Global exchange of quality data on health and well-being, leaving no one behind

Liesbeth Siderius

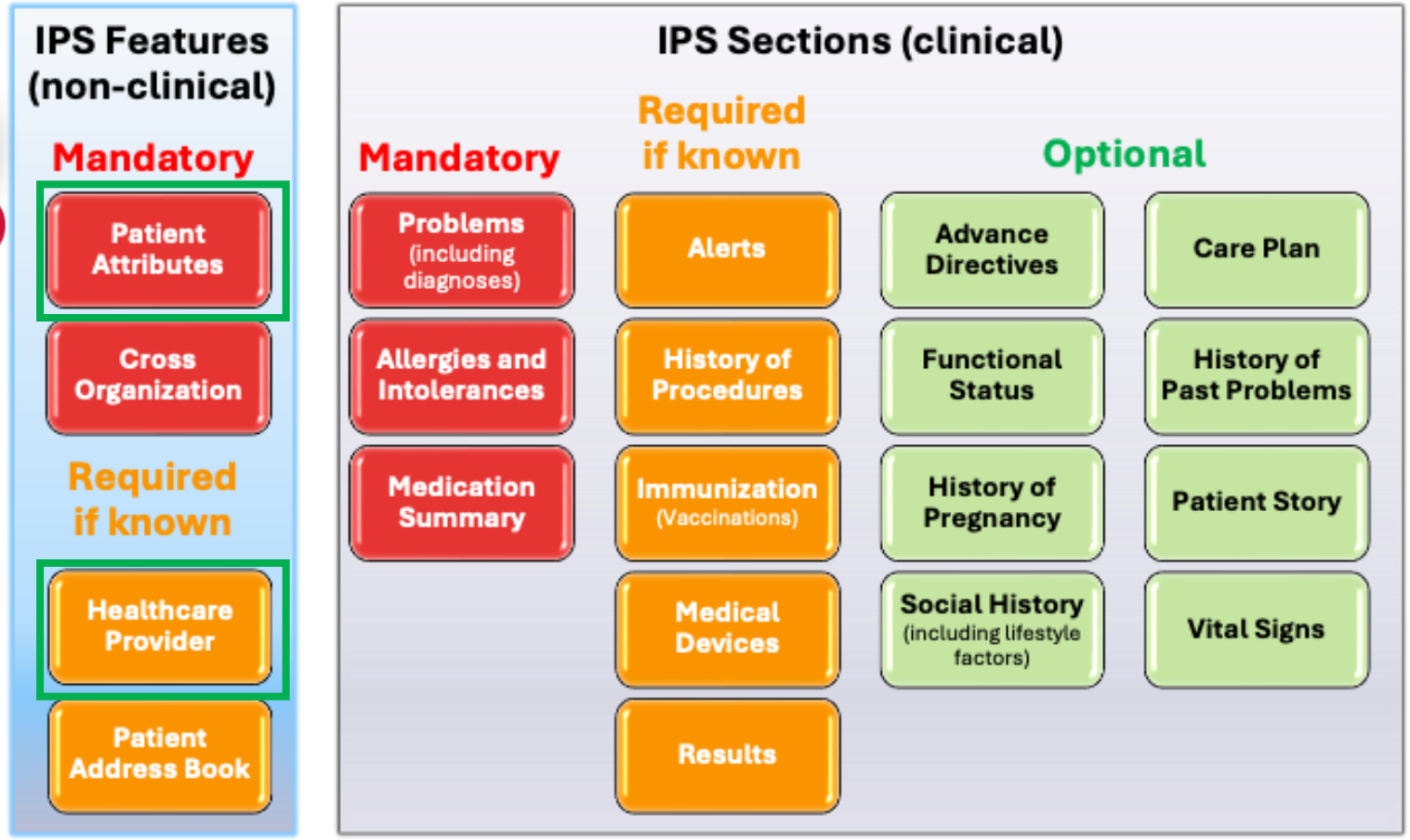
Rare Care World Foundation

Youth Health Almere

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

International Patient Summary updated October 2025

https://international-patient-summary.net/iso-27269/



- Short Stature
- Apgar score
- Vaccination
- Fetal alcohol-syndrome
- Rare disease
- Participation

