Interoperability of Primary Child Health Data

Global Digital Child Health

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Introduction

1. Promoting the dissemination and generation of knowledge on rare diseases in children depends on the availability and **interoperability of primary child health data.**
2. A lot of work has been going on in a variety of places to further this development, including presentations at **LOINC conferences** in Annecy and Atlanta, as well as discussions in **the X-eHealth** project and in the **IPS community**.
3. A first draft **HL7 FHIR** Implementation Guide for Rare Diseases in Child Health is available and will be a topic for discussion during this session. A strong connection is also felt with the CHOICE project (**Child Health Obstretics International Collaboration and Exploration**).

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

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

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

Paracetamol

Stofnaam	Paracetamol
Merknaam	Perfalgan, Apotel
ATC code	N02BE01

Real world data

ATC

New Therapeutics

ISO 3166-1

ICF

HL7/ FHIR

ISO9999

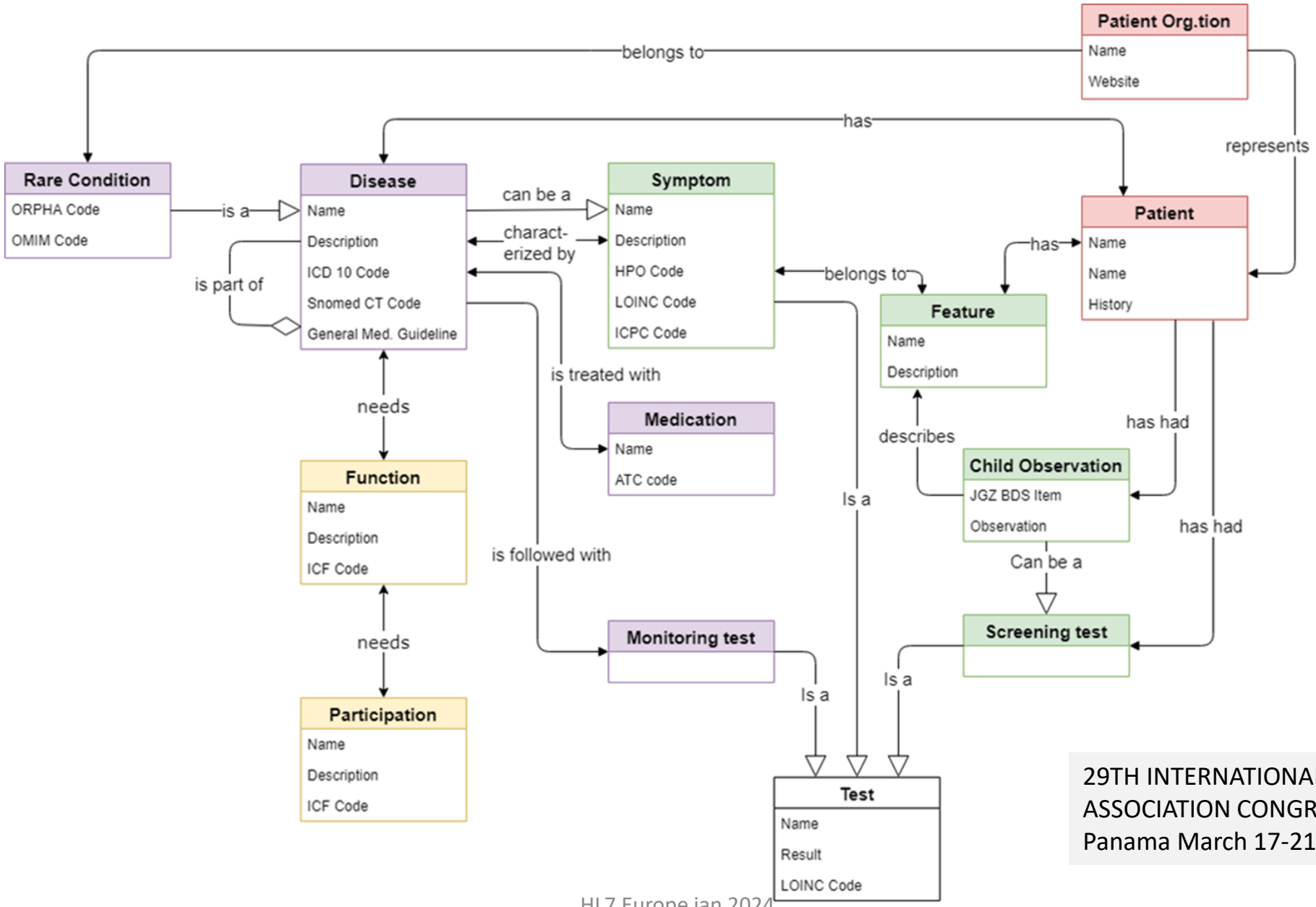
Registry

Data collection with systematically organised computer processable collection medial terms

Guideline

Social services and rehabilitation

Unified Modeling Language (UML)



29TH INTERNATIONAL PEDIATRIC ASSOCIATION CONGRESS Panama City, Panama March 17-21, 2019

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

Digital Modelling of Primary Child Health

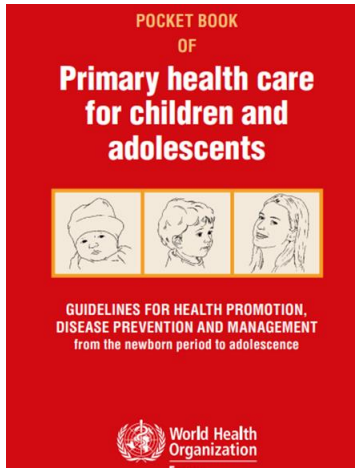
- Illustrative cases from The WHO Pocket Book on Primary health care for Children and Adolescents (WHO Europe, 2022)

- Proof of concept for a comprehensive implementation guide that harnesses the power of LOINC and **HL7/FHIR standards**,

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

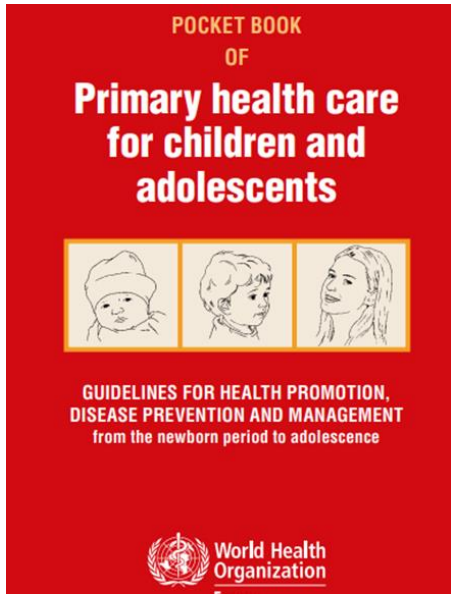
Digital Modelling of Primary Child Health

