

Een data model voor zeldzame ziekten

Liesbeth Siderius

Kinderarts, GGD IJsselland

Rare Disease Working Group

European Academy of Paediatrics

Shwachman Syndroom Support Holland



Stichting Shwachman syndroom

Support Holland



IJsselland



CONGRES
ARCHITECTUUR IN DE ZORG

Arnhem 2019



Zeldzaam is 'gewoon' in de kindergeneeskunde



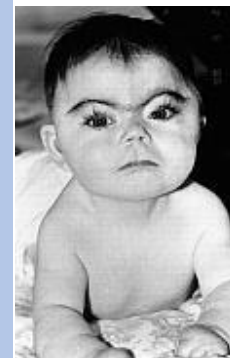
- In Europa is een ziekte definiëerd als zeldzaam als het minder dan **1: 2000** mensen treft.
- Er zijn tussen de **6000** en **8000** zeldzame aandoeningen: **5-8%** van een populatie
- 80% van de zeldzame aandoeningen hebben een genetische oorzaak, en zijn vaak **chronisch** en levensbedreigend
-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



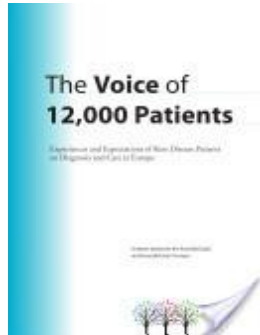
De last van zeldzame ziekten is aanzienlijk, met ongeveer 350 miljoen mensen aangedaan wereld wide.



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



Listen to the Voices of 12,000 Patients 2009

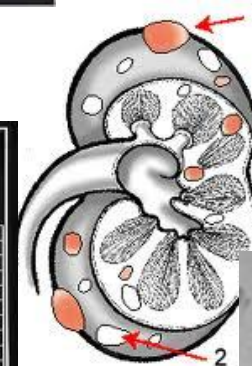
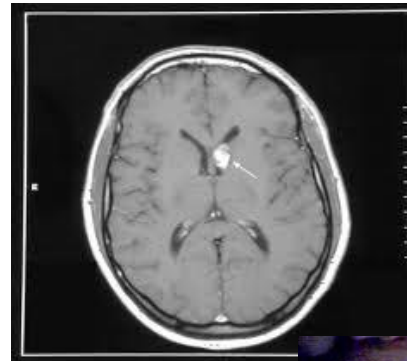
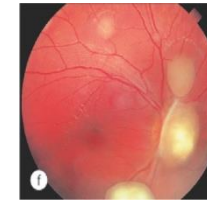


Na de **eerste symptomen** een gevecht om de diagnose dat jaren kan duren.

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



Tuberous sclerosis complex





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	
EMT	: is bevoegdheid

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

Onderzoek en/of onderzoeken worden in overleg met je arts

Nazorg

De nazorg is bedoeld als ondersteuning van de thalassituatie.

Het gaat daarbij vooral om de zelfredzaamheid en hulp bij financiële, psychologische en maatschappelijke problemen.

Instellingen

Algemeen Maatschappelijk Werk:

kan hulp bieden aan patiënt, ouders en familie op sociaal, juridisch en financieel gebied.

WMO (Vereenzamenende 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, tertiële preventie.

Heed 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.

