DIGITAL MODELLING OF PRIMARY CHILD HEALTH

FIRST 1000 DAYS

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Digital Modelling of Primary Child Health

Aim

- Identifying **pertinent child health data** points suitable for collection by parents and primary care providers.
- Developing **appropriate terminologies** for coding the collected data to ensure seamless interoperability between primary care and home-based records.
- Constructing an implementation guide adhering to the **FHIR standards** specifically for children during their formative years.

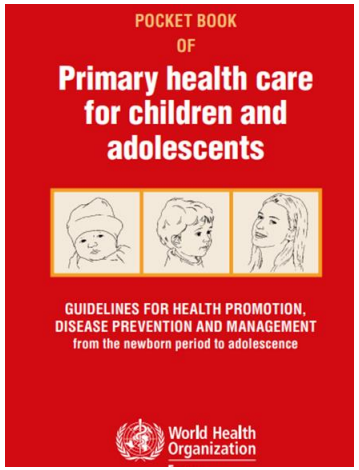
Method

An analysis of **WHO's Pocket-Book on Primary Health Care for children and adolescents** and recommendations on **home-based records** for maternal, newborn, and child health is conducted to ascertain relevant data points.

identify a **set of suitable terminologies** for data coding and explore the **FHIR** framework's standardization.

Digital Modelling of Primary Child Health

World Health Organization



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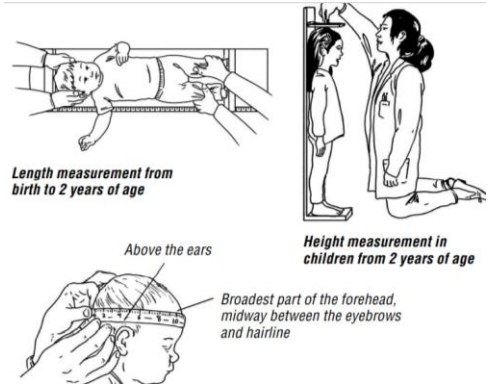
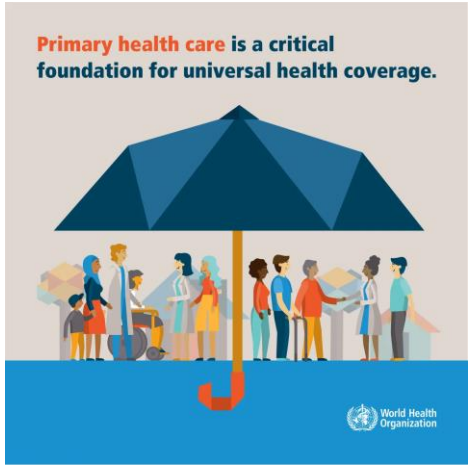




Digital Modelling of Primary Child Health

- The **LOINC** has a specific value in child health data since **numeric data** such as height, weight, head circumference measurements, and development can be followed in **time and place**.

- Six 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.

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

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

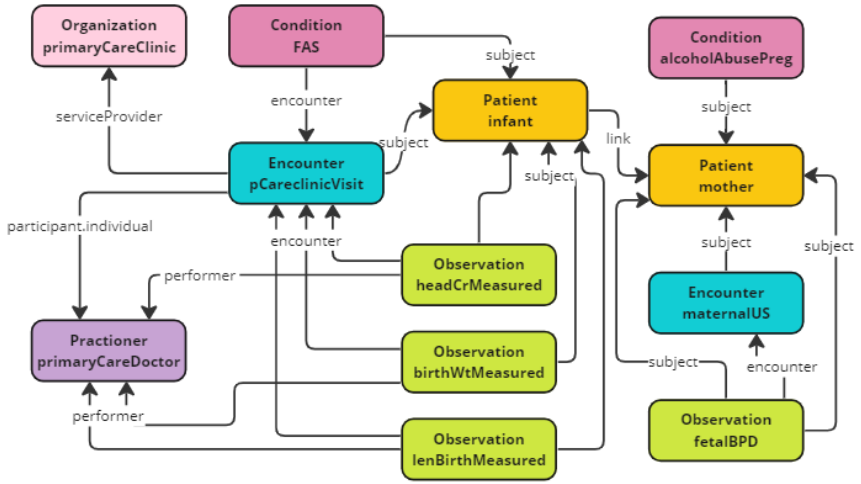
Probable Diagnosis: Fetal alcohol syndrome

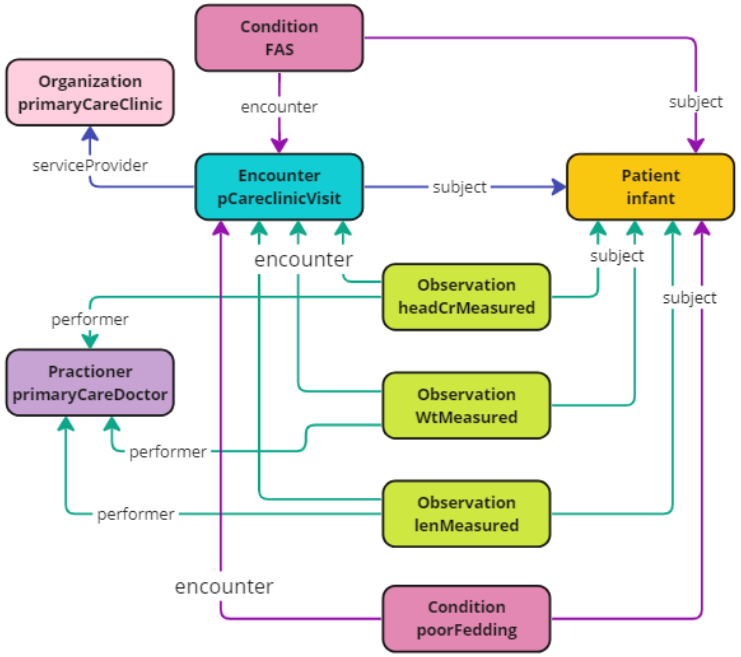
Ref https://www.mayoclinic.org/diseases-conditions/fetal-alcohol-syndrome/symptoms-causes/syc-20352901

