Een data model voor zeldzame ziekten

Liesbeth Siderius

Kinderarts, GGD IJsselland

Rare Disease Working Group

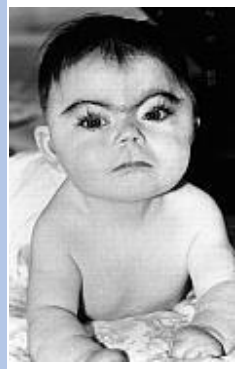
European Academy of Paediatrics

Shwachman Syndroom Support Holland

CONGRES
ARCHITECTUUR IN DE ZORG

Zeldzaam is ‘gewoon’ in de kindergeneeskunde

- In Europa is een ziekte definieerd als zeldzaam als het minder dan **1: 2000** mensen treft.
- Er zijn tussen de **6000** en **8000** zeldzame aandoeningen: **5-8%** van een populatie
- 80% van de zeldzame aandoeningen hebben een genetische oorzaak, en zijn vaak **chronisch** en levensbedreigend
- 50-75 % manifesteerd zijn op de **kinderleeftijd**, veel chronische aandoeningen op de kinderleeftijd zijn zeldzaam.
- < 5 jaar heeft 2-3 % een zeldzame aandoening



De last van zeldzame ziekten is aanzienlijk, met ongeveer 350 miljoen mensen aangedaan wereld wijd.

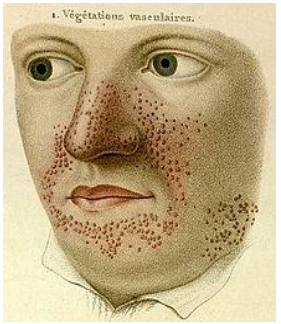
Dit is in dezelfde orde als de meer bekende ‘non-communicable´ zoals suikerziekte.

Listen to the Voices of 12,000 Patiënts 2009

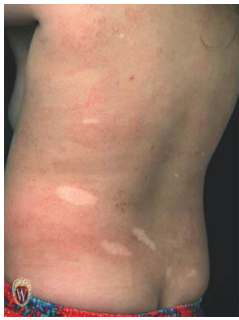
EURORDIS
RARE DISEASES EUROPE

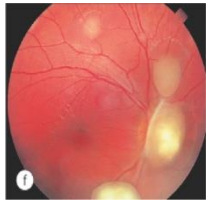
Na de **eerste symptomen** een gevecht om de diagnose dat jaren kan duren.

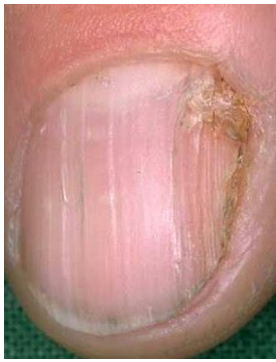
Na de diagnose geconfronteerd met **onvolledige gezondheid-** en **sociale zorg**.

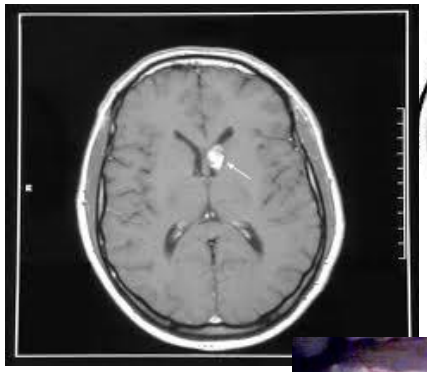


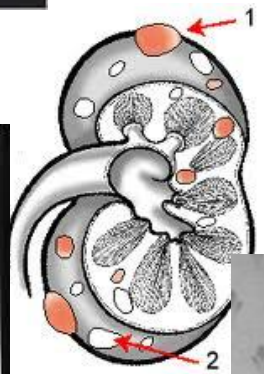
Tuberous sclerosis complex











Arnhem 2019

OSCAR NEDERLAND

OSCAR Nederland - De patiëntenorganisatie voor dragers en patiënten met sikkelcelziekte en thalassemie

Inleiding

Wat is thalassemie?
Thalassemie is een ziekte van de rode bloedcellen. De oorzaak is een kleine verandering in het erfelijk materiaal. Daardoor is het hemoglobine (de rode bloedkleurstof in de rode bloedcellen) anders dan bij gezonde mensen.

Begrippenlijst

Op alfabetische volgorde:

A
anemie : is een ander woord voor bloedarmoede.

B
E.M.T. : is beenmergtr

Onderzoek/ diagnostiek

Waarom onderzoek?
Om complicaties te voorkomen of zoveel mogelijk te beperken.
Meestal worden daarbij meerdere organen onderzocht.

Bij het onderzoek moet je te maken krijgen met verschillende specialisten:
hematoloog, cardioloog, orthopeed, oogarts, KNO-arts en natuurlijk verpleegkundigen.

Samen kan er meer

Nazorg

De nazorg is bedoeld als ondersteuning van de thuissituatie.
Het gaat daarbij vooral om de zelfredzaamheid en hulp bij financiële, psychologische en maatschappelijke problemen.

Instantie:
Algemeen Maatschappelijk Werk:
kan hulp bieden aan patiënt, ouders en familie op sociaal, juridisch en financieel gebied.

WMO (Wet Maatschappelijke Ondersteuning):

Preventie

Wat is preventie?

Preventie is:
1. het voorkomen van ziekten of aandoeningen,
2. het opsporen van ziekten, aandoeningen of afwijkingen in een zo vroeg mogelijk stadium,
3. het voorkomen van complicaties bij ziekten, aandoeningen of afwijkingen.

Er zijn 3 vormen van preventie: primaire preventie, secundaire preventie, tertiaire preventie.
Heud onderstaande punten goed in de gaten!!!
Zie voor de vetgedrukte woorden de begrippenlijst (blad 2).

1. **Primaire preventie**

Behandeling

Waarom behandeling ?
Om complicaties te voorkomen, te beperken of te genezen.
Is belangrijk voor een betere kwaliteit van leven.
De behandeling kan plaatsvinden in het ziekenhuis, de polikliniek of thuis.

Polikliniek behandeling en in de thuissituatie
Wat is belangrijk:
- Bij gebruik van medicijnen zoals **Hydrea** en/ of **Exjade** 3 keer per jaar bloedonderzoek, om complicaties te voorkomen (Hydrea verhoogt de aanmaak van rode bloedcellen, Exjade verlaagt het ijzergehalte in het bloed).

