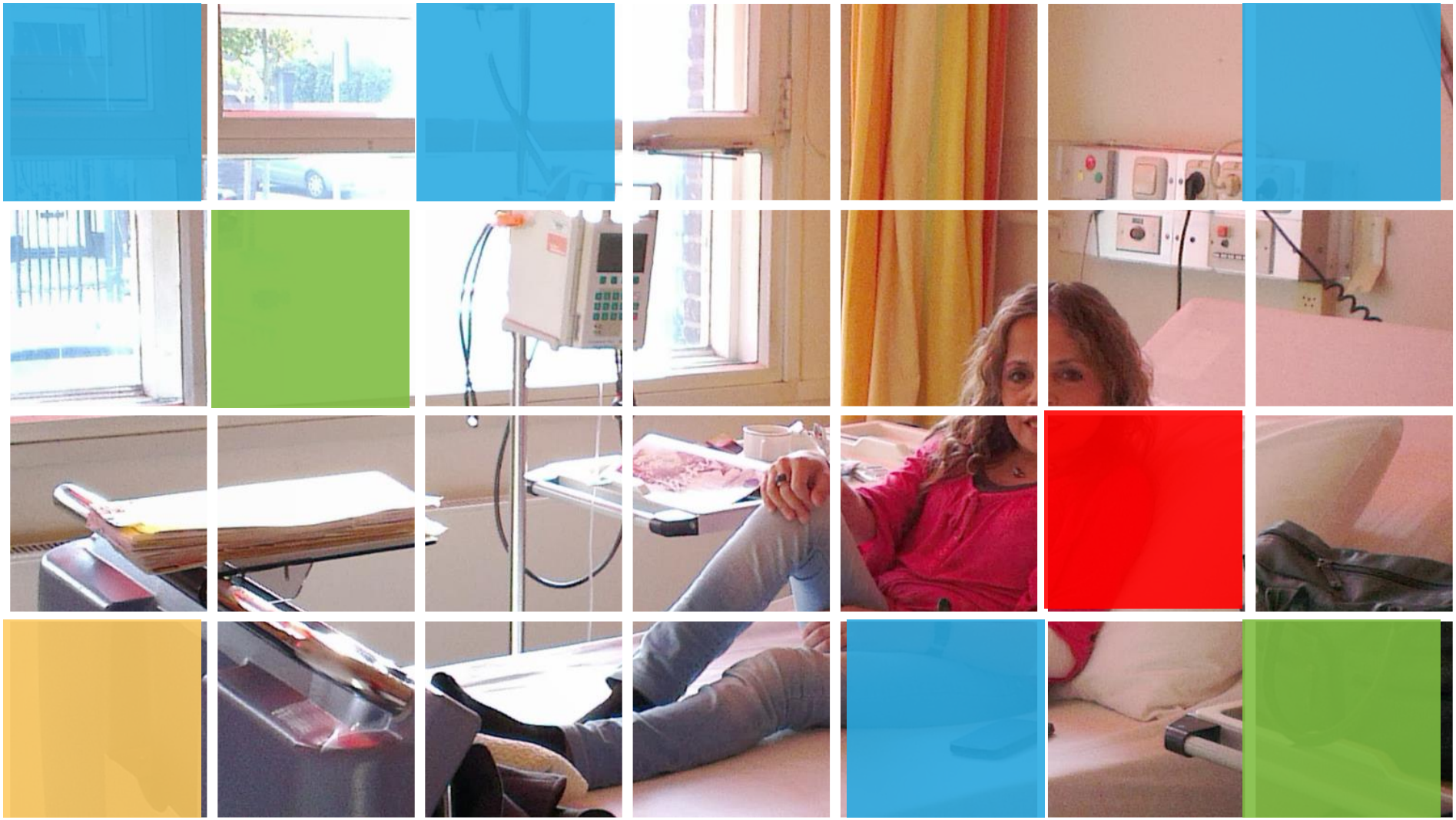


Importance of Psychology in Rare Diseases

Liesbeth Siderius, pediatrician

Introduction

- **(Dis)abled child**

Rare is common

- **The Diagnosis**

Early recognition

- **Universal Health Coverage**

Sustainable development goals

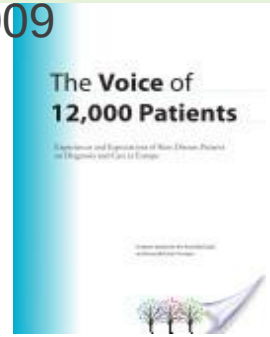
UNIVERSAL HEALTH COVERAGE DAY

- **Leave no child behind**

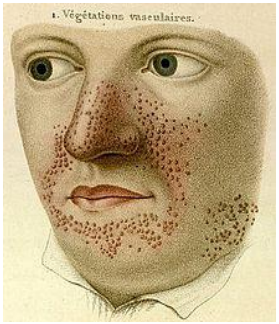
Global child health

Listen to the Voices of 12,000 Patiënts

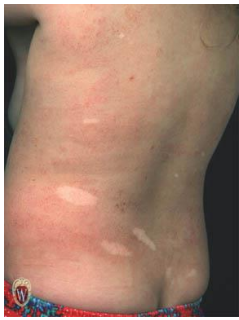
2009

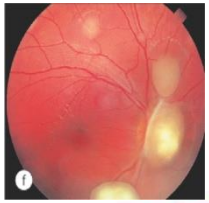


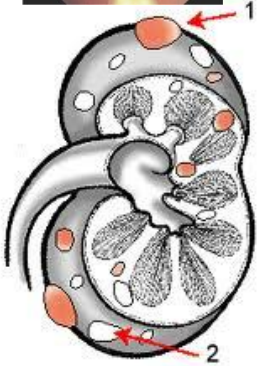
EURORDIS
Rare Diseases Europe

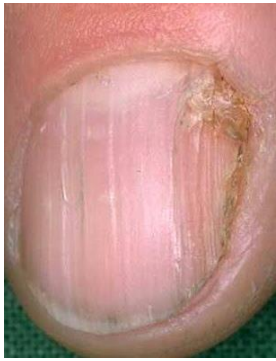