World Health Organization

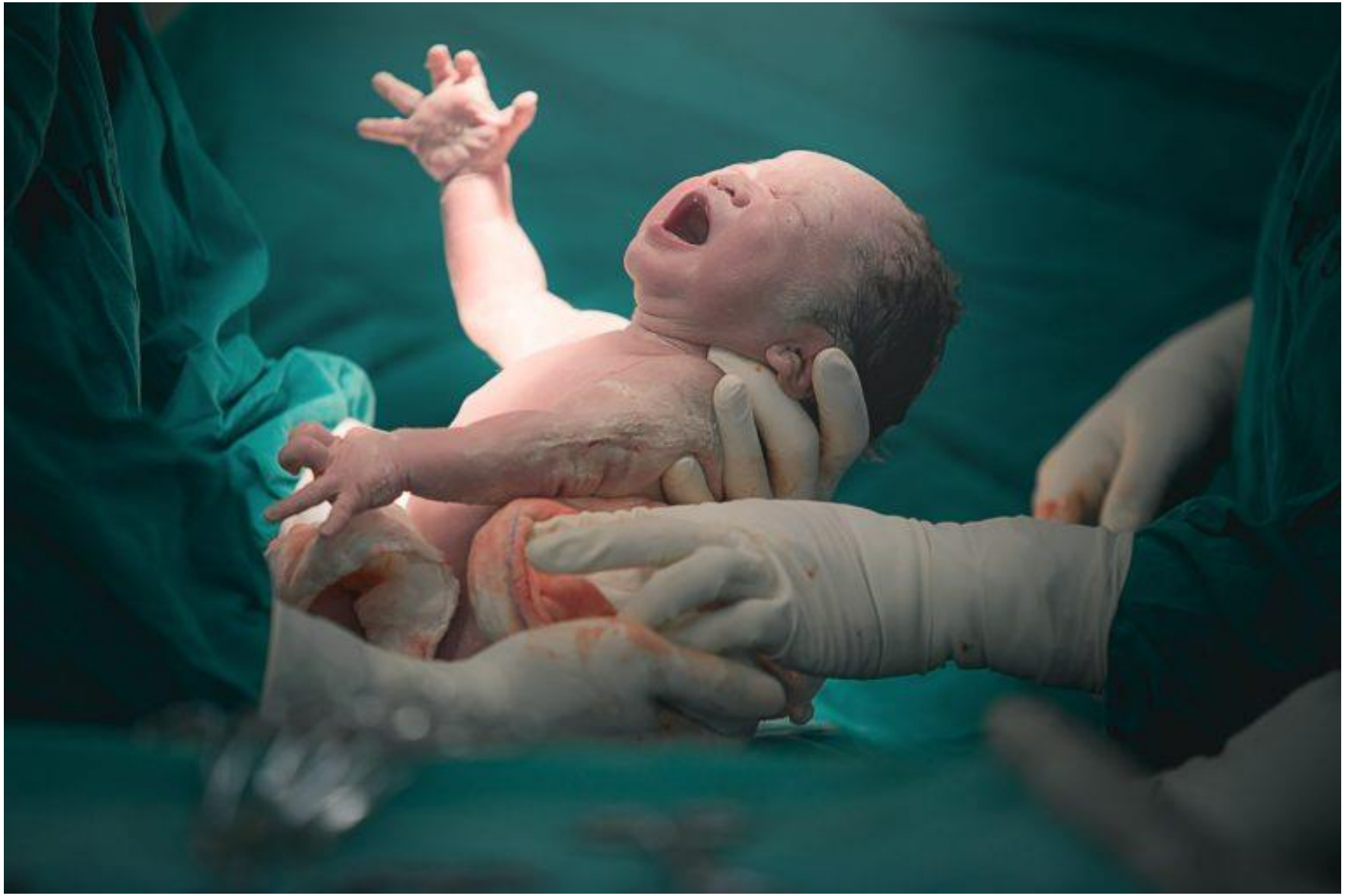


https://www.who.int/europe/publications/i/item/9789289057622

World Health Organization

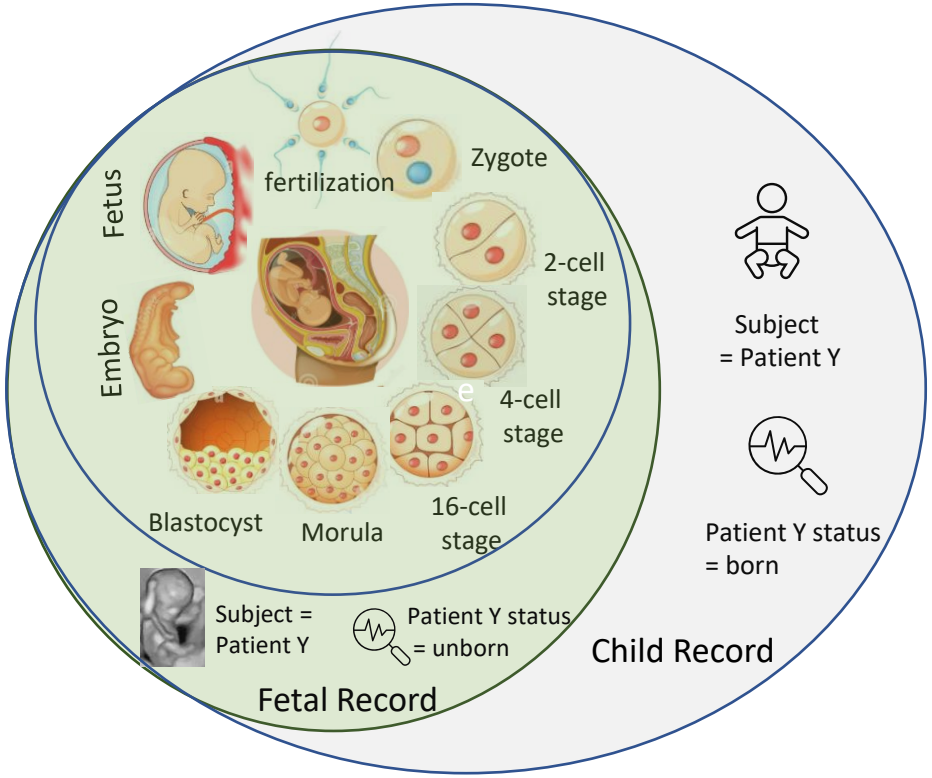


The health information system ensures the collection, analysis and use of data to ensure early, appropriate action to improve the care of every child



From : Biology Tutorials > Developmental Biology > Birth of a Human Baby

Child health record: Information requirements



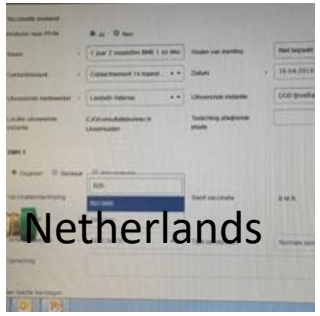
Note: the Australian Child Health Books is designed for health consumers. The contents are not intended for supporting clinical care. A different modelling approach will be required to support clinical care.
From CHOICE project (Child Health Obstretics International Collaboration and Exploration

Information requirements:

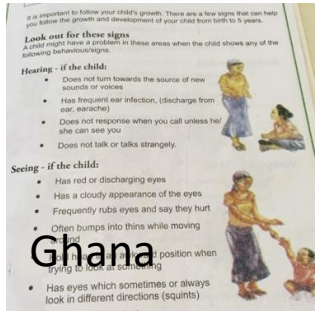
- Child: identifier and demographics
- Birth details (maternal, neonatal, APGAR score, discharge information …)
- Parent details and siblings
- Family health risk factors (e.g. deaf, eye low birth weight…)
- Growth charts
- Newborn checks: head, eyes, ears, mouth & palate, CVS, respiratory, abdomen and umbilicus, anus, genitalia, testes, Musculo-skeletal, hips, skin, reflex, other concern
- 1 – 4 weeks checks: health & safety (feeding, safe sleeping, growth (fontanelles, weight, height, head circumference), vaccination …); development (talking …), health assessment (eyes, CVS, etc) family (emotional health, smoking/alcohol/drugs …)
- 6 – 8 weeks checks: socio-emotional, language/comms, cognitive (learning, thinking, problem-solving), movement, developments …
- 4 months; 6 months; 12 months; 18 months; 2 years; 3 years; 4 years …

World Health Organization

Global Preventive Child Health Records



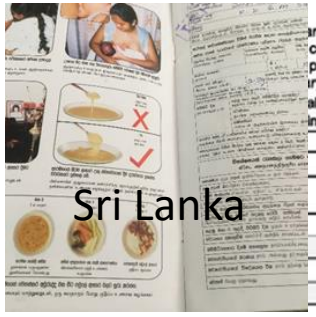
Netherlands



Ghana

Spain

Peru



Sri Lanka

Poland

cademy of Pediatrics
E 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.

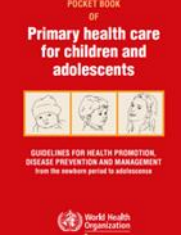
These guidelines represent a consensus by the American Academy of Pediatri
Bright Futures. The AAP continues to emphasize the great importance of conti
comprehensive health supervision and the need to avoid fragmentation of care
Refer to the specific guidance by age as listed in Bright Futures guidelines
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[a]	Newborn[a]	3-5 d[a]	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		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	

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



Case 1 Fetal Alcohol syndrome

Case History

WELL-CHILD VISIT: BIRTH – 72 HOURS

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
- Apgar score at 5 and 10 min of life (Table 5).

Two Month Old Girl at PCH Visit

Mother : Substance abuse (alcohol) prior and probably during pregnancy

Pregancy & **Fetus :** Delay in head growth by ultra sound observations at 28 weeks of pregnancy

Child at birth : Microcephaly at birth

At 2 months :

- Postnatal slowing of head growth
- Poor feeding and slow weight gain
- *Recognizable feature* - Small palpebral fissures, smooth philtrum and thin upper lip

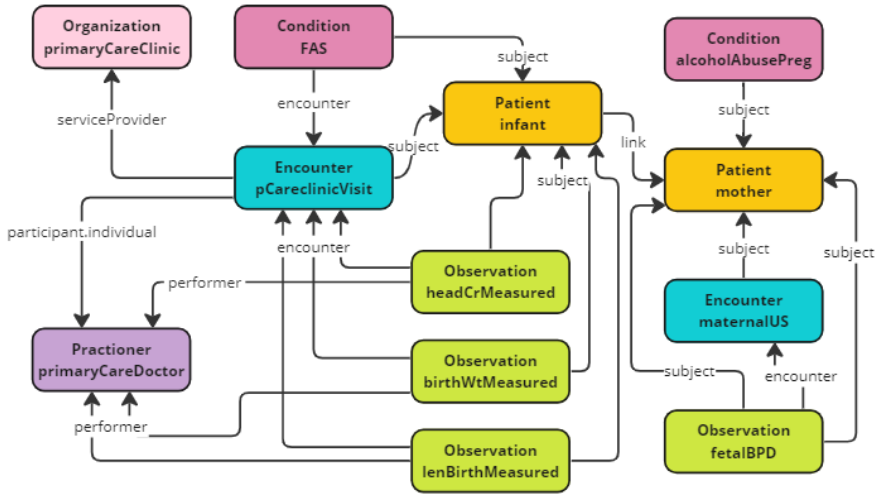
Probable Diagnosis: Fetal alcohol syndrome

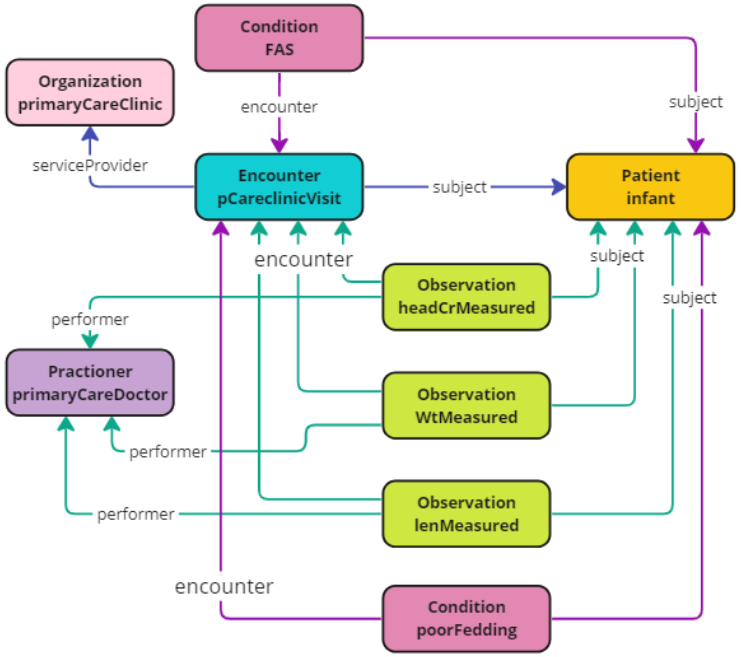
Ref https://www.mayoclinic.org/diseases-conditions/fetal-alcohol-syndrome/symptoms-causes/syc-20352901

Case 1 - Fetal Alcohol syndrome

Diagnosis in PMC

Maternal Record → Child Record - Primary Care

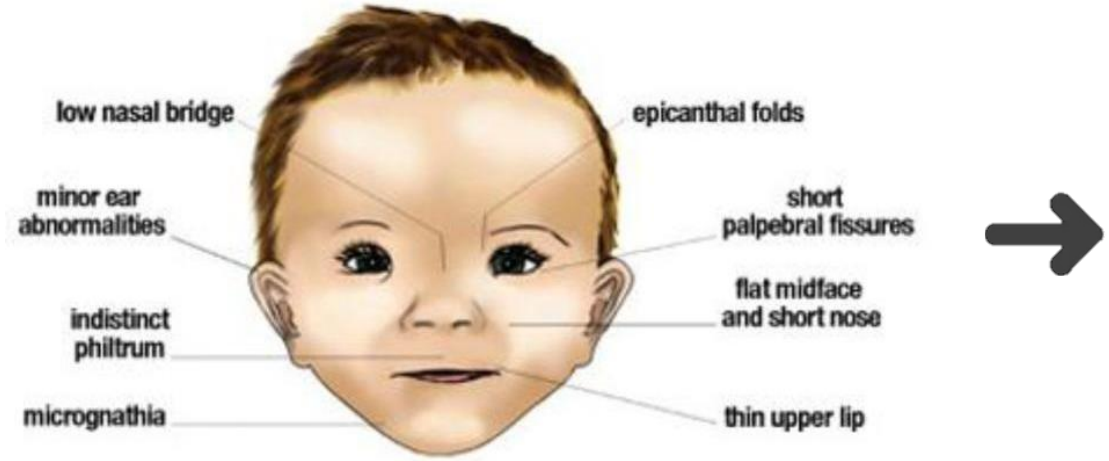




Terminologies

Fetal Head Diameter.biparietal US	LOINC	11820-8
Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
Birth weight Measured	LOINC	8339-4
Body height Measured --at birth	LOINC	89269-5
Body weight Measured	LOINC	3141-9
Body height Measured	LOINC	3137-7
Feeding disorder of infancy and childhood	IDC 10	F98.2
Alcohol Use Complicating Pregnancy	ICD 10-CM	O99.310
Fetal Alcohol Syndrome	ICD 10	Q86.0