Patient Informatie	Eerste lijn	Diagnose Collaborative care	Social Services
www.shwachman.nl	Groei achterstand Herhaalde infecties (LOINC)	Guideline SDS (Orphanetcode; SNOMED, ATC e.a.)	Vaak ziek, Moe, Klein (ICF)

Stichting Shwachman syndroom Support Holland

Diagnose
Hurler syndrome
PKU, Duchenne MD, FOP
Shwachman Diamond Syndroom

ICD - 10
Orphanetcode
OMIM
SNOMED -CT

Richtlijn
Integrale medische zorg

ATC
GS1
Nieuwe Therapie
ICF(-CY)
ISO9999

Sociale voorzieningen
en rehabilitatie

Data Verzameling
Data verzameling is systematisch georganiseerd computer geprogrameerd collection medial terms

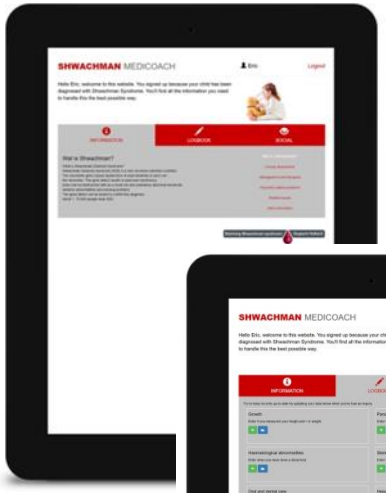
HL7
ISO 3166-1

Kenmerk in eerste lijn
Hielprik
Gehoor screening
Groei; Ontwikkeling

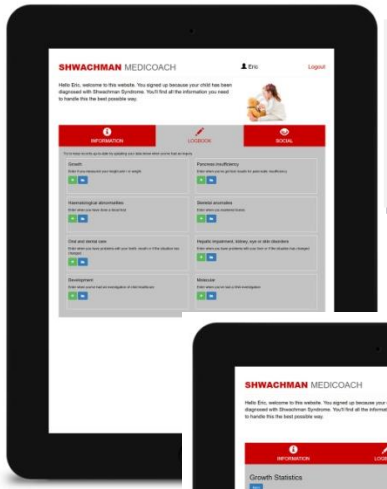
LOINC
ICPC
Nieuwe Diagnostiek

Arnhem 2019

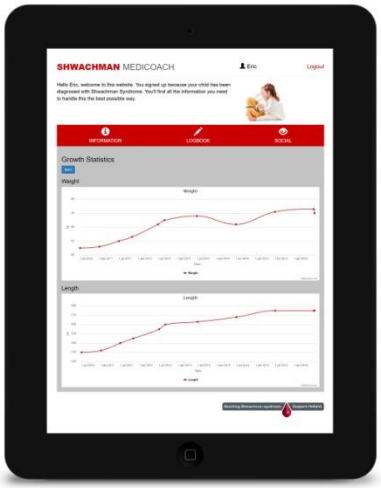
Co-management of people with Shwachman Diamond Syndrome in daily care generating comparable data



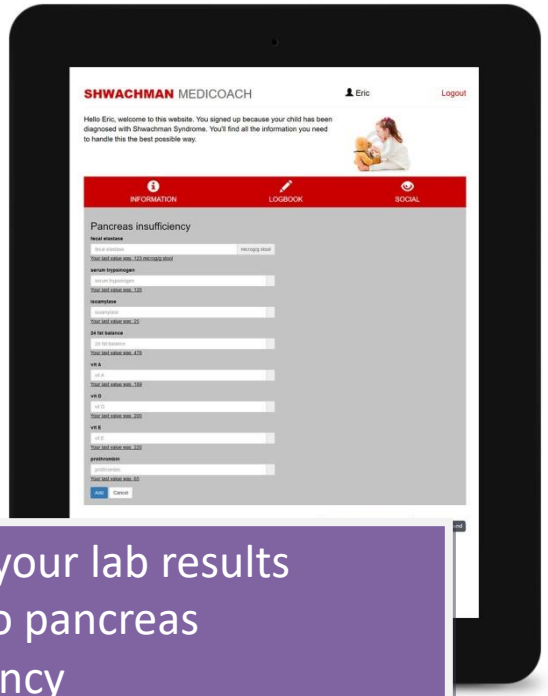
Information on SDS and the use of the application



Overview of the digital guideline



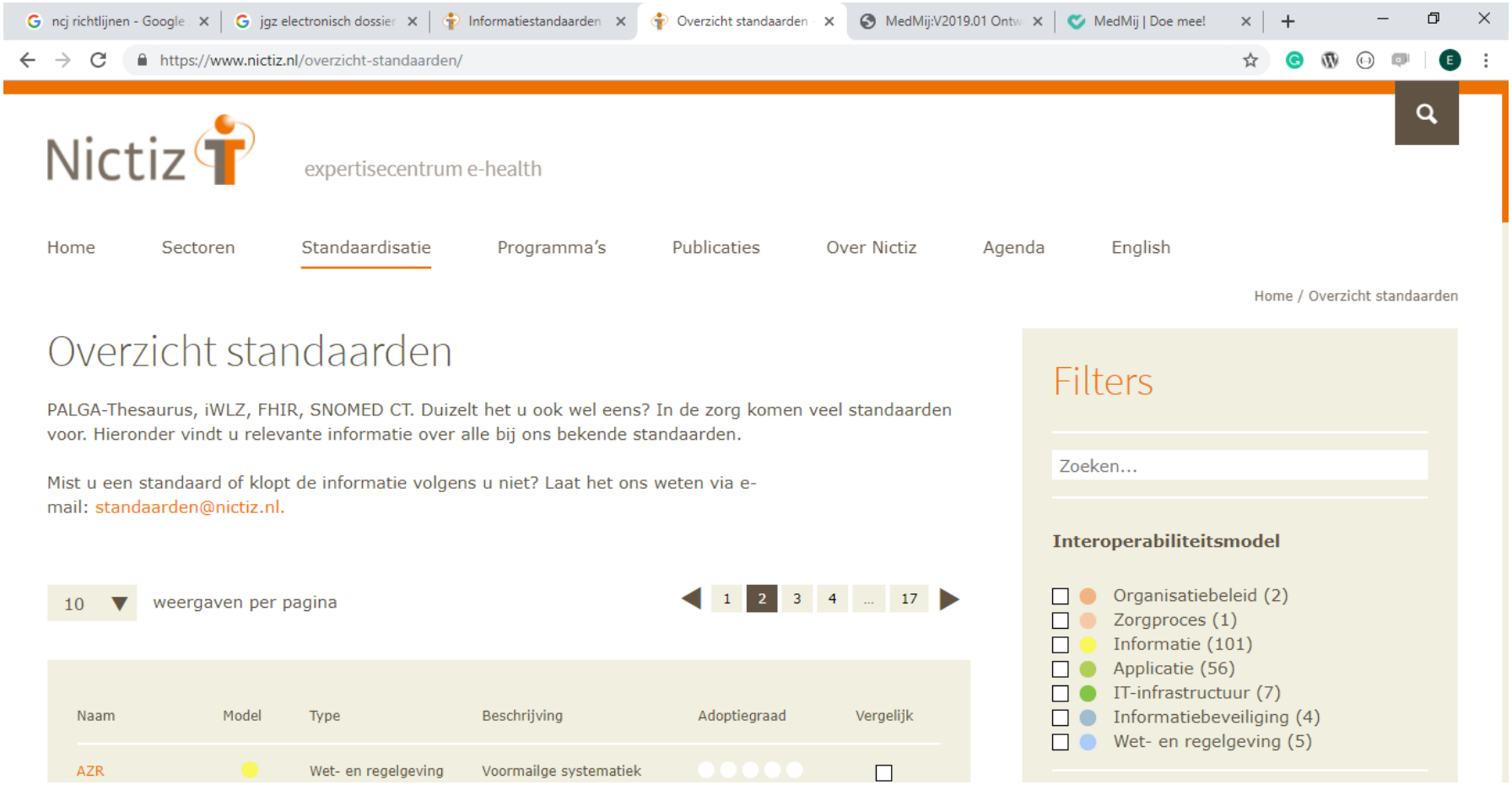
Collect weight and length data to create a SDS growth curve