Case 1 - Fetal Alcohol syndrome

Diagnosis in PMC

Maternal Record → Child Record - Primary Care

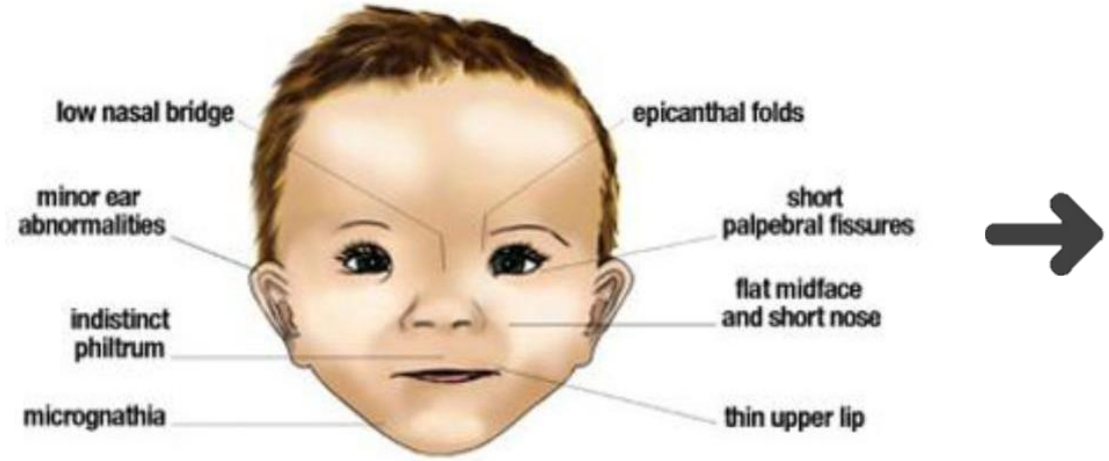




Terminologies

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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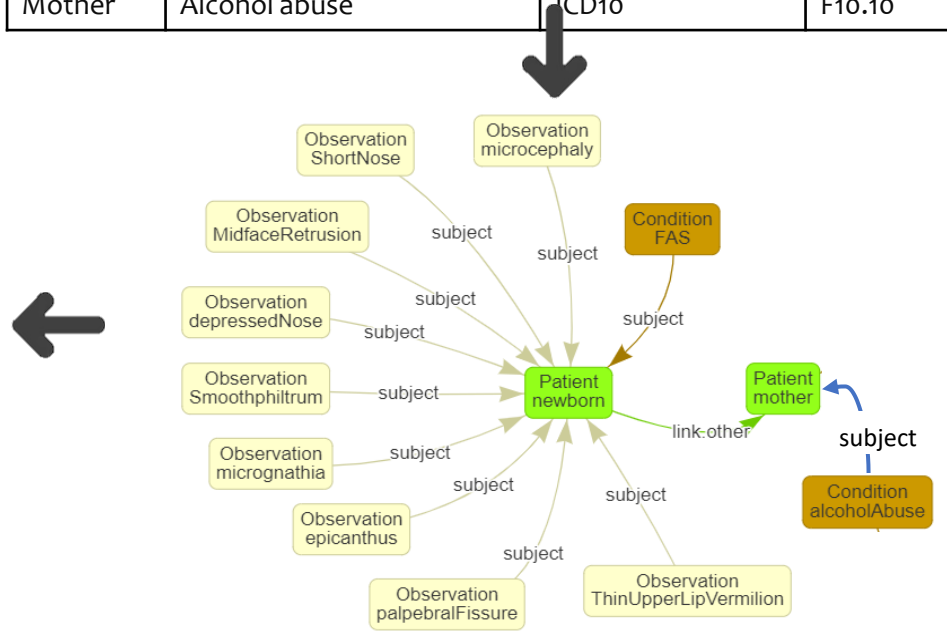
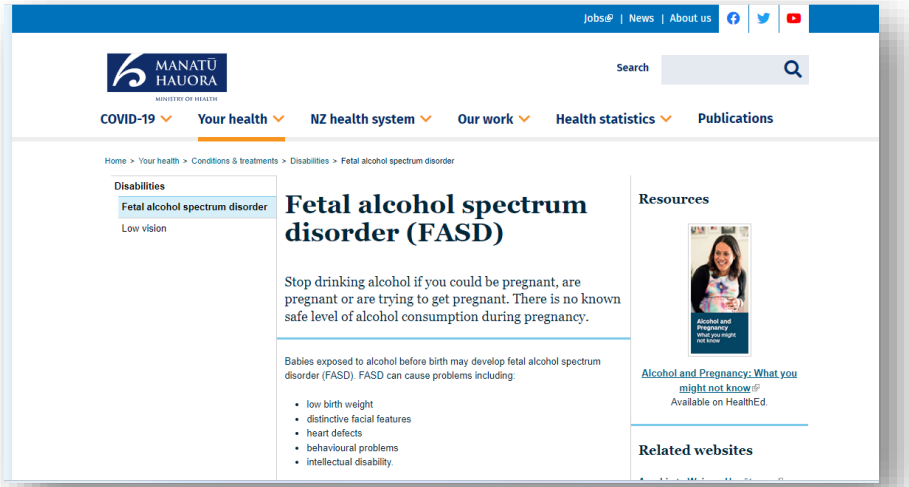
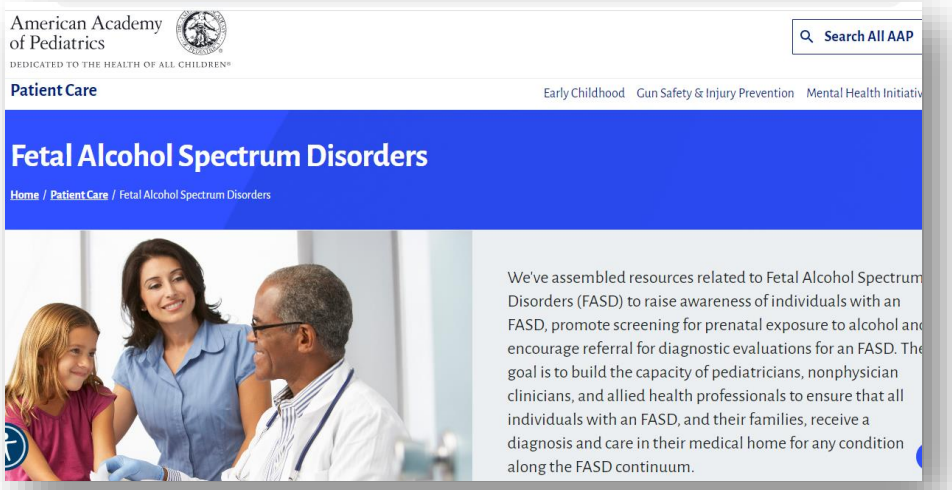