Phenotype Recording



Newborn	Microcephaly	Human Phenotype Ontology	HP:0000252
	Depressed nasal bridge		HP:0005280
	Smooth philtrum		HP:0000319
	Micrognathia		HP:0000347
	Epicanthus		HP:0000286
	Short palpebral fissure		HP:0012745
	Midface retrusion		HP:0011800
	Short nose		HP:0003196
	Thin upper lip vermilion		HP:0000219
	Fetal Alcohol Syndrome	ICD10	Q86.0
Mother	Alcohol abuse	ICD10	F10.10

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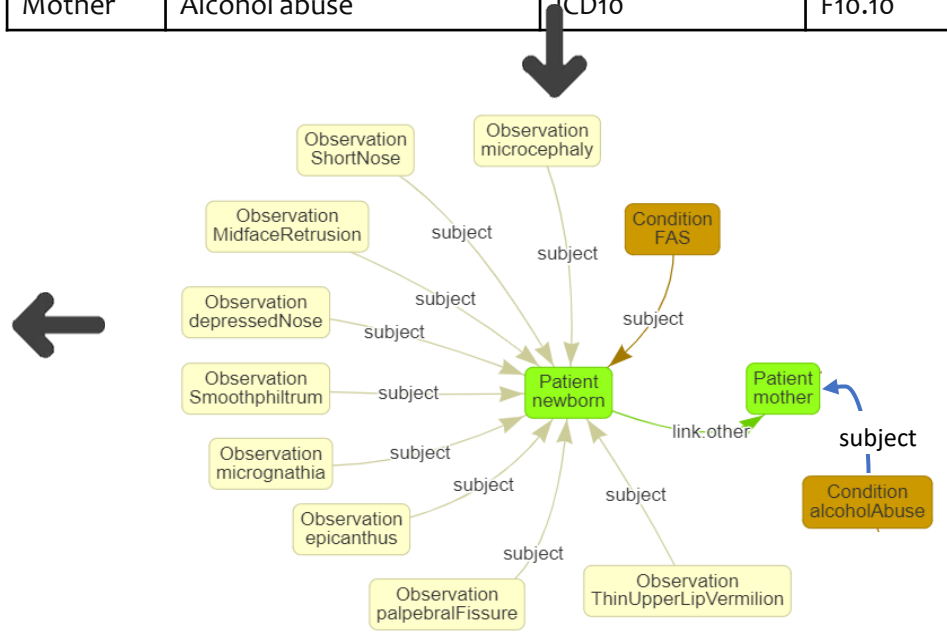
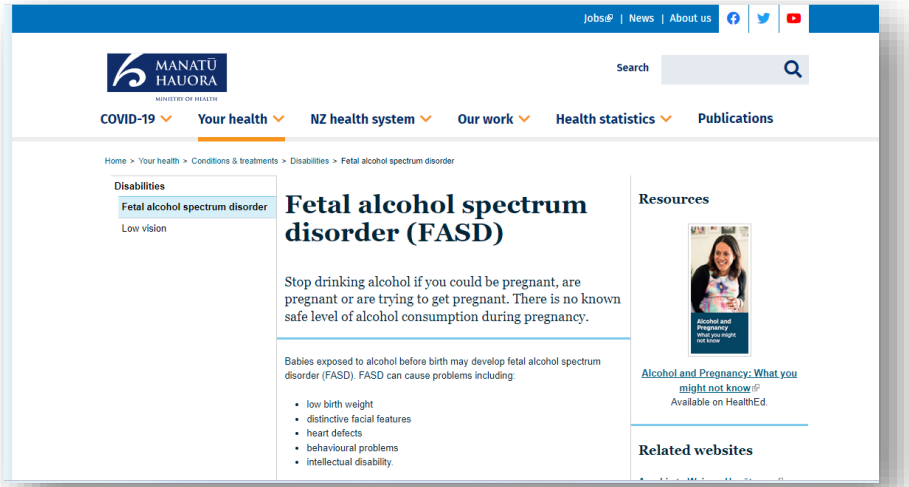
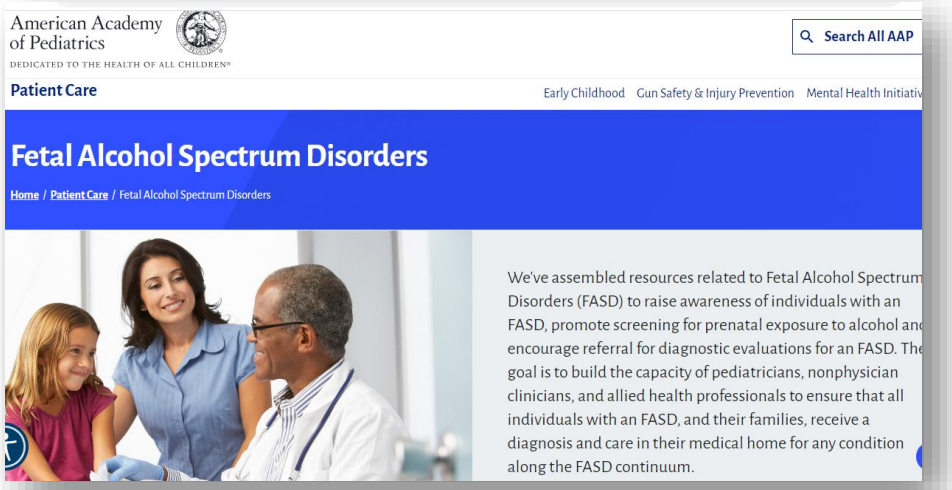
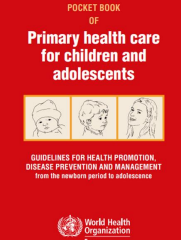


Fetal Alcohol syndrome







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

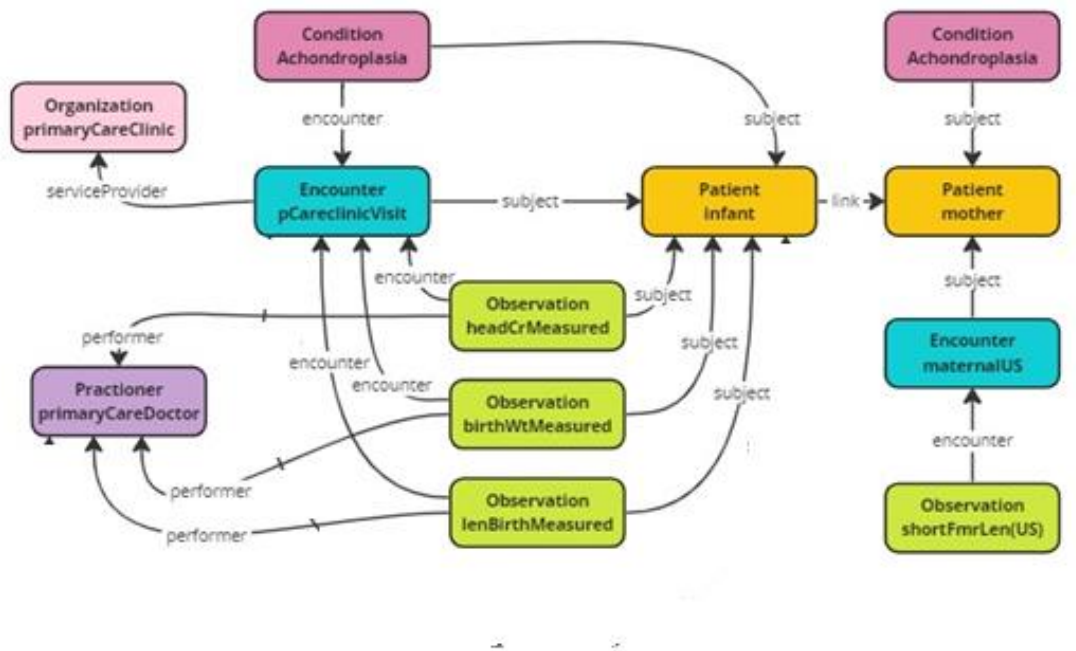
Case 2 - Maternal Achondroplasia

Management in Home

Date Flow

Maternal Record → Home Based Record

FHIR



Terminologies

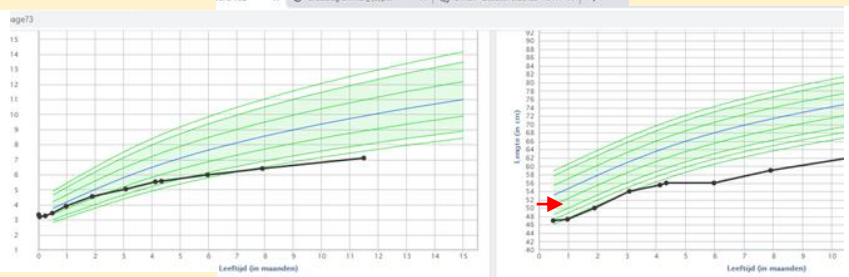
Femur Length US	LOINC	11963-6
Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
Birth weight Measured	LOINC	8339-4
Body height Measured --at birth	LOINC	89269-5
Achondroplasia	ICD 10	Q77.4

Maternal Achondroplasia

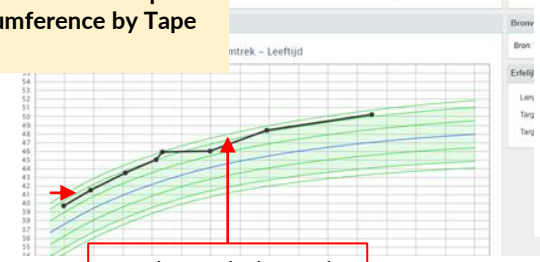
VOXZOGO™
(vosoritide) for injection

LOINC 29463-7 Body weight

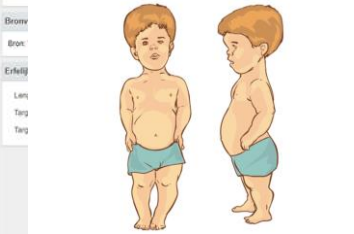
LOINC 8302-2 Body height



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



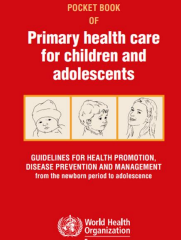
Hydrocephalus Risk



New Treatment

Achondroplasia-growth curve at each primary care visit

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



Case 3 – Birth with Congenital anomalies

Case History

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
- Apgar score at 5 and 10 min of life (Table 5).