ISO=
International Standards globally recognized guidelines and frameworks

Achondroplasia (disorder)
SCTID: 86268005

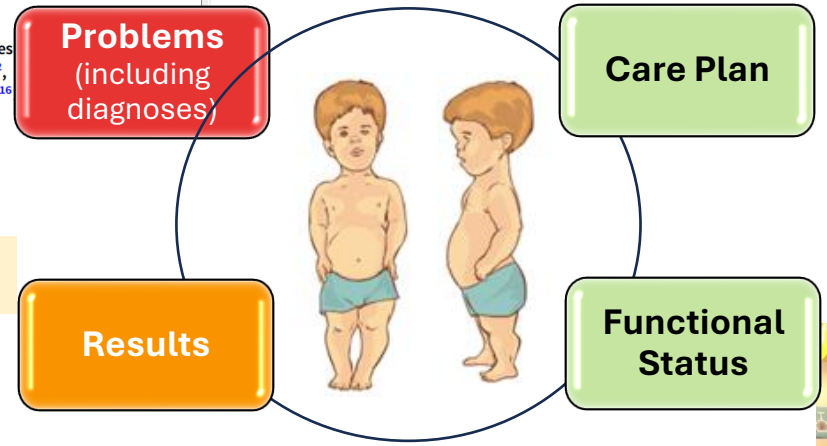
ICD11
LD24.00 Achondroplasia

European Journal of Endocrinology, 2026, 194, R17–R36
https://doi.org/10.1093/ejendo/lvag013
Published: 16 January 2026
Invited review

2026 EJE OXFORD

International guideline on genetic testing of children with short stature

Andrew Dauber[1†], Alexander A.L. Jorge[2†], Ola Nilsson[3], Olaf M. Dekkers[4,5,6], Jes
Irene Netchine[9], Philippe Backeljauw[10], Jeffrey Baron[11], Debora R. Bertola[12],
Peter Clayton[13], Justin H. Davies[14], Thomas Edouard[15], Thomas Eggermann[16]



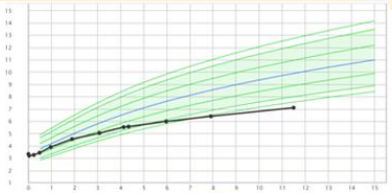
POSITION STATEMENT Open Access

The first European consensus on principles of management for achondroplasia

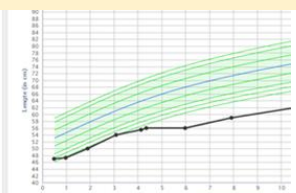
Valerie Cormier-Daire[1], Moeenaldeen AlSayed[2,3], Tawfeg Ben-Omran[4], Sérgio Bernardo de Sousa[5,6], Silvio Boero[7], Svein O. Fredwall[8], Encarna Guillen-Navarro[9], Melita Irving[10], Christian Lampe[11], Mohamad Maghnie[12], Geert Mortier[13], Zagorka Peijin[14] and Klaus Mohnike[15*]

2021

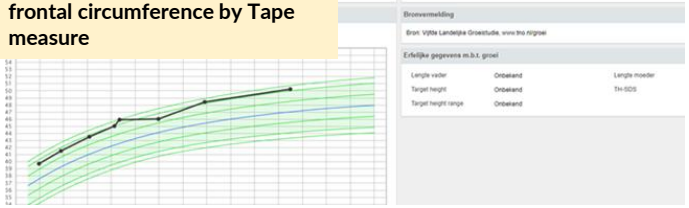
LOINC 29463-7 Body weight



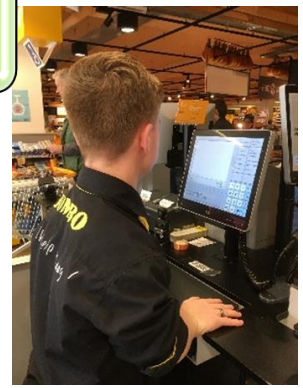
LOINC 8302-2 Body height



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



SUSTAINABLE DEVELOPMENT GOALS



ICF-d 850 Employment

Problems
(including diagnoses)