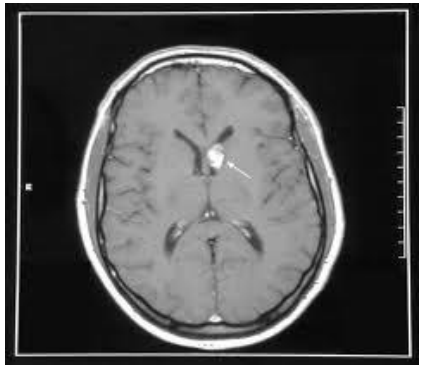
Tuberous sclerosis complex



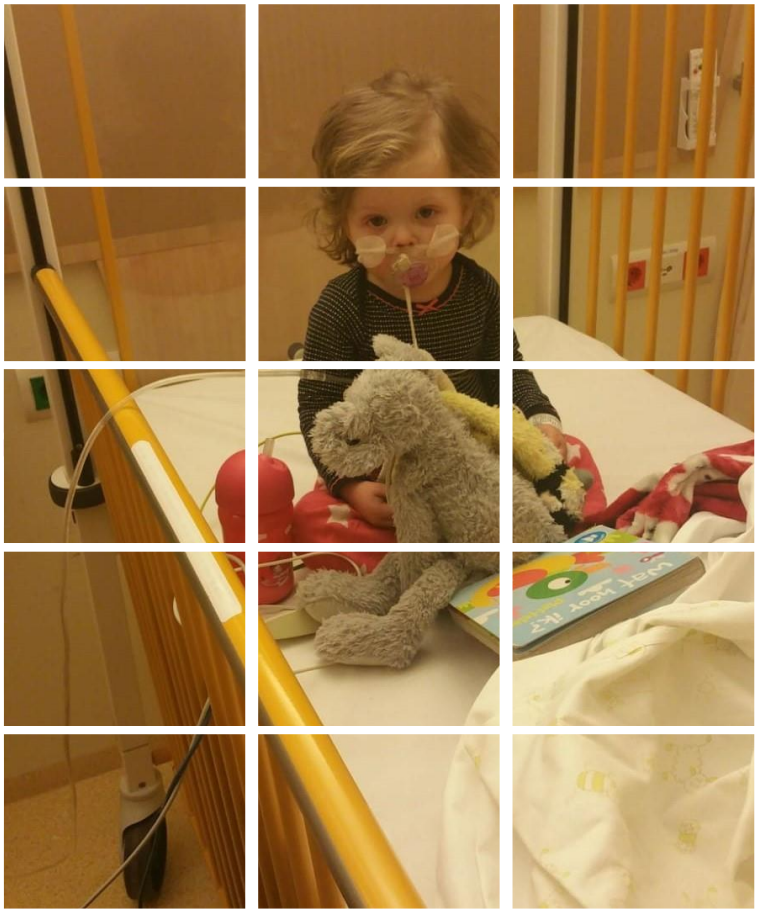








After first symptoms,
battle for diagnosis,
which can last for years.
After the diagnosis,
faced with
inadequate health and social care.



Disabling and Rare Conditions

70 % 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

72% of genetic origin

Shwachman Diamond Syndrome

Disabled child

Children living with the diagnosis

- autism,
- developmental delay,
- cerebral palsy,
- hearing deficits and
- visual impairment

may very well have a

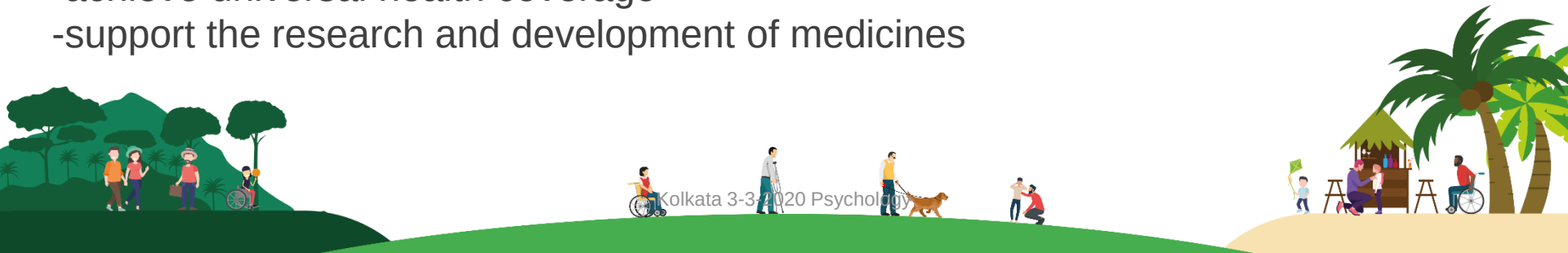
rare condition with

➢ specific health risks and treatment.

