# Paediatric care for the child with a rare condition, where East and West meet

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The Netherlands

# Disclosure Statement

I declare that I have no potential conflict of interest.


# Rare is Common in Pediatrics

- A disease or disorder is defined as rare in Europe when it affects less than **1 in 2000.**
- There are between **6000** and 8000 rare diseases: **5-8%** population
- 80% of rare diseases are of genetic origin, and are often **chronic** and **life-threatening**
- 50-75 % manifests in **childhood**, almost each chronic condition in childhood,
- < 5 y 2-3 % have a rare disease

The burden of rare disease is considerable, with around 350 million affected individuals worldwide.
This is in the same range as main and more 'visible' non-communicable diseases such as diabetes.


The goals of the NGO Committee for Rare Diseases are fully aligned with many of the 17 Sustainable Development Goals (SDGs), and support the UN's vision to create a world where every single human can lead a dignified life.

2016

The United Nations Sustainable Development Goals (SDG)
#3 "Ensure healthy lives and promote well-being for all ages".

The United Nations has emphasized the need to:
-end preventable deaths of new-borns and children under five
-end avoidable mortality caused by non-communicable diseases
-achieve universal health coverage
-support the research and development of medicines


# 1 End Poverty in all its forms everywhere

# 3 Ensure healthy lives and promote well-being for all at all ages

# 4: Ensure inclusive and equitable quality education and promote lifelong learning opportunities for all

# 5: Achieve gender equality and empower all women and girls

#10: Reduce inequality within and among countries

#17: Revitalize the global partnership for sustainable development

NGO COMMITTEE FOR RARE DISEASES

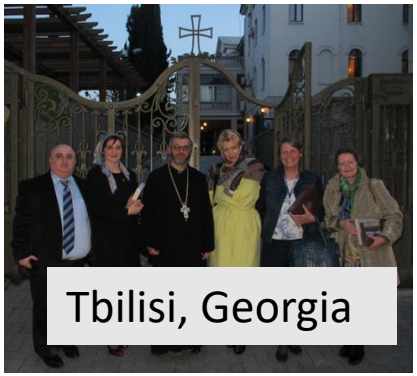
17 PARTNERSHIPS FOR THE GOALS

The NGP **Committee for Rare Diseases** aims to:
act as an open forum to collect, share and disseminate information and research on the global dimensions of rare diseases encourage the dialogue between public, private and civil society groups boost the role of science, technology and innovation as an enabler of the 2030 agenda.

*As pediatricians we share information, on children with rare and disabling conditions and contribute to research, training……*

Panama

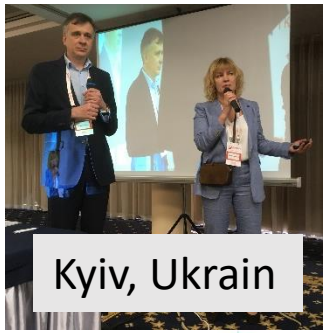

Vilnius, Lithuania


Tbilisi, Georgia

Rare Best Practices


Colombo, Sri Lanka


Xi An, China


Kyiv, Ukrain



NGO COMMITTEE FOR RARE DISEASES

10 REDUCED INEQUALITIES

Persons living with a rare disease tend to remain a **marginalised and invisible population**, with **little information** available about their diseases and very few treatment options. They suffer inequality in accessing health care services and treatment, and in the prices they have to pay, due to their social status or their country of origin.

## *As pediatricians we inform and support patient organisations and individual families*

AlportS Ohrid

Developmental disabilities, Kolkata

DucheneMD Gdansk

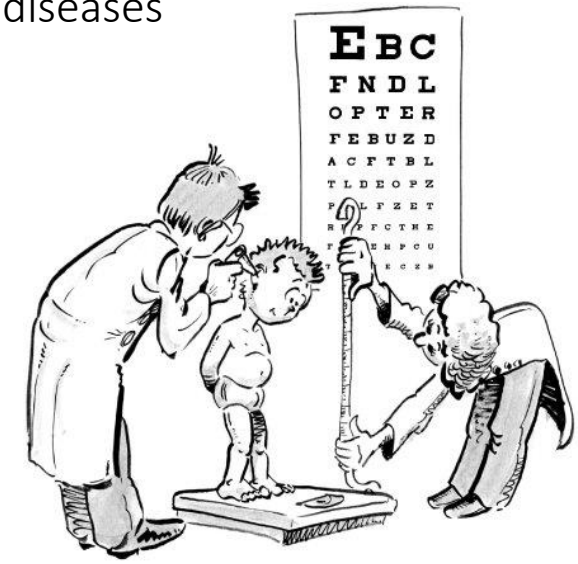
ShwachmanDS Hannover



NGO COMMITTEE FOR RARE DISEASES

The United Nations has emphasized the need to:
-end preventable deaths of new-borns and children under five
-end avoidable mortality caused by non-communicable diseases
-achieve universal health coverage
-support the research and development of medicines