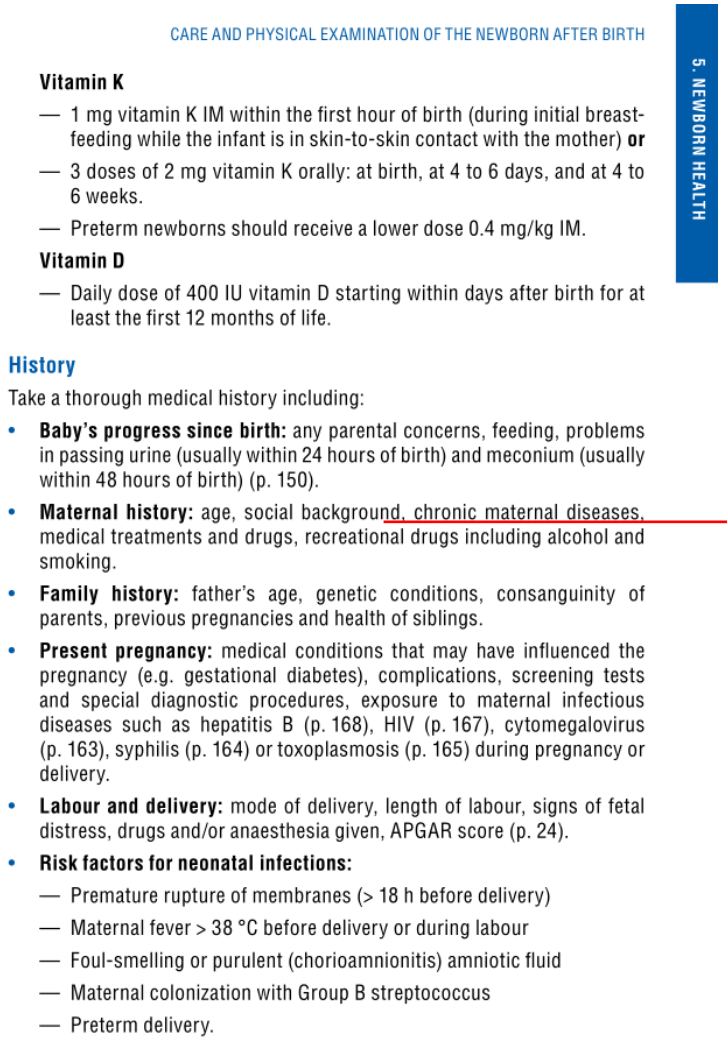
Fetal Alcohol syndrome





Case 2 - Maternal Achondroplasia

Case History



CARE AND PHYSICAL EXAMINATION OF THE NEWBORN AFTER BIRTH

5. NEWBORN HEALTH

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.