NGO Committee for Rare Diseases

The NGO Committee for Rare Diseases aims to promote multi-stakeholder collaboration and actions for rare diseases within the United Nations system.
It is established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO) and acts as a forum of interested parties such as NGOs from the field of rare diseases and beyond; United Nations bodies and agencies; as well as individual experts.

The United Nations Sustainable Development Goals (SDG) 2016
#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



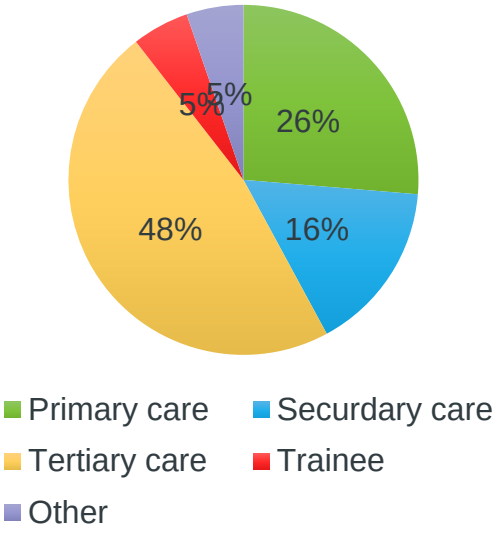
1 End Poverty in all its forms everywhere
3 Ensure healthy lives and promote well-being for all at all ages
4: Ensure inclusive and equitable quality education and promote lifelong learning opportunities for all
5: Achieve gender equality and empower all women and girls
#10: Reduce inequality within and among countries
#17: Revitalize the global partnership for sustainable development

NGO COMMITTEE FOR RARE DISEASES


#ADay4All
Most rare and disabling conditions
manifest in early childhood
EAP

Universal Health Coverage, leave no child behind

Pediatricians working in:



European Pediatric Network Rare Diseases

Questionnaire
October- November 2019

Responce 38 ; 24 countries

EAP Rare Diseases

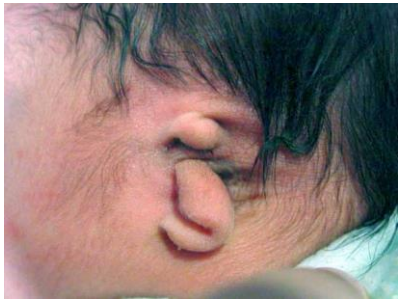
EUROPEAN PAEDIATRIC ASSOCIATION (EPA / UNEPSA)

ECPCP

What can we do?

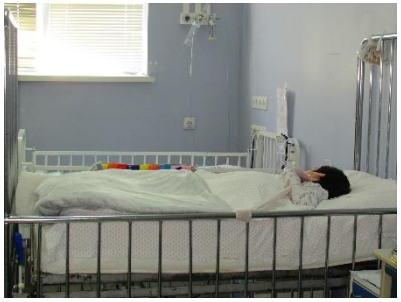
The Netherlands

Day Centre
Coffin Lowrey Syndrome



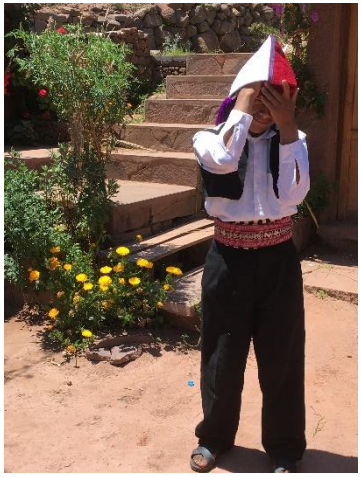
Sri Lanka

Pediatric Clinic
To be diagnosed



Georgia

Abandoned
Undiagnosed



South America

Living with family
In rural village
‘ Birth trauma’