www.childneurologyfoundation.org/disorder/spasticity/

History of Past Problems

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

History of Past Problems

Results

Apgar Score LOINC and SNOMED CT

Apgar score
Evaluation of Newborn infant based on GUIDELINE

LOINC

LOINC CODE
48332-1

LONG COMMON NAME
10 minute Apgar panel

Reference Information

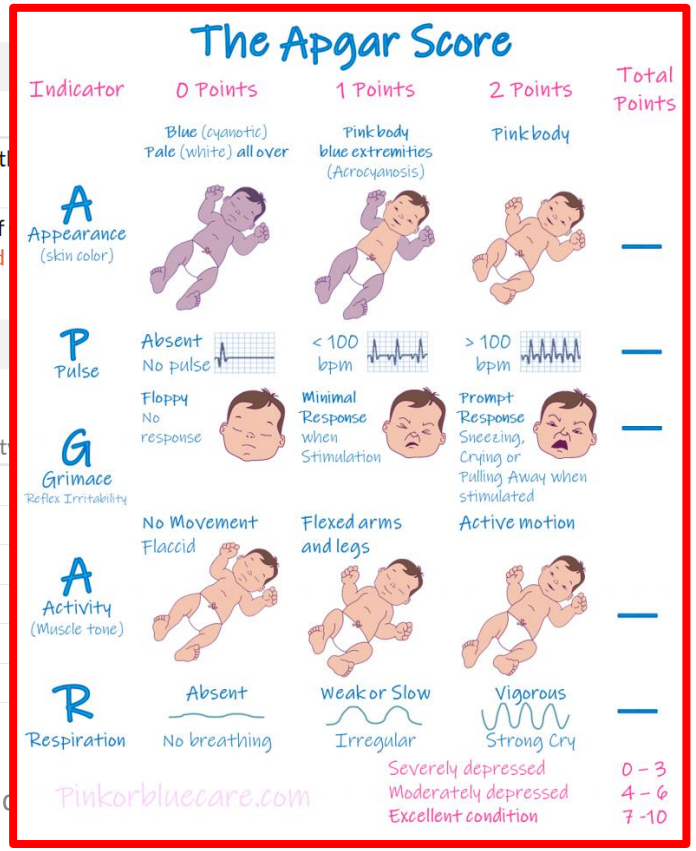
Type	Source	Reference
Article	NCBI PubMed	Apgar V. A proposal for a new method of evaluation of the newborn infant. Curr Res Anest
Article	NCBI PubMed	American Academy of Pediatrics, Committee on Fetus and Newborn; American College of Obstetric Practice. The Apgar score. Pediatrics. 2006 Apr;117(4):1444-7. Link to PubMed

Panel Hierarchy

Details for each LOINC in Panel | LHC-Forms

LOINC	Name	R/O/C	Cardinalit
48332-1	10 minute Apgar panel		
32401-2	10 minute Apgar Color		1..1
32402-0	10 minute Apgar Heart rate		1..1
32404-6	10 minute Apgar Reflex irritability		1..1
32403-8	10 minute Apgar Muscle tone		1..1
32405-3	10 minute Apgar Respiratory effort		1..1
9271-8	10 minute Apgar Score		1..1

Global exchange of c



NHS Digital SNOMED CT Browser

© SNOMED International 2017 v1.36.4 - Hosted and maintained by NHS Digital

	(observable entity)
Apgar score 4 (finding)	Apgar score 4 (finding)
Apgar score 5 (finding)	Apgar score 5 (finding)
Apgar score 0 (finding)	Apgar score 0 (finding)
Apgar score 8 (finding)	Apgar score 8 (finding)
Apgar score 9 (finding)	Apgar score 9 (finding)
Apgar score 3 (finding)	Apgar score 3 (finding)
Apgar score 1 (finding)	Apgar score 1 (finding)
Apgar score 7 (finding)	Apgar score 7 (finding)
Apgar score 6 (finding)	Apgar score 6 (finding)
Apgar score 10 (finding)	Apgar score 10 (finding)
Apgar score at 5 minutes	Apgar score at 5 minutes (observable entity)
Apgar score at 20 minutes	Apgar score at 20 minutes (observable entity)
Apgar score at 10 minutes	Apgar score at 10 minutes (observable entity)
Apgar score at 15 minutes	Apgar score at 15 minutes (observable entity)

Immunization
(Vaccinations)

DPT vaccination ATC and SNOMED CT

Anatomical Therapeutic Chemical (ATC) Classification

Date and Place vaccination

Bacterial and viral vaccines, combined

- J07CA01 : Diphtheria-poliomyelitis-tetanus
- J07CA02 : Diphtheria-pertussis-poliomyelitis-tetanus
- J07CA03 : Diphtheria-rubella-tetanus
- J07CA04 : Hemophilus influenzae B and poliomyelitis
- J07CA05 : Diphtheria-hepatitis B-pertussis-tetanus
- J07CA06 : Diphtheria-hemophilus influenzae B-pertussis-poliomyelitis-tetanus
- J07CA07 : Diphtheria-hepatitis B-tetanus
- J07CA08 : Hemophilus influenzae B and hepatitis B
- J07CA09 : Diphtheria-hemophilus influenzae B-pertussis-poliomyelitis-tetanus-hepatitis B
- J07CA10 : Typhoid-hepatitis A
- J07CA11 : Diphtheria-Hemophilus influenzae B-pertussis-tetanus-hepatitis B
- J07CA12 : Diphtheria-pertussis-poliomyelitis-tetanus-hepatitis B
- J07CA13 : Diphtheria-hemophilus influenzae B-pertussis-tetanus-hepatitis B-meningococcus

