

Sustainable Development Goals, Universal Health Coverage and the Disabled Child

Liesbeth Siderius, at ICORD, Jerusalem, November 13, 2019



Rare is Common Just normal People



22qdeletion

The Family



Skeletal Dysplasia

At work



**Fibrodysplasia
Ossificans Progressiva**

Studi
Animal Science



Introduction



- **Sustainable Development Goals, United Nations 2015**

Rare is common, in pediatrics

- **Universal Health Coverage, United Nations, 2019**

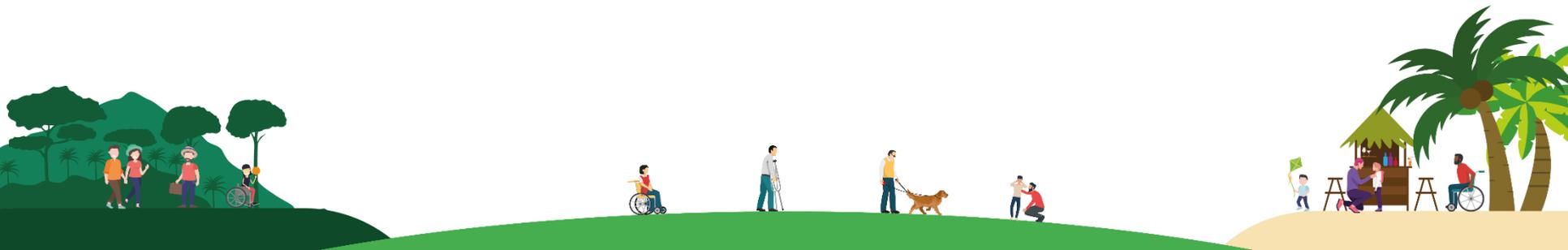
Pediatric Support

- **Disabled child**

Early recognition

- **Digital Health for all**

Interoperability



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Rare common in pediatrics

> 50 % manifests in childhood

Health and well being for all

< 5 year 2-3 % have a rare disease

Early recognition

chronic and life-threatening

Reduce inequalities

80% of genetic origin

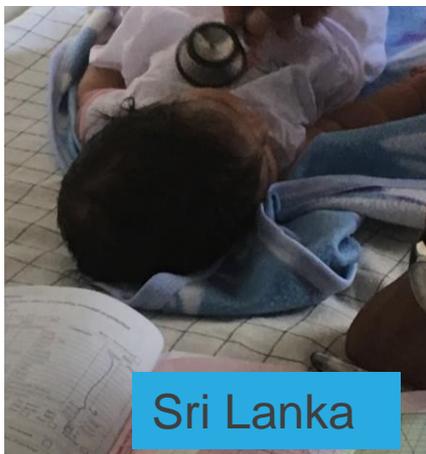




The United Nations emphasizes the need to:

- end preventable deaths of new-borns and children under five
- end avoidable mortality caused by non-communicable diseases
- achieve **universal health coverage**

World Wide children are measured, developmental screened, and vaccinated





At least half of the world's people do not have access to essential health services.

UHC ensures that

NO ONE IS LEFT BEHIND.



**Accessible
Diagnostics**



Quality Information



**Training
Primary Care**



Data exchange

12.12.18
UHC



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World Health Organization

Home-based records

Early recognition

WHO recommendations on home-based records for maternal, newborn and child health*

Web annex A. Evidence base (GRADE and CERQual profiles)

* The full guideline document is available at:
<http://apps.who.int/iris/bitstream/handle/10665/271427/99831493032-4eng.pdf>



Stages of Growth (Development Milestones)

It is important to follow your child's growth. There are a few signs that can help you follow the growth and development of your child from birth to 5 years.

Look out for these signs
A child might have a problem in these areas when the child shows any of the following behaviours/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 respond 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.

Ghana



Coordinated Care

-developmental delay

-early loss of teeth

Exome screening, genes related to developmental delay

Coffin Lowry Syndrome

-progressive kyphosis/scoliosis

-sensorineural hearing defect

-cardiac evaluation

-**sudden loss of muscle tone** induced by unexpected tactile or auditory stimuli and epilepsy.



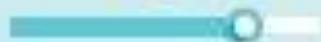
What EU citizens expect...