Leave no one behind

When new techniques available and affordable for all

Importance of a diagnosis

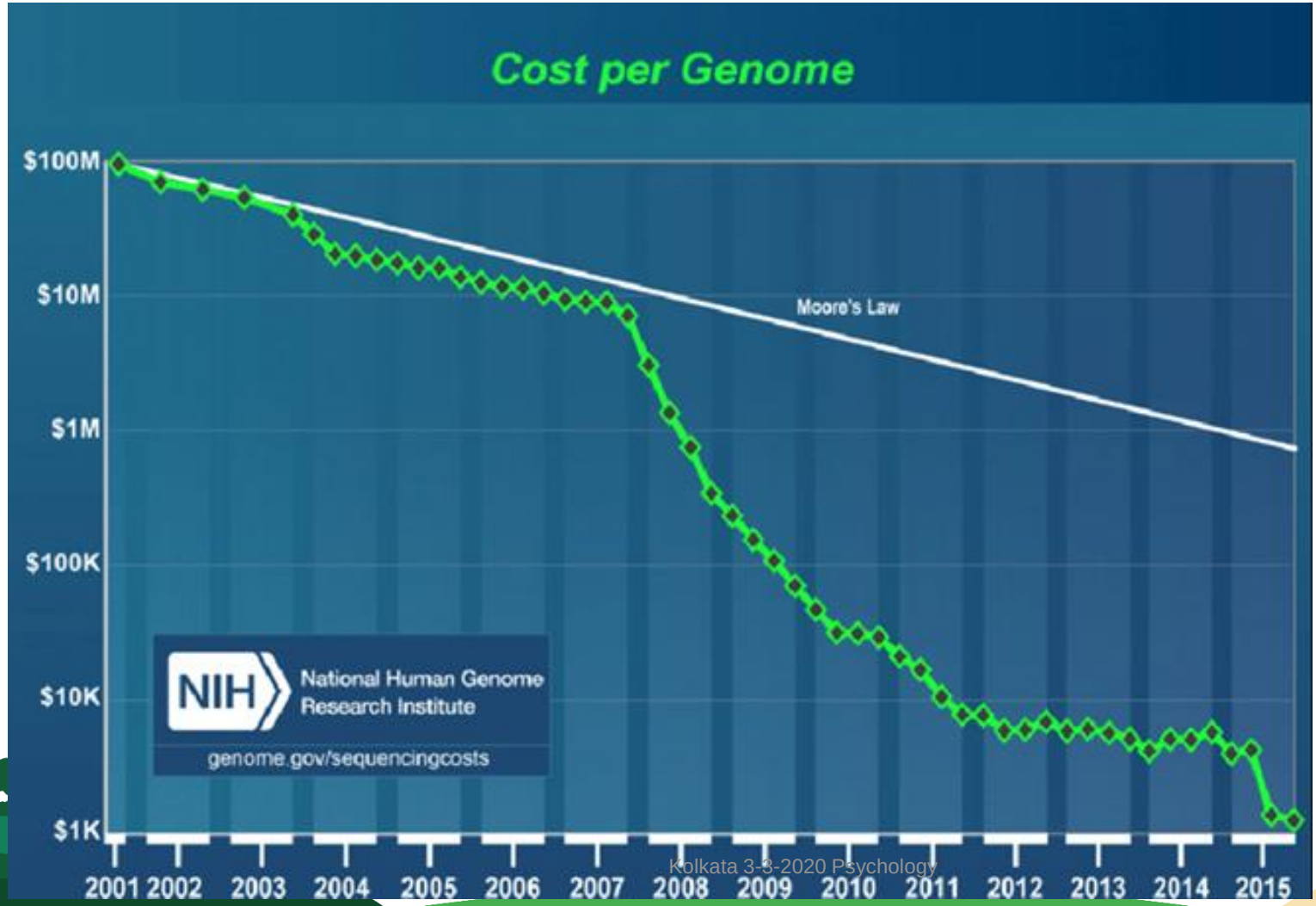
Physical and mental disabilities reported due to **birth trauma**; may be caused by **GATAD2b gene** mutation.

.

Most parts of the world these diagnostics are not available, and pricing of foreign laboratories unaffordable.