SNOMED CT

	Corynebacterium diphtheriae antigens (procedure)
First diphtheria, pertussis, and tetanus vaccination	Administration of first dose of vaccine product containing only Bordetella pertussis and Clostridium tetani and Corynebacterium diphtheriae antigens (procedure)
Third diphtheria, pertussis, and tetanus vaccination	Administration of third dose of vaccine product containing only Bordetella pertussis and Clostridium tetani and Corynebacterium diphtheriae antigens (procedure)
Second diphtheria, pertussis, and tetanus vaccination	Administration of second dose of vaccine product containing only Bordetella pertussis and Clostridium tetani and Corynebacterium diphtheriae antigens (procedure)
Requires diphtheria, tetanus and pertussis vaccination	Requires diphtheria, tetanus and pertussis vaccination (finding)

Administration of first dose of vaccine product containing only Bordetella pertussis and Clostridium tetani and Corynebacterium diphtheriae antigens (procedure)

SCTID: 866158005

866158005 | Administration of first dose of vaccine product containing only Bordetella pertussis and Clostridium tetani and Corynebacterium diphtheriae antigens (procedure) |

en Administration of first dose of vaccine product containing only Bordetella pertussis and Clostridium tetani and Corynebacterium diphtheriae antigens (procedure)

en Administration of first dose of diphtheria, pertussis, and tetanus vaccine

en Administration of first dose of

ıtional User Guide Contact Us

Social History (including lifestyle factors)

Mother/ Woman

History of Past Problems

Child

Jobs | News | About us

MANATŪ HAUORA
MINISTRY OF HEALTH

Search

COVID-19 | Your health | NZ health system | Our work | Health statistics | Publications

Home > Your health > Conditions & treatments > Disabilities > Fetal alcohol spectrum disorder

Disabilities
- Fetal alcohol spectrum disorder
- Low vision

Fetal alcohol spectrum disorder (FASD)

Stop drinking alcohol if you could be pregnant, are pregnant or are trying to get pregnant. There is no known safe level of alcohol consumption during pregnancy.

Babies exposed to alcohol before birth may develop fetal alcohol spectrum disorder (FASD). FASD can cause problems including:

- low birth weight
- distinctive facial features
- heart defects
- behavioural problems
- intellectual disability.

Resources

Alcohol and Pregnancy: What you might not know
Available on HealthEd.

Related websites

healthdirect

Free Australian health advice you can count on.

Home | COVID-19 | Health topics A-Z | Medicines | Symptom checker | Servi

Fetal alcohol spectrum disorder

5-minute read

Print | Share | Save

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN®

Patient Care | Early Childhood | Gu

Fetal Alcohol Spectrum Disorders

Home / Patient Care / Fetal Alcohol Spectrum Disorders

We've assembled res
Disorders (FASD) to r
FASD, promote scree
encourage referral fo
goal is to build the ca
clinicians, and allied
individuals with an F
diagnosis and care in
along the FASD conti

NHS

Search

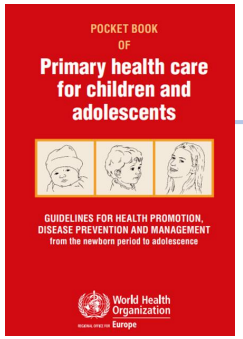
Health A-Z | Live Well | Mental health | Care and support

Home › Health A to Z

Foetal alcohol spectrum disorder

If you drink alcohol during pregnancy you risk causing harm to your baby. Sometimes this can result in mental and physical problems in the baby, called foetal alcohol spectrum disorder (FASD).

FASD can happen when alcohol in the mother's blood passes to her baby through the placenta.



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

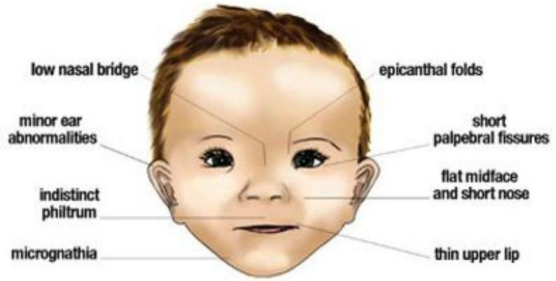
Probable Diagnosis: Fetal alcohol syndrome

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

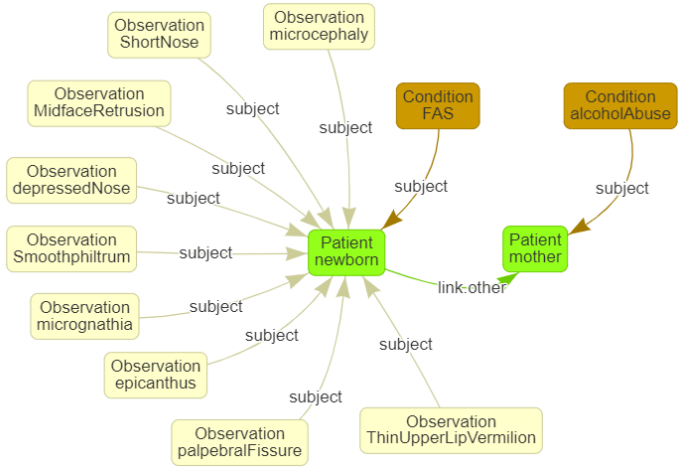
Fetal alcohol syndrome

History of Past Problems

Problems (including diagnoses)