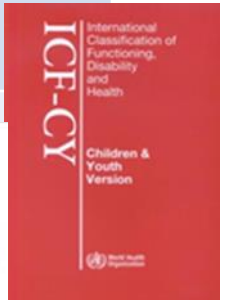
Register your lab results related to pancreas insufficiency

Vroeg signaleren in de Eerste Lijn 2015-2016

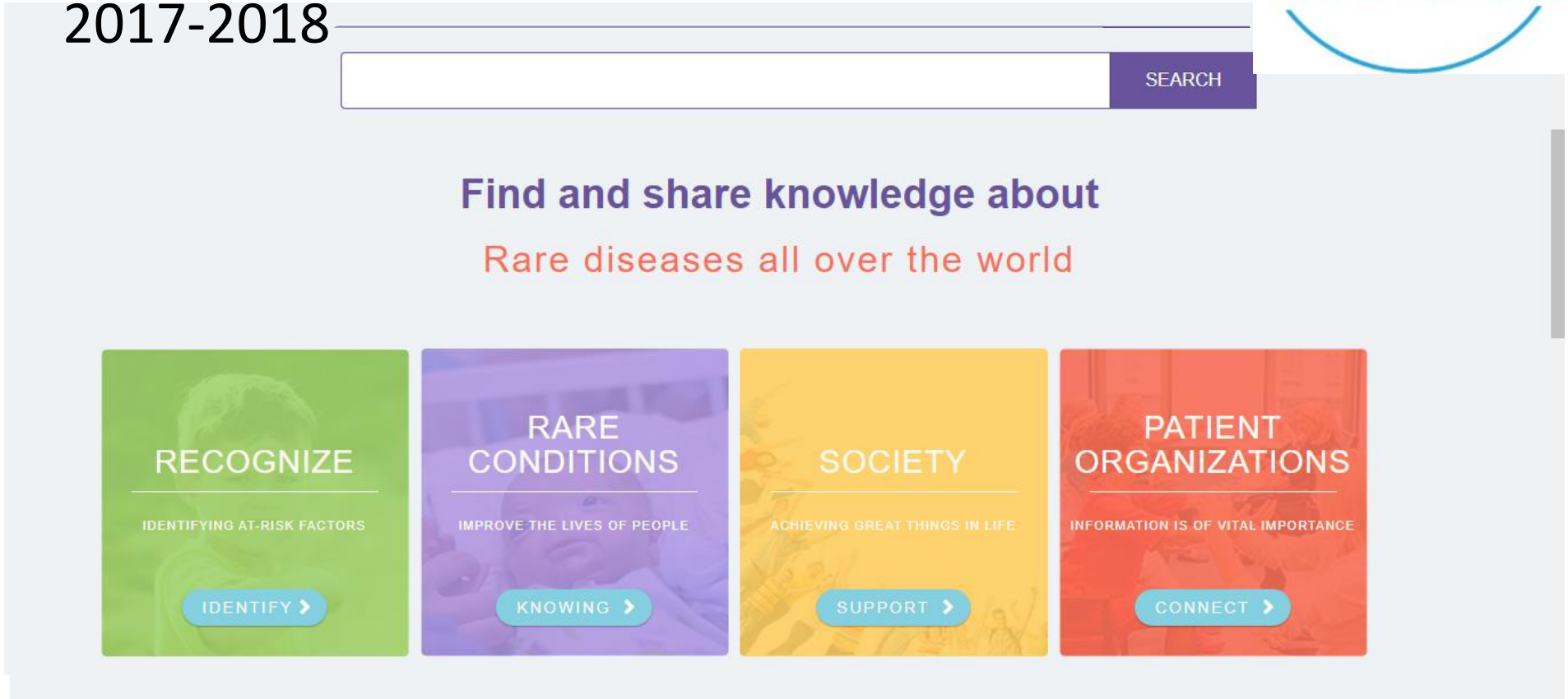
Met dank aan:



code	betekenis
ICPC	International Classification of Primary Care
HPO	Human Phenotype Ontology
LOINC	Standard for identifying health measurements, observations, and documents
ICD	International Classification of Diseases
ORPHAnet	Classification of rare diseases
ATC	Anatomical Therapeutic Chemical Classification System
ICF	International Classification of Functioning, Disability and Health
SNOMED	Clinical health terminology

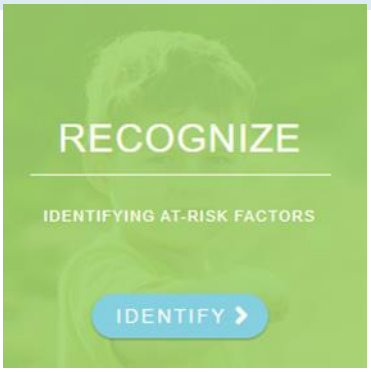


Project “Stem van de Patient”
Door en voor mensen met een (zeldzame) chronische aandoening
2017-2018

