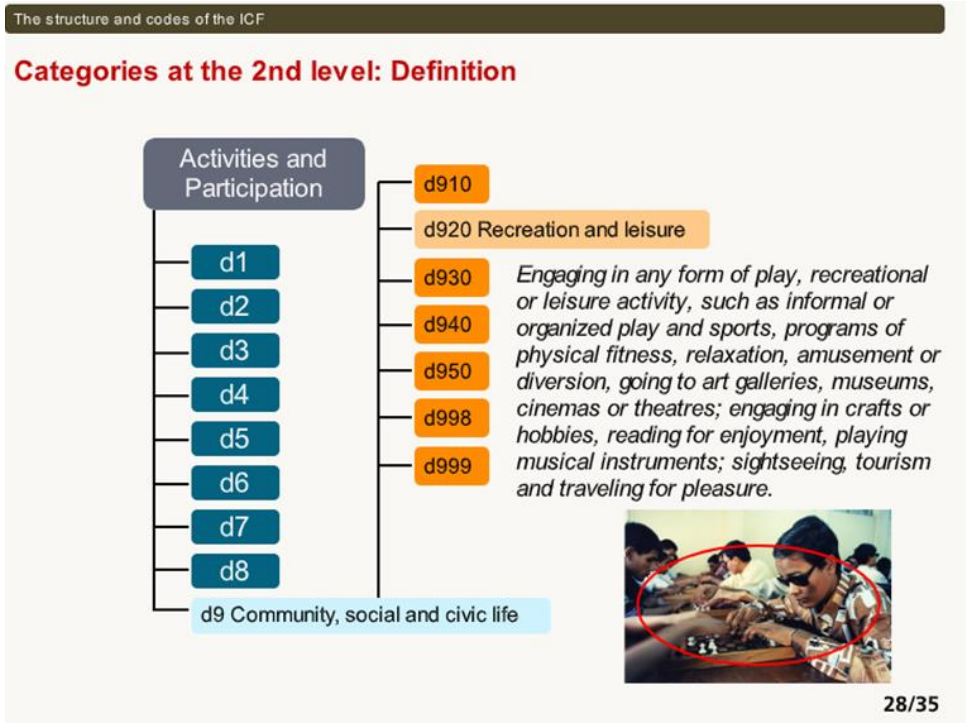
ANNALS OF THE NEW YORK ACADEMY OF SCIENCES
Issue: Annals *Meeting Reports*

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]

ICF d 920.0

Recreation and leisure

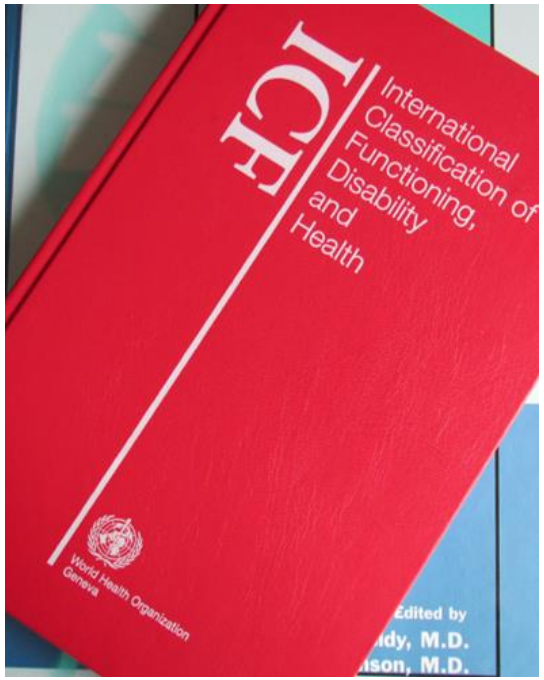


Indian Mother and Childcare
Kolkata, 2020

World Health Organization

ICF : INTERNATIONL CLASSIFICATION of FUNCTIONING. DISABILITY and HEALTH

Gowers's Sign



ICF-b 730 Muscular Power Functions

Boys with Duchenne Muscular Dystrophy Poland

Growing up with Shwachman Diamond syndrome
International Classification of Function (ICF)

Materials & Methods

Representatives of the Dutch SDS patient organisation selected 12 categories from the domain activities and participation of the ICF core-set autism brief and included these items in a questionnaire.