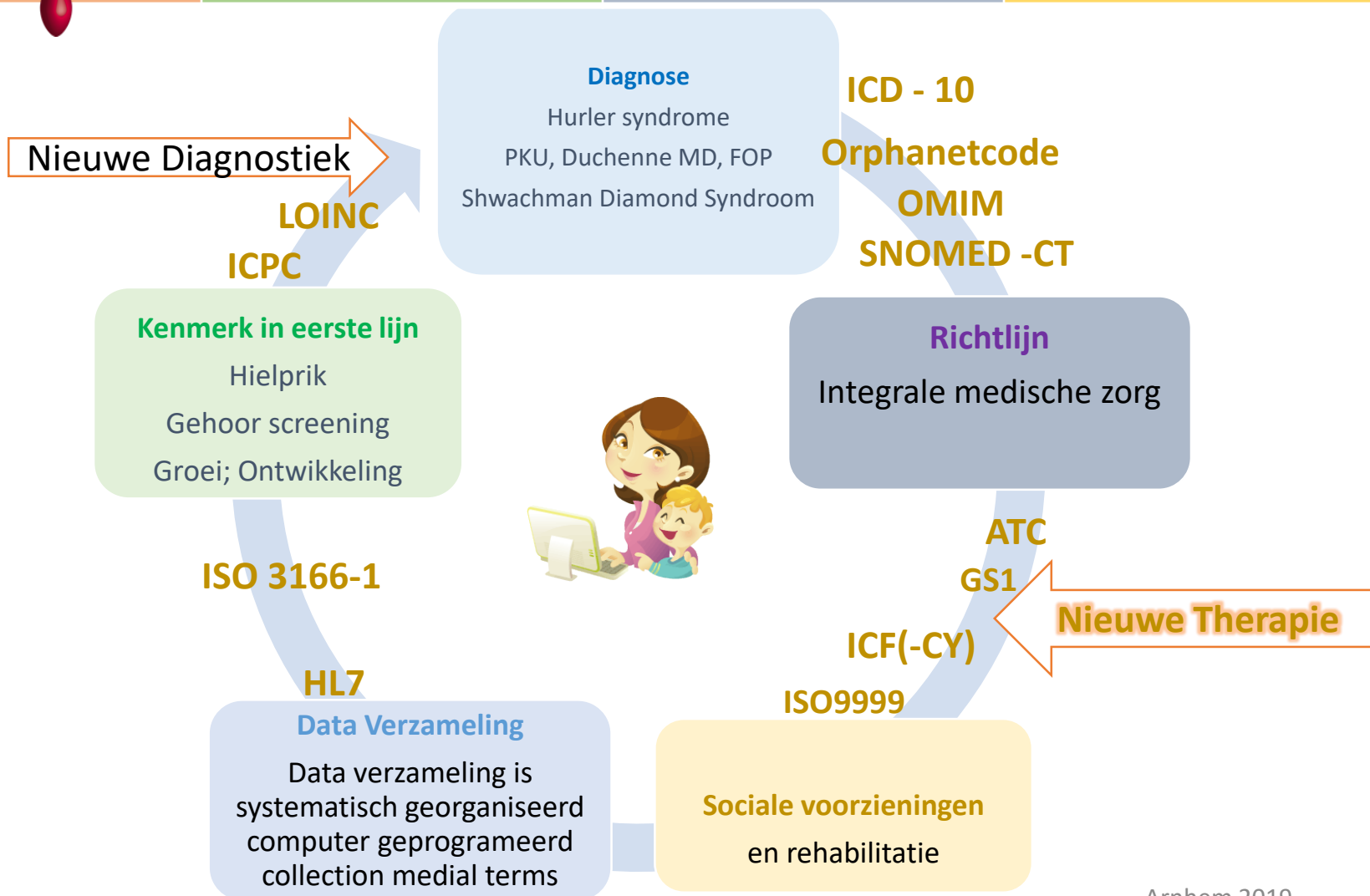
Is belangrijk voor een betere kwaliteit van leven. De behandeling kan plaatsvinden in het ziekenhuis, de polikliniek of thuis.

Poliklinische behandeling en in de thalassituatie

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 Stichting Shwachman syndroom Support Holland	Groei achterstand Herhaalde infecties (LOINC)	Guideline SDS (Orphanetcode; SNOMED, ATC e.a.)	Vaak ziek, Moe, Klein (ICF)



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

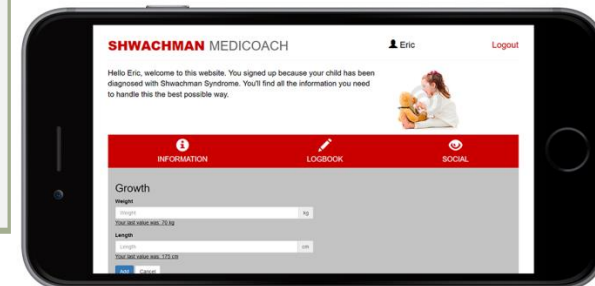
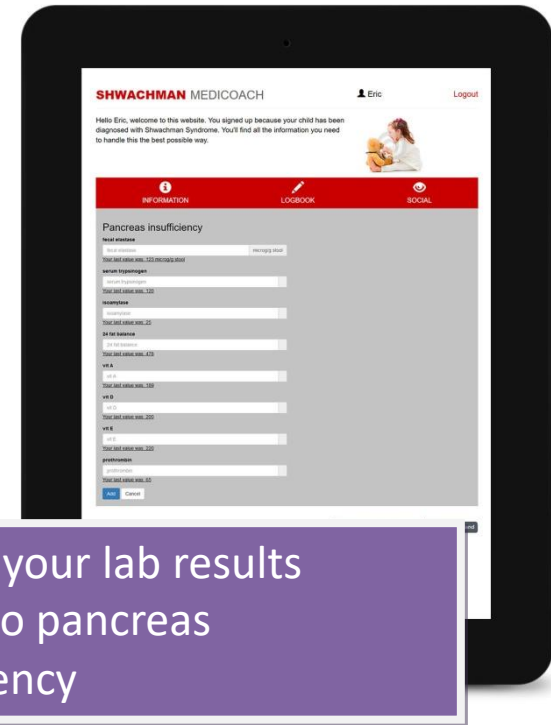
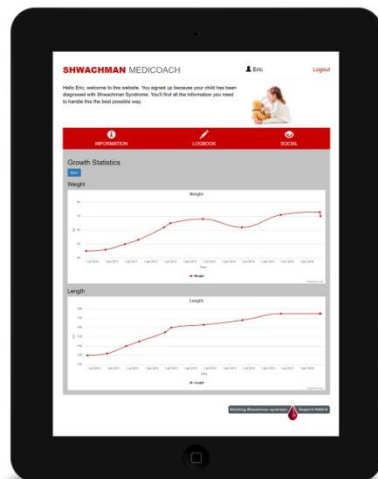
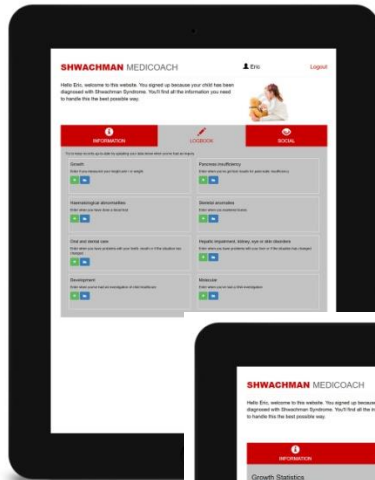
Information on SDS and the use of the application