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

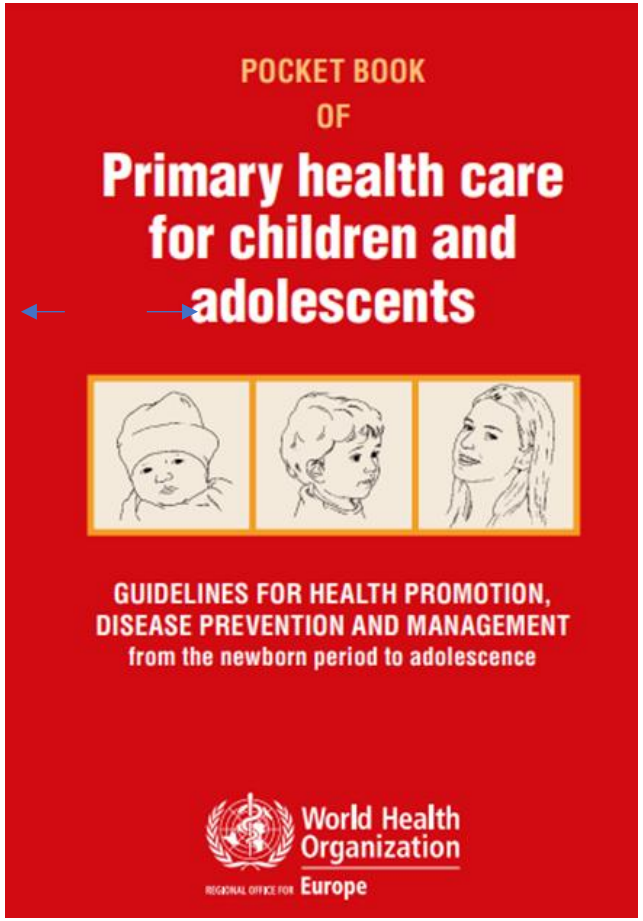


{ "resourceType": "Bundle", "type": "collection", "entry": [{ "fullUrl": "http://clinfhir.com/fhir/Patient/cfsb1693121208440", "resource": { "resourceType": "Patient", "id": "cfsb1693121208440", "name": [{ "text": "MJ", "given": ["M"], "family": "J" }], "birthDate": "1991-01-01", "gender": "female" } }, { "fullUrl": "http://clinfhir.com/fhir/Condition/cfsb1693124146803", "resource": { "resourceType": "Condition", "id": "cfsb1693124146803", "subject": { "reference": "Patient/cfsb1693121208440" }, "code": { "text": "Alcohol abuse, uncomplicated" }, "clinicalStatus": { "coding": [{ "code": "active", "system": "http://terminology.hl7.org/CodeSystem/condition-clinical", "display": "Active" }], "text": "confirmed" } } }, { "fullUrl": "http://clinfhir.com/fhir/Patient/cfsb1693127581588", "resource": { "resourceType": "Patient", "id": "cfsb1693127581588", "identifier": [{ "system": "http://www.maternalhosp.com", "value": "010" }], "link": [{ "type": "seealso", "other": { "reference": "Patient/cfsb1693121208440" } }] } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693127612403", "resource": { "resourceType": "Observation", "id": "cfsb1693127612403", "status": "final", "code": { "coding": [{ "code": "HP:0005280", "system": "http://human-phenotype-ontology.org" }], "text": "Depressed nasal bridge" }, "subject": { "reference": "Patient/cfsb1693127581588" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693127863888", "resource": { "resourceType": "Observation", "id": "cfsb1693127863888", "status": "final", "code": { "coding": [{ "code": "HP:0000319", "system": "http://human-phenotype-ontology.org" }], "text": "Smooth philtrum" }, "subject": { "reference": "Patient/cfsb1693127581588" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693127972179", "resource": { "resourceType": "Observation", "id": "cfsb1693127972179", "status": "final", "code": { "coding": [{ "code": "HP:0000347", "system": "http://human-phenotype-ontology.org" }], "text": "Micrognathia" }, "subject": { "reference": "Patient/cfsb1693127581588" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693128028899", "resource": { "resourceType": "Observation", "id": "cfsb1693128028899", "subject": { "reference": "Patient/cfsb1693127581588" }, "status": "final", "code": { "coding": [{ "code": "HP:0000286", "system": "http://human-phenotype-ontology.org" }], "text": "Epicanthus" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693128090244", "resource": { "resourceType": "Observation", "id": "cfsb1693128090244", "subject": { "reference": "Patient/cfsb1693127581588" }, "status": "final", "code": { "coding": [{ "code": "HP:0012745", "system": "http://human-phenotype-ontology.org" }], "text": "Short palpebral fissure" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693128176077", "resource": { "resourceType": "Observation", "id": "cfsb1693128176077", "subject": { "reference": "Patient/cfsb1693127581588" }, "status": "final", "code": { "coding": [{ "code": "HP:0011800", "system": "http://human-phenotype-ontology.org" }], "text": "Midface retrusion" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693128235547", "resource": { "resourceType": "Observation", "id": "cfsb1693128235547", "subject": { "reference": "Patient/cfsb1693127581588" }, "status": "final", "code": { "coding": [{ "code": "HP:0003196", "system": "http://human-phenotype-ontology.org" }], "text": "Short nose" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693128297601", "resource": { "resourceType": "Observation", "id": "cfsb1693128297601", "subject": { "reference": "Patient/cfsb1693127581588" }, "status": "final", "code": { "coding": [{ "code": "HP:0000219", "system": "http://human-phenotype-ontology.org" }], "text": "Thin upper lip vermilion" } } }, { "fullUrl": "http://clinfhir.com/fhir/Condition/cfsb1693128532718", "resource": { "resourceType": "Condition", "id": "cfsb1693128532718", "subject": { "reference": "Patient/cfsb1693127581588" }, "code": { "text": "Fetal alcohol syndrome" }, "clinicalStatus": { "coding": [{ "code": "active", "system": "http://terminology.hl7.org/CodeSystem/condition-clinical", "display": "Active" }], "text": "active" } } }, { "fullUrl": "http://clinfhir.com/fhir/Observation/cfsb1693129051982", "resource": { "resourceType": "Observation", "id": "cfsb1693129051982", "subject": { "reference": "Patient/cfsb1693127581588" }, "status": "final", "code": { "coding": [{ "code": "HP:0000252", "system": "http://human-phenotype-ontology.org" }], "text": "Microcephaly" } }] }