Results

The table shows ICF ≥6 quotations from ≥ 3 respondents; the most frequent are on top. Not only activities and participation categories were used frequently, but also functions and environmental factors

Activities and participation	Functions
d920 Recreation & leisure	b152 Emotional functions
d240 Handling stress and other psychological demands	b126 Temperament and personality functions
d850 Remunerative employment	b455 Exercise tolerance functions
d570 Looking after one's health	b125 Dispositions and intra-personal functions
d475 Driving	
d310 Understand spoken messages	**Environmental factors**
d720 Complex interpersonal interactions	e310 Immediate family
d610 Acquiring a place to live	e330 Peoples in positions of authority
d750 Informal social relationships	e355 Health professionals
d640 Doing housework	e360 Other professionals
d710 Basic interpersonal interactions	e120 Transportation
d230 Carrying out daily routine	
d210 Undertaking a single task	

Conclusion

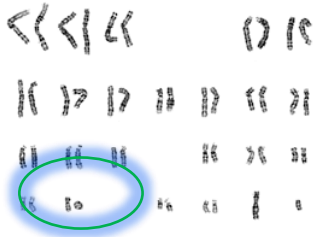
Understanding the positive / neutral and negative aspects of living with a rare condition may help parents and communities to support growing up towards a fulfilled life.
Incorporating the **ICF in personal digital health records promotes health and well-being at all ages (Sustainable Development Goal #3, United Nations**)

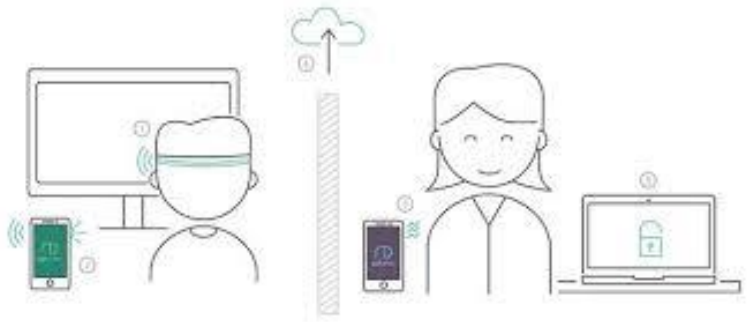
Epilepsy – assistive products- Health Technology Assessment

ICF d132
Acquiring Information

-

Mosaic ring chromosome 20





ICS › 11 › 11.180 › 11.180.01

ISO 9999:2016

Assistive products for persons with disability — Classification and terminology

EPIHUNTER

Health Technology Assessment (HTA)

ISO/TS 82304-2:2021
Health software Part 2: Health and wellness apps
Quality and reliability

Our FHIR SDK for Android Developers

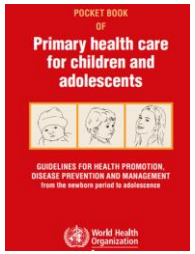
Android Developers · Follow
Published in Android Developers · 3 min read · Mar 24, 2022