*As pediatricians we provide (Preventive )Child Health Care*





Georgia


Sweden


Lithuania


Sri Lanka

Netherlands

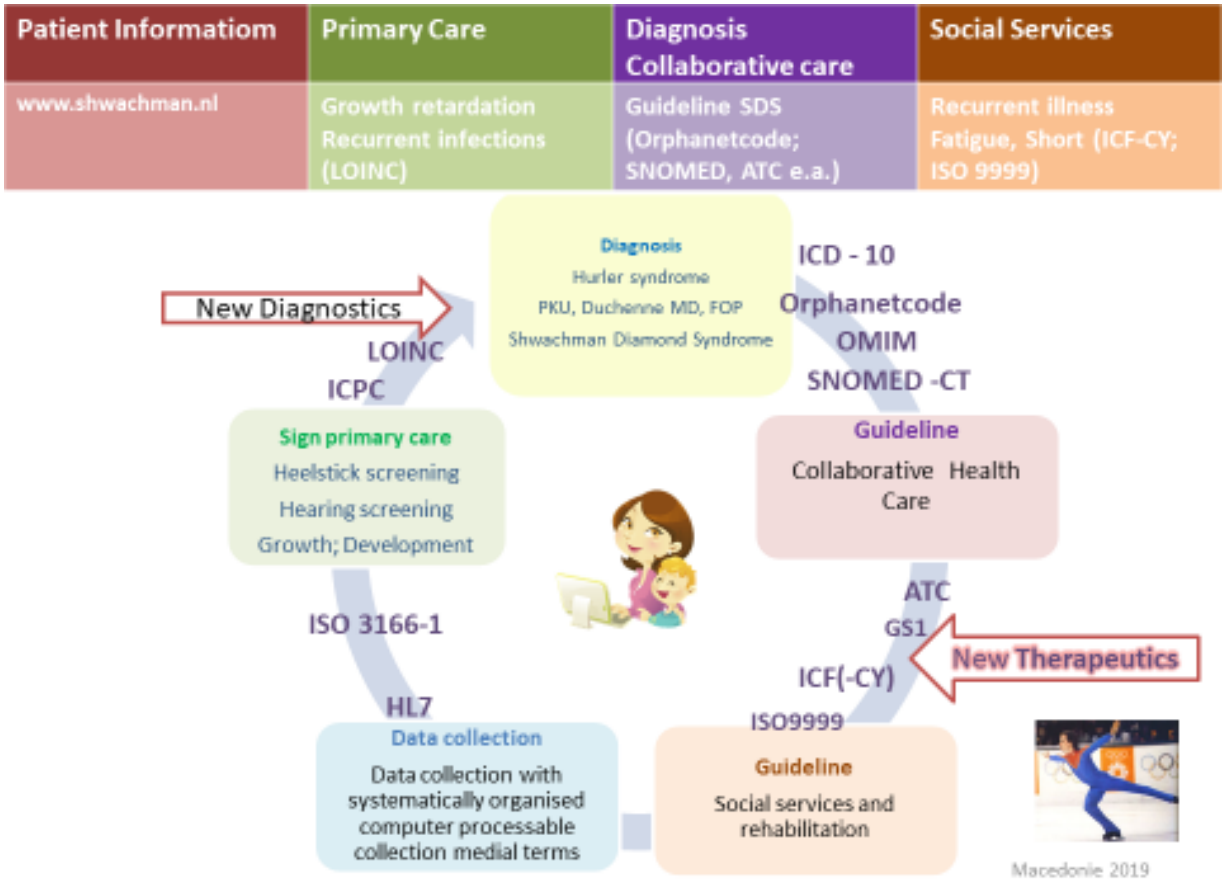
# What can we, pediatricians, do more or better?

Global Child Health Schemes

Implement New Diagnostics

Inform Society

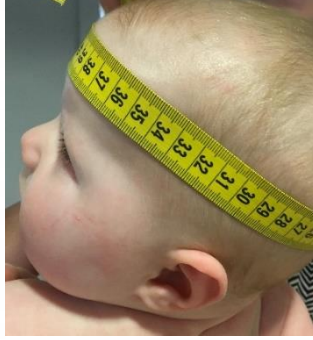
Digital Child Health Data



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

3 GOOD HEALTH AND WELL-BEING

Models of Child Health Appraised
(A Study of Primary Healthcare in 30 European countries)



## 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[2] | Newborn[3] | 3-5 d[4] | By 1 mo | 2 mo | 4 mo | 6 mo | 9 mo | 12 mo | 15 mo | 18 mo | 24 mo | 30 mo | 3 y | 4 y | 5 y |
| **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] | | • | • | • | • | • | • | • | • | • | • | • | • | • | • | • |

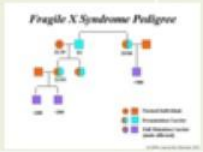
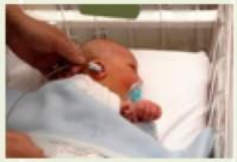
# Global Child Health Schemes