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



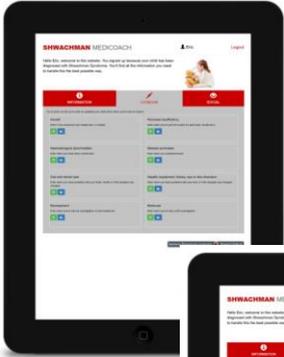
EUROPE twitter



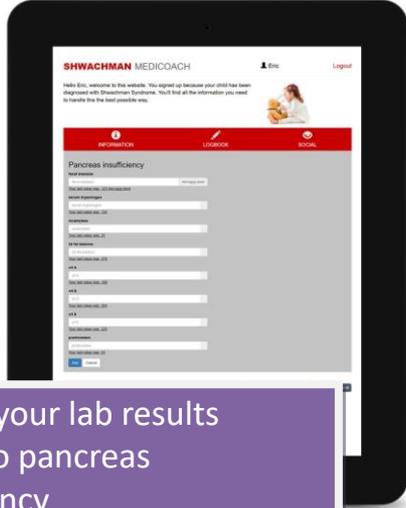
Co-management for people with Shwachman Diamond Syndrome



Information on SDS and the use of the application



Overview of the digital guideline



Register your lab results related to pancreas insufficiency



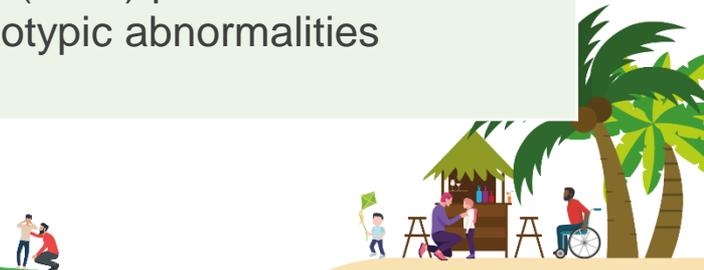
Collect weight and length data to create a SDS growth curve



Codification	Meaning
ICD & Orpha code	International Code of Diseases / Orphanet code
ICF (-CY) 	The <i>International Classification of Functioning, Disability and Health for Children and Youth (ICF-CY)</i> is a derived version of the <i>International Classification of Functioning, Disability and Health (ICF, WHO, 2001)</i> 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.
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.
HPO 	The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease.



World Health Organization



Patient Information	Primary Care	Diagnosis Collaborative care	Social Services
www.shwachman.nl https://rarecare.world	Growth retardation Recurrent infections (LOINC)	Guideline SDS (Orphanetcode; SNOMED, ATC e.a.)	Recurrent illness Fatigue, Short (ICF-CY; ISO 9999)

Stichting Shwachman syndroom Support Holland

New Diagnostics

HPO
LOINC
ICPC

Diagnosis
 Hurler syndrome
 PKU, Duchenne MD, FOP
 Shwachman Diamond Syndrome

ICD - 10
Orphacode
 OMIM
 SNOMED -CT
 DCOM

Sign primary care
 Heelstick screening
 Hearing screening
 Growth; Development

Guideline
 Collaborative Health
 Care

**Interoperable data
 model**

ISO 3166-1



New Therapeutics

ATC
 ICF
 ISO9999

HL7/ FHIR

Registry
 Data collection with
 systematically organised
 computer processable
 collection medial terms

Guideline
 Social services and
 rehabilitation



LOINC

The international standard for identifying health measurements, observations, and documents.

Interoperable codes in care

```
LocalCode^LocalName^CodeSystem|LOINCcode^LOINCname^CodeSystem
OBX|2|NM|123^WBC^HSP_A^26464-8^Leukocytes [# /volume] in Blood^LN|10.8|K/MM3|F|
OBX|3|NM|234^RBC^HSP_A^26453-1^Erythrocytes [# /volume] in Blood^LN|4.82|MIL/MM3|F|
OBX|4|NM|545^HGB^HSP_A^718-7^Hemoglobin [mass/volume] in Blood^LN|15.1|G/DL|F|
OBX|5|NM|456^HCT^HSP_A^20570-8^Hematocrit [Volume Fraction] of Blood^LN|45|%|F|
```

Notice how the result value and units have their own places in the message



Goldenhar s
Abnormal ear

HP:0008551



FOP

Abnormal toe

HP:0010109



Coffin Lowry s
Tapered fingers

HP:0001182

**Measuring
Head
Circumference**

LOINC
8287-5



Lower limb
asymmetry

HP:0100559

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Shwachman DS

ATC

.A09AA02
Pancreatine



Building the Rare Disease

knowledge and information ecosystem



Thalassemia

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)