Cleft lip and palate

- Refer for surgical closure. Closure of the lip can be done at 6 months and of the palate at 1 year of age. The lip may be repaired earlier if it is safe to give an anaesthetic and the repair is technically possible.
- Closely monitor feeding and growth. Babies with isolated cleft lip can feed normally, whereas cleft palate is associated with feeding difficulties.
- Provide feeding advice to the caregivers: feed with expressed breast milk from a cup and spoon or bottles; a special teat may be used. The technique of feeding is to deliver a bolus of milk over the back of the tongue into the pharynx with a spoon, pipette or some other pouring device. The baby will then swallow normally. Refer if feeding or weight gain is not satisfactory.
- Note that sleep-related upper airway obstruction can cause hypoxaemia and growth failure. If suspected, refer for specialist treatment.

Breech Delivery with Congenital Anomalies

Mother : Breech delivery

Child at birth : Birth Weight

Physical examination at 5 hours after birth :

- Congenital anomaly visible at birth: Cleft palate | Microtia

PCH rerefers to:

- Cleft palate team | Auditory screening (microtia risk of hearing deficit) | Genetic test: Genetic cleft lip palate | Ultrasound hip (risk hip Dysplasia)

PCH Information for Home based record : Feeding difficulty:

Feed with expressed breast milk from a cup and spoon or bottles; a special teat may be used. The technique of feeding is to deliver a bolus of milk over the back of the tongue into the pharynx with a spoon, pipette or some other pouring device. The baby will then swallow normally.

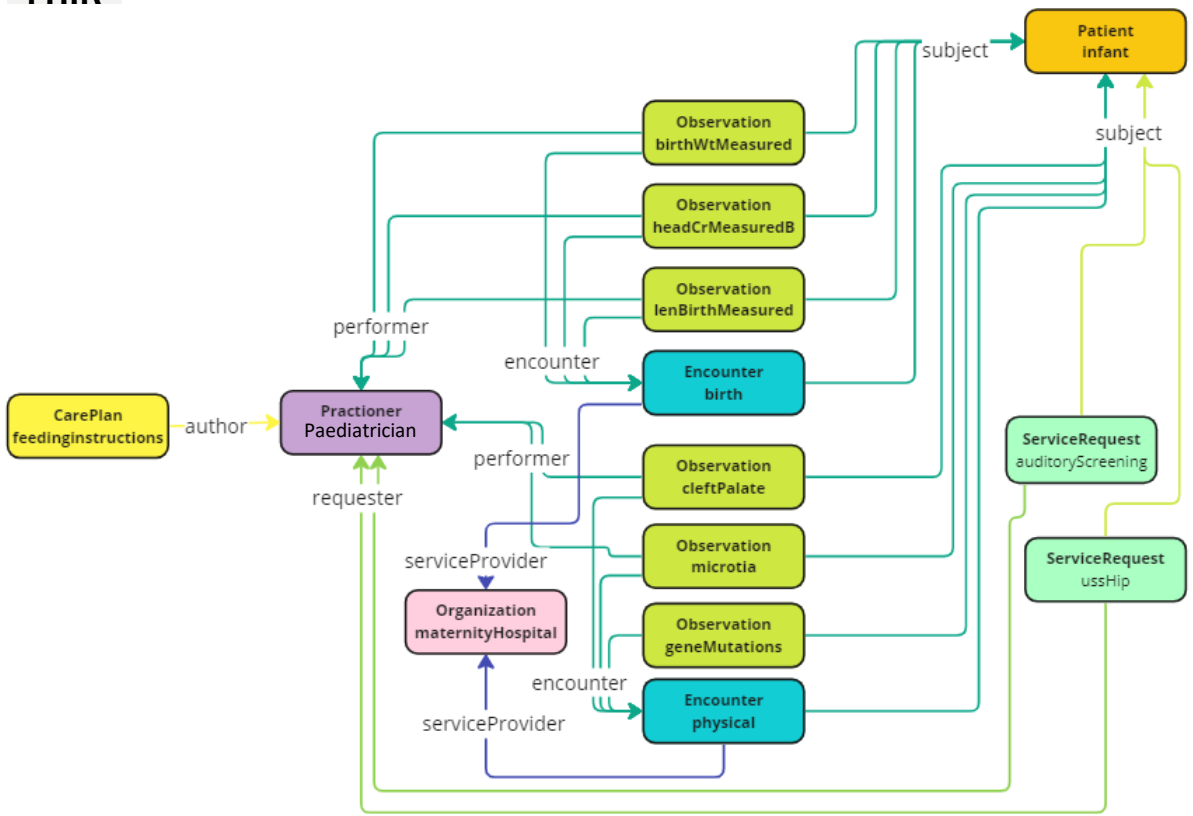
Case 3 - Breech-Cleft Palate-Microtia

Diagnosis

Date Flow

Hospital Record → Home Based Record

FHIR



Terminologies

Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
Birth weight Measured	LOINC	8339-4
Body height Measured --at birth	LOINC	89269-5
Gene studied [ID]	LOINC	48018-6
Breech Delivery	ICD 10	O80.1
Cleft Palate	ICD 10	Q35
Microtia	ICD 10	Q17.2
Feeding difficulties	ICF	d550

Assess to genetic testing

OMIM

612290

MICROTIA, HEARING IMPAIRMENT, AND CLEFT PALATE

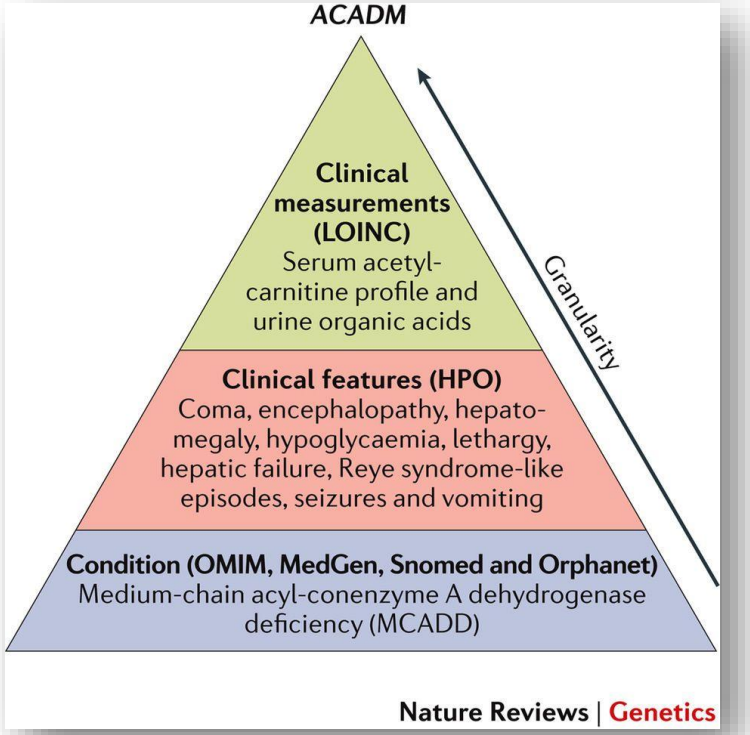
Other entities represented in this entry:

MICROTIA WITH OR WITHOUT HEARING IMPAIRMENT, INCLUDED

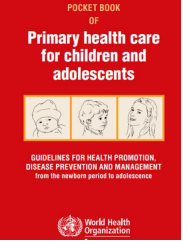
Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
7p15.2	?Microtia, hearing impairment, and cleft palate (AR)	612290	AD, AR	3	HOXA2	604685
7p15.2	Microtia with or without hearing impairment (AD)	612290	AD, AR	3	HOXA2	604685

https://www.omim.org/entry/612290



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



Case 4-Neonatal Jaundice & Hyperbilirubinaemia

Case History

NEONATAL JAUNDICE

Table 25. Bilirubin thresholds for management of babies ≥ 35 weeks' gestational age

Age	35 to < 38 weeks with risk factors	35 to < 38 weeks without risk factors; ≥ 38 with risk factors	≥ 38 weeks without risk factors
24 h	140 µmol/L (8 mg/dL)	170 µmol/L (10 mg/dL)	200 µmol/L (12 mg/dL)
48 h	190 µmol/L (11 mg/dL)	220 µmol/L (13 mg/dL)	260 µmol/L (15 mg/dL)
72 h	230 µmol/L (13.5 mg/dL)	260 µmol/L (15 mg/dL)	310 µmol/L (18 mg/dL)
96 h	250 µmol/L (14.5 mg/dL)	290 µmol/L (17 mg/dL)	340 µmol/L (20 mg/dL)
≥ 120 h	260 µmol/L (15 mg/dL)	310 µmol/L (18 mg/dL)	360 µmol/L (21 mg/dL)

Two Day Old Neonate

Mother : Pregnancy duration 36+2 weeks

Child at birth : Birth weight 2900 gram | Breast feeding

Physical examination : Jaundice

Laboratory test : Bilirubin

If the bilirubin is above the threshold (Table 25 from the book): refer urgently to hospital for phototherapy or exchange transfusion.

To Home Based record:

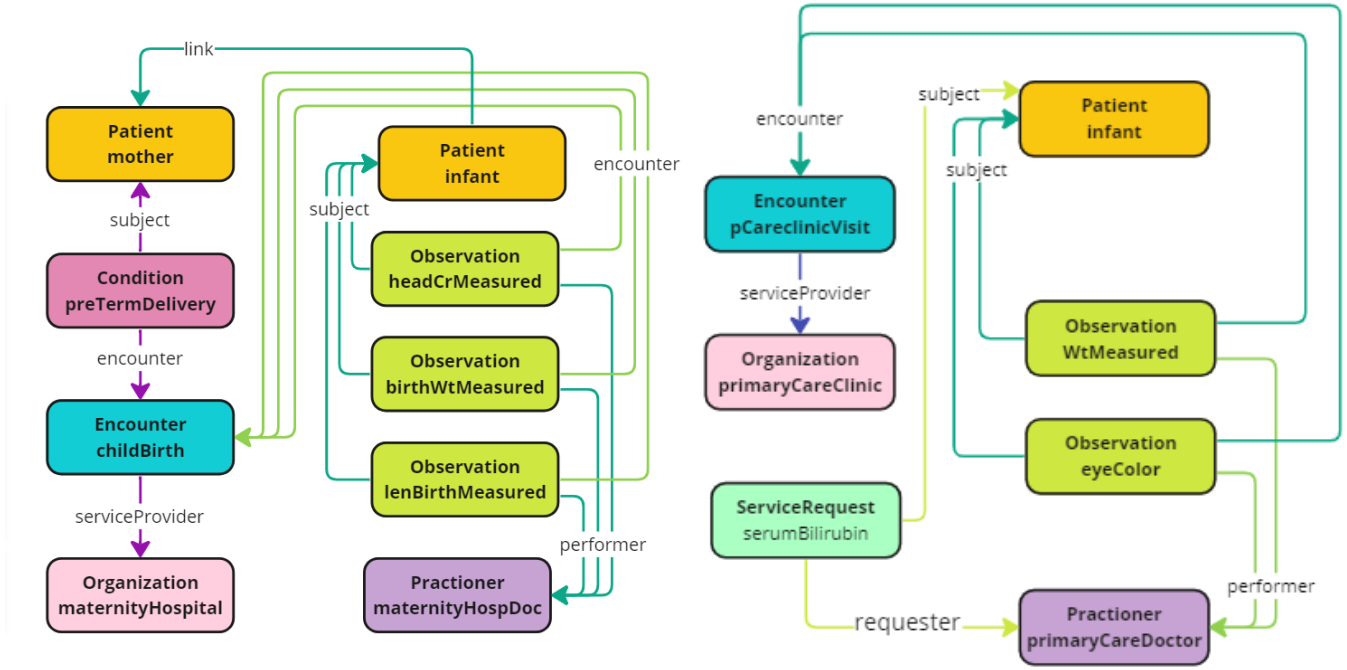
Counsel to continue breastfeeding to ensure adequate hydration and address breastfeeding problems, if needed