Global Pediatric Digital Health

Problems (including diagnoses)

Care Plan



Problems (including diagnoses)

Shwachman Diamond Syndrome From Feature to Medical Guideline

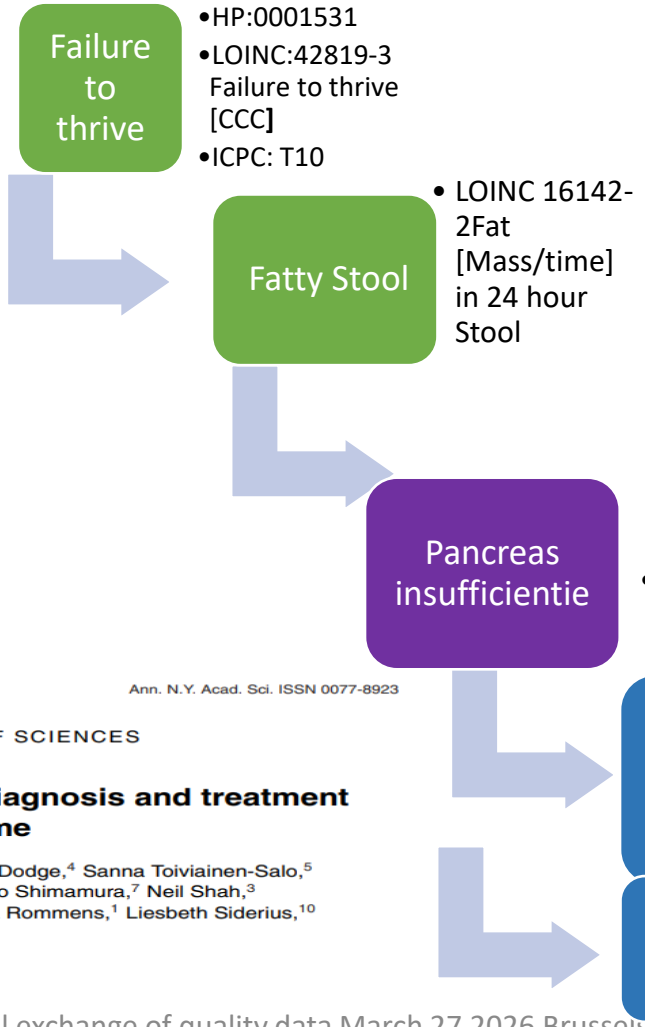
Feature

- Fatty Stool
- Growth Retardation
- Common infections

Shwachman Diamond Syndrome-

Management

- Pancreas insufficiency
- Neutropenia
- Skeletal Dysplasia
- Autisme like



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]

HL7® FHIR®

Problems (including diagnoses)

Shwachman Diamond Syndrome

ICD-11

International Classification of Diseases for Mortality and Morbidity Statistics

Eleventh Revision

World Health Organization

Selected: 3A70.0

Matching Terms

Shwachman-Diamond syndrome *

Description

A disease caused by determinants in the antenatal period leading to the inability of stem cells to generate new characterised by low levels of red blood cells, white blood cells, platelets. This disease may present with pallor, infection or increased bruising or bleeding.