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

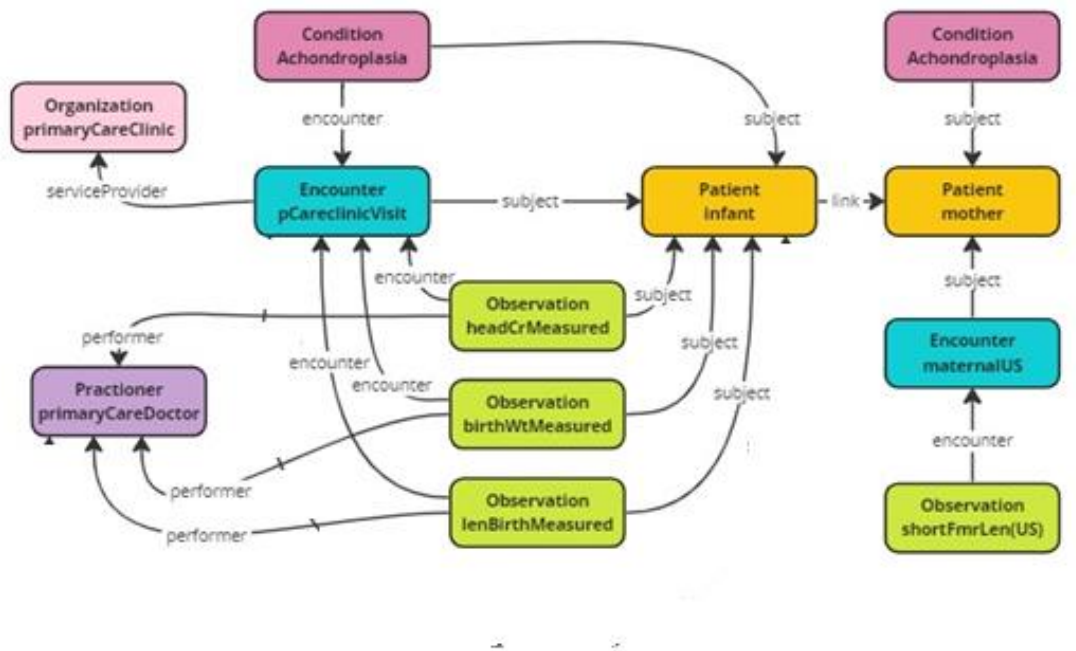
Case 2 - Maternal Achondroplasia

Management in Home

Date Flow

Maternal Record → Home Based Record

FHIR



Terminologies

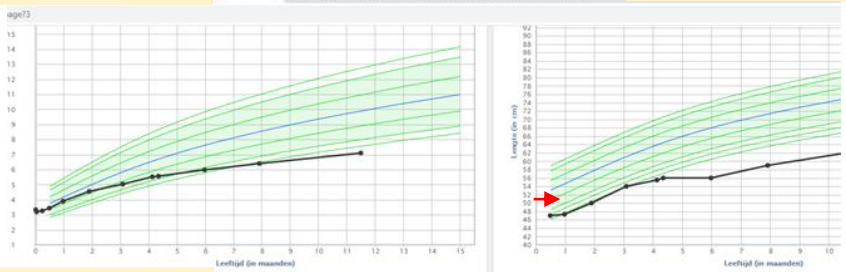
Femur Length US	LOINC	11963-6
Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
Birth weight Measured	LOINC	8339-4
Body height Measured --at birth	LOINC	89269-5
Achondroplasia	ICD 10	Q77.4

Maternal Achondroplasia

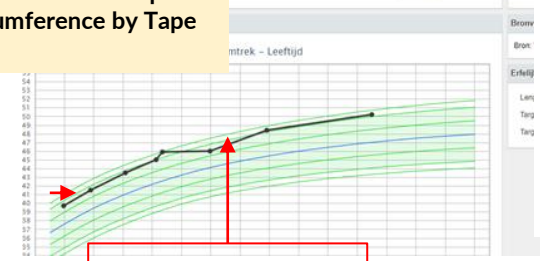
VOXZOGO™
(vosoritide) for injection

LOINC 29463-7 Body weight

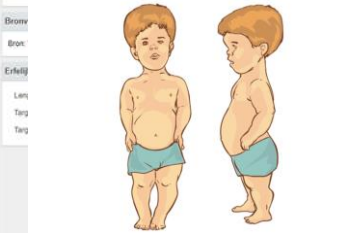
LOINC 8302-2 Body height



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.