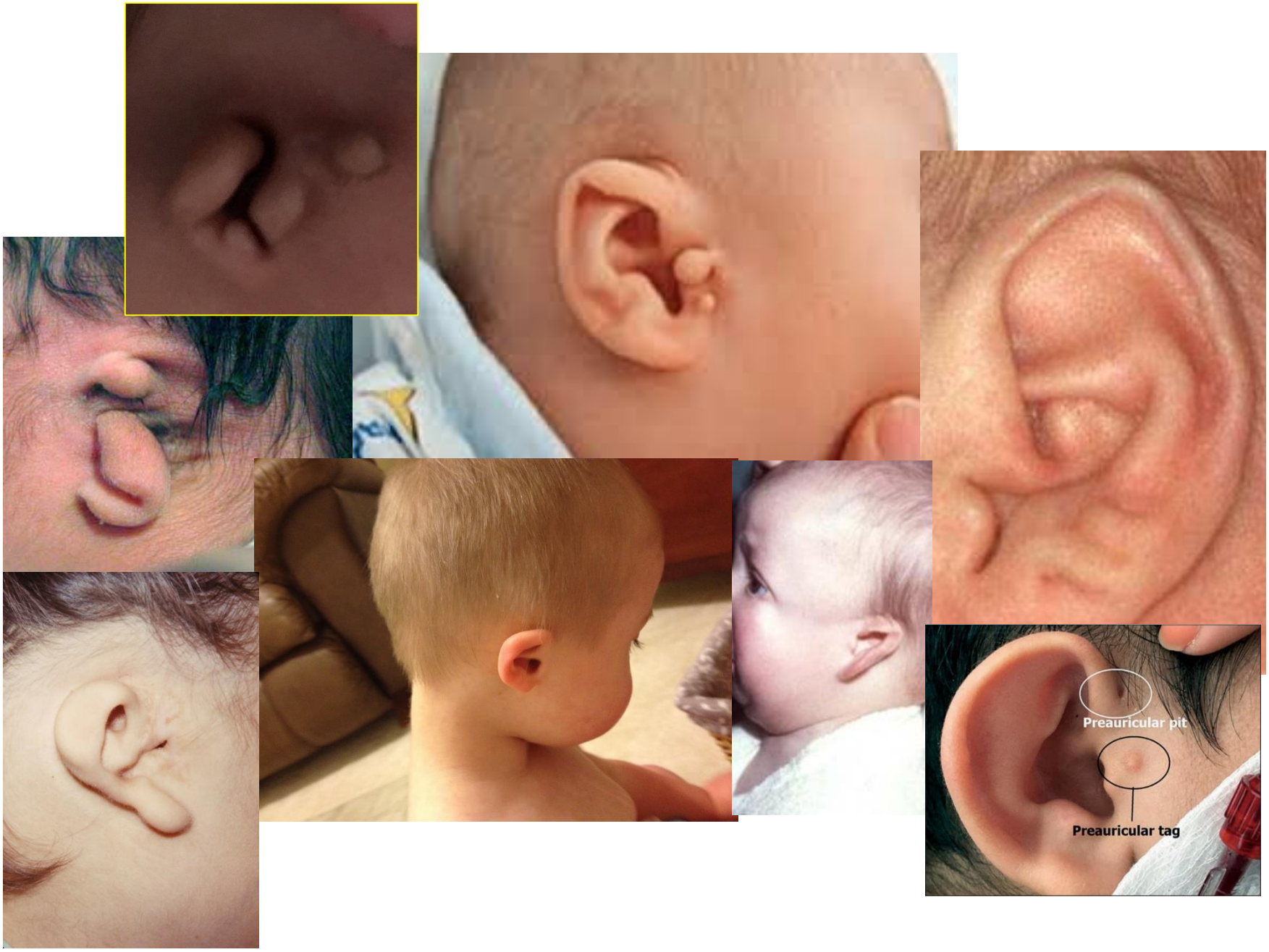
- Collaborate on a minimal global set for measurements, observations and surveillances

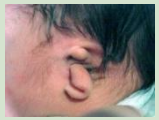
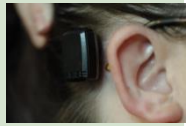
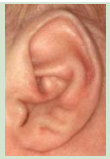
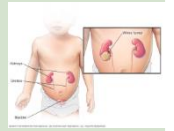
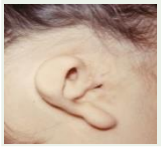
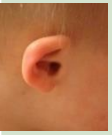
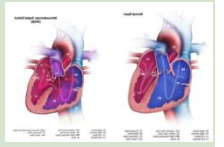
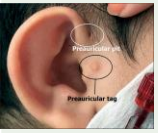
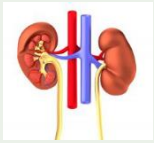
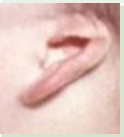
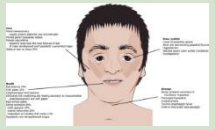
EURORDIS
Rare Diseases Europe

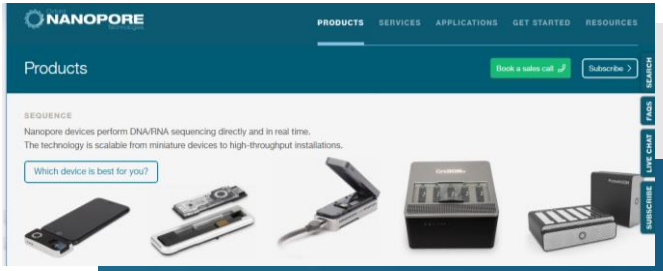
*'After first symptoms, **battle for diagnosis**, which can last for years'*

# 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<br>• Neurofibromatosis |
| Heel stick screening | neonatal | National program | • Thallasemia<br>• Phenyl Keton Uria |
| Hearing screening<br>*Otoacoustic emission*<br>*Automated Auditory Brainstem Response*  | neonatal | Hearing deficit 0,2%<br>> 76% cause identified<br>39% - 60% genetic ,<br>30% aquered (cytomegalovirus)<br>Other causes | • Usher syndrome<br>50% hereditary forms combining deafness and blindness<br>prevalence of 3 to 6.2 per 100,000 |
| Congenital anomaly  | neonatal | Club feet<br>1/700-1/1000 of liveborns | 20% associated with<br>• distal arthrogryposis,<br>• congenital myotonic dystrophy,or other genetic |