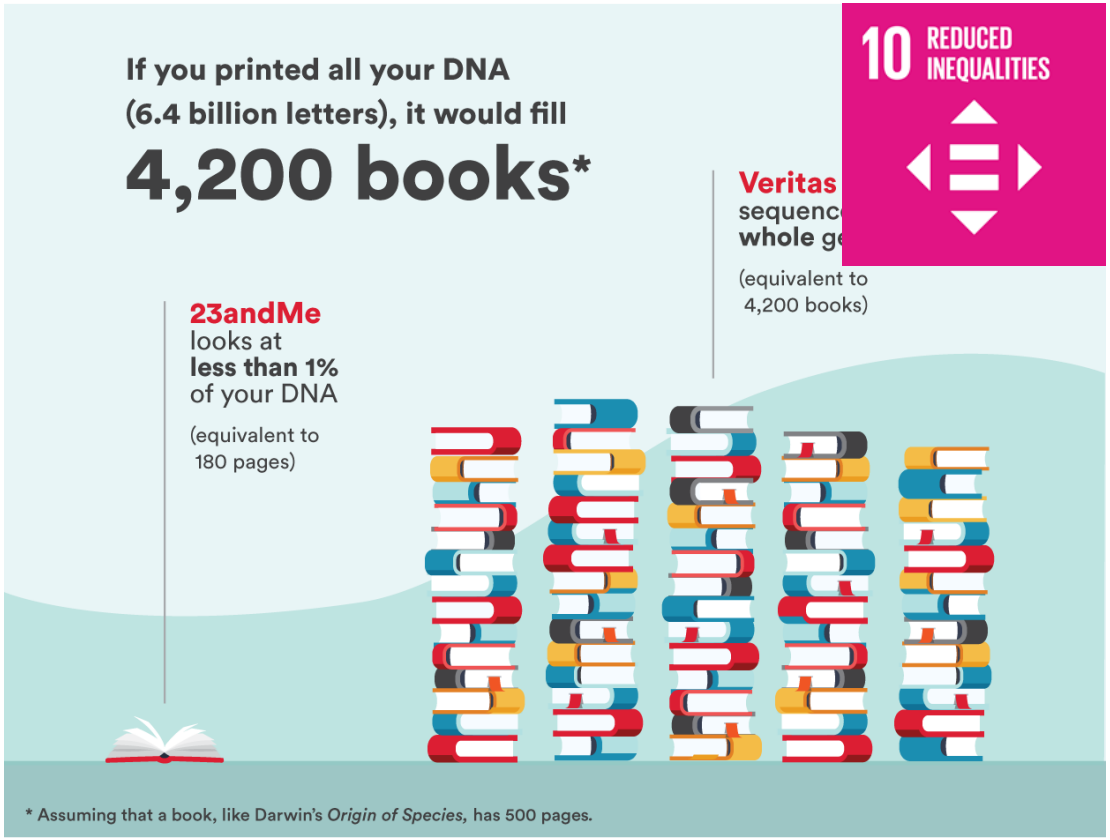
The Netherlands, South America , 2019

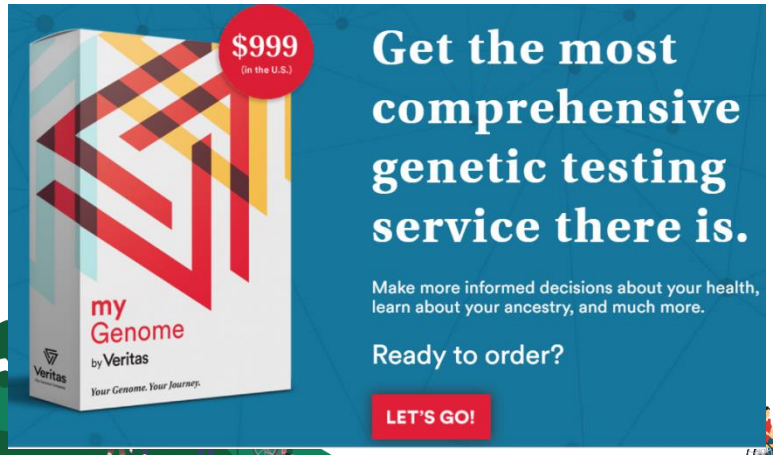
3 GOOD HEALTH AND WELL-BEING



Whole Genome Sequencing,
or WGS for short, is literally knowing all the letters of a person's DNA in the proper sequence. But knowing the letters is just the first part of the equation.
The tricky part is interpreting, or analyzing, what those letters mean.

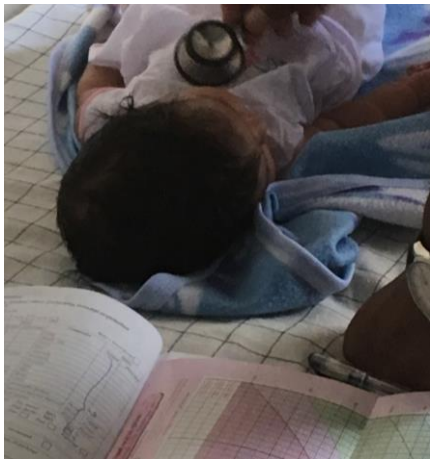
www.veritasgenetics.com/myGenome





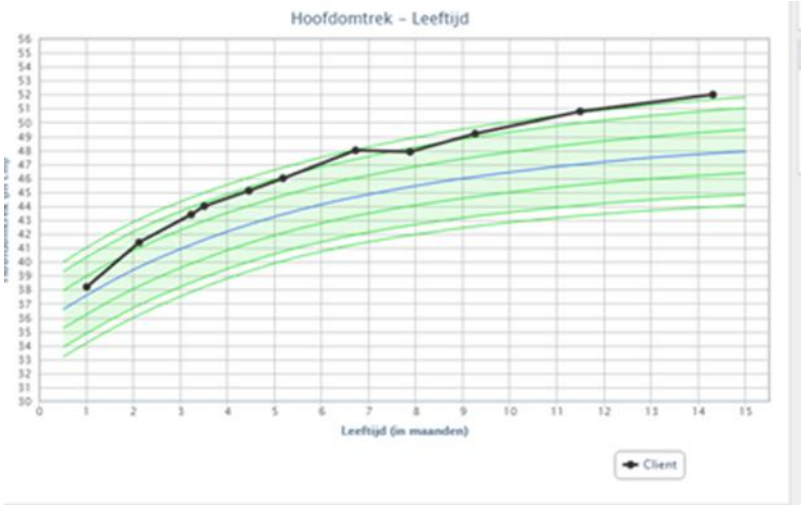
World wide children are measured, examined, developmental screened, and vaccinated in

Preventive child health



Global Child Health

Measure
Head circumference



The Netherlands

DCC gene mutation p.(Val1117Met) p.(Thr1339Ile)
Pathogenic mutation ?
DCC gene mutations are associated with congenital mirror movement disorder

Causes of Autism

WHOLE – EXOME sequenc

In this study, we performed whole-exome sequencing on 120 Autism Spectrum Disorder cases and identified three missense mutations in coding regions of the MECP2 gene. > RETT Syndrome

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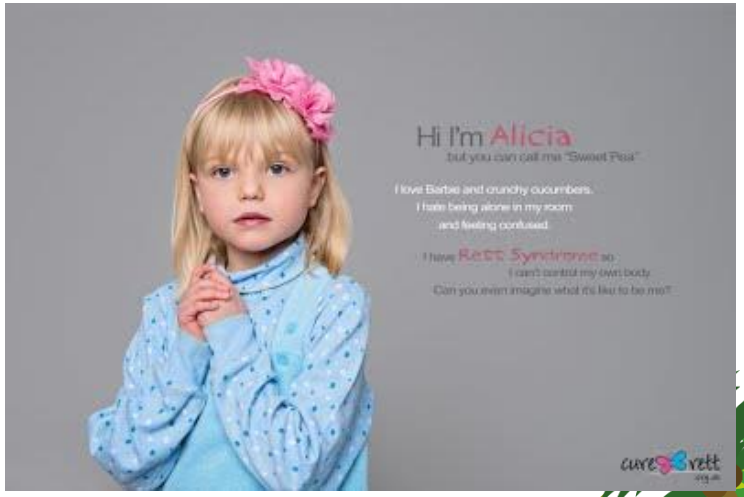
Molecular Autism
Home | About | Articles | Submission Guidelines