Overview of the digital guideline

Register your lab results related to pancreas insufficiency

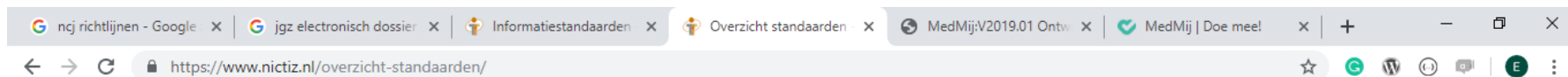
Collect weight and length data to create a SDS growth curve

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Vroeg signaleren in de Eerste Lijn 2015-2016

Met dank aan:



Home Sectoren Standaardisatie Programma's Publicaties Over Nictiz Agenda English

Home / Overzicht standaarden

Overzicht standaarden

PALGA-Thesaurus, iWLZ, FHIR, SNOMED CT. Duizelt het u ook wel eens? In de zorg komen veel standaarden voor. Hieronder vindt u relevante informatie over alle bij ons bekende standaarden.

Mist u een standaard of klopt de informatie volgens u niet? Laat het ons weten via e-mail: standaarden@nictiz.nl.

10 ▼ weergaven per pagina

◀ 1 2 3 4 ... 17 ▶

Naam	Model	Type	Beschrijving	Adoptiegraad	Vergelijk
AZR	●	Wet- en regelgeving	Voormalige systematiek	● ● ● ● ●	□

Filters

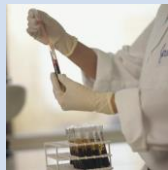
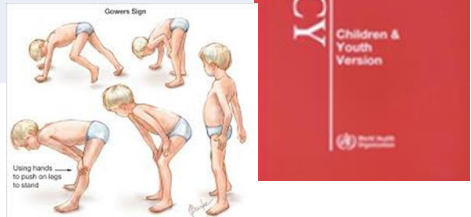
Zoeken...

Interoperabiliteitsmodel

- ☐ ● Organisatiebeleid (2)
- ☐ ● Zorgproces (1)
- ☐ ● Informatie (101)
- ☐ ● Applicatie (56)
- ☐ ● IT-infrastructuur (7)
- ☐ ● Informatiebeveiliging (4)
- ☐ ● Wet- en regelgeving (5)



World Health Organization

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



Find and share knowledge about
Rare diseases all over the world

 RECOGNIZE IDENTIFYING AT-RISK FACTORS IDENTIFY >	 RARE CONDITIONS IMPROVE THE LIVES OF PEOPLE KNOWING >	 SOCIETY ACHIEVING GREAT THINGS IN LIFE SUPPORT >	 PATIENT ORGANIZATIONS INFORMATION IS OF VITAL IMPORTANCE CONNECT >
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Ministerie van Volksgezondheid,
Welzijn en Sport

Arnhem 2019

Eerste Symptomen Preventieve Jeugd Gezondheid Zorg

RECOGNIZE

IDENTIFYING AT-RISK FACTORS

IDENTIFY >

ontwikkeling

Lichamelijk onderzoek

Vaccinatie

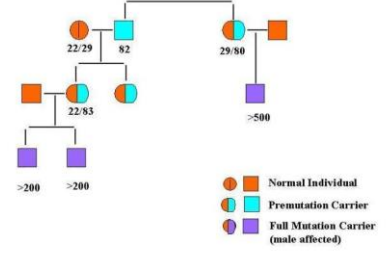
Zwangerschap

Heel prik

Thuis

Groei

Fragile X Syndrome Pedigree



© 2005, Laurie Ann Demme, M.D.

