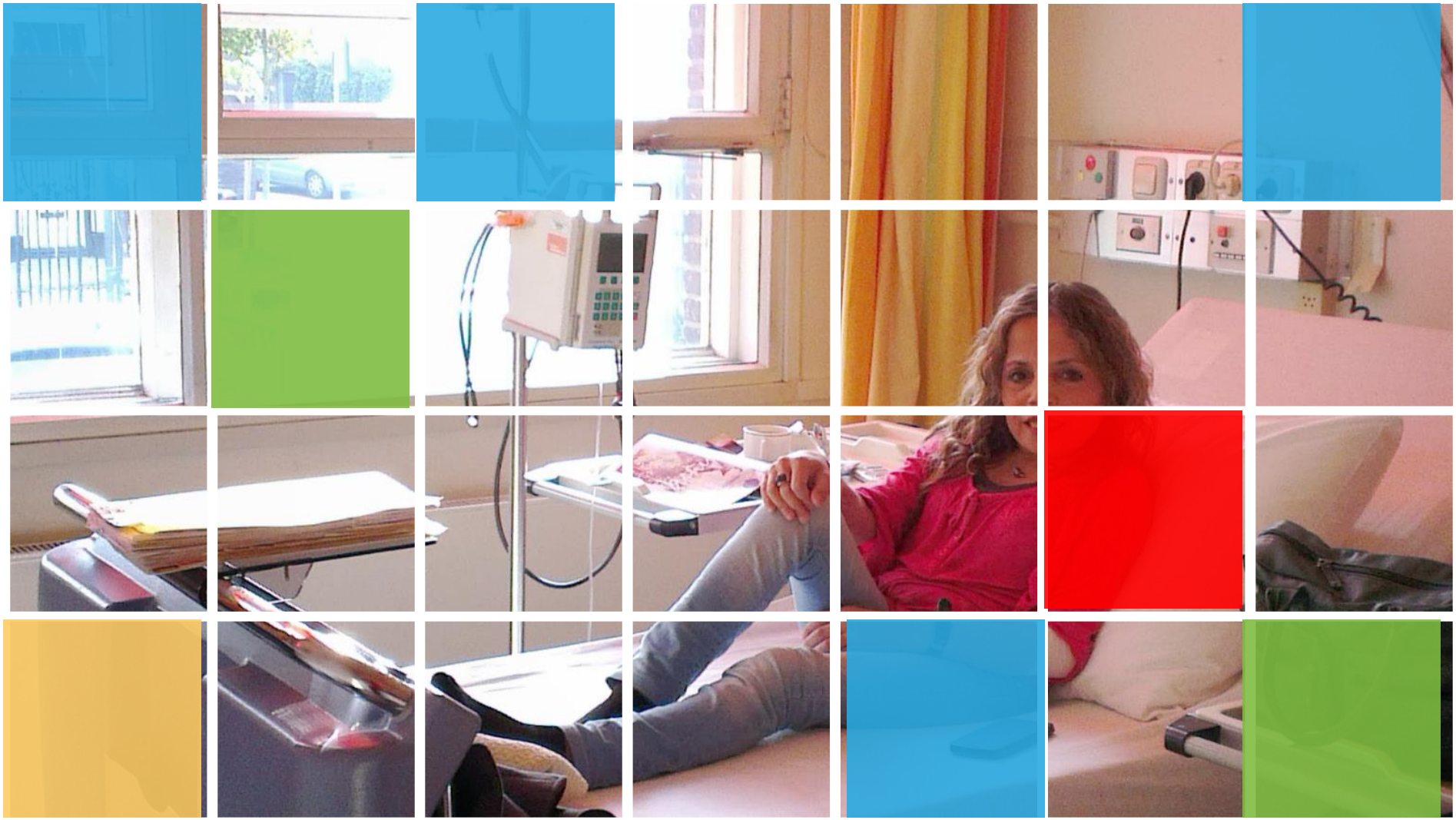


Sustainable Development Goals, Universal Health Coverage and the Disabled Child

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

Rare is Common
Just normal People

22qdeletion

The Family

Skeletal Dysplasia

At work

Fibrodysplasia Ossificans Propressiva

Studiy
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

ICORD, Jerusalem, November 13th 2019

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

Accessible Diagnostics

Quality Information

Training Primary Care

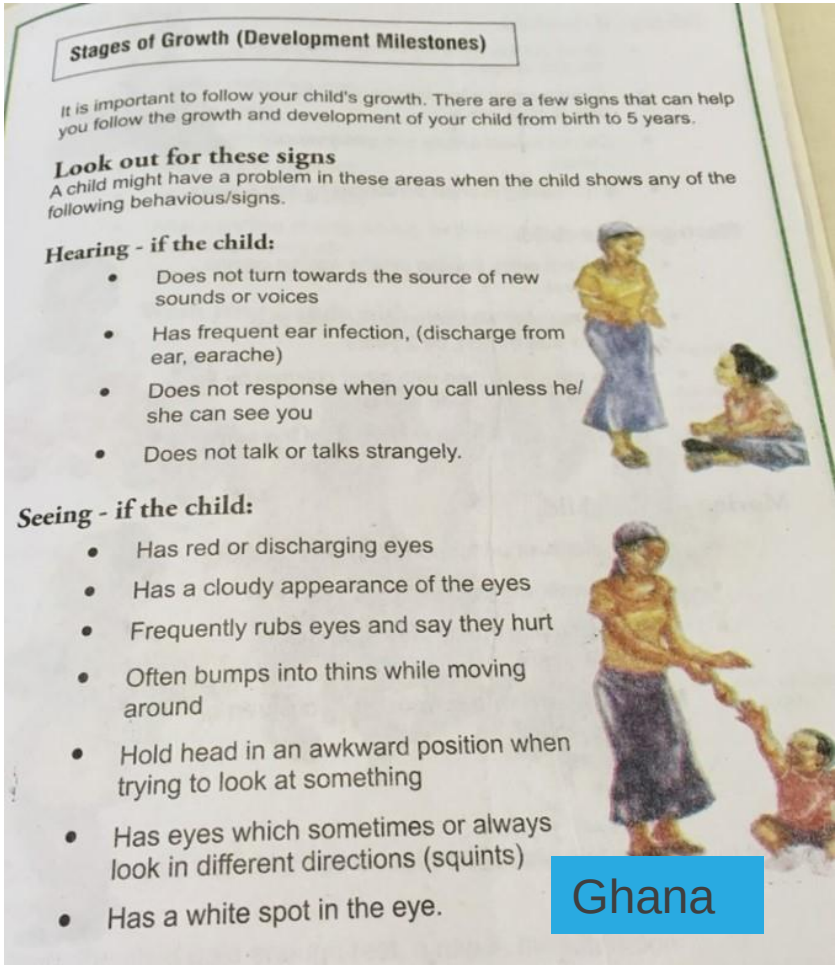
Data exchange

ICORD, Jerusalem, November 13th 2019