Case 4-Neonatal Jaundice & Hyperbilirubinaemia

Diagnosis

Date Flow

FHIR



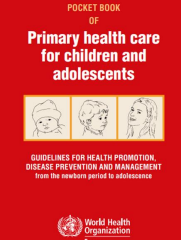
Terminologies

Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
Body height Measured --at birth	LOINC	89269-5
Body height Measured --at birth	LOINC	89269-5
Scleral icterus (finding)	SNOMED CT	246975001
Preterm spontaneous labour with preterm delivery	ICD 10	O60.1
Neonatal jaundice from other and unspecified causes	ICD 10	P59
Neonatal bilirubin panel [Mass/volume] - Serum or Plasma	LOINC	50189-0

The need for neonatal jaundice screening awareness in the Pakistani population: short communication

- Educating the mothers on screening for early detection of neonatal jaundice and seeking medical treatment in a country like Pakistan, which is considered a high-risk population, is crucial.
- Also, as most females give birth at home, hence, midwives' knowledge about neonatal jaundice also needs to be improved.

Naeem H, Ullah K, Ochani S, Naeem K, Ahmad HB, Hasibuzzaman MA. The need for neonatal jaundice screening awareness in the Pakistani population: short communication. Ann Med Surg (Lond). 2023 Jul 3;85(8):4187-4189. doi: 10.1097/MS9.0000000000000960. PMID: 37554868; PMCID: PMC10406009.



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 >

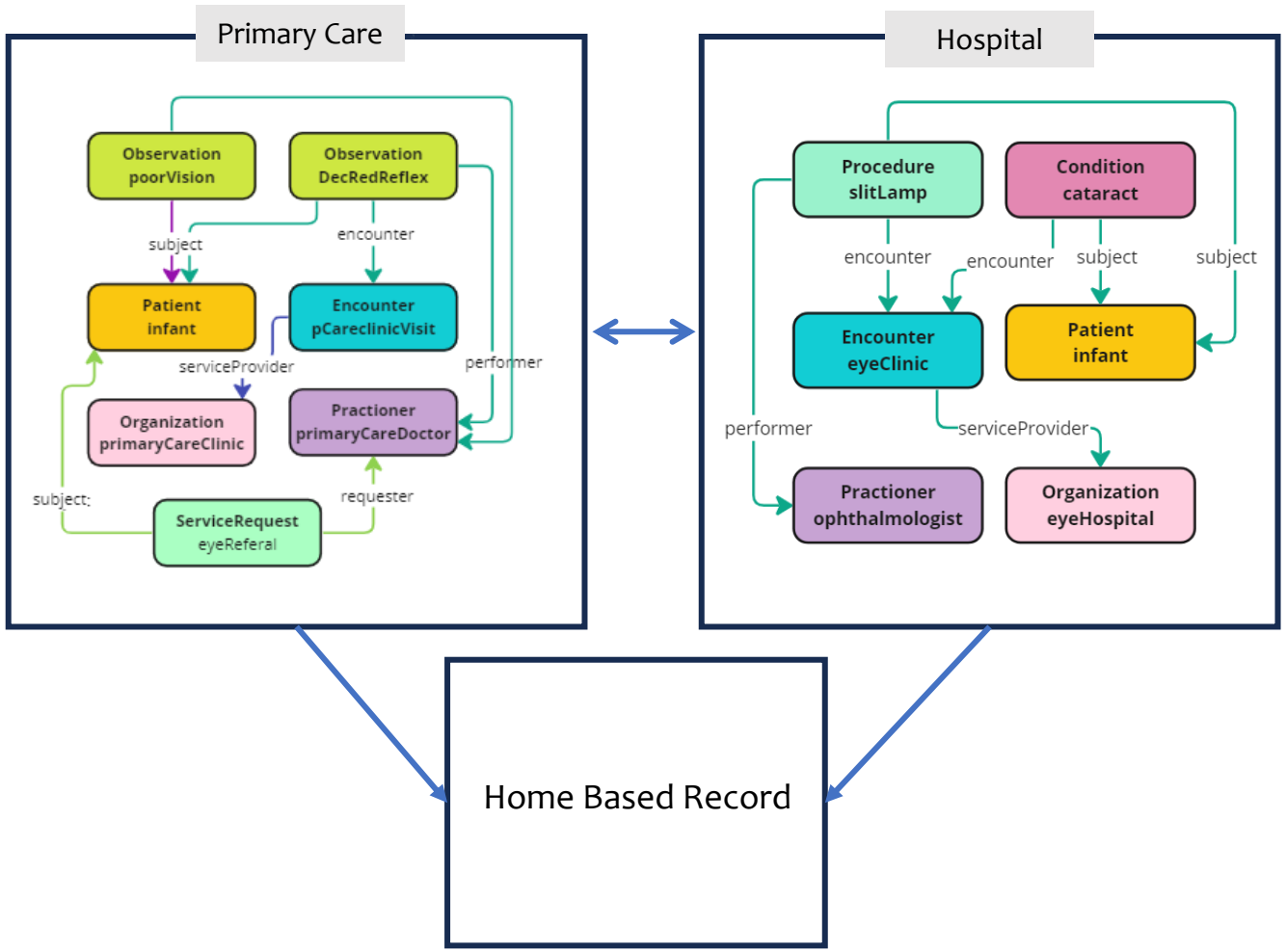
Diagnosis : Infantile cataract

Case 5- Juvenile Cataract

Diagnosis

Date Flow

Primary Care → Hospital Record → Home Based Record



Terminologies

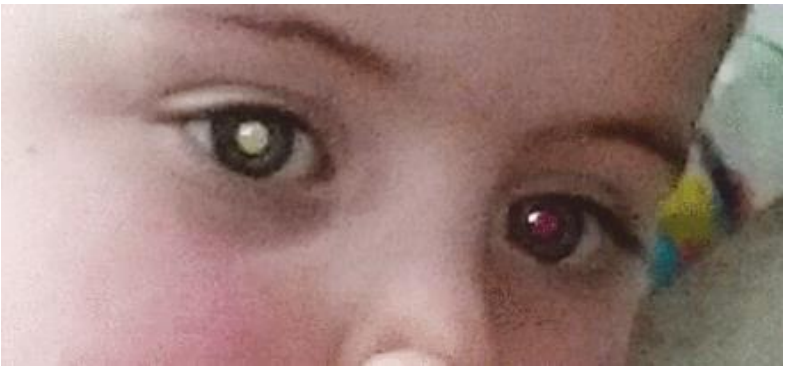
Red reflex absent	SNOMED CT	247079003
Abnormal vision	SNOMED CT	7973008
Study observation Left optic lens Slit lamp biomicroscopy	LOINC	79866-0
Infantile, juvenile and presenile cataract	ICD 10	H26.0

Visual impairment

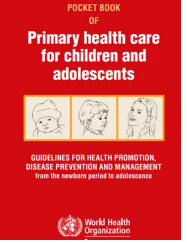
Infantile cataracts remain one of the **most treatable causes of lifelong visual impairment**.

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.



Case 6 - 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**

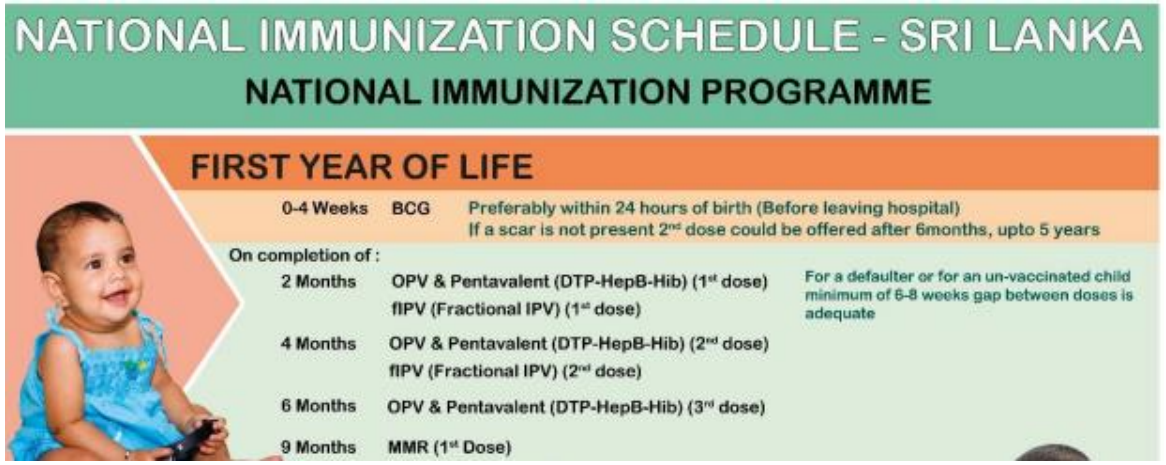
Laboratory test : Hemoglobine | Microcosis red blood cells