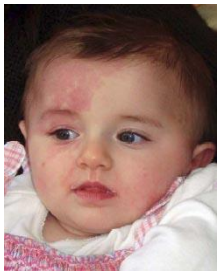


Ministerie van Volksgezondheid, Welzijn en Sport

Eerste Symptomen Preventieve Jeugd Gezondheid Zorg



ontwikkeling

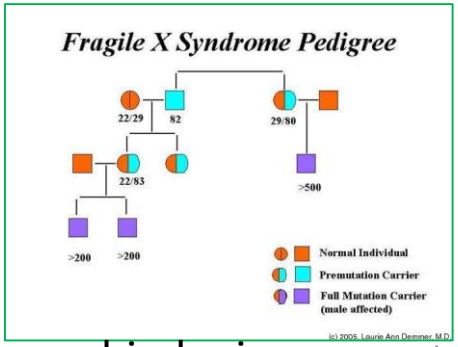


Lichamelijk onderzoek

Zwangerschap

Vaccinatie

Groei

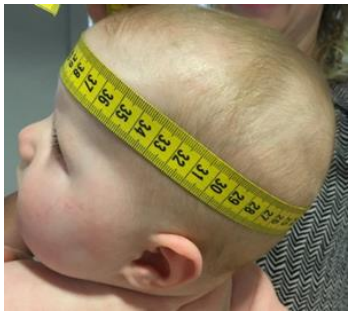


Familie geschiedenis

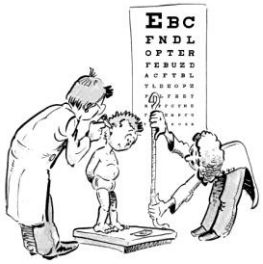
Heel prik

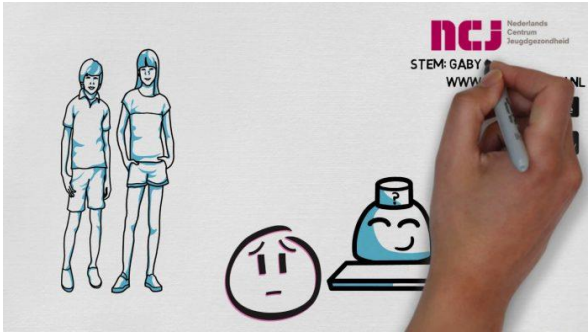
Thuis

Eerste Symptomen Preventieve Jeugd Gezondheid Zorg



Fibrodysplasia Ossificans Progressiva
1:2.000000
SPIER wordt BOT





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

2015 Recommendations for Preventive Pedi

Bright Futures/American Academy of P

Each child and family is unique; therefore, these Recommendations for Preventive Pediatric Health Care are designed for the care of children who are receiving competent parenting, have no manifestations of any important health problems, and are growing and developing in satisfactory fashion. Additional visits may become necessary if circumstances suggest variations from normal.

Developmental, psychosocial, and chronic disease issues for children and adolescents may require frequent counseling and treatment visits separate from preventive care visits.

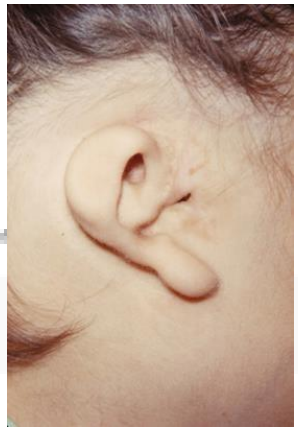
These guidelines represent a consensus by the American Academy Bright Futures. The AAP continues to emphasize the great importa comprehensive health supervision and the need to avoid fragmenta

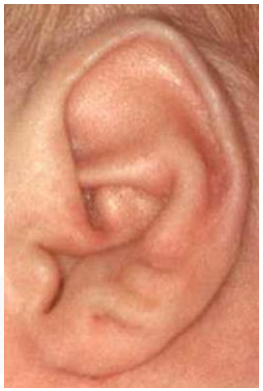
Refer to the specific guidance by age as listed in Bright Futures Shaw JS, Duncan PM, eds. *Bright Futures Guidelines for Health Su and Adolescents*. 3rd ed. Elk Grove Village, IL: American Academy

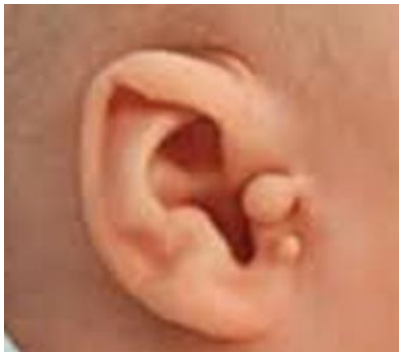
	INFANCY								EARLY CHILDHOOD							
AGE[1]	Prenatal[4]	N														
HISTORY Initial/Interval	●															
MEASUREMENTS																
Length/Height and Weight																
Head Circumference																
Weight for Length																
Body Mass Index[5]																
Blood Pressure[6]																
SENSORY SCREENING																
Vision																
Hearing																
DEVELOPMENTAL/BEHAVIORAL ASSESSMENT																
Developmental Screening[9]																
Autism Screening[10]																
Developmental Surveillance																
Psychosocial/Behavioral Assessment																
Alcohol and Drug Use Assessment[11]																
Depression Screening[12]																
PHYSICAL EXAMINATION[13]		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●

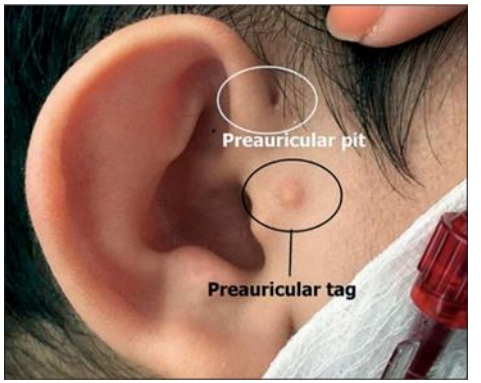
Nederlands Centrum Jeugdgezondheid

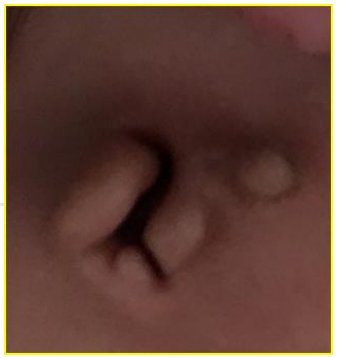
Basis data set Electronisch Dossier



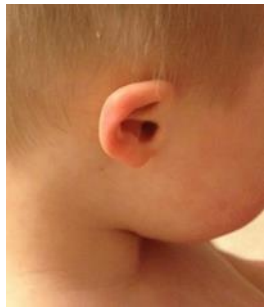


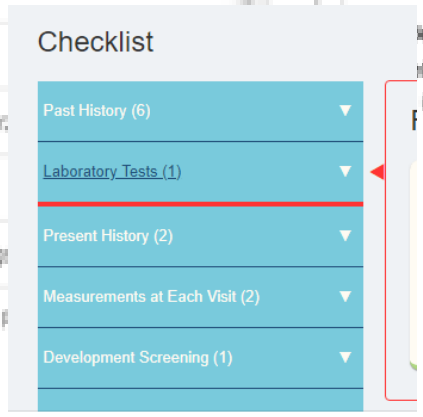


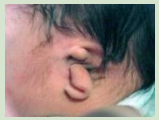
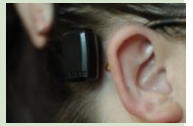
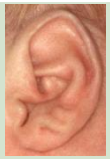
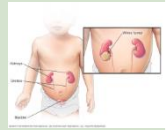
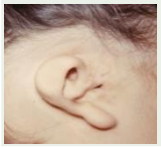
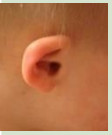
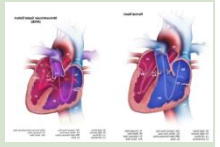
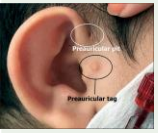
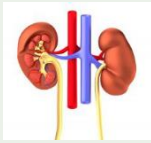
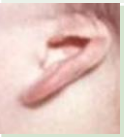
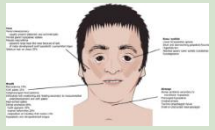




Microtia
(klein oor)
HP:0008551



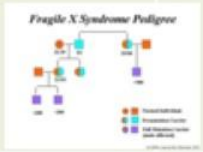
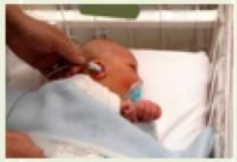