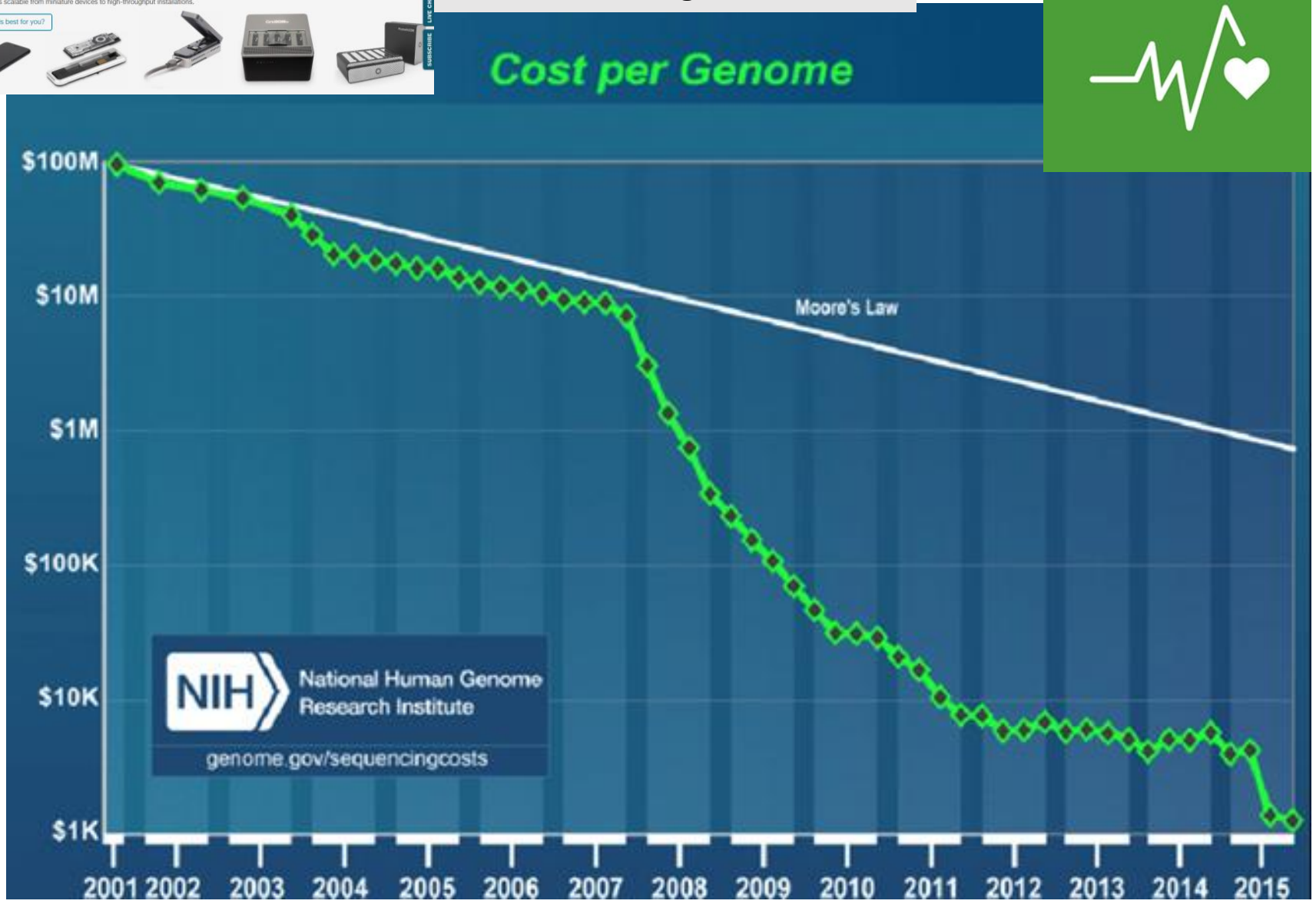

Preauricular pit
Preauricular tag

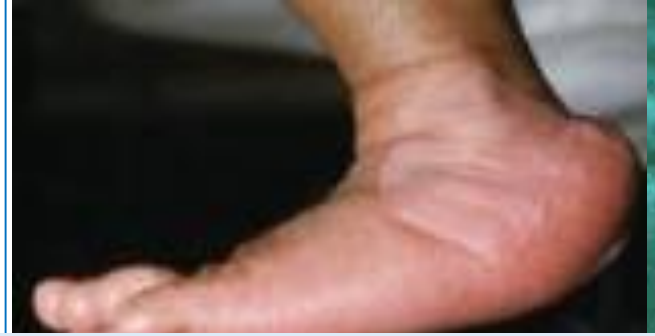
| | Disease | Associated RISK |
|---|---|---|
|  | Microtia | Conductive Hearing loss  |
|  | Ear Tags | The A.A.P. endorses universal **hearing screening** regardless of the presence or absence of preauricular skin. No recommendations or guidelines about the role of renal ultrasonography.  |
|  | Beckwith Wiedemann Syndrome | Most of the **tumors** associated with BWS occur in the first 8–10 years of life with very few being reported beyond this age; most common are Wilms tumor and hepatoblastoma.  |
|  | Goldenhar Syndrome | **Hemivertebrae**  |
|  | Down syndrome | Congenital **cardiac anomalies**  |
|  | Brachio Oto Renal Syndrome | **Renal disease**  |
|  | Treacher Collins syndrome | **Early operations focus on maintaining the airway, protecting the eyes, and auditory neurological development.**  |