Referal to Thalassemia clinic

Parents are advised about routine vaccinations

Cascade Screening of Family

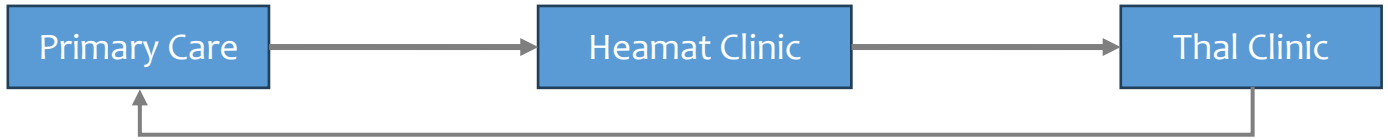
Diagnosis : Beta Thalassemia



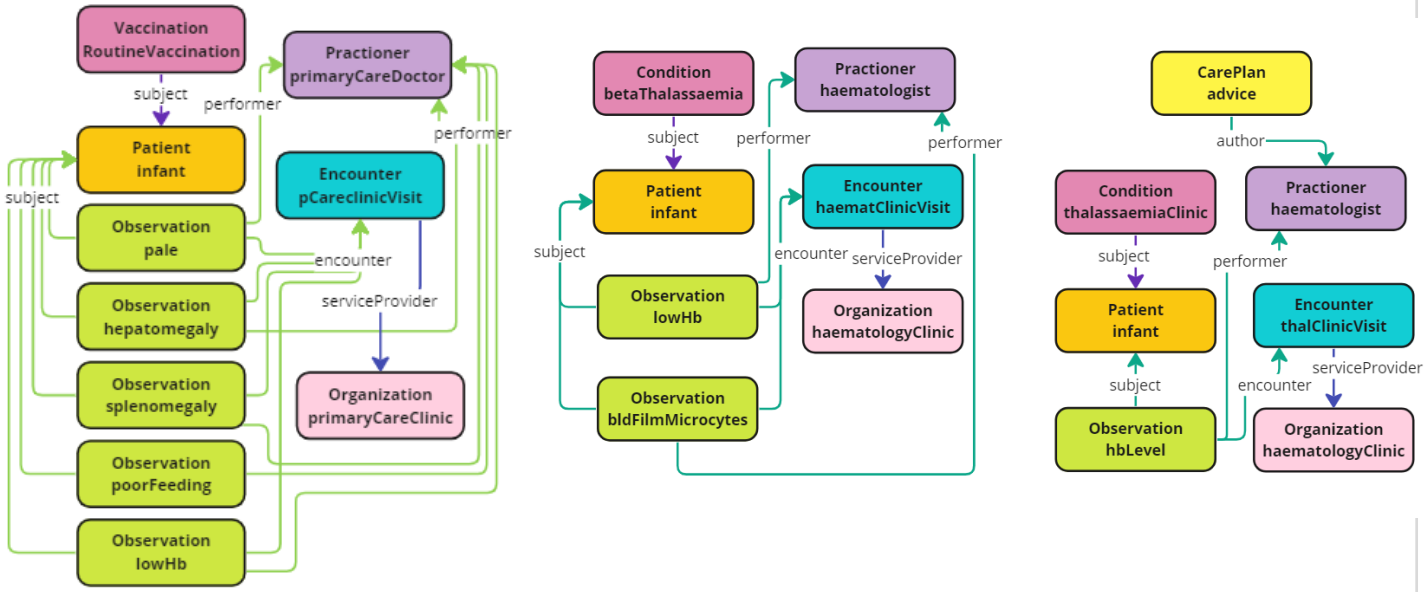
Case6 - Beta Thalassemia

Diagnosis

Date Flow



FHIR

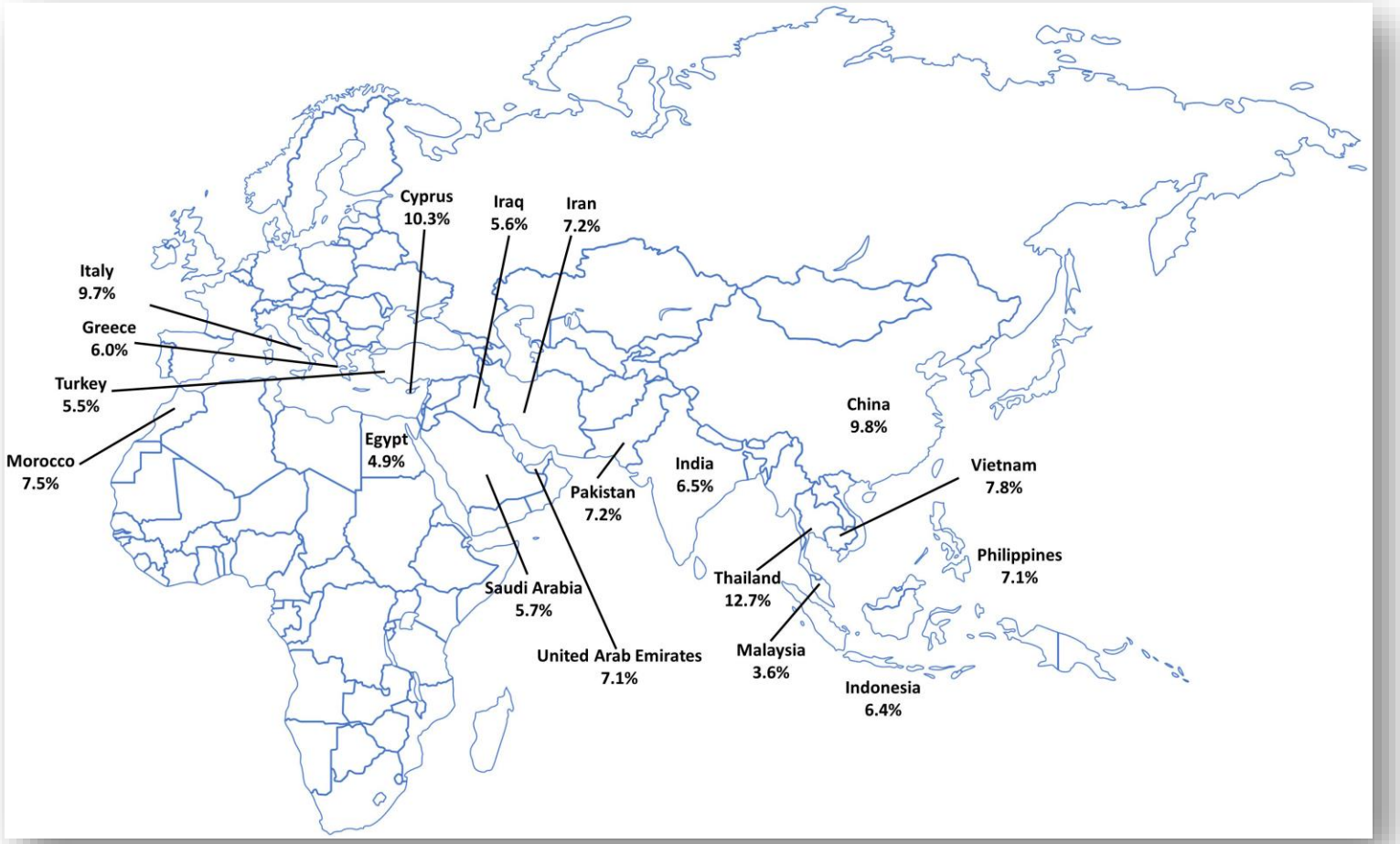


Terminologies

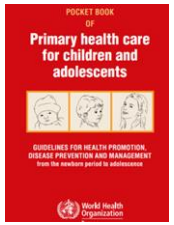
diphtheria-poliomyelitis-tetanus	ATC	J07CA01
Pallor	SNOMED CT	1237486008
Hepatomegaly	SNOMED CT	80515008
Splenomegaly	SNOMED CT	16294009
Haemoglobin concentration in blood	LOINC	718-7
Microcytes in blood film	LOINC	741-9
Feeding disorder of infancy and childhood	IDC 10	F98.2
Beta Thalassaemia	ICD 10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

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



Case 7 – Down Syndrome

Case History

7.5 Down syndrome

Down syndrome (trisomy 21) is the most common genetic syndrome. It is due to extra genetic material from chromosome 21 and associated with increased maternal age. Down syndrome causes a distinct facial appearance and, in most cases, early developmental problems and intellectual disability. It may be associated with thyroid or heart disease.

associations and websites.

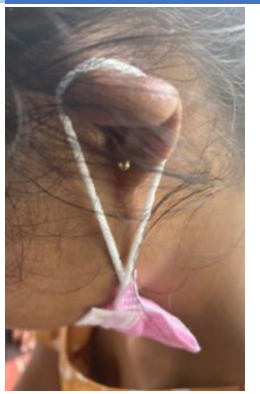
Immunization

- Provide immunizations as per local routine immunization schedule (p. 69).

Monitoring

- Monitor for associated problems or conditions:
  - Hearing and vision problems (p. 80)
  - Duodenal atresia
  - Hypogonadism in male patients
  - Congenital heart disease (p. 159)
  - Hypothyroidism
  - Coeliac disease
  - Acute myeloid leukaemia
  - Immunodeficiency (bacterial or fungal infections)
  - Psoriasis and eczema.

574



Diagnosis : Down Syndrome

2 year old girl in Ingiriya, Sri Lanka

Diagnosed with **Down Syndrome**
ICD Q90. 9
The mother knows about the international Down syndrome quideline through her **facebook group**

she asks the primary care physision to check for thyriod functions

ICD E03.9 – **Hypothyroidism**
LOINC 3015-5 Thyrotropin [Units/volume] in Blood
LOINC 32215-6 Thyroxine (T4) free index in Serum or Plasma by calculation Active Component