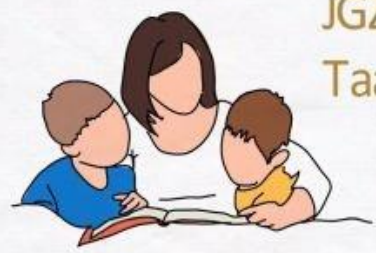
Eerste Symptomen Preventieve Jeugd Gezondheid Zorg



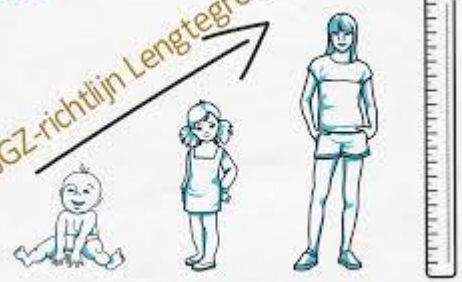
**Fibrodysplasia Ossificans
Progressiva
1:2.000000
SPIER wordt BOT**



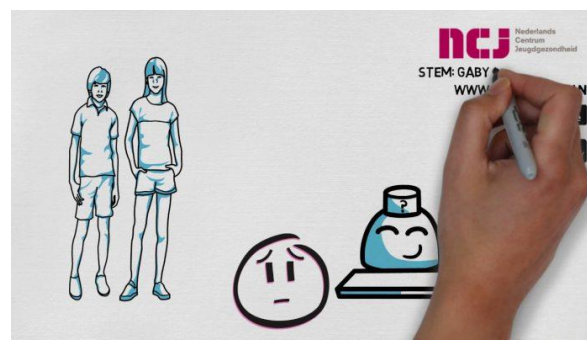
JGZ-richtlijn Taalontwikkeling



JGZ-richtlijn Lengtegroei



STEM GABY



2015 Recommendations for Preventive Pediatric Health Care

Bright Futures/American Academy of Pediatrics

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.

These guidelines represent a consensus by the American Academy of Pediatrics and the American Academy on Child and Adolescent Psychiatry. The AAP continues to emphasize the great importance of comprehensive health supervision and the need to avoid fragmented care. Refer to the specific guidance by age as listed in Bright Futures Guidelines for Health Supervision and Adolescent Care, 3rd ed. Elk Grove Village, IL: American Academy of Pediatrics; 2014.

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

	INFANCY										EARLY CHILDHOOD									
AGE ¹	Prenatal ²	Newborn ³	1 mo	2 mo	3 mo	4 mo	5 mo	6 mo	9 mo	12 mo	15 mo	18 mo	24 mo	30 mo	36 mo	48 mo	60 mo	72 mo	84 mo	96 mo
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 EXAMINATION¹³																				

- Created date
- Relevance ▼
- Name

Current Filter

- Type: Feature
- Clear my filters
- (-) Feature (3)

Type

- Symptom (3)
- Code (2)
- Social Support (2)
- General Medical Guideline (1)
- News (1)
- Rare Condition (1)
- (-) Feature (3)

Rare Condition

Feature

... sometimes curved inward. May be a feature of for example Limbs Feet Movement restriction Congenital hallux valgus

Rare Condition

Fibrodysplasia Ossificans Progressiva

Symptom

Congenital hallux valgus

Abnormality

Physical Exam

Limbs

Feet

Fibrodysplasia Ossificans Progressiva

Feature

... Oculo-Auriculo-Vertebral Spectrum (Goldenhar syndrome) Fibrodysplasia Ossificans Progressiva Auriculo-Vertebral Spectrum (Goldenhar Syndrome) Fibrodysplasia Ossificans Progressiva

Rare Condition

Oculo-Auriculo-Vertebral Spectrum (Goldenhar Syndrome)

Fibrodysplasia Ossificans Progressiva

RECOGNIZE

IDENTIFYING AT-RISK FACTORS

IDENTIFY >

Basis data set Electronisch Dossier

Contactmoment	
Vragenlijst peuters	MIE
Activiteit	
Groei	
Terugkerende anamnese	
Functioneren	
Algemene indruk	
Ontvangen zorg	
Huid Haar Nagels	
Hoofd Hals	
Gehooronderzoek	
Oogonderzoek	
Romp	
Hartonderzoek	
Heupen	
Bewegingen	
Genitalia	

met gevraagd geen bijzondere

Hoofd Hals

Schedel

OREN

Bijzonderheden uiterlijk

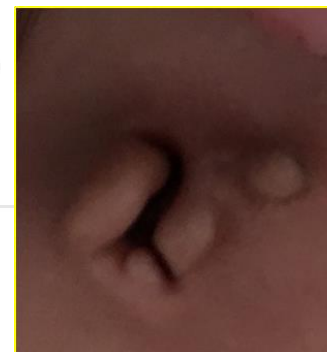
oor rechts



- ☐ Afwijkende vorm kraakbeinig deel van het oor
- ☐ Afwijkende stand
- ☐ Lage-implantatie
- ☐ Resten van kieuwboogspijten
- ☐ Bij-oorje
- ☐ Anders