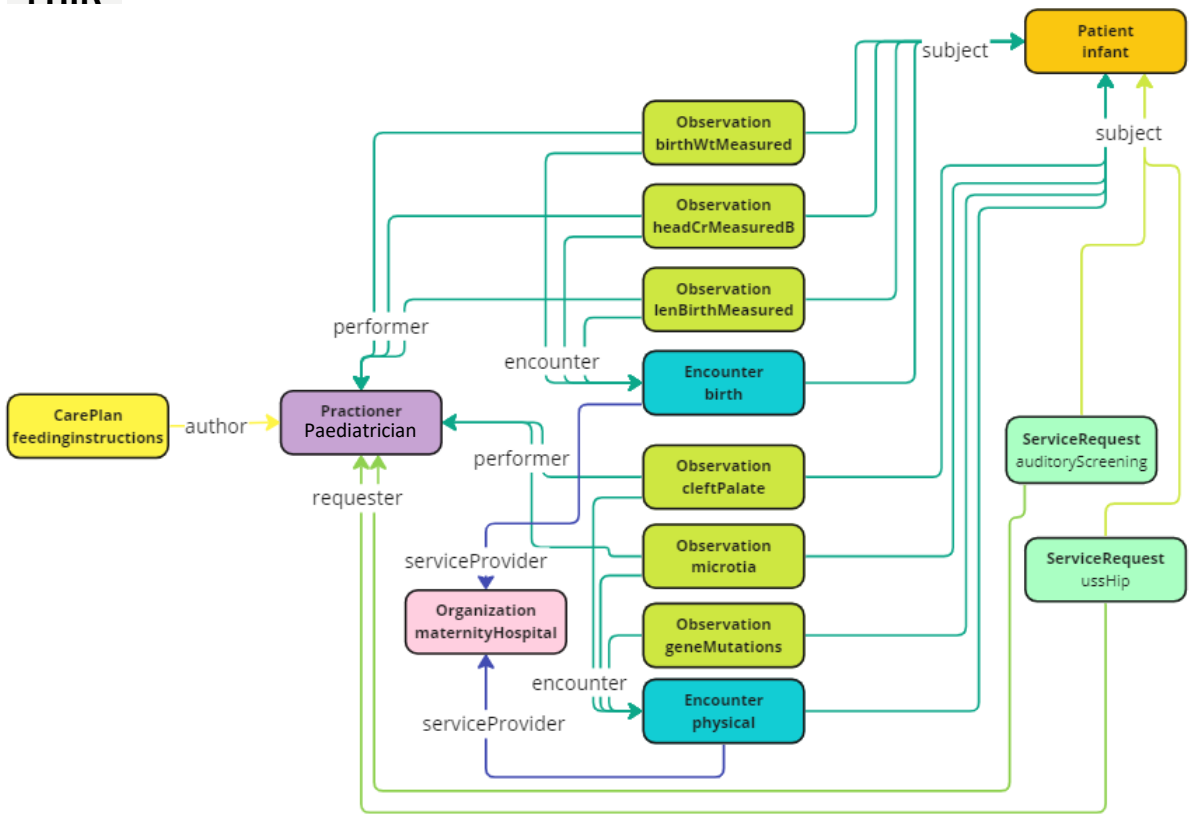
Case 3 - Breech-Cleft Palate-Microtia

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

World Health Organization

OMIM

612290

MICROTIA, HEARING IMPAIRMENT, AND CLEFT PALATE

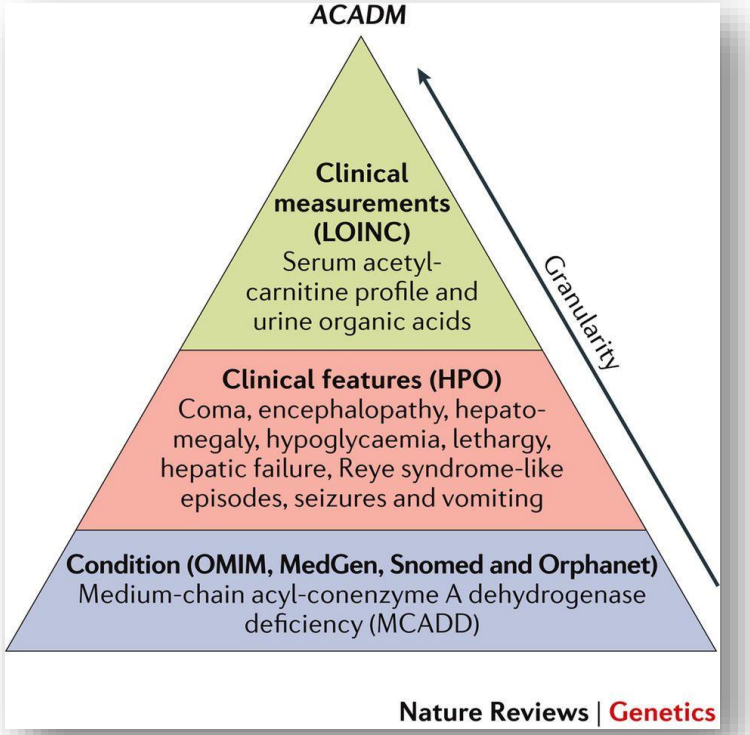
Other entities represented in this entry:

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:

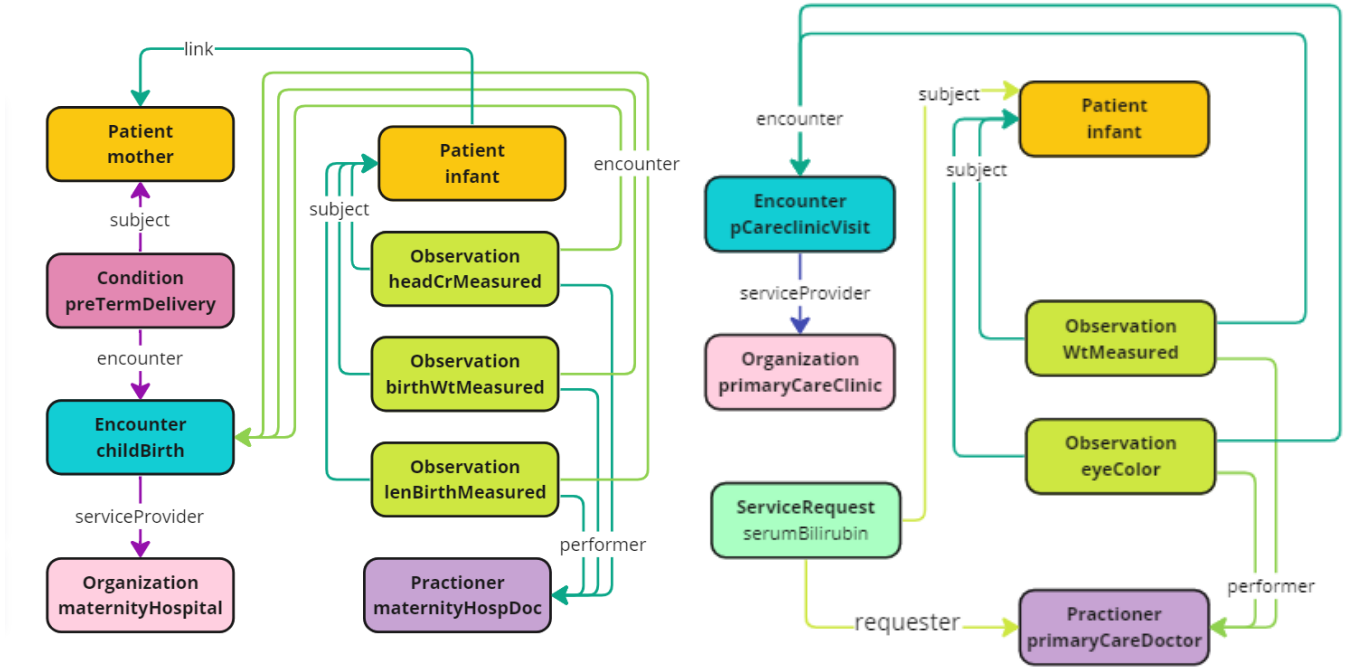
Counsel to continue breastfeeding to ensure adequate hydration and address breastfeeding problems, if needed