Abstract
Background
Methods
Results
Conclusions
Discussion
Declarations
References

Research | Open Access

Identification of autism-related *MECP2* mutations by whole-exome sequencing and functional validation

Zhu Wen, Tian-Lin Cheng, Gai-zhi Li, Shi-Bang Sun, Shun-Ying Yu, Yi Zhang, Ya-Song Du and Zilong Qiu

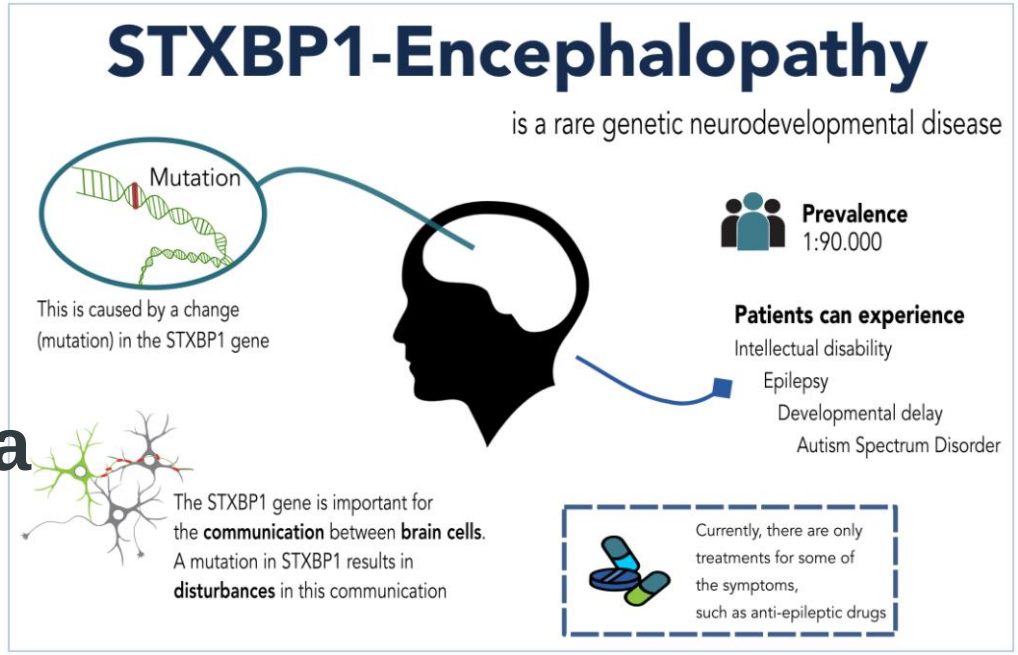


1 month hospitalized

Seizures : abnormal EEG generalized epilepsy

MRI normal

STXBP1; epileptic encephalopathy type 4 OMIM #612164/ Otahara syndrome



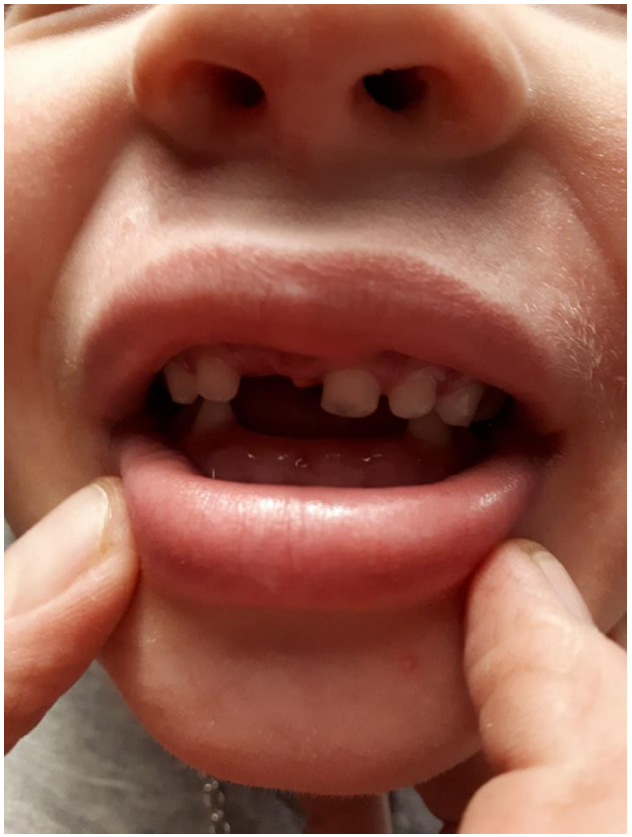
Coordinated Care

-developmental delay

-**early loss of teeth**

Exoom screening, genes related to developmental delay:

Gene RPSKA3: mutation c.1198C>T



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.