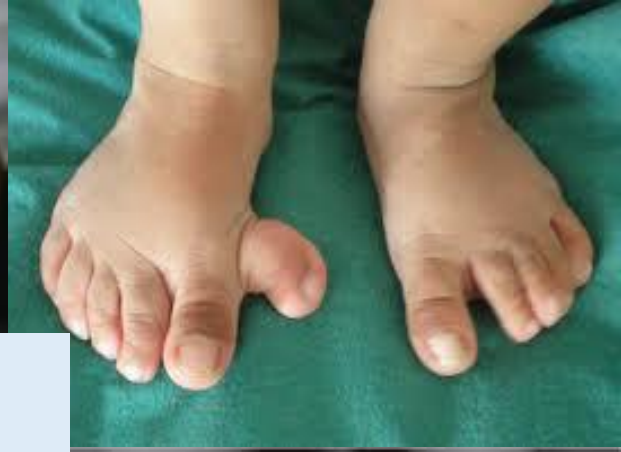



Point of care, bloodspot
3 hours, whole genome

3 GOOD HEALTH AND WELL-BEING

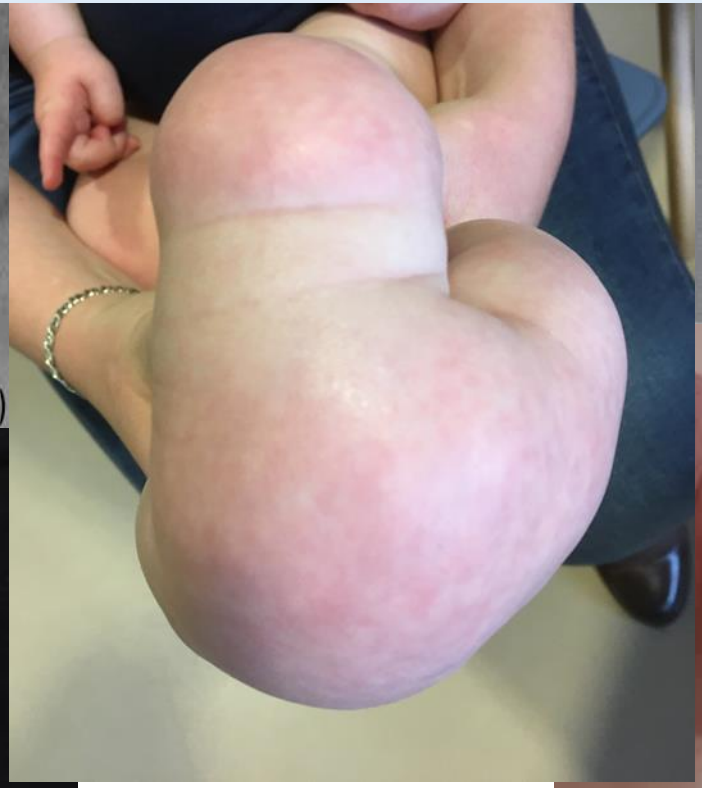


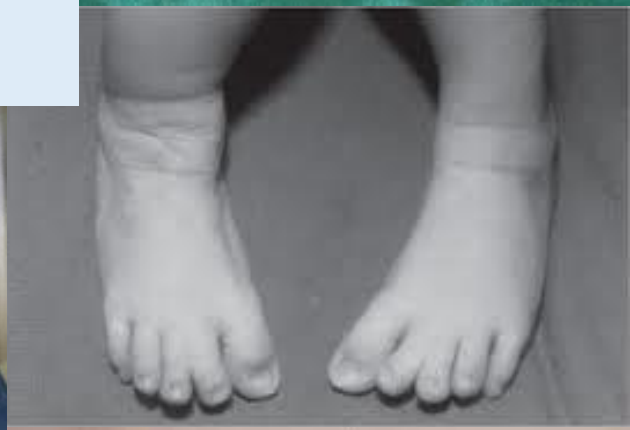


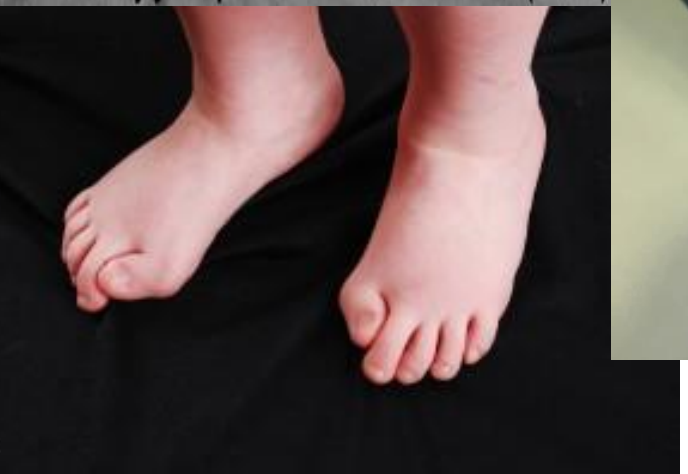


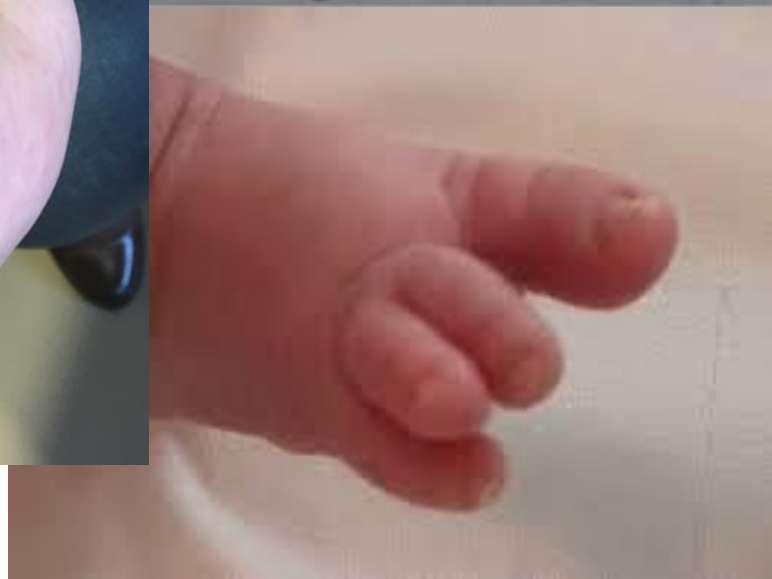
**Implement New Diagnostics**

- Translate new genetic diagnostics in daily practice management







# Girl 11 month
# Vaccination DKTP Hib HepB + Pneu



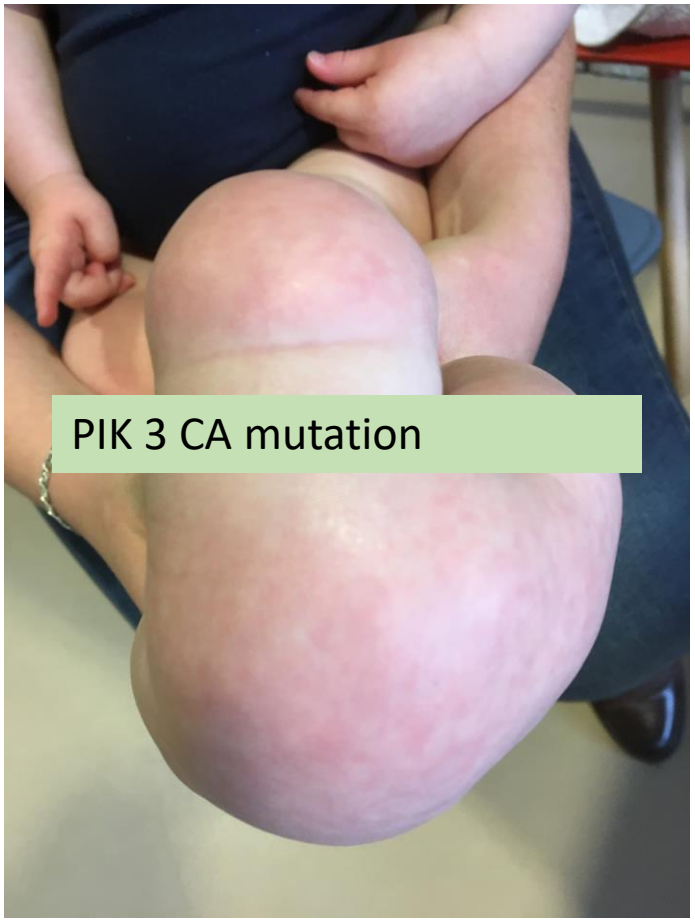