World Health Organization

Home-based records

Early recognition



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

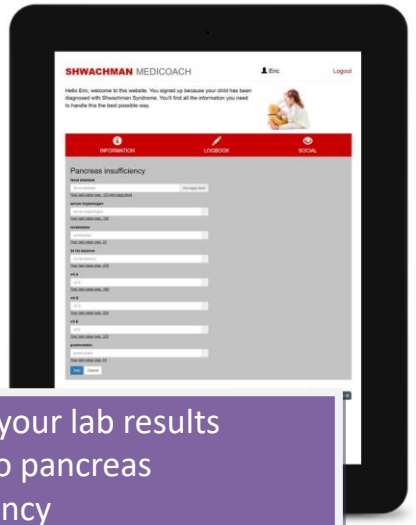
EUROPE twitter

Co-management for people with Shwachman Diamond Syndrome

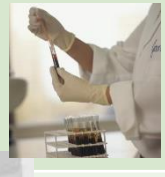
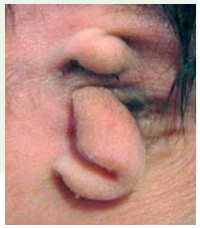
Information on SDS and the use of the application

Overview of the digital guideline

Collect weight and length data to create a SDS growth curve



Register your lab results related to pancreas insufficiency

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.
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