	Disease	Associated RISK
	Microtia (klein oor)	**Conductief gehoorsverlies** 
	Oor Aanhangsel	De Amerikaanse Kinderartsen (AAP) bevelen een gehoorscreening aan ongeacht de aanwezigheid van extra huid. 
	Beckwith Wiedemann Syndroom	De meeste **tumoren** geassocieerd met BWS treden in de eerste t 8–10 jaar Van het leven op. Meest voorkomende nier- en lever tumor 
	Goldenhar Syndroom	**Hemivertebrae, wervel afwijking** 
	Down syndroom	**Aangeboren hart afwijkingen** 
	Brachio Oto Renal Syndroom	**Nierafwijking** 
	Treacher Collins syndroom	**Vroege operaties gericht op openhouden luchtwegen, beschermen van ogen, gehoorscreening en neurologische ontwikkeling.** 

EURORDIS
Rare Diseases Europe

*'After first symptoms, **battle for diagnosis**, which can last for years'*

Identifying children with a disability through primary care, child health surveillance, and vaccination programs

Preventive Child Health	age	feature	Rare Disease
Family history 	neonatal	X linked and autosomal recessive & dominant	• Fragile X • Neurofibromatosis
Heel stick screening	neonatal	National program	• Thallasemia • Phenyl Keton Uria
Hearing screening *Otoacoustic emission* *Automated Auditory Brainstem Response* 	neonatal	Hearing deficit 0,2% > 76% cause identified 39% - 60% genetic , 30% aquered (cytomegalovirus) Other causes	• Usher syndrome 50% hereditary forms combining deafness and blindness prevalence of 3 to 6.2 per 100,000
Congenital anomaly 	neonatal	Club feet 1/700-1/1000 of liveborns	20% associated with • distal arthrogryposis, • congenital myotonic dystrophy,or other genetic

Building the Rare Disease

knowledge and information ecosystem

Home / Rare conditions

Alport Syndrome

Alport syndrome is a hereditary glomerulonephritis resulting in renal failure. In addition, there is often (sensorineural) deafness and a congenital anomaly of the eye, a lenticonus. Alport syndrome is a genetic disease of collagen a3a4a5(IV). The collagen a3a4a5(IV) hetero-trimer is a major…

Bardet Biedl Syndrome

Bardet–Biedl syndrome (BBS) consists of eye disease(rod-cone dystrophy), skeletal abnormality (polydactyly, brachydactyly, and syndactyly), obesity, reduced intelligence, renal dysfunction, and male genital anomaly (hypogonadism). Some features manifest at in the first decade of life with…

Coffin Lowry Syndrome

Coffin-Lowry syndrome is a rare genetic disorder characterized by mental retardation; abnormalities of the head and facial (craniofacial) area; large, soft hands with short, thin (tapered) fingers; short stature and/or various skeletal abnormalities. Characteristic facial features may…

Fibrodysplasia Ossificans Progressiva

Fibrodysplasia Ossificans Progressiva is a rare genetic disorder in which connective tissue and muscle tissue are gradually ossified. Extra-skeletal bone formation causes progressive loss of mobility. The process starts in early childhood and often forms a pattern starting with the neck and…

Oculo-Auriculo-Vertebral Spectrum (Goldenhar Syndrome)

In the total spectrum of OAVS are many syndromes. One of them is Goldenhar Syndrome (GHS). Goldenhar syndrome is a rare congenital defect in the craniofacial spectrum and is characterized by incomplete development in the formation of one side of the face and body. Mainly affected are Ear, Eye and…

Building the Rare Disease

knowledge and information ecosystem

Thalassemia, 3500 patients have been identified in Sri Lanka.

thalassemia SEARCH

Thalassemia Major

Find and share knowledge about

Rare diseases all over the world

ATC

- L01XX05 Hydroxycarbamide (Hydroxyurea) (1)
- V03AC01 Deferoxamine (1)
- V03AC02 Deferipron (1)
- V03AC03 Deferasirox (1)

ICD

- D57 Sickle-cell disorders (1)

ICPC Reference

- B78.01 Thalassemia (1)
- B87 Splenomegaly (1)

LOINC

- 718-7 Hemoglobin in blood (1)
- 20567-4 Ferritin in Serum or Plasma (1)
- 46740-7 Hemoglobin disorders newborn screen interpretation (1)
- 53857-9 Hemoglobin F (1)

OMIM

fractures or vertebral deformities. **Thalassemia** major or Beta **Thalassemia** …
Rare Condition
Thalassemia major or Beta Thalassemia

Large spleen

Feature

… costal margin. A large spleen is a feature of for example **Thalassemia** Infections Niemann-Pick disease Gaucher disease
Splenomegaly Splenomegaly in **thalassemia** **Thalassemia** major or Beta **Thalassemia** …
Rare Condition
Thalassemia major or Beta Thalassemia
Symptom
Splenomegaly in thalassemia
Abnormality
Splenomegaly

Carrier screening thala

Symptom

… Carrier screening **thalassemia** Related family members with elevated HbA2 In carrier screening for the classical beta-**thalassemia** trait, the hallmark is the presence of an … 2 (α 2 δ 2). Another way of identifying people with **thalassemia** major is neonatal screening. Neonatal screening …
Rare Condition
Thalassemia major or Beta Thalassemia
Disease
Hemoglobinopathies

application programming interface (API)

Building the Rare Disease

knowledge and information ecosystem

থ্যালাসেমিয়া প্রধান বা বিটা থ্যালাসেমিয়া

থ্যালাসেমিয়া জেনেটিকালি (উত্তরাধিকারসূত্রে) রক্তের রোগের একটি গ্রুপ যা সাধারণ এক বৈশিষ্ট্যতে ভাগ করে; হিমোগ্লোবিনের ত্রুটিপূর্ণ উত্পাদন যা প্রোটিন যা লাল রক্ত কোষগুলিকে বহন ও অক্সিজেন সরবরাহ করতে সক্ষম করে। ত্রুটিপূর্ণ হিমোগ্লোবিন সংশ্লেষণের বিভিন্ন পদ্ধতি রয়েছে এবং অতএব, বহু ধরনের থ্যালাসেমিয়া রয়েছে।

বিটা থ্যালাসেমিয়া হিমোগ্লোবিনের বিটা গ্লবিন শৃঙ্খলার অনুপস্থিতি বা হ্রাস সংশ্লেষণের কারণে ঘটে। বিটা থ্যালাসেমিয়া বৈশিষ্ট্য বা বিটা থ্যালাসেমিয়া নাবিক ব্যক্তিরা বিটা থ্যালাসেমিয়া বা বিটা-থ্যালাসেমিয়ার একজন ক্যারিয়ারের হেটারজাইজাস। বিটা থ্যালাসেমিয়া মেজারের ব্যাক্তি হ'ল বিটা থ্যালাসেমিয়ার জন্য হোমজাইজাস এবং এভাবে ত্রুটিপূর্ণ জিনের দুটি কপি রয়েছে এবং এই রোগটি বিকাশ করে: থ্যালাসেমিয়া প্রধান। জিনের সম্পূর্ণ অনুপস্থিতিটি β0 থ্যালাসেমিয়া হিসাবে বর্ণনা করা হয় এবং β + হিসাবে সংশ্লেষকে হ্রাস করা হয়। বিটা-গ্লোবিনের হ্রাস α গ্লবিন চেইনগুলিতে আপেক্ষিক অতিরিক্ত বাড়ায়।