Curr Top Microbiol Immunol. Author manuscript; available in PMC 2011 Aug 31.
Published in final edited form as:
Curr Top Microbiol Immunol. 2010; 347: 21–41.
doi: 10.1007/82_2010_68
PMCID: PMC31
NIHMSID: NIHMS3
PMID: 205

Oncogenic Mutations of PIK3CA in Human Cancers

Yardena Samuels and Todd Waldman



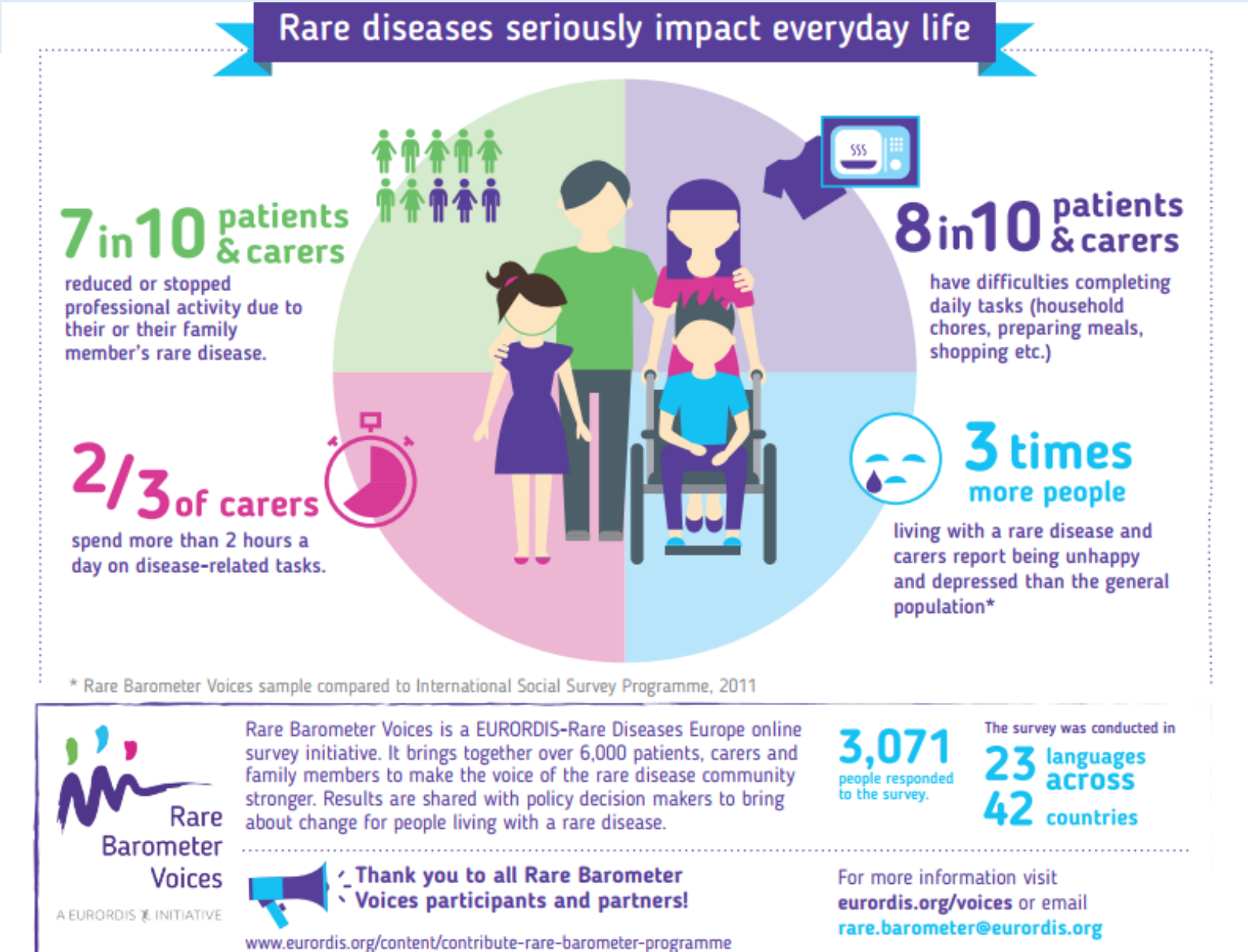
Abstract




What is the clinical diagnosis?
Are there medical guidelines?
What social support is available?