Kolkata, 2020

Cameroon's Children

Child Health & Rights CHIFA .org

17 January 2024 CHIFA message from Cameroon on child health pocket handbook

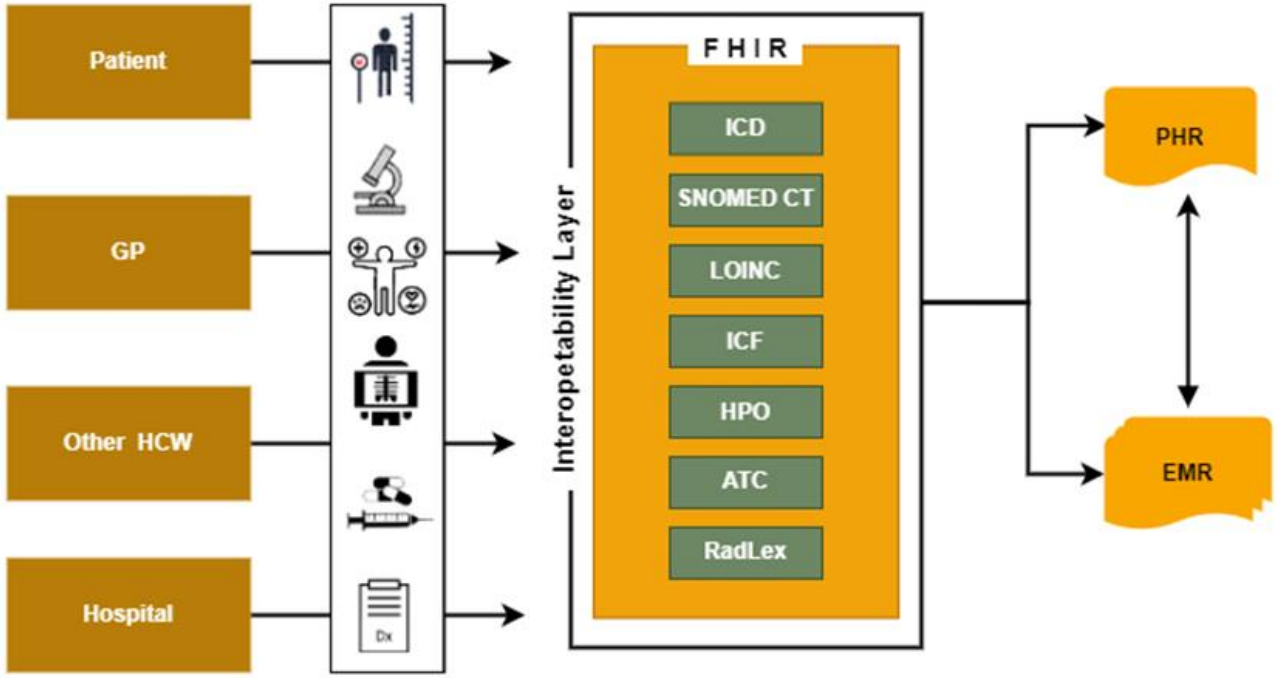


"sharing it to some medical and child protection whatsapp groups around Cameroon and particularly in conflict affected regions.
People are subject to solar light for charging of phones."

Conflicts, wars, disasters hinder our work globally. Lets work to mitigate these factors.

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".

Front. Pediatr., 22 December 2023
Sec. Children and Health
Volume 11 - 2023 | https://doi.org/10.3389/fped.2023.1264829



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

International Patient Summary

HL7 International

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.

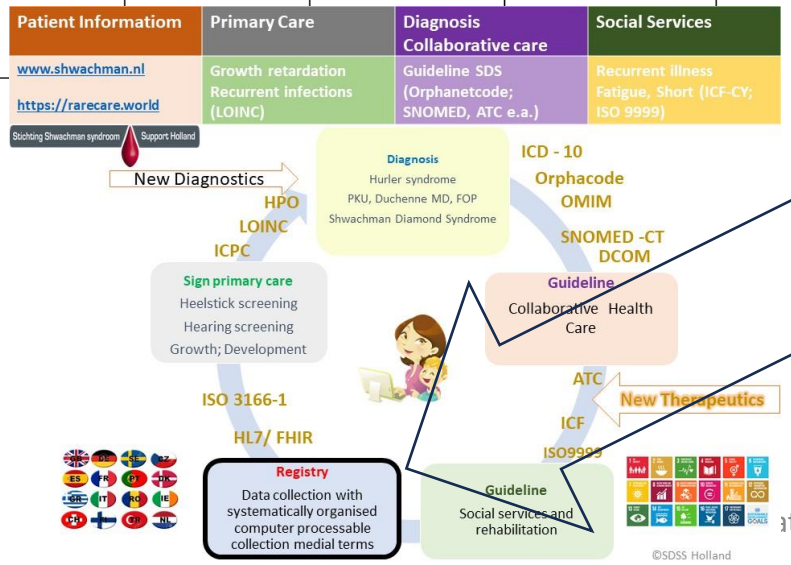
Set of common data elements for rare diseases registration

EUROPEAN COMMISSION
JOINT RESEARCH CENTRE
Directorate F – Health and Food
Unit F.1 – Disease Prevention

EUROPEAN PLATFORM ON RARE DISEASE REGISTRATION (EU RD Platform)

SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION

GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
1. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	• String	https://eu-rd-platform.jrc.ec.europa.eu/spider
2. Personal information	2.1.	Date of birth	Patient's date of birth	• Date (dd/mm/yyyy)	
	2.2.	Sex	Patient's sex at birth	• Female • Male • Undetermined • Foetus (Unknown)	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	• Alive • Dead • Lost in follow-up • Opted-out	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	• Date (dd/mm/yyyy)	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	• Date (dd/mm/yyyy)	
5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
	5.2.	Age at diagnosis	Age at which diagnosis was made	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
6. Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis retained by the specialised centre	Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code	
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of	http://www.hgvs.org
	6.3	Undiagnosed case	How the undiagnosed case i defined		
7. Research	7.1.	Agreement to be contacted for research purposes	Patient's permission exists fo being contacted for researc purposes		
	7.2.	Consent to the reuse of data	Patient's consent exists fo his/her data to be reused fo other research purposes		
	7.3.	Biological sample	Patient's biological sample available for research	• YES • NO	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	• YES (if appropriate use link) • NO	https://directory.bbmri-eric.eu
8. Disability	8.1.	Classification of functioning/disability	Patient's disability profile according to International Classification of Functioning and Disability (ICF)	• Disability profile / Score	



ORPHA
ICD 9
ICD10

HGVS Human Genome Variety
HPO
HGNC Human Genome Nomenclature
OMIM

ICF

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

HL7 International

HL7 Terminology (THO)

3.1.0 - Publication

Home | HL7 Code Systems | Value Sets | Concept Maps | External Identifier Systems | External Code Systems | Documentation | Downloads

Table of Contents > **Artifacts Summary** > **Passport Numbers Namespace for LITHUANIA**

This page is part of the HL7 Terminology (v3.1.0: Release) based on FHIR R4. The current version which supercedes this version is 5.2.0. For a full list of available versions, see the Directory of published versions

Narrative Content | XML | JSON | TTL | History

9.3899.9 : Passport Numbers Namespace for LITHUANIA - JSON Representation

Active as of 2022-02-07

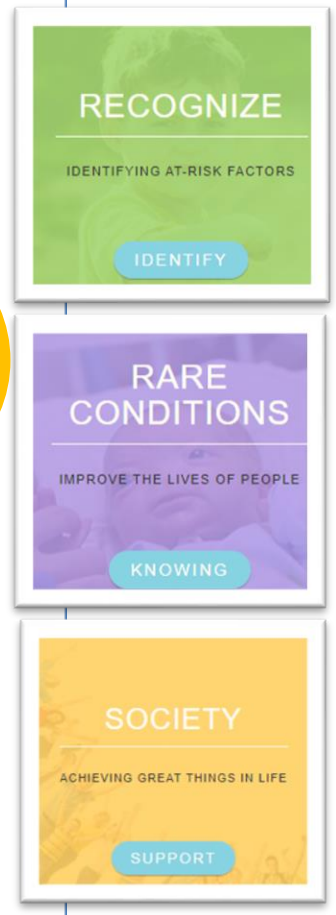
Raw json | Download


```
{
  "resourceType" : "NamingSystem",
  "id" : "passportNumNS-LTU",
  "text" : {
    "status" : "generated",
    "div" : "<div xmlns=\"http://www.w3.org/1999/xhtml\"><h3>Summary</h3><table class=\"gric
  },
  "extension" : [
    {
      "url" : "http://hl7.org/fhir/tools/StructureDefinition/extension-title",
      "valueString" : "Passport Numbers Namespace for LITHUANIA"
```

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

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

Just normal People

22qdeletion

The Family

Skeletal Dysplasia

At work

Fibrodysplasia Ossificans Propressiva

Studiy
Animal Science

Thank

- European Pediatric Rare Disease Network

John Dodge, U.K.
Lali Margvelashvili, Georgia
Velibor Tasic, N- Macedonia
David Neubauer, Slovenia
Arunas Valiulis, Lithuania
Lina Jankauskaite, Lithuania
Jola Wierzba, Poland
Jernej Zavrsnik, Slovenia

- Consensus in Pediatrics and Child Health

Manual Katz, Israel

- Forum Rare Diseases, Sri Lankan Pediatric Society

Sahan Damsiri Perera, IT Expert, Sri Lanka
Anjan Bhattacharya, India
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

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