The Netherlands

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

Kolkata, India

Co - Management

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

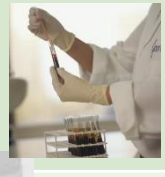
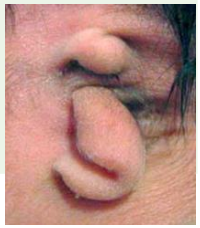
ISO 3166-1

HL7/ FHIR

ATC
New Therapeutics
ICF
ISO9999

Registry
Data collection with systematically organised computer processable collection medial terms

Guideline
Social services and rehabilitation

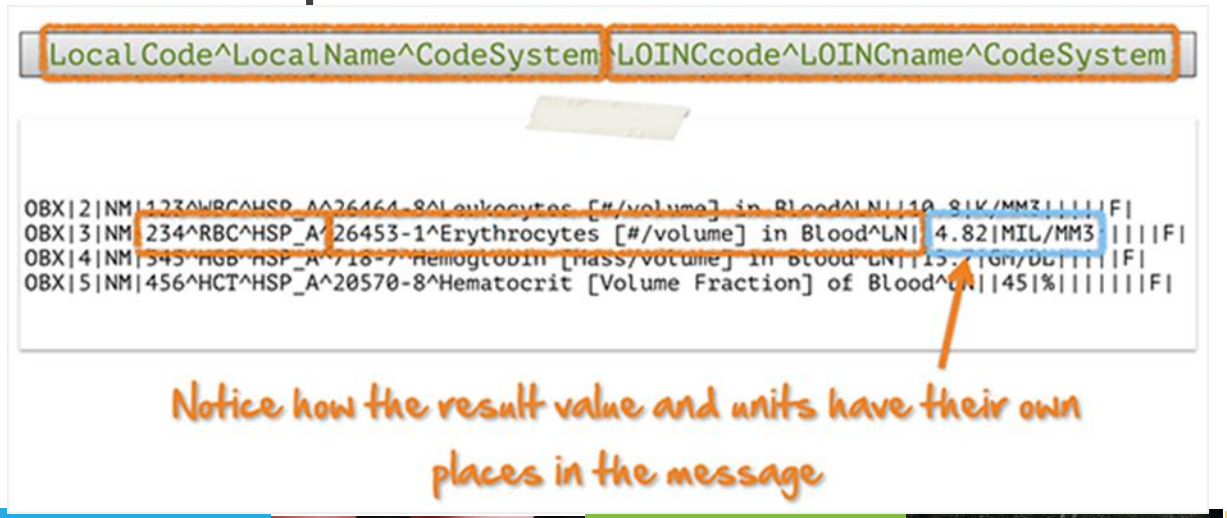
Codification	Meaning
ICD & Orpha code	International Code of Diseases / Orphanet code
ICF (-CY) 	The *International Classification of Functioning, Disability and Health for Children and Youth (ICF-CY)* is a derived vers the *International Classification of Functioning, Disability a Health* (ICF, WHO, 2001) designed to record characteris 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

LOINC

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

Interoperable codes in care



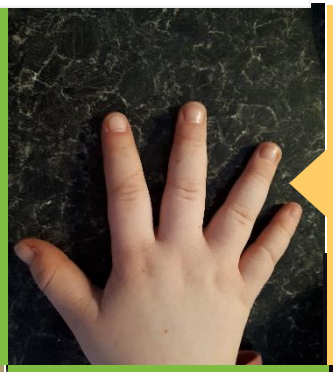
Goldenhar s
Abnormal ear

.

HP:0008551

FOP
Abnormal toe

HP:0010109



Coffin Lofry s
Tapered fingers

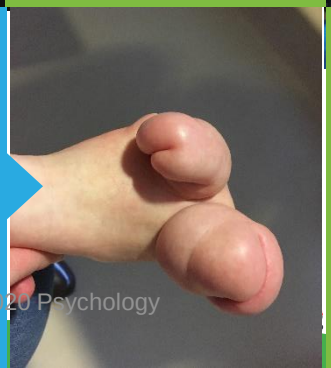
HP:0001182

Measuring Head Circumference

LOINC
8287-5

Lower limb asymmetry

HP:0100559



Shwachman DS

ATC

. A09AA02
Pancreatine

(Dis) Abled People in Society

22qdeletion

The Family

Skeletal Dysplasia

At work

Fibrodysplasia Ossificans Propressiva

Studies
Animal Science

Anal atresia 1:5000, after operation parents daily wash the bowel, child will not easily be toilet trained if at all

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

A EURORDIS INITIATIVE

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.

3,071 people responded to the survey.

The survey was conducted in **23** languages across **42** countries

Thank you to all Rare Barometer Voices participants and partners!