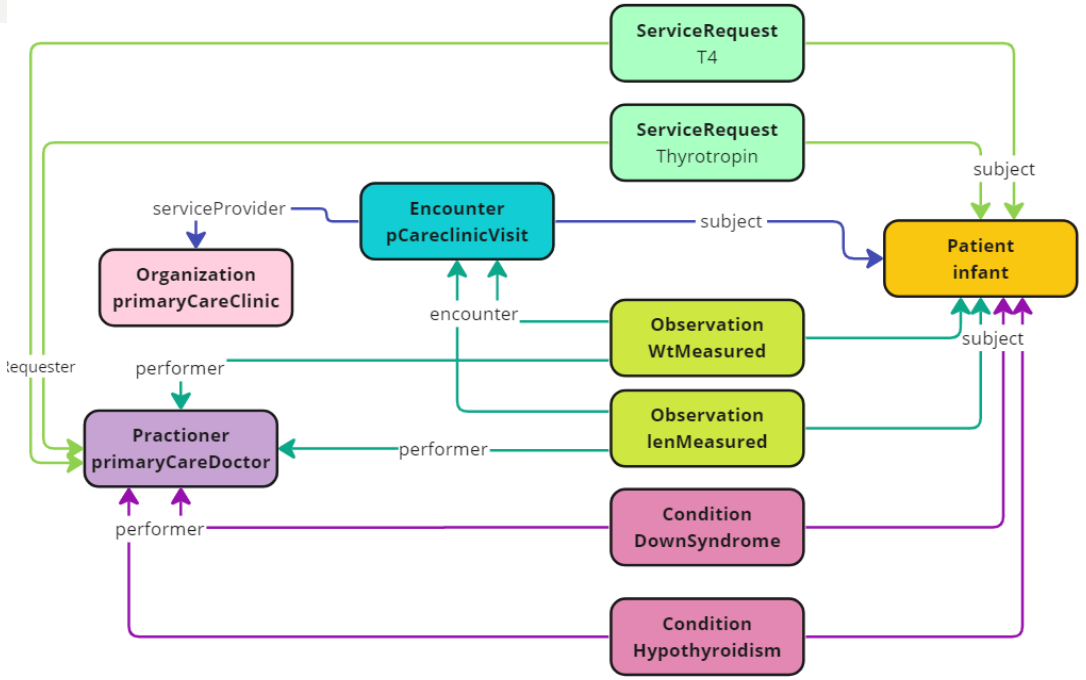
Case 7 – Down Syndrome

Diagnosis

Date Flow

Primary Care → Home Based Record

FHIR



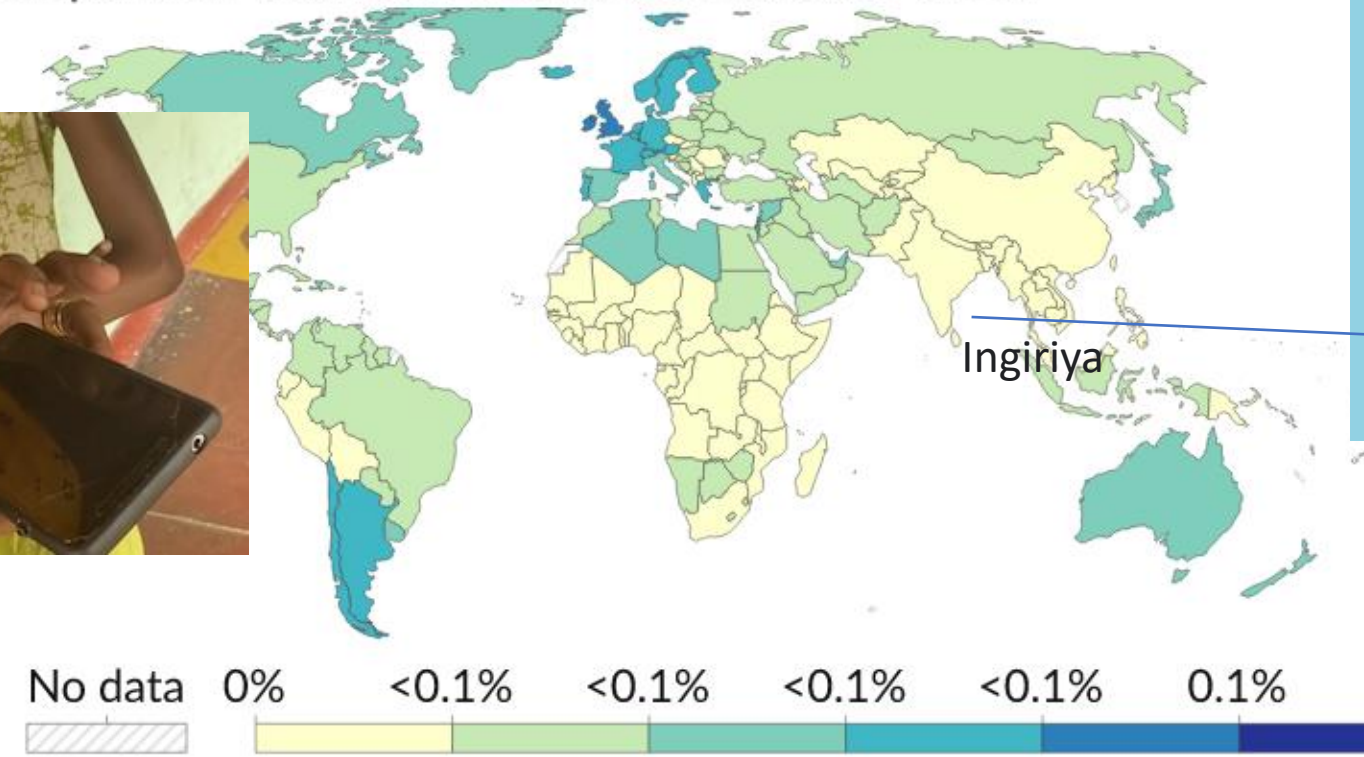
Terminologies

Thyroxine (T4)	LOINC	**32215-6**
Thyrotropin [Units/volume] in Blood	LOINC	3015-5
Body height Measured	LOINC	3141-9
Body Height Measures	LOINC	3137-7
Hypothyroidism	IDC 10	E03.9
Down Syndrome	ICD 10	Q90.0

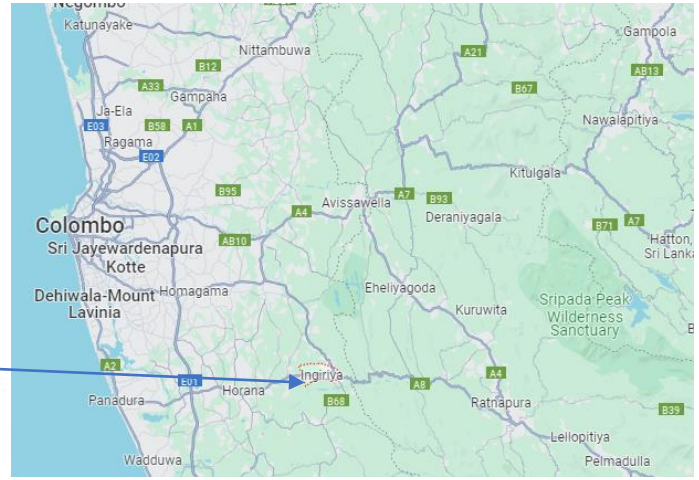
Share of the population with down syndrome, 2019

Our World in Data

Share of the population with down syndrome, measured as the age-standardized prevalence for comparison between countries and over time.



Ingiriya



Data source: IHME, Global Burden of Disease (2019) OurWorldInData.org/burden-of-disease | CC BY

https://ourworldindata.org/grapher/share-with-down-syndrome

From Feature to Medical Guideline

Feature

- Fatty Stool
- Growth Retardation
- Common infections

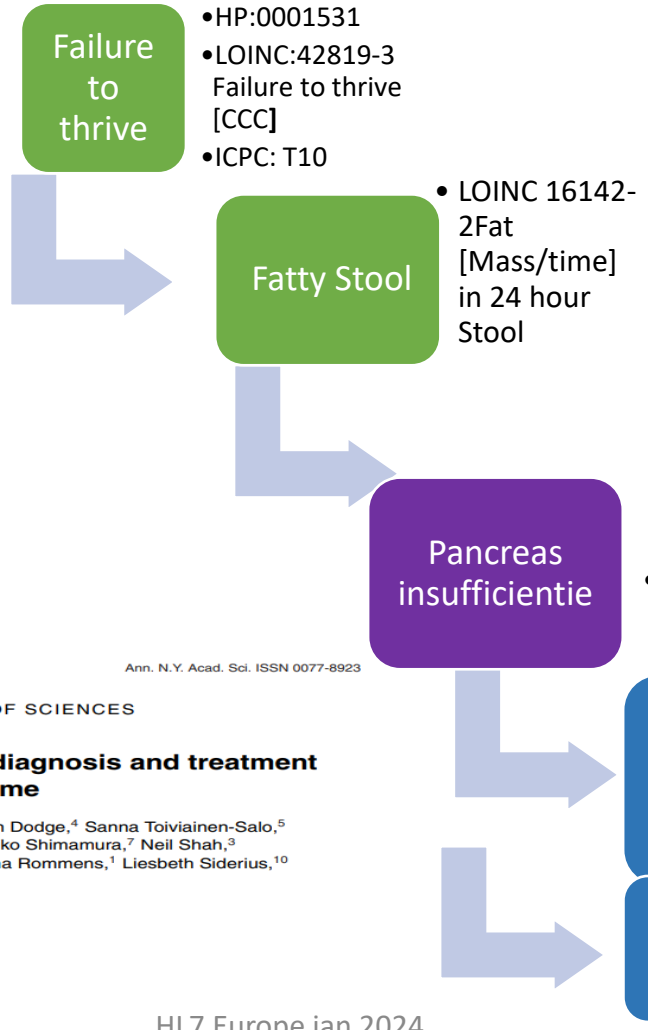
Shwachman

Diamond

Syndrome-

Management

- Pancreas insufficiency
- Neutropenia
- Skeletal Dysplasia
- Autisme like



2022

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

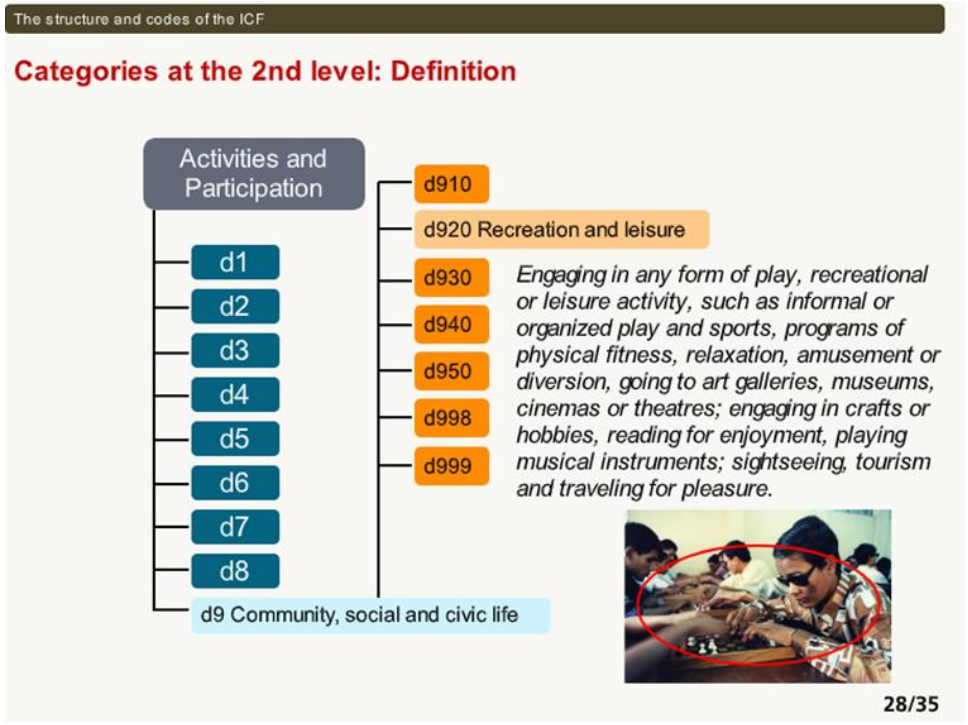
ANNALS OF THE NEW YORK ACADEMY OF SCIENCES
Issue: Annals *Meeting Reports*

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



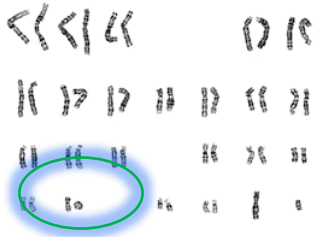
Indian Mother and Childcare
Kolkata, 2020

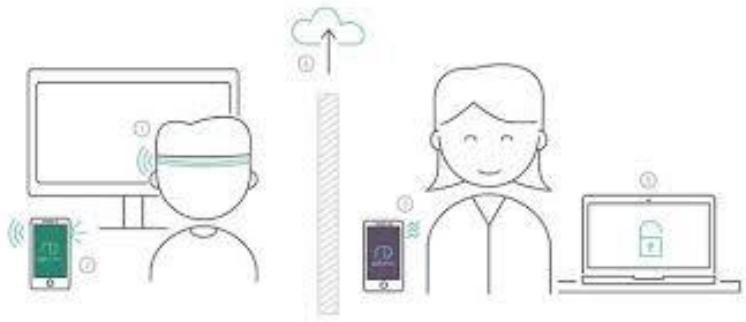
Epilepsy – assistive products- Health Technology Assesment

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)

Universal Health Coverage

What can we do, together?