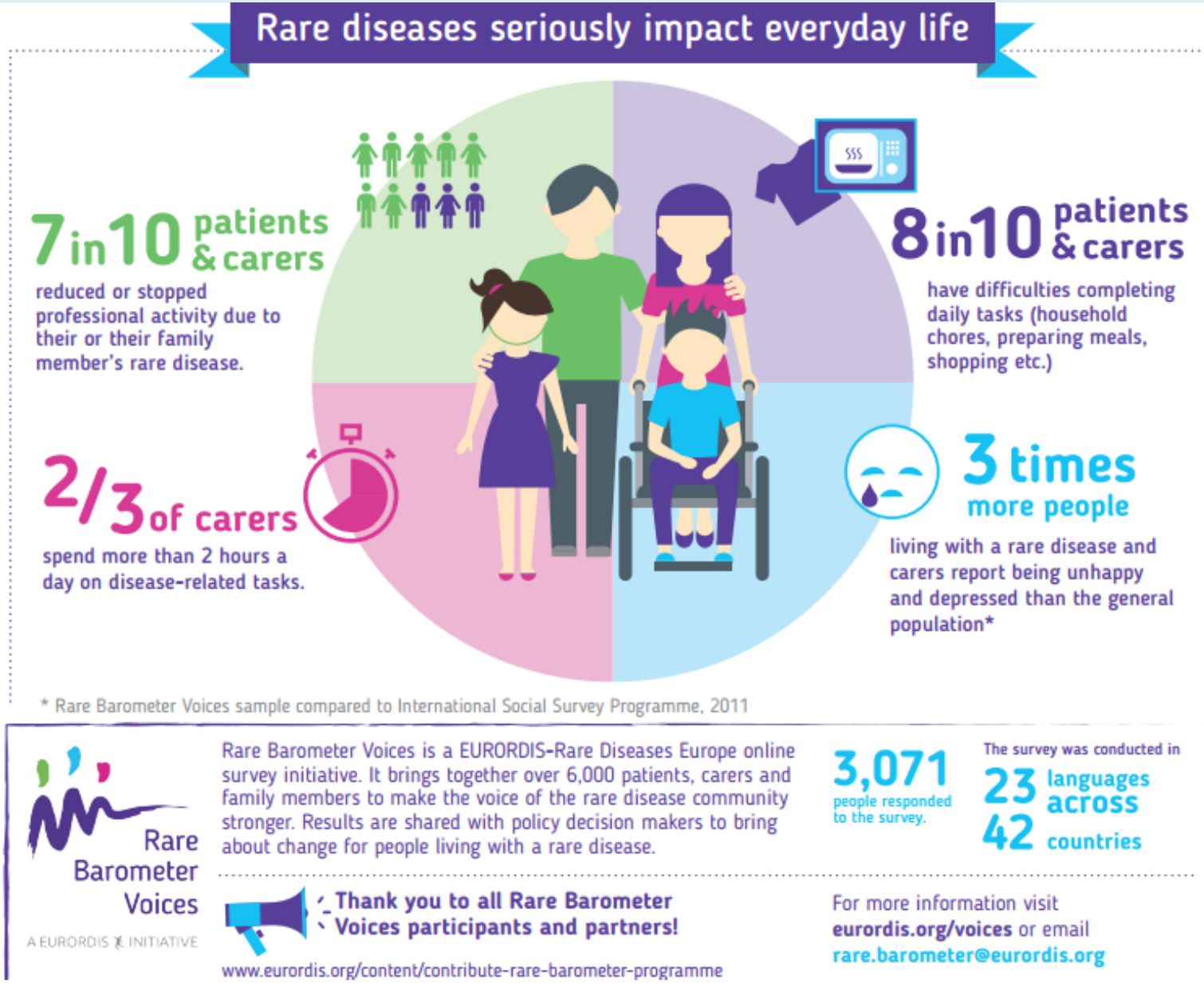
OMIM

613985 BETA-THALASSEMIA

ORPHA

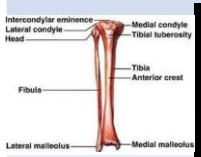
ORPHA:231214 Beta-thalassemia major

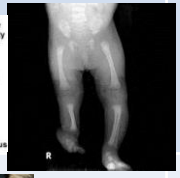
Societial Support



Fibula Hypoplasie –
ICF International Classification, Functioning, Disability and Health

First Feature



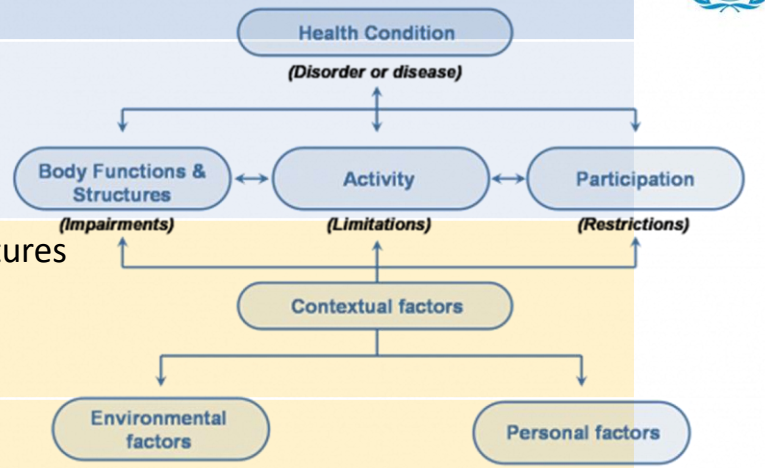


Diagnosis
Fibula Hypoplasia

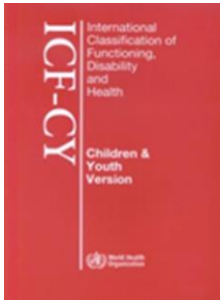
Medical Guideline

ICF : Body Functions & Structures

ICF: Activity
Participation



World Health Organization



Building the Rare Disease

knowledge and information ecosystem

Home / Social support / 360 Using communication devices and techniques

Social Support

Using devices, techniques and other means for the purposes of communicating, such as calling a friend on the telephone. Inclusions: using telecommunication devices, using writing machines and communication techniques.

Using communication devices and techniques → using devices, techniques and other means for the purpose of communicating.