Inclusions

familial hypoplastic anaemia
Constitutional medullar aplasia
Fanconi anaemia

Exclusions

Congenital amegakaryocytic thrombocytopenia (3B64.01)

Coded Elsewhere

Congenital hypoplastic anaemia (KA8C)
Noonan syndrome (LD2F.15)

Related categories in maternal chapter J

Anaemia complicating pregnancy, childbirth or the puerperium / Congenital aplastic anaemia (JB64.0/3A70.0)

Related categories in perinatal chapter K

Congenital hypoplastic anaemia (KA8C)

SNOMED CT

The global language of healthcare

Shwachman syndrome (disorder)

SCTID: 89454001

89454001 | Shwachman syndrome (disorder) |

en Shwachman syndrome (disorder)
en Shwachman syndrome
en Congenital lipomatosis of pancreas
en Metaphyseal chondrodysplasia with pancreatic insufficiency AND neutropenia
en Metaphyseal chondrodysplasia, Shwachman type
en Metaphyseal dysplasia with malabsorption and neutropenia
en Schwachman's syndrome
en Schwachman-Bodian syndrome
en Schwachman-Diamond syndrome
en Schwachmann-Diamond syndrome
en Shwachman's syndrome
en Shwachman-Diamond syndrome

Interprets → Neutrophil count
Has interpretation → Below reference range

Occurrence → Congenital
Finding site → Pancreatic structure
Pathological process → Pathological developmental process

Occurrence → Congenital
Finding site → Bone structure
Associated morphology → Dysplasia
Pathological process → Pathological developmental process

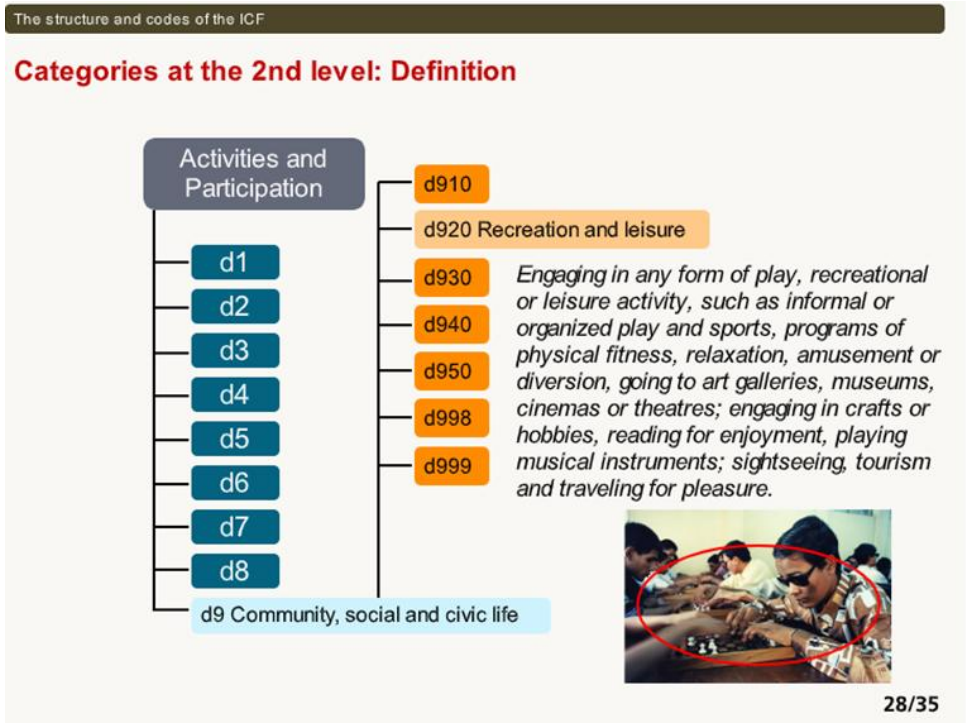
Pathological process → Abnormal immune process