Digital Health Europe

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

DigitalHealthEurope recommendations on the European Health Data Space

3 May 2022

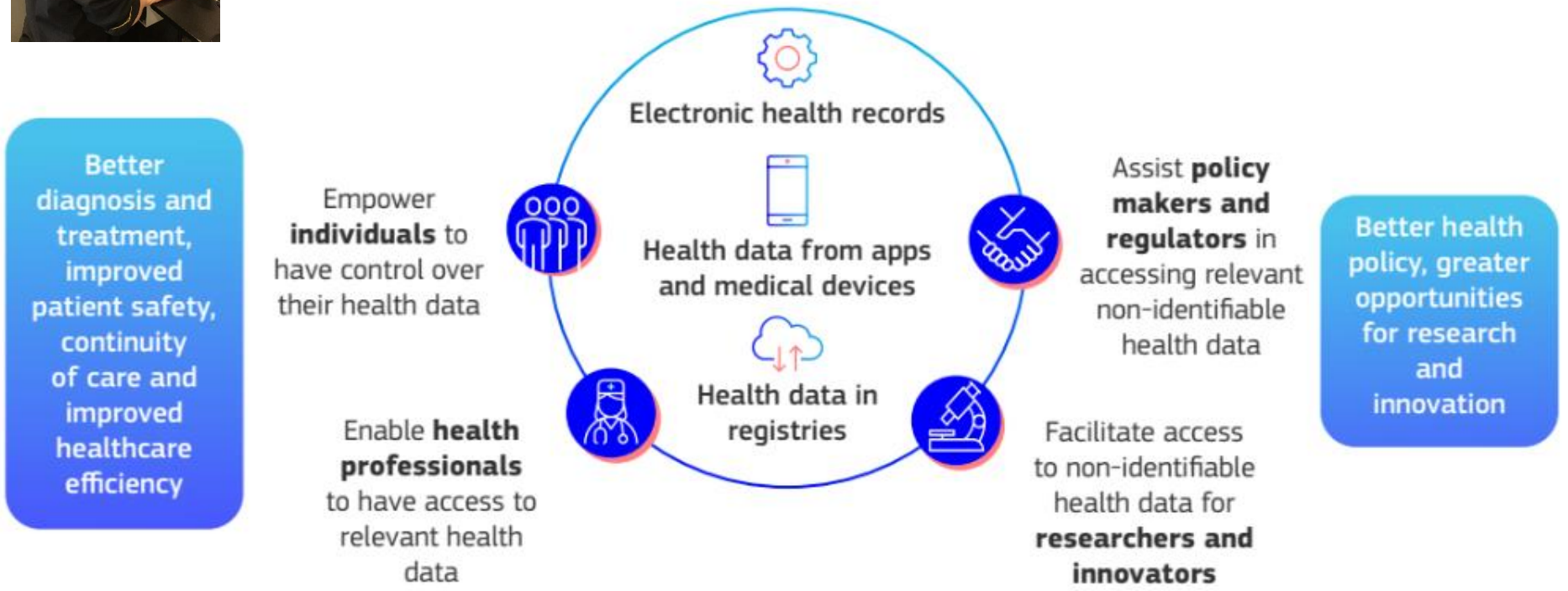
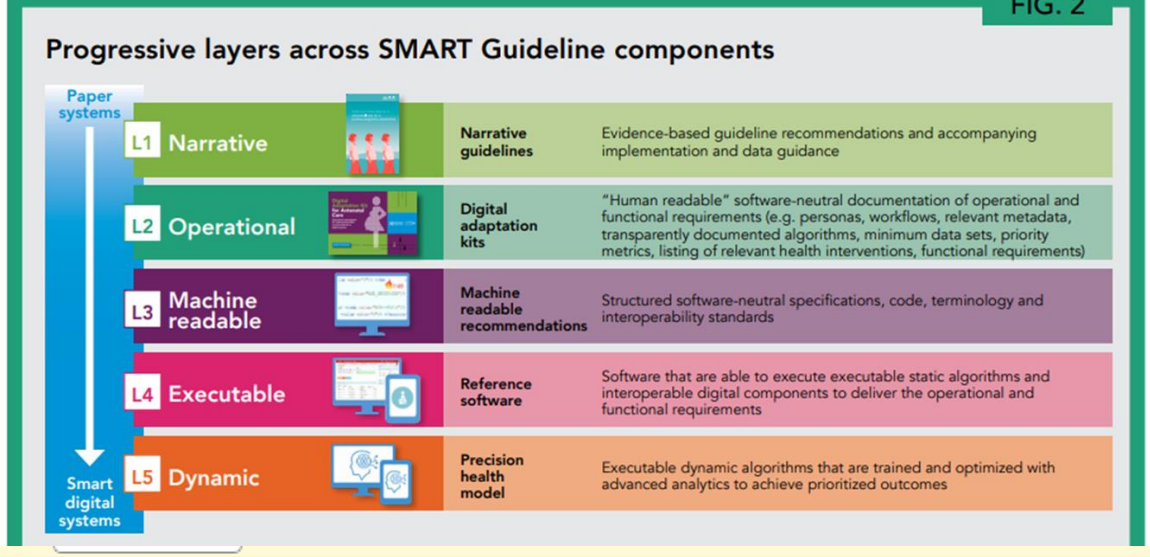


FIG. 2

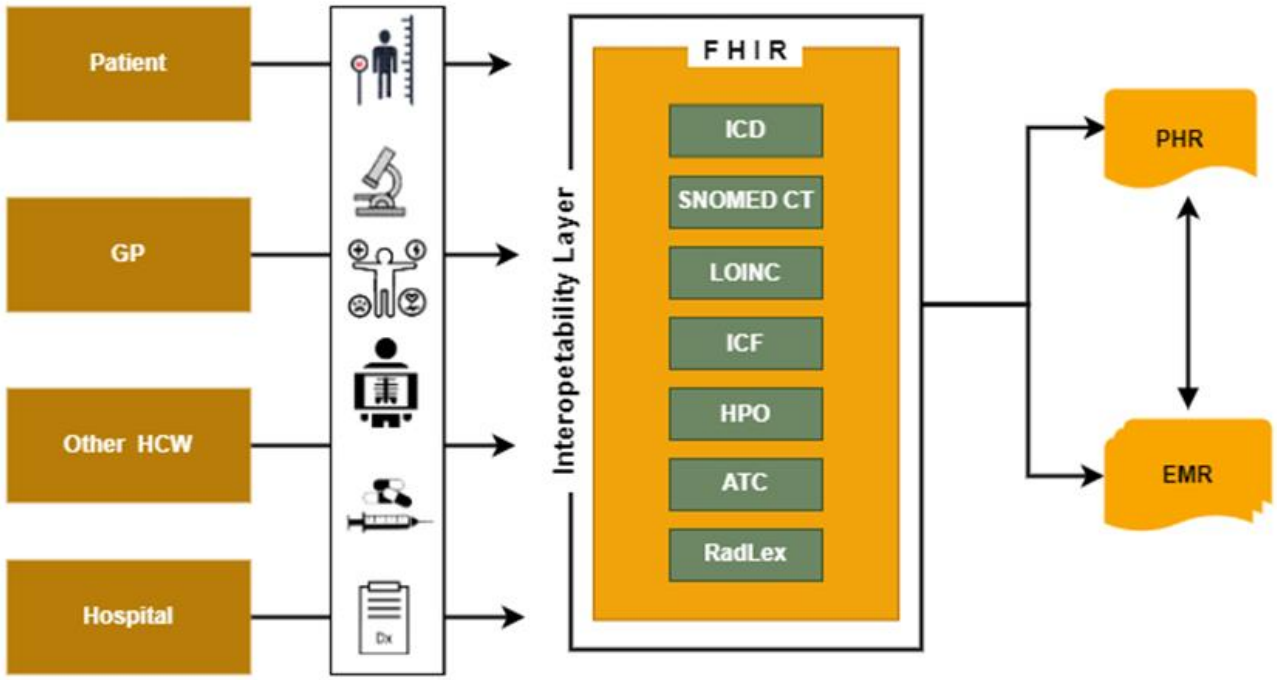


J145 | N/A

	A	B	Q	R	S	T	U	V	W	X
1		[ANC] Activity ID	ICD-11 Code	ICD-11 URI	ICD-11 Comments / Considerations	ICD-10 Code	ICD-10 Comments / Considerations	LOINC version 2.68 Code	LOINC version 2.67 Comments / Considerations	ICHI (Beta 3) Code
46		**ANC.B6. Collect woman's profile and history**	JA25.3	http://id.who.int/icd/entity/250375350	Code title: Eclampsia, time period unspecified	O15.9	Code title: Eclampsia, unspecified as to time period	58297-3	Code LongName: Diagnosis of high blood pressure during pregnancy only	Not classifiab ICHI
47		**ANC.B6. Collect woman's profile and history**	QE10	http://id.who.int/icd/entity/499098434	Code title: Hazardous alcohol use	Z72.1	Code title: Alcohol use	74205-6	Code LongName: Alcohol use [NTDS]	Not classifiab ICHI
48		**ANC.B6. Collect woman's profile and history**	KD3B.Z	Parent URI for residual code KD3B.Z http://id.who.int/icd/entity/861066692	Code title: Unspecified time of fetal death, cause not specified	P95	Code title: Fetal death of unspecified cause	Not classifiable in LOINC		Not classifiab ICHI
		ANC.B6. Collect woman's profile and history	8A68.Z	Parent URI for residual code 8A68.Z	Code title: Type of seizure, unspecified	R56.8	Code title: Other and unspecified convulsions	45662-4	Code LongName: Seizure disorder [Minimum Data Set] Best fit	Not classifiab ICHI

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

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

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

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.

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Acknowledgements