This occurs in Oculo-Auriculo-Vertebral-syndrome

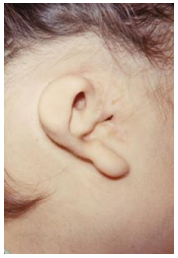
Oculo Auriculo Vertebral Syndroom

ICF-d Reference

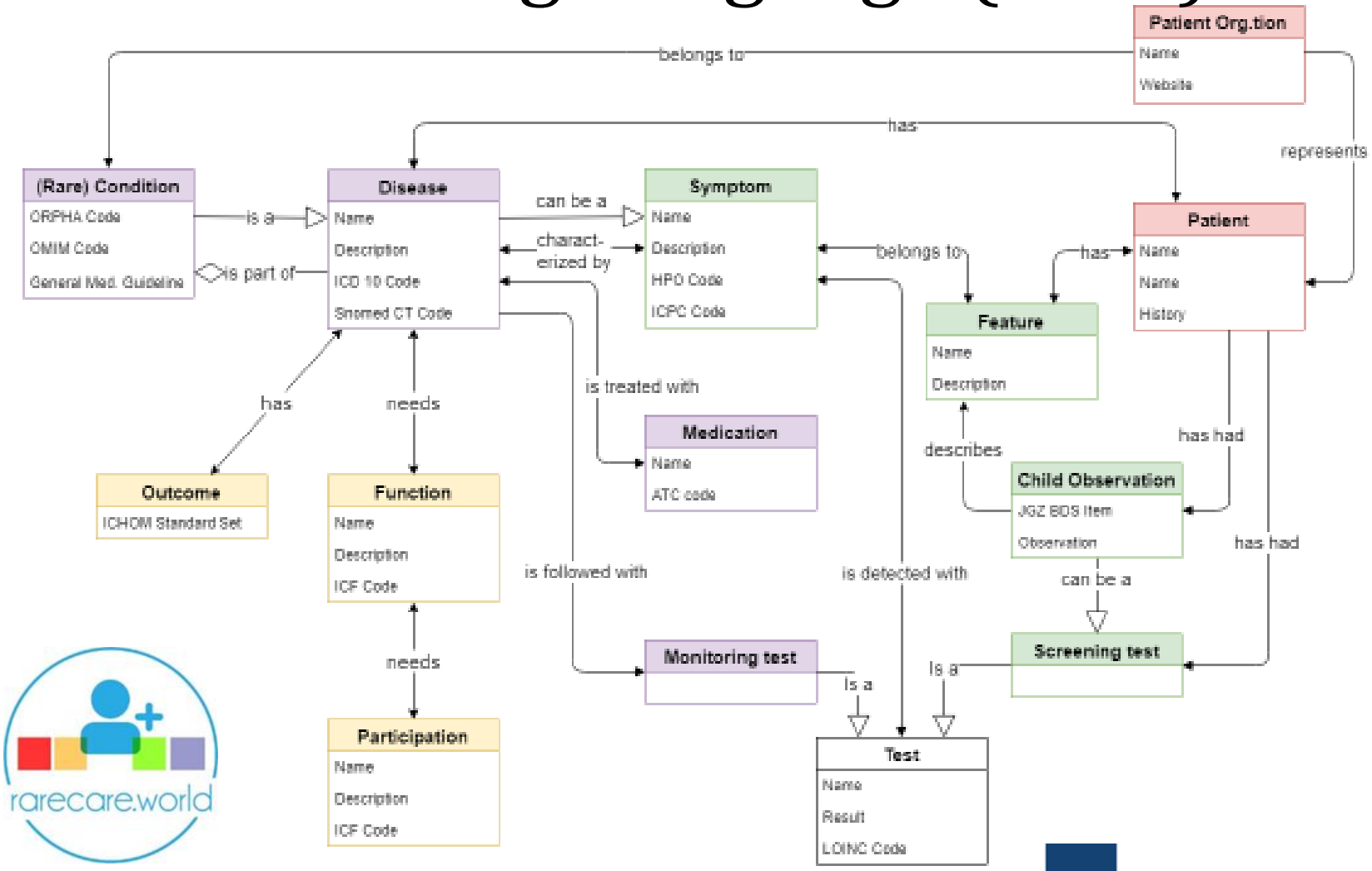
d 360*

Disability

b 230 Hearing functions



Digital Child Health
Unified Modeling Language (UML)



Ministerie van Volksgezondheid, Welzijn en Sport

Digitale gegevens uitwisseling

90% agree — To access their own health data (requiring interoperable and quality health data)

80% agree — To share their health data (if privacy and security are ensured)

80% agree — To provide feedback on quality of treatments

Arnhem 2019

Kolkata, India

Een persoonlijk gezondheid dossier
op basis van internationale classificaties/ terminologieën