HL7/ FHIR

ATC
New Therapeutics
ICF
ISO9999

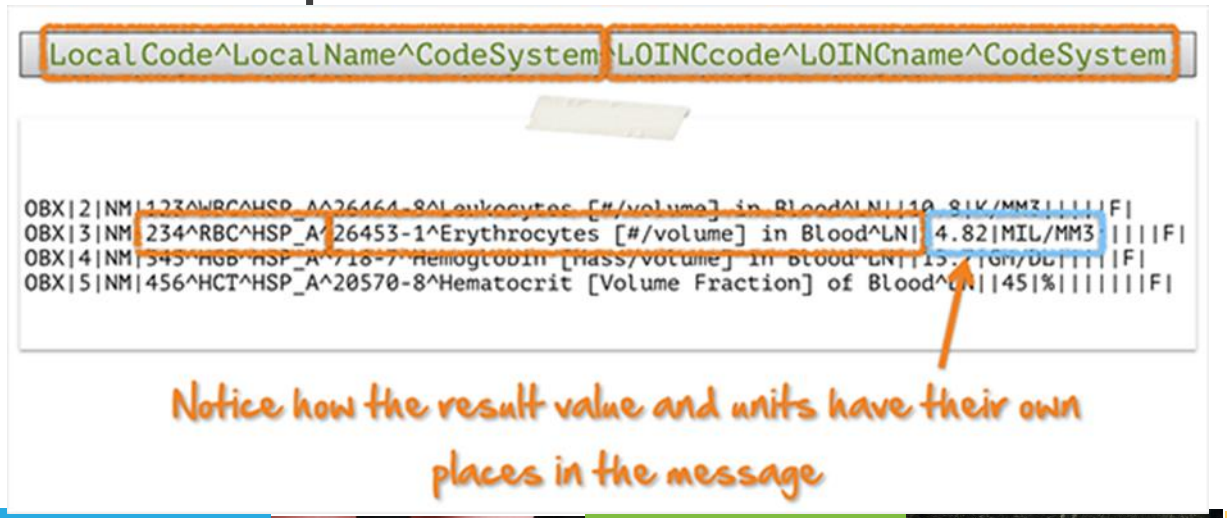
Registry
Data collection with systematically organised computer processable collection medial terms

Guideline
Social services and rehabilitation

LOINC

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

Interoperable codes in care



Goldenhar s

Abnormal ear

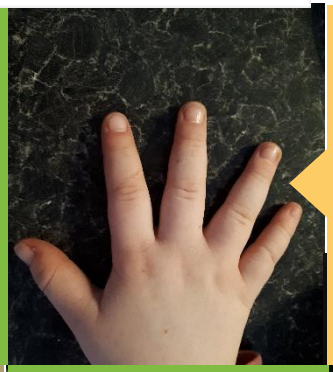
.

HP:0008551

FOP

Abnormal toe

HP:0010109



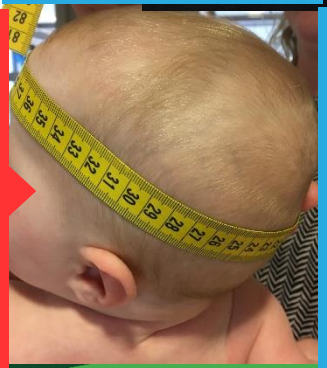
Coffin Lofry s

Tapered fingers

HP:0001182

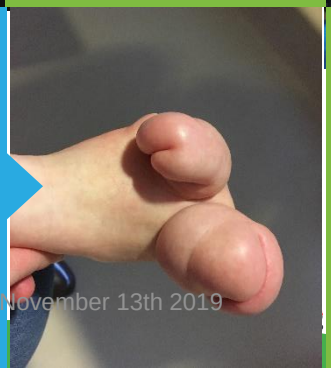
Measuring Head Circumference

LOINC 8287-5



Lower limb asymmetry

HP:0100559



Shwachman DS

ATC

. A09AA02 Pancreatine

Building the Rare Disease

knowledge and information ecosystem

rarecare.world

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

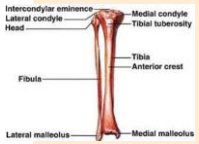
Carrier screening thalassemia

Symptom

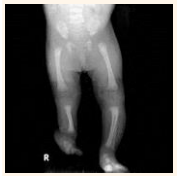
... Carrier screening **thalassemia** Related family members with elevated HbA2 In carrier screening for the classical beta-**thalassemia** trait, the hallmark is the presence of an ... 2 (α 2 δ 2). Another way of identifying people with **thalassemia** major is neonatal screening. Neonatal screening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