Interprets → Body height
Has interpretation → Below reference range

Functional Status

ICF d 920.0

Recreation and leisure



Indian Mother and Childcare
Kolkata, 2020

Open Access FHIR RESTfull API Library

Mother and Child Health

- Growth & Development
- Conditions

Computable clinical guidelines

- Thalassemia
- Shwachman Diamond Syndrome

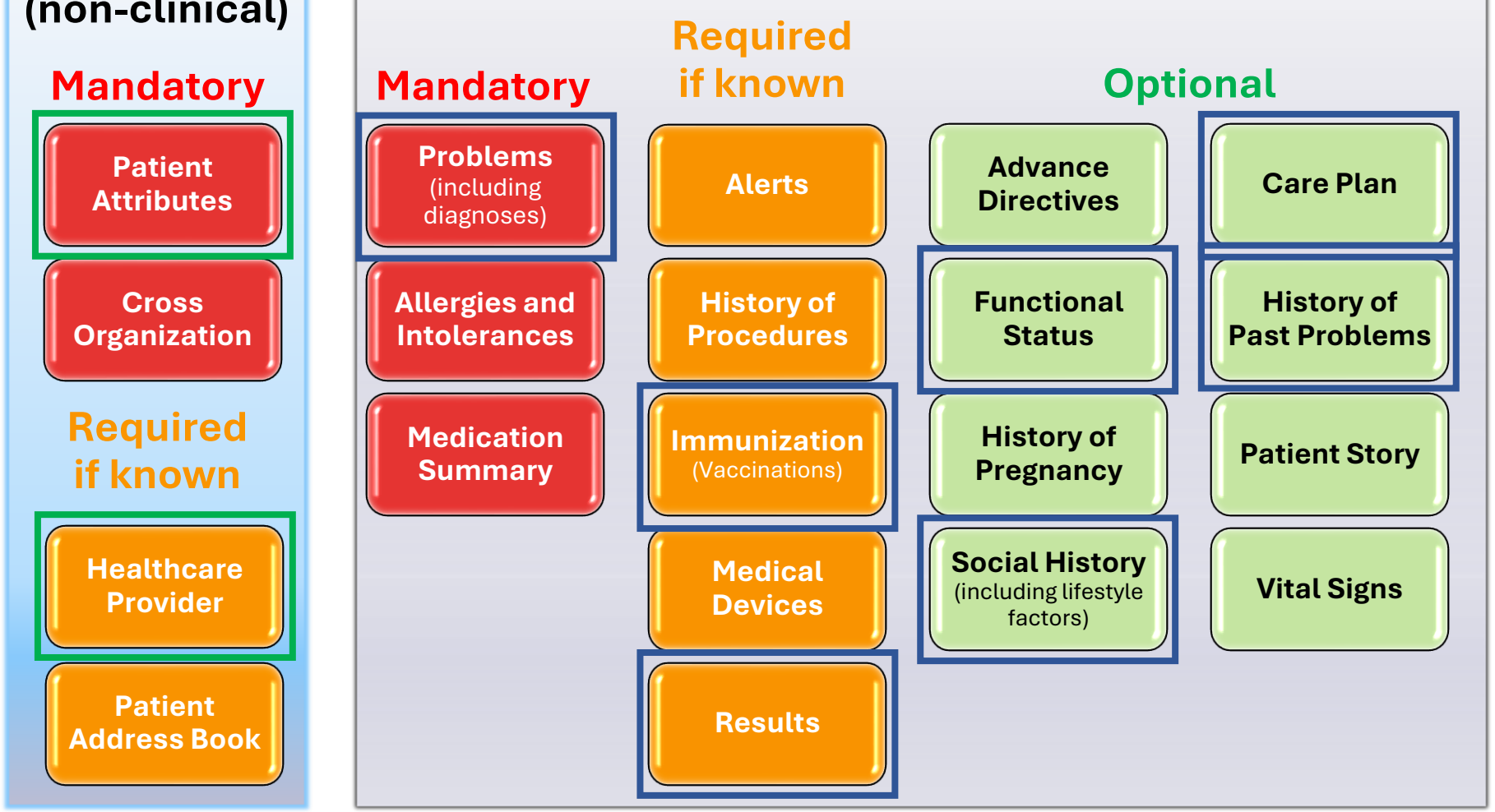
Immunizations

- Vaccination schemes

Social Support

- ICF
- ISO 9999

ISO 27269:2025 – The International Patient Summary



Thank you
- 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
- EAP IT network
- HL7 Child+Health+Obstetrics+International+Collaboration+and+Exploration

Anjan Bhattacharya, ICF expert India
Sahan Damsiri Perera, IT Expert, Sri Lanka/ Australia
Marc de Graauw, IT Expert, Netherlands
Martin Postma, IT Expert, Netherlands
Robert Stegwee, IT Expert, the Netherlands

- People with a rare condition and their families

Paulo Gonçalves, Portugal

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

e.siderius@kpnplanet.nl