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 thalassemia

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

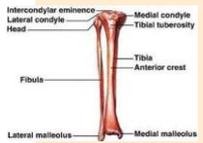
Hemoglobinopathies

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



First Feature



Diagnosis
Fibula Hypoplasia



Medical Guideline



ICF : Body Functions & Structures



ICF: Activity Participation

ICS > 11 > 11.180 > 11.180.01

ISO 9999:2016

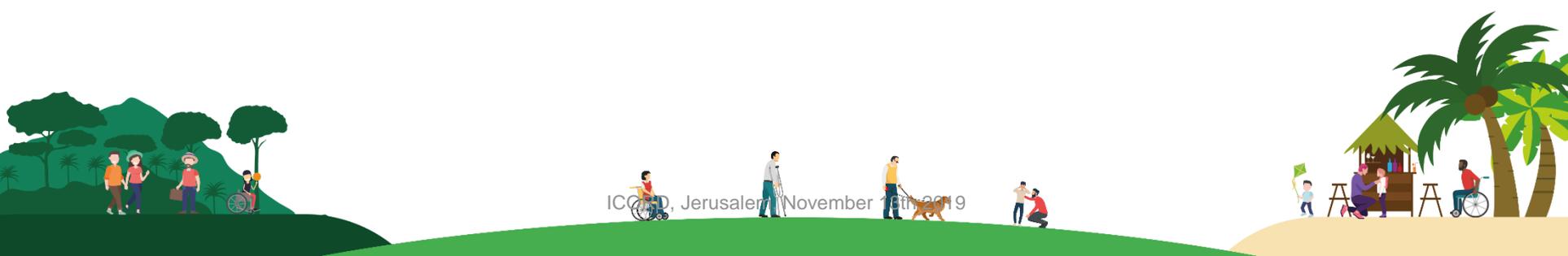
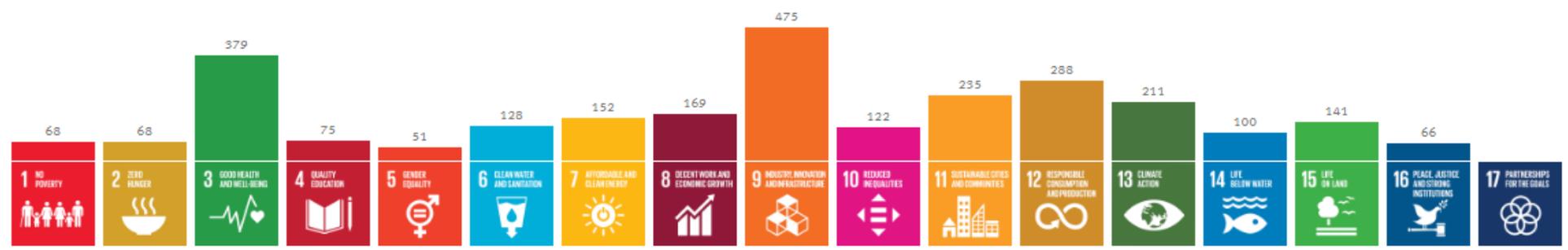
Assistive products for persons with disability – Classification and terminology



ISO= International Standards globally recognized guidelines and frameworks

IMPACT AT A GLANCE

ISO contributes to all of the SDGs. Here you can see the number of ISO standards that are directly applicable to each Goal.



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 >

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 $\beta 0$ থ্যালাসেমিয়া হিসাবে বর্ণনা করা হয় এবং $\beta +$ হিসাবে সংশ্লেষকে হ্রাস করা হয়। বিটা-গ্লোবিনের হ্রাস α গ্লবিন চেইনগুলিতে আপেক্ষিক অতিরিক্ত বাড়ায়।

OMIM

613985 BETA-THALASSEMIA

ORPHA

ORPHA:231214 Beta-thalassemia major

10 Rare Diseases	Primary nature	Open information	Code
Anal Atresia	C	 	ICD
Craniosynostosis Treacher Collins			Code Dutch child health
Cystic Fibrosis			ICD Orphacode
Duchenne Muscular Dystrophy			ICF - CY, LOINC
Fibrodysplasia Ossif			Code Dutch child health
Glaucoma / C Hurler S corne			ICD
Neurofibromatosis			Code Dutch child health
Rett syndrome			ICF- CY cm
Shwachman Diamond Syndrome	G. neutro		Kg, cm, LOINC
Sickle Cell Disease	Heel Stick Sc	 	ICD, LOINC, orphacode

Global consensus in
CHILD HEALTH

Thank

- European Pediatric Rare Disease Network

John Dodge, U.K.

Lali Margvelashvili, Georgia

Velibor Tasic, N- Macedonia

David Neubauer, Slovenia

Arunas Valiulis, Lithuania

Jola Wierzba, Poland

- Consensus in Pediatrics and Child Health

Manual Katz, Israel

- Forum Rare Diseases, Sri Lankan Pediatric Society

Anjan Bhattacharya, India

- People with a rare condition and their families



Stichting Shwachman syndroom Support Holland



ICOPD, Jerusalem, November 7-8, 2019



Imagine... all just ordinary people, sharing all the world



Goldenhar syndrome

Speaker at Digital Health Congress



Complex Chromosome abn

Mother



Thalassemia

Patient Advocate

e.siderius@kpnplanet.nl

ICD-10, Jerusalem, November 7-8, 2019