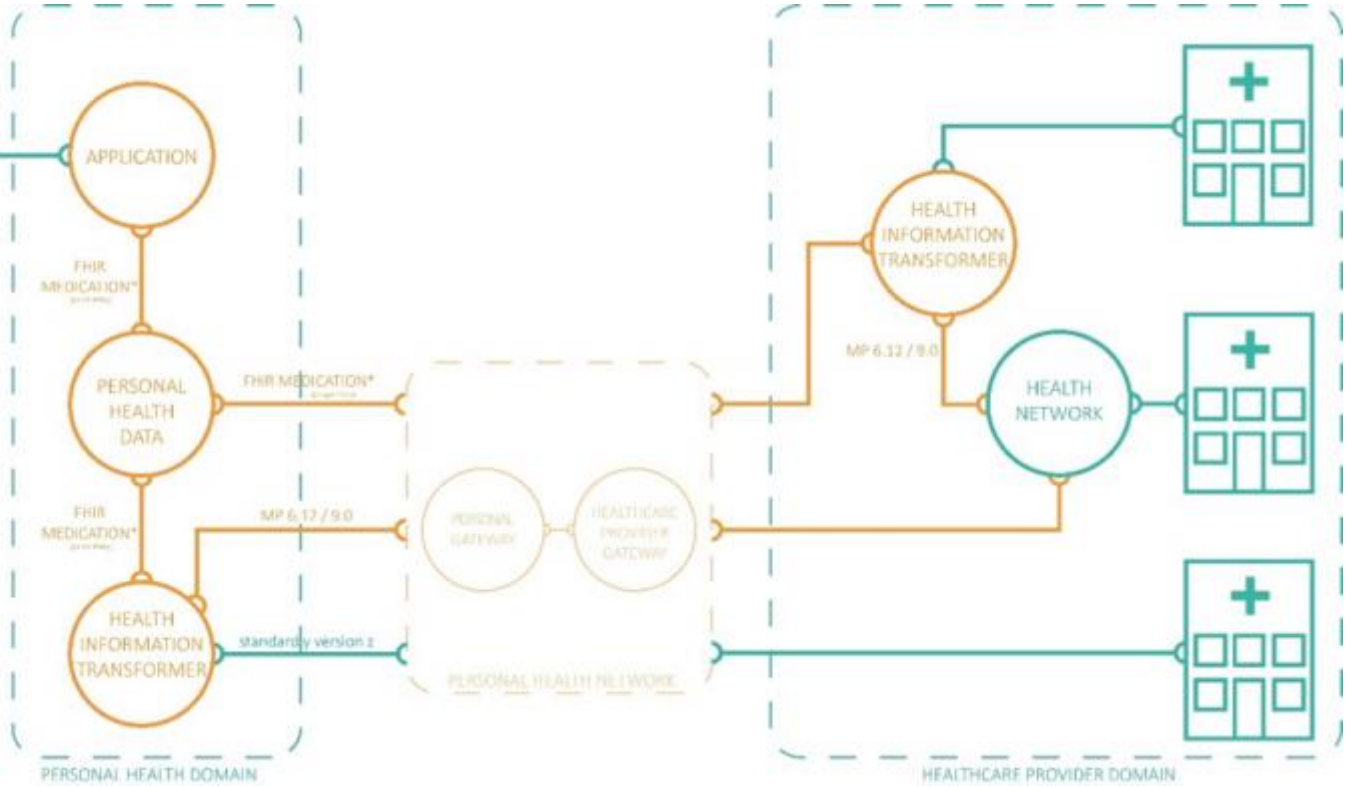
MedMij

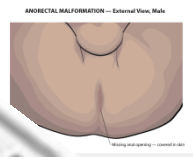
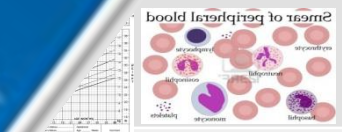
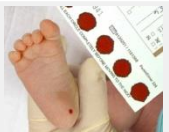
Grip op je eigen gezondheidsgegevens

PATIENT ORGANIZATIONS
INFORMATION IS OF VITAL IMPORTANCE
CONNECT

LOINC® from Regenstrief

751-8 Neutrofielen in bloed



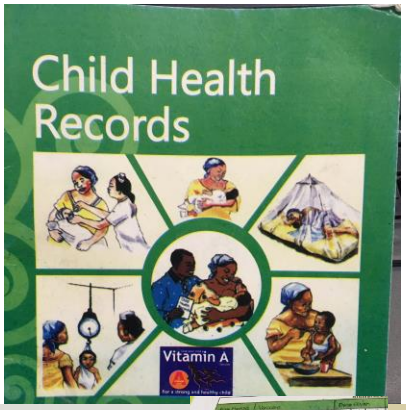
10 Rare Diseases	Prim… …eature	Open information	Code
Anal Atresia	C…	 	ICD
Craniosynostosis Treacher Collins			Code Dutch child health
Cystic Fibrosis			ICD Orphacode
Duchenne Muscular …			ICF - CY, …OINC
Fibrodysplasi… Progressiva			…ode Dutch child health
Glaucoma / … Hurler S cornea…			ICD
Neurofibromatosis			Code Dutch child health
Rett syndrome	H…		ICF- CY cm
Shwachman Diamond Syndrome	Growt… neutrophi…	 	Kg, cm, LOINC
Sickle Cell Disease	Heel Stick Scree…		ICD, LOINC, orphacode

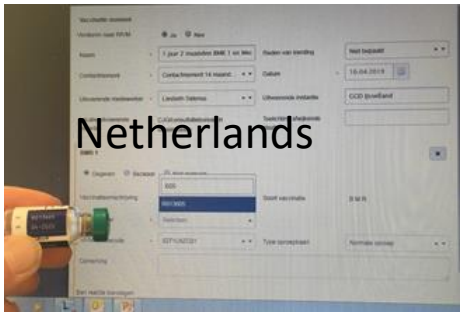
Global Consensus in CHILD HEALTH

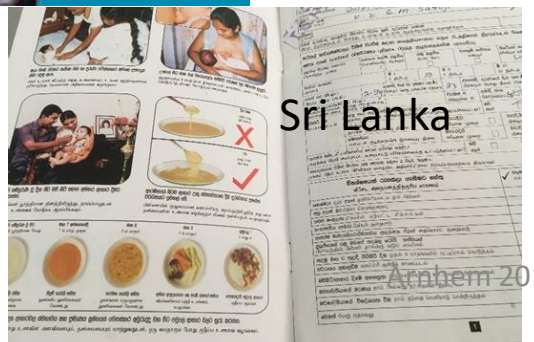
Global Preventive Child Health Checks
Home-based records

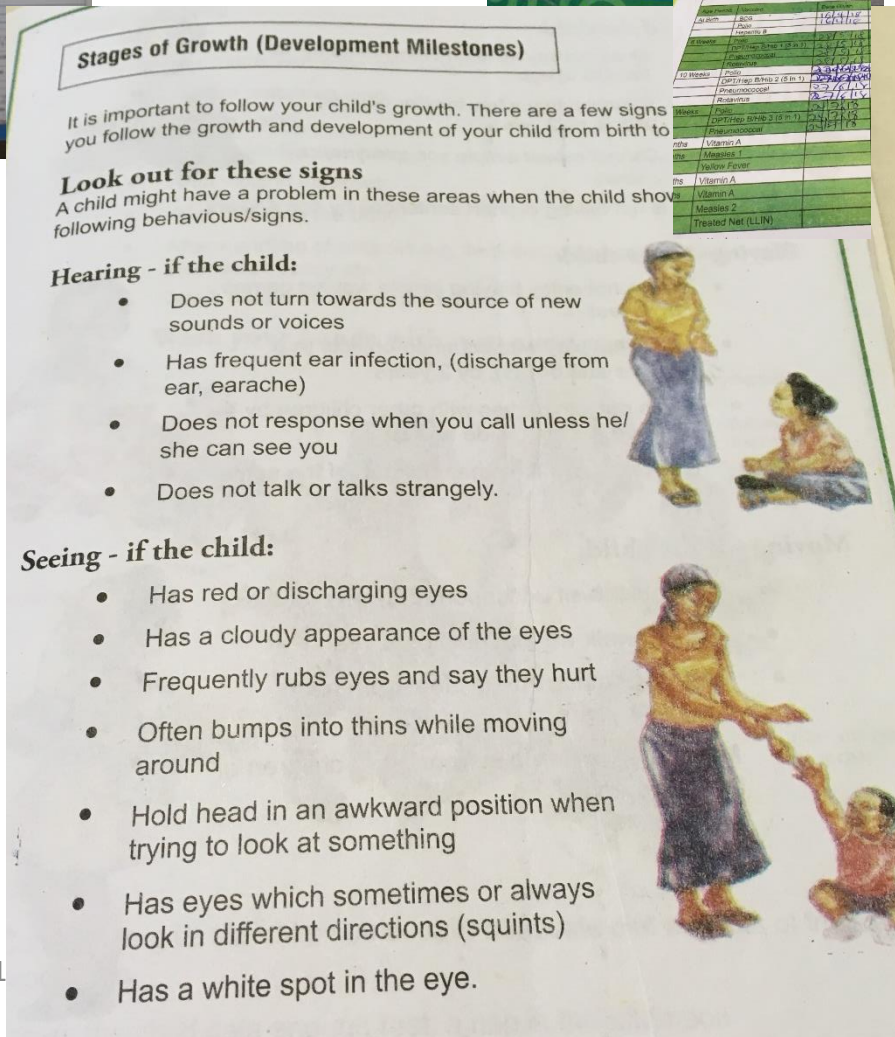
- Growth, developement, physical examination,
- Family history, heel stickscreening, hearing
- Vaccination

E- Health









CONGRES
ARCHITECTUUR IN DE ZORG

Dank

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