Microtia
(klein oor)
HP:0008551



Checklist

- Past History (6) ▼
- Laboratory Tests (1) ▼
- Present History (2) ▼
- Measurements at Each Visit (2) ▼
- Development Screening (1) ▼

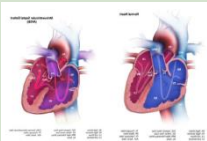
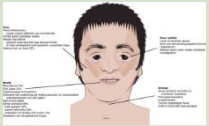
Bijzonderheden
uiterlijk rechts



IDENTIFY >

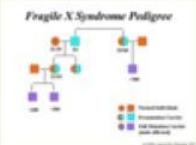
- ☐ Inbreuk
- ☐ Perforatie
- ☐ Loopoor
- ☐ Beluchtingbuisjes
- ☐ Anders

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	Disease	Associated RISK
	Microtia (klein oor)	Conductief gehoorsverlies 
	Oor Aanhangel	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. 

'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	- Thalassemia - Phenyl Keton Uria
Hearing screening <i>Otoacoustic emission</i> 	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
<i>Automated Auditory Brainstem Response</i>			
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 $\alpha3(\text{IV})$. The collagen $\alpha3(\text{IV})$ 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...

RARE
CONDITIONS

IMPROVE THE LIVES OF PEOPLE

KNOWING >

Building the Rare Disease

knowledge and information ecosystem

Thalassemia, 3500 patients have been identified in Sri Lanka.

thalassemia

Thalassemia Major

SEARCH

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 Nieman 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 ($\alpha 2 \delta 2$). Another way of identifying people with **thalassemia** major is neonatal screening. Neonatal screening ...

Rare Condition

Thalassemia major or Beta Thalassemia

Disease

Hemoglobinopathies

Amhem 2019



application
programming
interface (API)



Building the Rare Disease

knowledge and information ecosystem

[Home](#) / [Rare condition](#) / থ্যালাসেমিয়া প্রধান বা বিটা থ্যালাসেমিয়া

থ্যালাসেমিয়া প্রধান বা বিটা থ্যালাসেমিয়া

থ্যালাসেমিয়া জেনেটিকালি (উত্তরাধিকারসূত্রে) রক্তের রোগের একটি গ্রুপ যা সাধারণ এক বৈশিষ্ট্যে 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 ব্যক্তির বিটা থ্যালাসেমিয়া বা বিটা-থ্যালাসেমিয়ার একজন ক্যারিয়ারের হেটারজাইজাস। বিটা থ্যালাসেমিয়া মেজারের ব্যক্তি হ'ল বিটা থ্যালাসেমিয়ার জন্য হোমজাইজাস এবং এভাবে ত্রুটিপূর্ণ জিনের দুটি কপি রয়েছে এবং এই রোগটি বিকাশ করে: থ্যালাসেমিয়া প্রধান। জিনের সম্পূর্ণ অনুপস্থিতিটি β^0 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

Rare diseases seriously impact everyday life

7 in 10 patients & carers

reduced or stopped professional activity due to their or their family member's rare disease.



8 in 10 patients & carers

have difficulties completing daily tasks (household chores, preparing meals, shopping etc.)



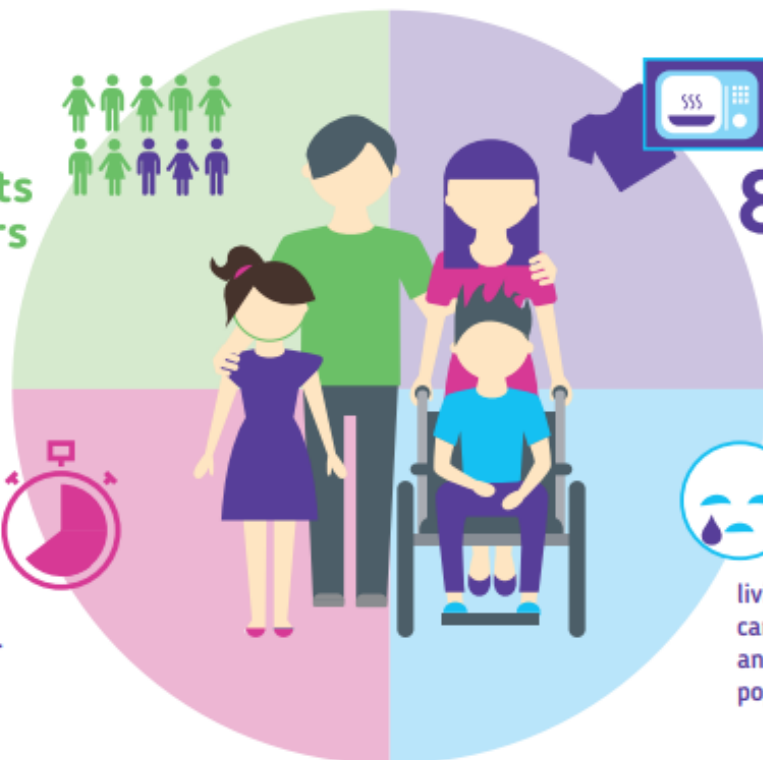
2/3 of carers

spend more than 2 hours a day on disease-related tasks.