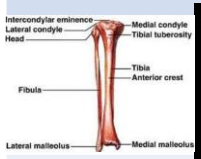
**Inform Society**

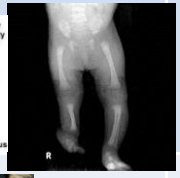
- Learn about the daily life with a rare condition
- Support solutions to help overcome inequalities



# Fibula Hypoplasia – ICF International Classification, Functioning, Disability and Health

**First Feature**



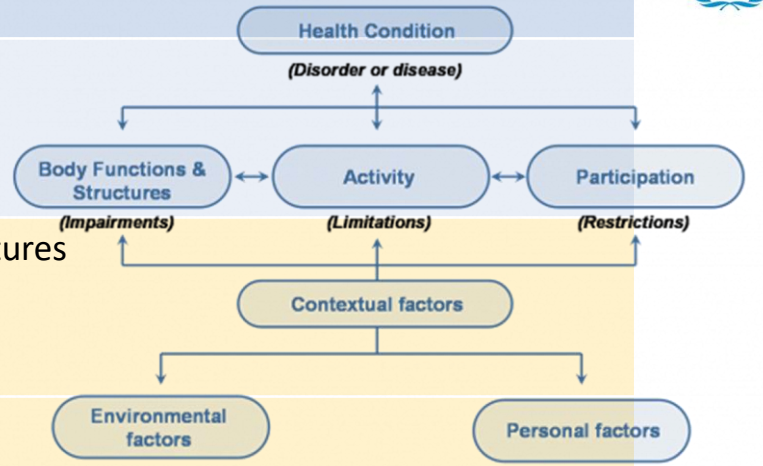



Diagnosis
Fibula Hypoplasia

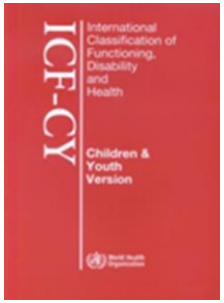
Medical Guideline

ICF : Body Functions & Structures

ICF: Activity
Participation




World Health Organization



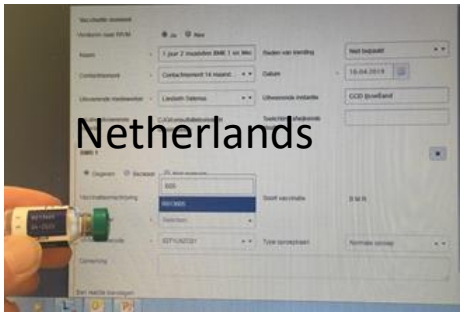
# Global Preventive Child Health Checks
# Home-based records

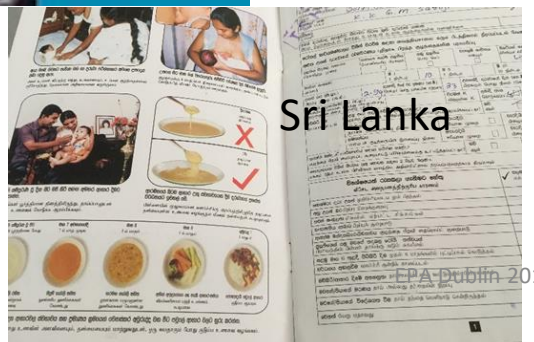
- Growth, developement, physical examination,
- Family history, heel stickscreening, hearing
- Vaccination