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 >

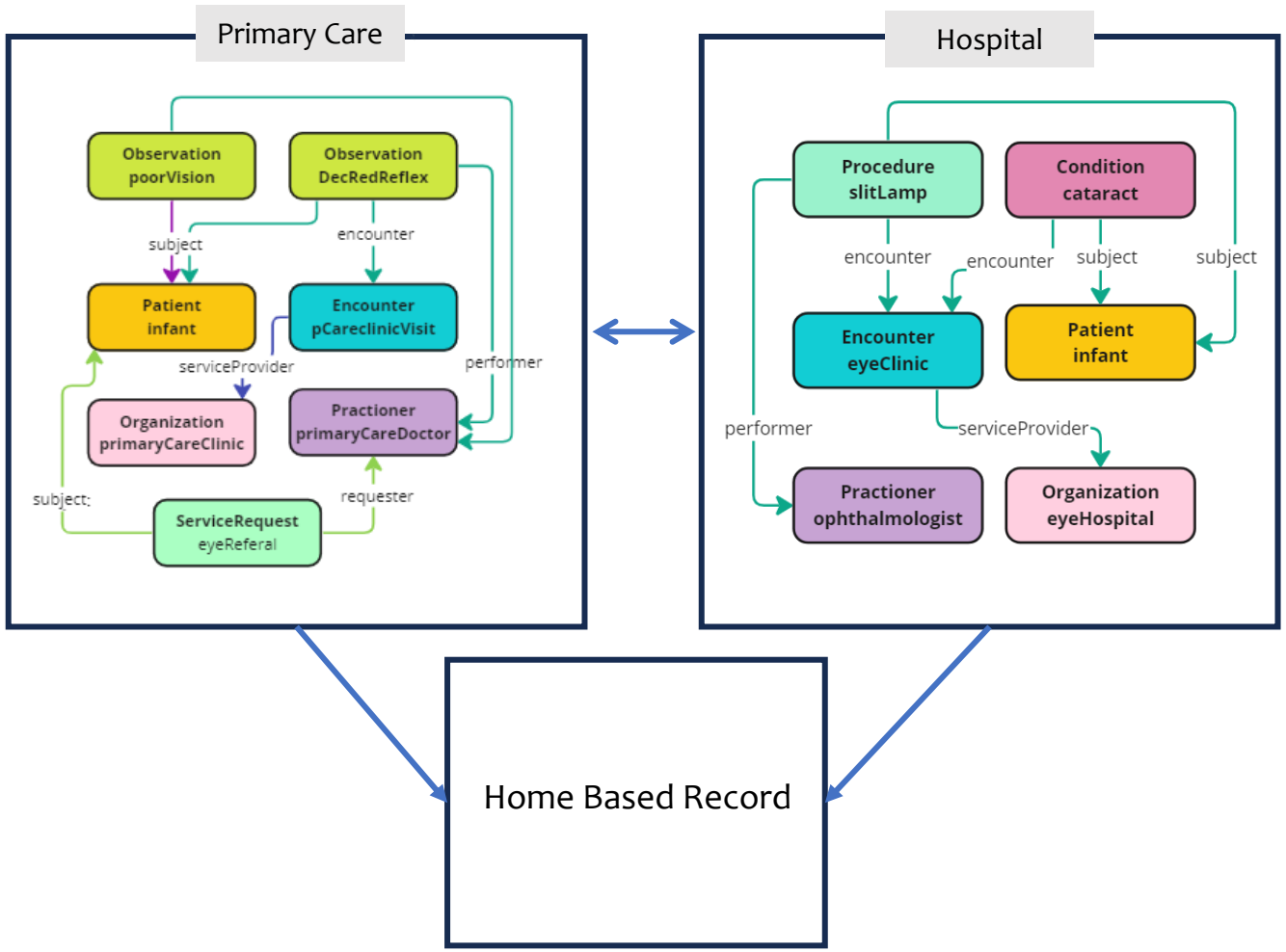
Diagnosis : Infantile cataract

Case 5- Juvenile Cataract

Diagnosis

Date Flow

Primary Care → Hospital Record → Home Based Record



Terminologies

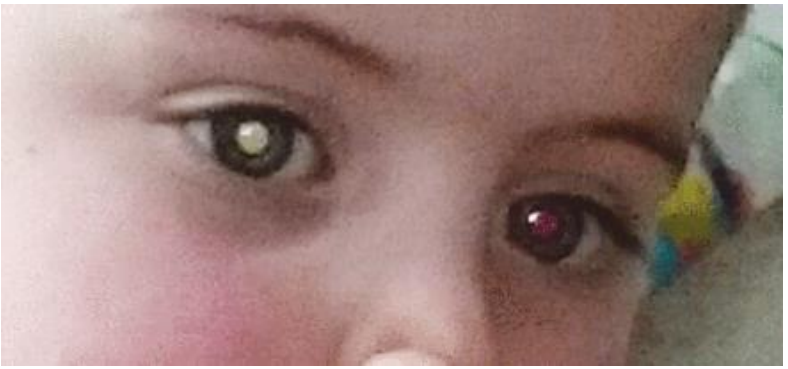
Red reflex absent	SNOMED CT	247079003
Abnormal vision	SNOMED CT	7973008
Study observation Left optic lens Slit lamp biomicroscopy	LOINC	79866-0
Infantile, juvenile and presenile cataract	ICD 10	H26.0