3 times more people

living with a rare disease and carers report being unhappy and depressed than the general population*



* Rare Barometer Voices sample compared to International Social Survey Programme, 2011



Rare Barometer Voices is a EURORDIS-Rare Diseases Europe online survey initiative. It brings together over 6,000 patients, carers and family members to make the voice of the rare disease community stronger. Results are shared with policy decision makers to bring about change for people living with a rare disease.



Thank you to all Rare Barometer Voices participants and partners!

www.eurordis.org/content/contribute-rare-barometer-programme

3,071
people responded to the survey.

The survey was conducted in
23 languages across
42 countries

For more information visit
eurordis.org/voices or email
rare.barometer@eurordis.org

SOCIETY

ACHIEVING GREAT THINGS IN LIFE

SUPPORT >

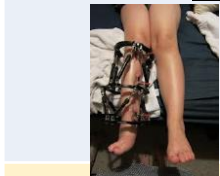
Fibula Hypoplasie – ICF International Classification, Functioning, Disability and Health



First Feature



Diagnosis
Fibula Hypoplasia



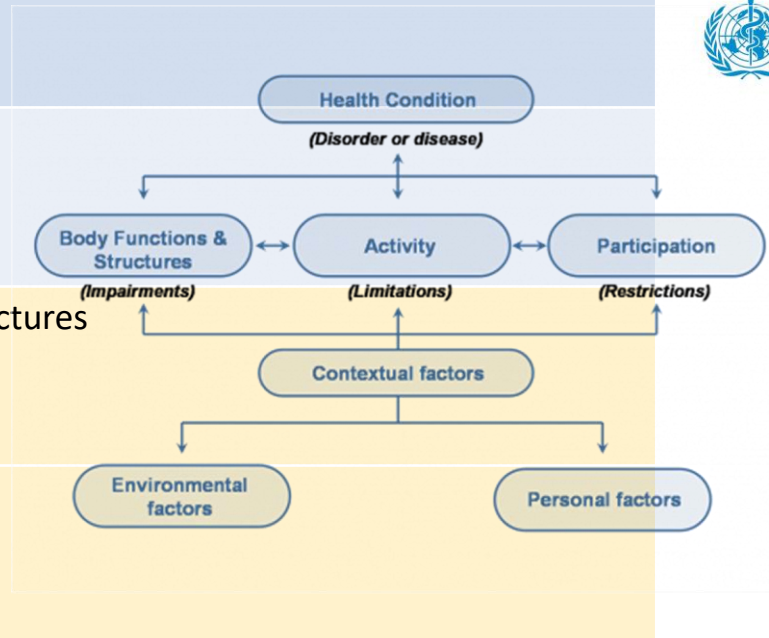
Medical Guideline



ICF : Body Functions & Structures



ICF: Activity
Participation



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

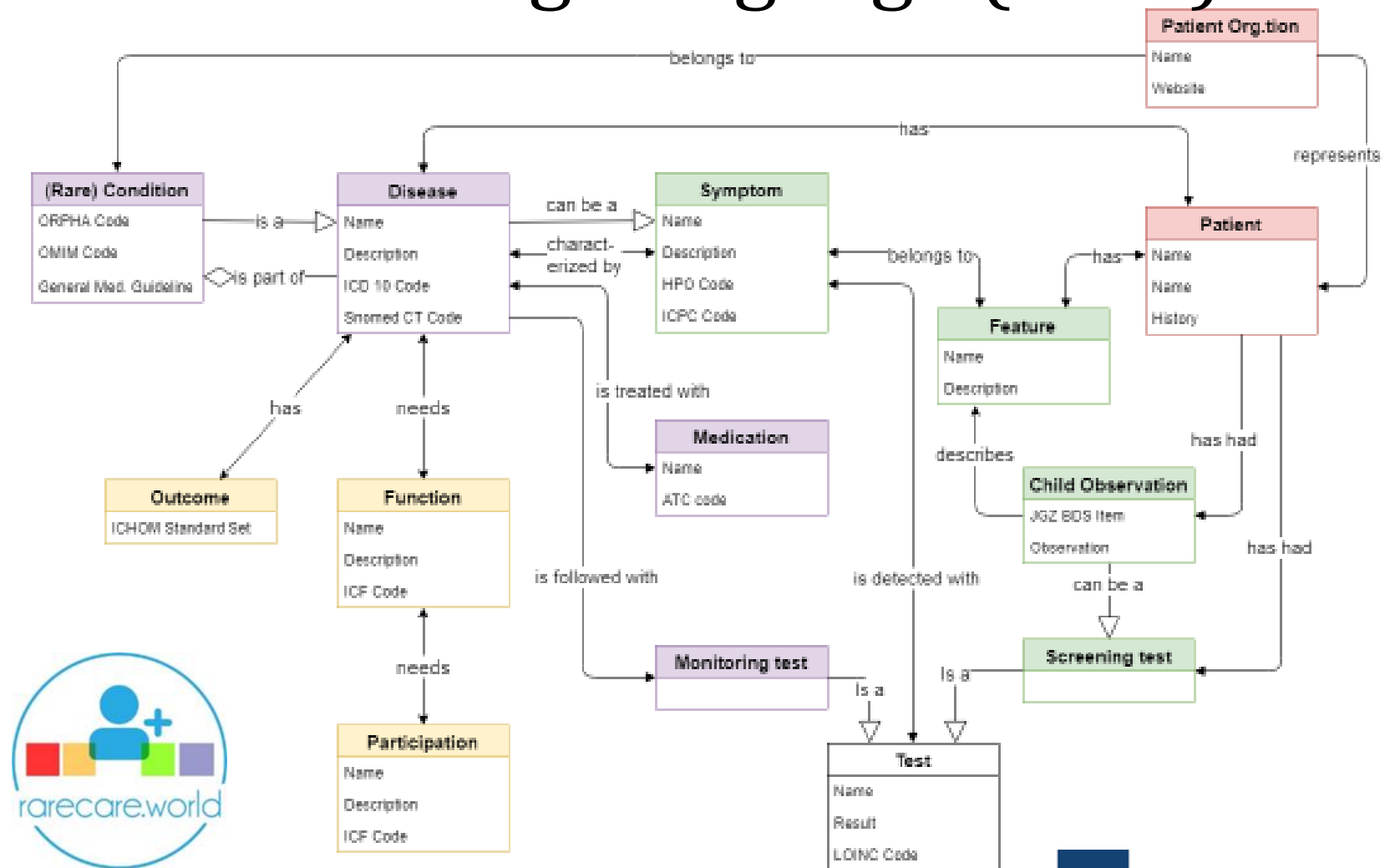
d 360*

Disability

[b 230 Hearing functions](#)



Digital Child Health Unified Modeling Language (UML)



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



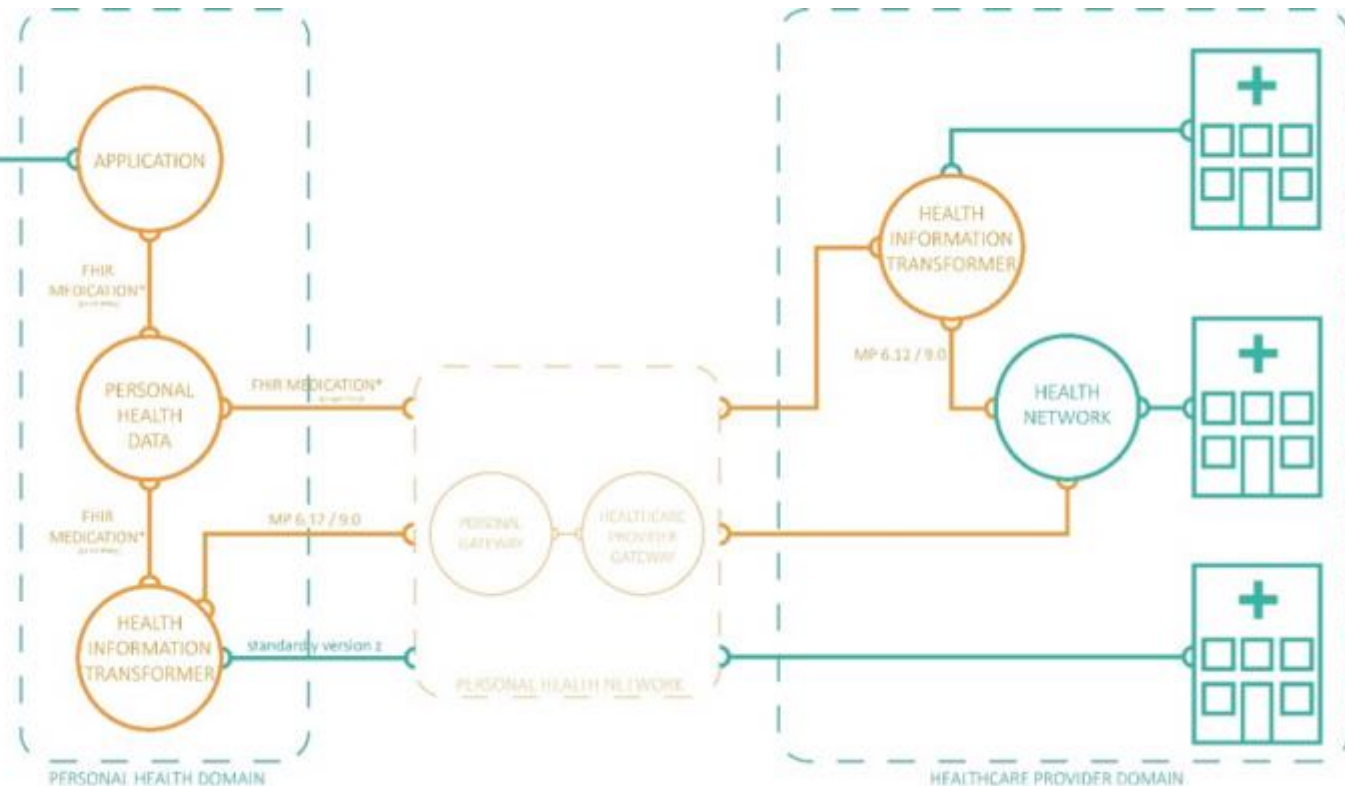
Een persoonlijk gezondheid dossier op basis van internationale classificaties/ terminologieën