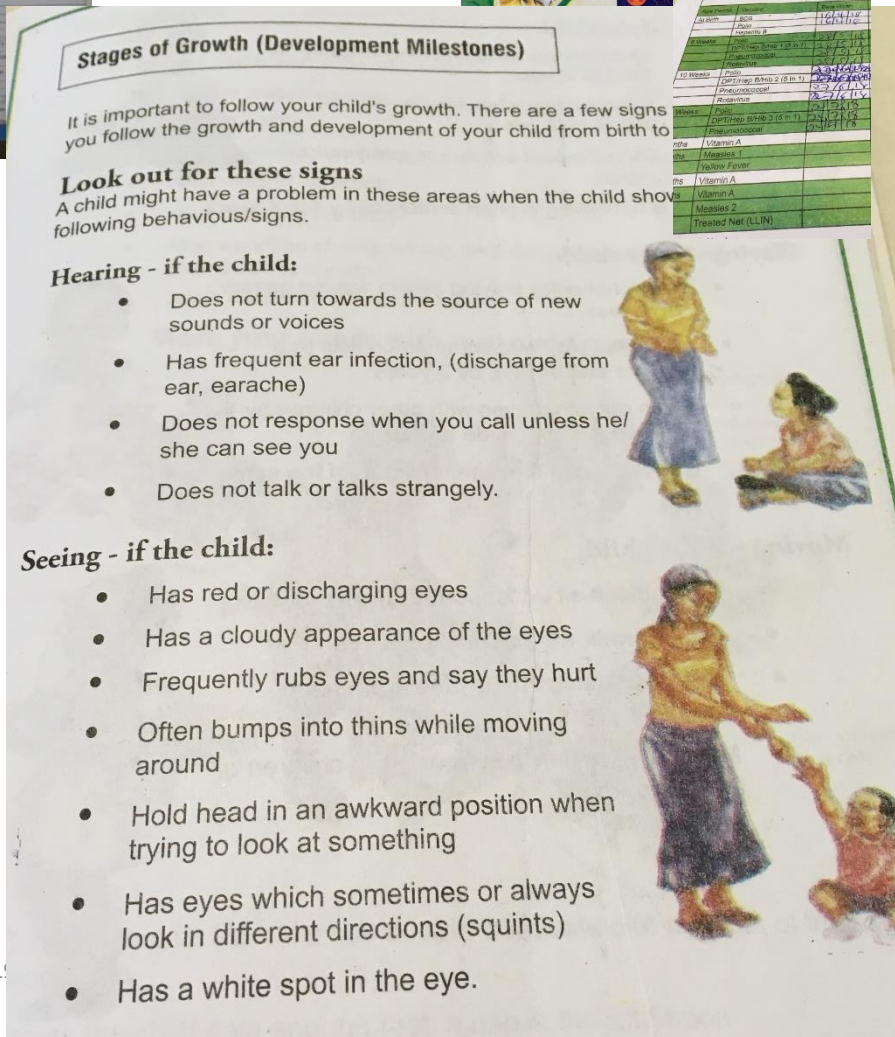
E- Health







**Digital Child Health Data**

- Learn about e-health to be an equal partner in global digital health


**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



10 REDUCED INEQUALITIES

17 PARTNERSHIPS FOR THE GOALS

| Codification | Meaning |
|---|---|
| ICD & Orpha code | International Code of **Diseases** / Orphanet code |
| ICF (-CY)  | The *International Classification of Functioning, **Disability** and Health for Children and Youth (ICF-CY)* is a derived version of the *International Classification of Functioning, Disability and Health* (ICF, WHO, 2001) designed to record characteristics of the developing child and the influence of environments surrounding the child . |
| LOINC  | A universal code system for **tests,** measurements, and observations. |
| ICPC | ICPC-2 classifies patient data and clinical activity in the domains of General/Family Practice and primary care, taking into account the frequency distribution of problems seen in these domains. |
| ATC  | The purpose of the ATC/DDD system is to serve as a tool for **drug** utilization research in order to improve quality of drug use. |
| ISO 9999  | ISO 9999:2011 establishes a classification of **assistive products**, especially produced or generally available, for persons with disability. |

World Health Organization


| Patient Informatiom | Primary Care | Diagnosis<br>Collaborative care | Social Services |
|---|---|---|---|
| www.shwachman.nl | Growth retardation<br>Recurrent infections<br>(LOINC) | Guideline SDS<br>(Orphanetcode;<br>SNOMED, ATC e.a.) | Recurrent illness<br>Fatigue, Short (ICF-CY;<br>ISO 9999) |

**Diagnosis**

Hurler syndrome

PKU, Duchenne MD, FOP

Shwachman Diamond Syndrome

ICD - 10

Orphanetcode

OMIM

SNOMED -CT

New Diagnostics

LOINC

ICPC

**Sign primary care**

Heelstick screening

Hearing screening

Growth; Development

**Guideline**

Collaborative Health Care

ISO 3166-1

HL7

ATC

GS1

New Therapeutics

ICF(-CY)

ISO9999

**Data collection**

Data collection with systematically organised computer processable collection medial terms

**Guideline**

Social services and rehabilitation

## What can we, pediatricians, do more or better?

**Global Child Health Schemes**

- Collaborate on a minimal global set for measurements, observations and surveillances

**Implement New Diagnostics**

- Translate new genetic diagnostics in daily practice management

**Inform Society**

- Learn about the daily life with a rare condition
- Support solutions to help overcome inequalities

**Digital Child Health Data**

- Learn about e-health to be an equal partner in global digital health


9th Europaediatrics 13-15 June Dublin, Ireland 2019

rarecare.world



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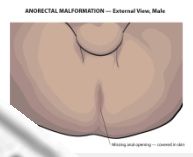
| 10 Rare Diseases | Prima…re | Open information | Code |
|---|---|---|---|
| Anal Atresia | C… |   | ICD |
| Craniosynostosis<br>Treacher Collins | | | Code Dutch child health |
| Cystic Fibrosis | | | ICD<br>Orphacode |
| Duchenne Muscular D… | | | ICF - CY, LOINC |
| Fibrodysplasia Ossifi… | | | …e Dutch child …ealth |
| Glaucoma / C…<br>Hurler S cornea | | | ICD |
| Neurofibromatosis | |   | Code Dutch child health |
| Rett syndrome | | | ICF- CY<br>cm |
| Shwachman Diamond Syndrome | G…<br>neutr… |   | Kg, cm, LOINC |
| Sickle Cell Disease | Heel Stick Scr… |  | ICD, LOINC, orphacode |

Global Consensus in CHILD HEALTH

Child and adolescent **global** health challenges; The first **1000 days** and life long effects; Co-creating integrated care for children with **complex care** needs: lessons from Europe; Maternal impact and **epigenetic** influences; Clinical and molecular features of twenty children with **hyper-IgE** syndrome caused by STAT3 gene mutation in mainland; Clinical and molecular features of twenty children with hyper-IgE syndrome caused by **STAT3 gene** mutation in mainland ; New developments and treatments in paediatric haematology and oncology**; Home based** parent held medical records in EuropeUpdate on **sickle cell** disease and its management; Signs and symptoms of **paediatric cancer:** clues for early diagnosis; The **genomics** revolution and its clinical implications; Expanding newborn **screening**, the endless possibilities...; **Immunodeficiency**; Metabolic and rare diseases: new effective treatments; Nephrology new treatments; **Cystic fibrosis** - new treatments; **Genetic** screening; Latest developments in **autism** genomics and future directions; **Cholesterol** screening; Infants with complex needs/life-limiting conditions; Children's **medicine**: development and regulation; The incidence of **congenital heart** disease in Baku-Azerbaijan. Prospective epidemiology study; Is **down syndrome** related arthritis (DA) a distinct disease from Juvenile Idiopathic Arthritis (JIA)?; The population incidence of childhood **gonadoblastoma** over 20 years in the Republic of Ireland; Re-interrogation of **whole exome** sequencing data in developmental epileptic encephalopathies; A retrospective study of **myeloid leukaemia** in children with down syndrome in Ireland; 114 cases of chronic **granulomatous** disease in mainland China; Inhibitors in Albanian children with **hemophilia A**; **Von Willebrand** disease reclassification in a national paediatric comprehensive care centre; National newborn screening for **cystic fibrosis**: genetic data from the first 6 years

>29 presentations Rare Diseases


Health is a human right for everyone, at every age.
LOVE SAFE
Give blood for those who give life
Depression Let's talk
TOBACCO BREAKS HEARTS
June 11, 2019
World Health Organization

**The *Convention of the Rights of the Child*** and the ***Convention of the Rights of Persons with a disability*** highlight how **children with a disability have the same rights as other children.**

Children with disabilities and their family have ordinary needs and must have access to **mainstream programs** and services.