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

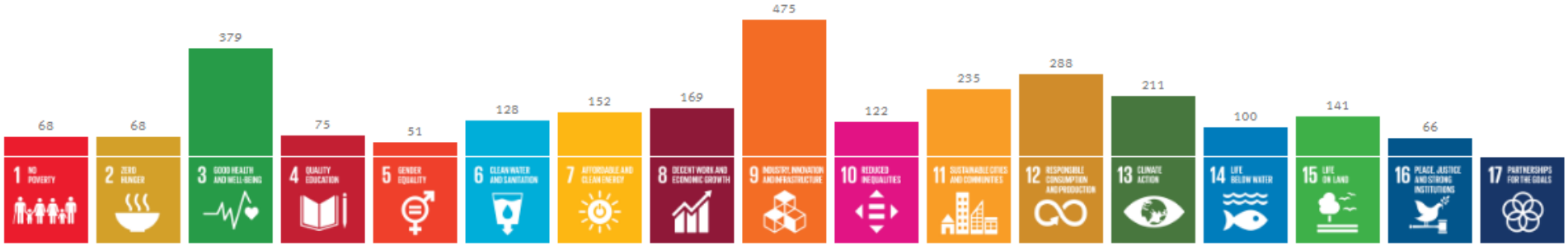
Assistive products for persons with disability — Classification and terminology

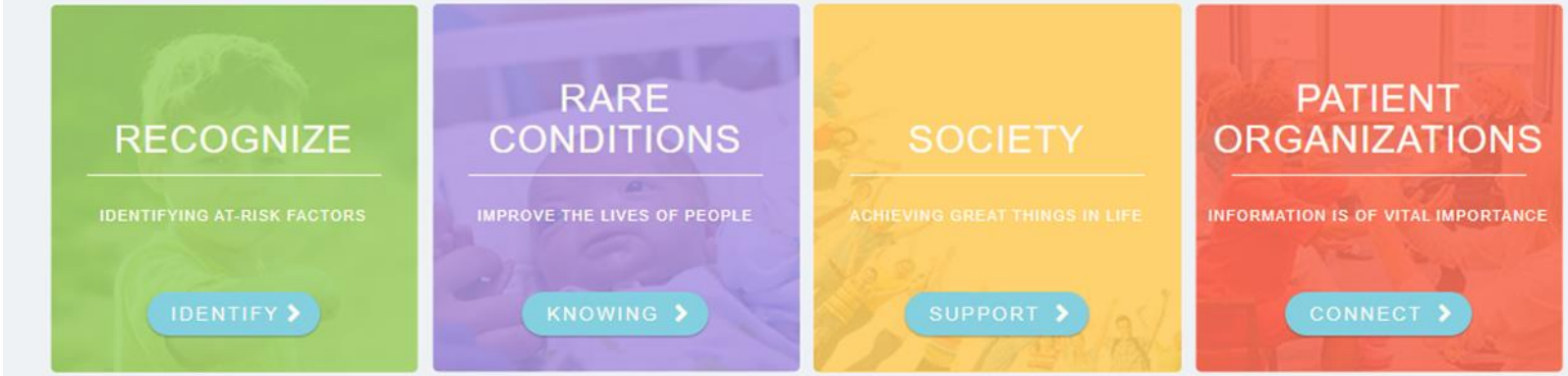
ISO= International Standards globally recognized guidelines and frameworks

ISO | Standards | All about ISO | Taking part | Store | EN

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.





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 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।

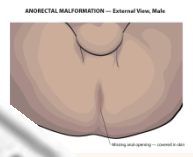
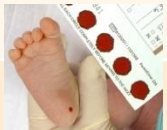
বিটা থ্যালাসেমিয়া হিমোগ্লোবিনের বিটা গ্লবিন শৃঙ্খলার অনুপস্থিতি বা হ্রাস সংশ্লেষণের কারণে ঘটে। বিটা থ্যালাসেমিয়া বৈশিষ্ট্য বা বিটা থ্যালাসেমিয়া নাবিক ব্যক্তিরা বিটা থ্যালাসেমিয়া বা বিটা-থ্যালাসেমিয়ার একজন ক্যারিয়ারের হেটারজাইজাস। বিটা থ্যালাসেমিয়া মেজারের ব্যাক্তি হ'ল বিটা থ্যালাসেমিয়ার জন্য 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

10 Rare Diseases	Prima…ature	Open information	Code
Anal Atresia	C…	 	ICD
Craniosynostosis Treacher Collins			Code Dutch child health
Cystic Fibrosis			ICD Orphacode
Duchenne Muscula… Dystrophy			ICF - CY, LOINC
Fibrodysplasia Ossi…			…e Dutch …ild health
Glaucoma / C… Hurler S corn…			ICD
Neurofibromatosis			Code Dutch child health
Rett syndrome			ICF- CY cm
Shwachman Diamond Syndrome	G… neutr…	  	Kg, cm, LOINC
Sickle Cell Disease	Heel Stick S…		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

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