Grip op je eigen
gezondheidsgegevens

LOINC®
from Regenstrief

751-8 Neutrofielen in bloed



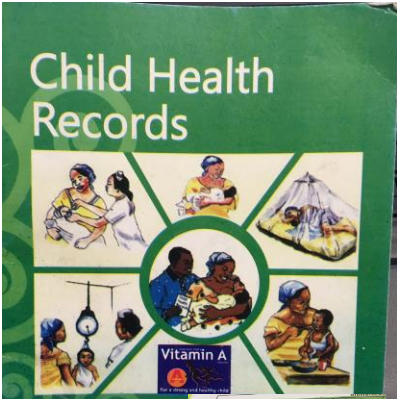
10 Rare Diseases	Primary Feature	Open information	Code
Anal Atresia	C	 	ICD
Craniosynostosis			Code Dutch child health
Treacher Collins			ICD
Cystic Fibrosis			Orphacode
Duchenne Muscular D			ICF - CY, LOINC
Fibrodysplasia			Code Dutch child health
Progressive			ICD
Glaucoma / c			Code Dutch child health
Hurler S cornea			ICF- CY cm
Neurofibromatosis			Kg, cm, LOINC
Rett syndrome			ICD, LOINC, orphacode
Shwachman Diamond Syndrome	Growth, neutrophils	 	
Sickle Cell Disease	Heel Stick Screen		

Global Consensus in
CHILD HEALTH

Global Preventive Child Health Checks

Home-based records

- Growth, development, physical examination,
 - Family history, heel stickscreening, hearing
 - Vaccination
- E- Health



Stages of Growth (Development Milestones)

It is important to follow your child's growth. There are a few signs you follow the growth and development of your child from birth to

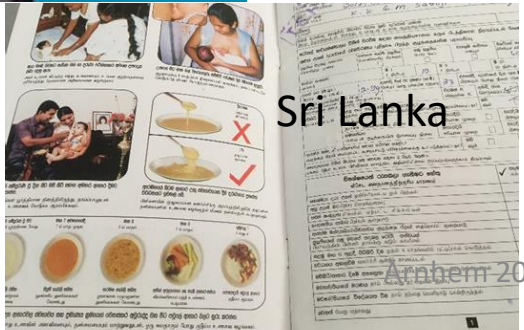
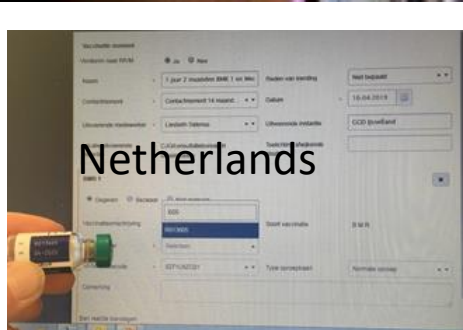
Look out for these signs
A child might have a problem in these areas when the child shows following behaviour/signs.

Hearing - if the child:

- Does not turn towards the source of new sounds or voices
- Has frequent ear infection, (discharge from ear, earache)
- Does not response when you call unless he/she can see you
- Does not talk or talks strangely.

Seeing - if the child:

- Has red or discharging eyes
- Has a cloudy appearance of the eyes
- Frequently rubs eyes and say they hurt
- Often bumps into things while moving around
- Hold head in an awkward position when trying to look at something
- Has eyes which sometimes or always look in different directions (squints)
- Has a white spot in the eye.





CONGRES
ARCHITECTUUR IN DE ZORG

Dank

e.siderius@kpnplanet.nl