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**

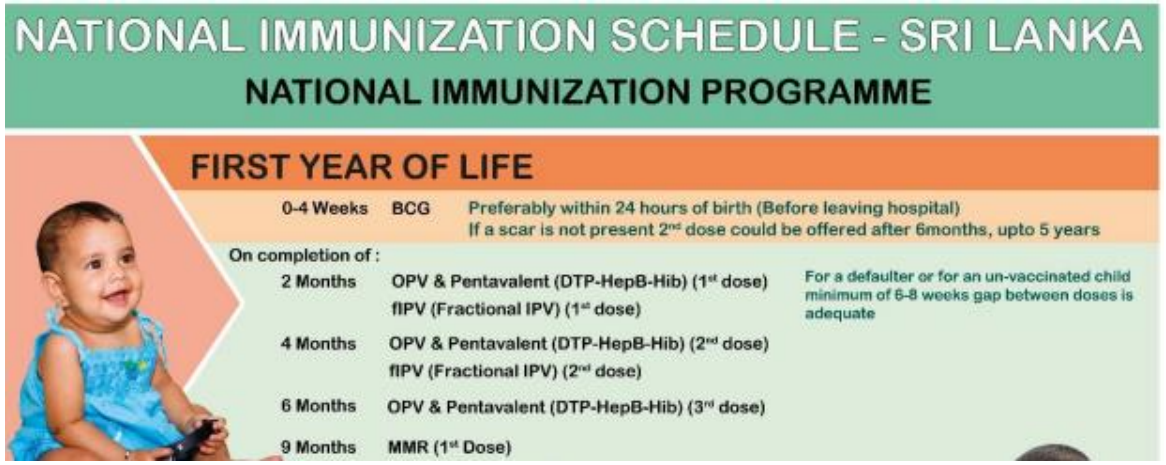
Laboratory test : Hemoglobine | Microcosis red blood cells

Referal to Thalassemia clinic

Parents are advised about routine vaccinations

Cascade Screening of Family

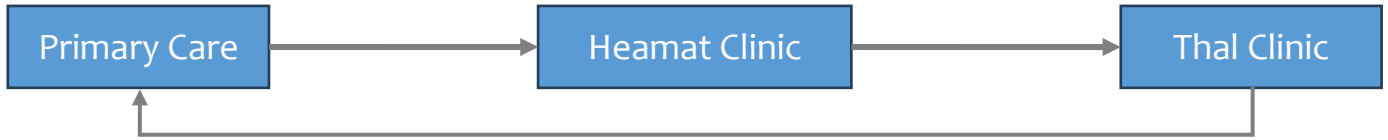
Diagnosis : Beta Thalassemia



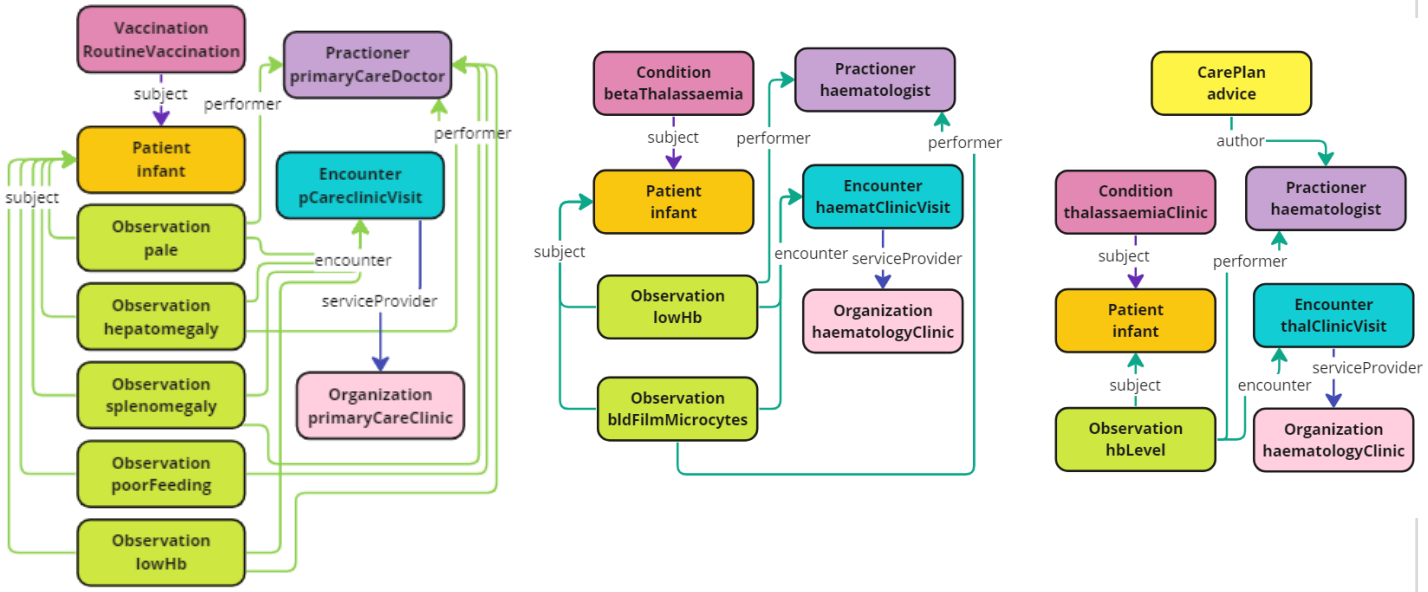
Case6 - Beta Thalassemia

Diagnosis

Date Flow



FHIR

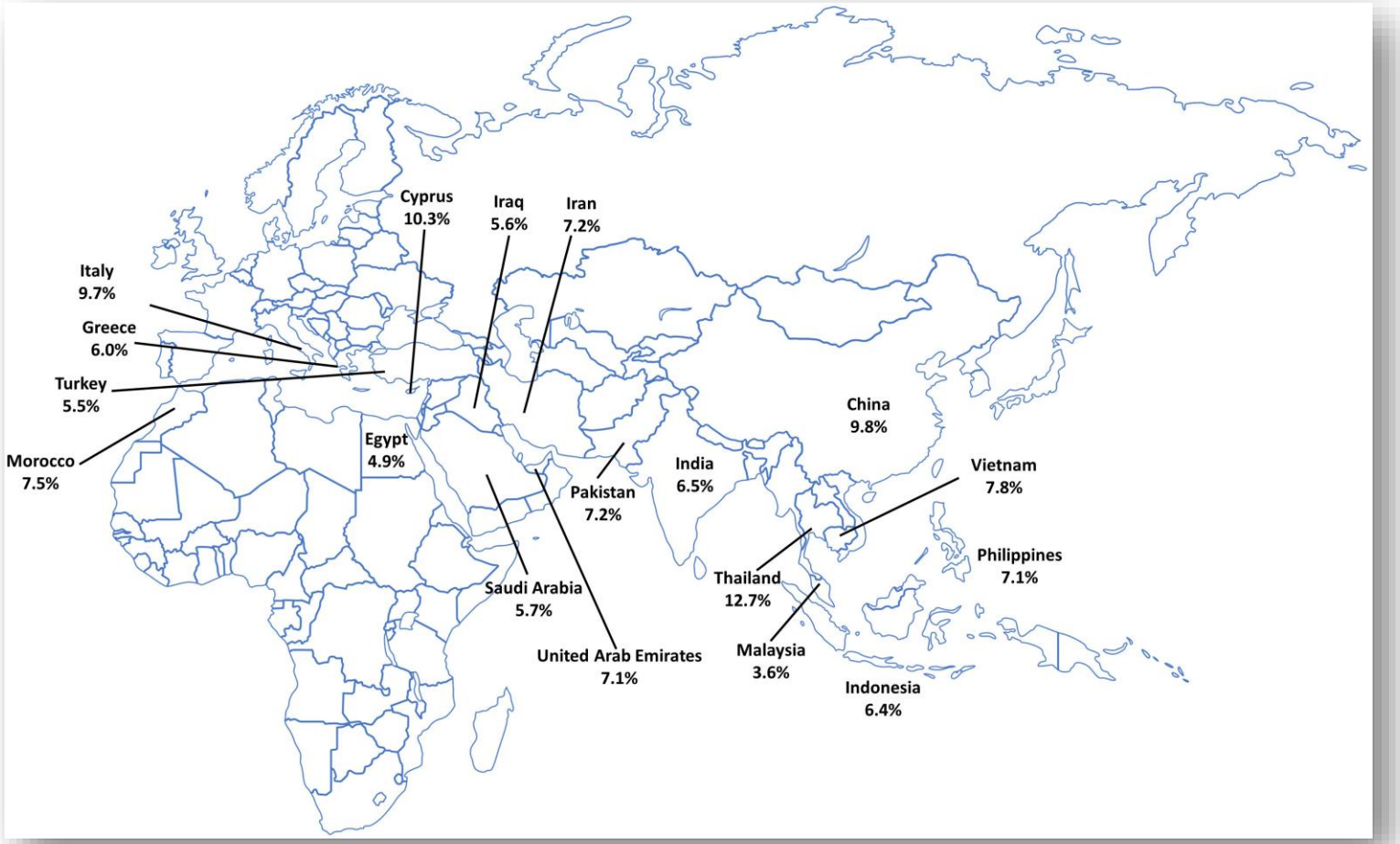


Terminologies

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

Codes Identified for Global Child Health

1	Fetal Head Diameter.biparietal US	LOINC	11820-8
2	Head Occipital-frontal circumference by Tape measure	LOINC	8287-5
3	Birth weight Measured	LOINC	8339-4
4	Body height Measured --at birth	LOINC	89269-5
5	Body weight Measured	LOINC	3141-9
6	Body height Measured	LOINC	3137-7
7	Femur Length US	LOINC	11963-6
8	Gene studied [ID]	LOINC	48018-6
9	Neonatal bilirubin panel [Mass/volume] - Serum or Plasma	LOINC	50189-0
10	Study observation Left optic lens Slit lamp biomicroscopy	LOINC	79866-0
11	Haemoglobin concentration in blood	LOINC	718-7
12	Microcytes in blood film	LOINC	741-9
13	Pallor	SNOMED CT	1237486008
14	Hepatomegaly	SNOMED CT	80515008
15	Splenomegaly	SNOMED CT	16294009
16	Red reflex absent (situation)	SNOMED CT	247079003

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We are going to have a great time learning together!

Acknowledgements