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

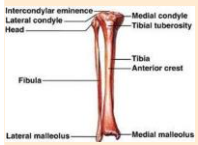
For more information visit **eurordis.org/voices** or email **rare.barometer@eurordis.org**

Fibula Hypoplasia

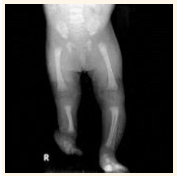
ICF International Classification, Functioning, Disability and Health

First Feature

World Health Organization



Diagnosis
Fibula Hypoplasia



Medical Guideline

ICF : Body Functions & Structures

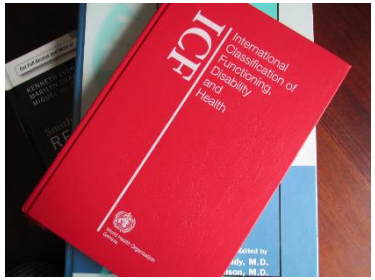
ICF: Activity Participation

ICS › 11 › 11.180 › 11.180.01

ISO 9999:2016

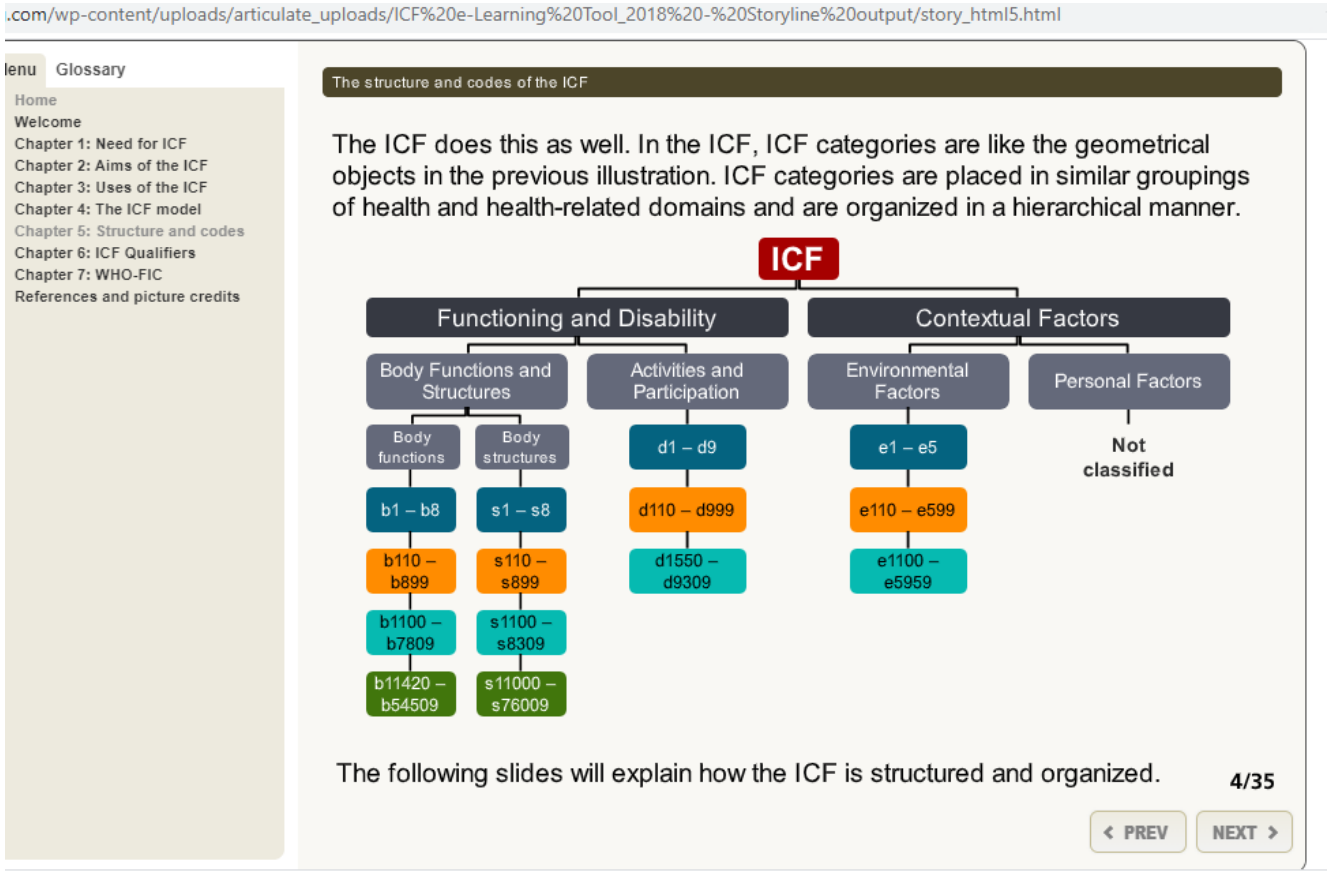
Assistive products for persons with disability — Classification and terminology

ICF Classifications



ICF Chapters	
Function	Scale of the severnes of the disturbance of the function: b167.3 severe disturbance of mental functions related to language
Structure	Scale of anatomic structure s 730.3 severe disturbance of anatomic structure upper arm
Activities/Participation	Performance and ability
Environment	Scale Restricting and supportive factors
Participation (KOLKATA)	Scale of personal factors

https://www.icf-elearning.com



https://www.icf-elearning.com

rning.com/wp-content/uploads/articulate_uploads/ICF%20e-Learning%20Tool_2018%20-%20Storyline%20output/story_html5.html

Menu Glossary

- Home
- Welcome
- Chapter 1: Need for ICF
- Chapter 2: Aims of the ICF
- Chapter 3: Uses of the ICF
- Chapter 4: The ICF model
- Chapter 5: Structure and codes
- Chapter 6: ICF Qualifiers
- Chapter 7: WHO-FIC
- References and picture credits

The structure and codes of the ICF

Categories at the 2nd level: Definition

Activities and Participation

- d1
- d2
- d3
- d4
- d5
- d6
- d7
- d8
- d9 Community, social and civic life

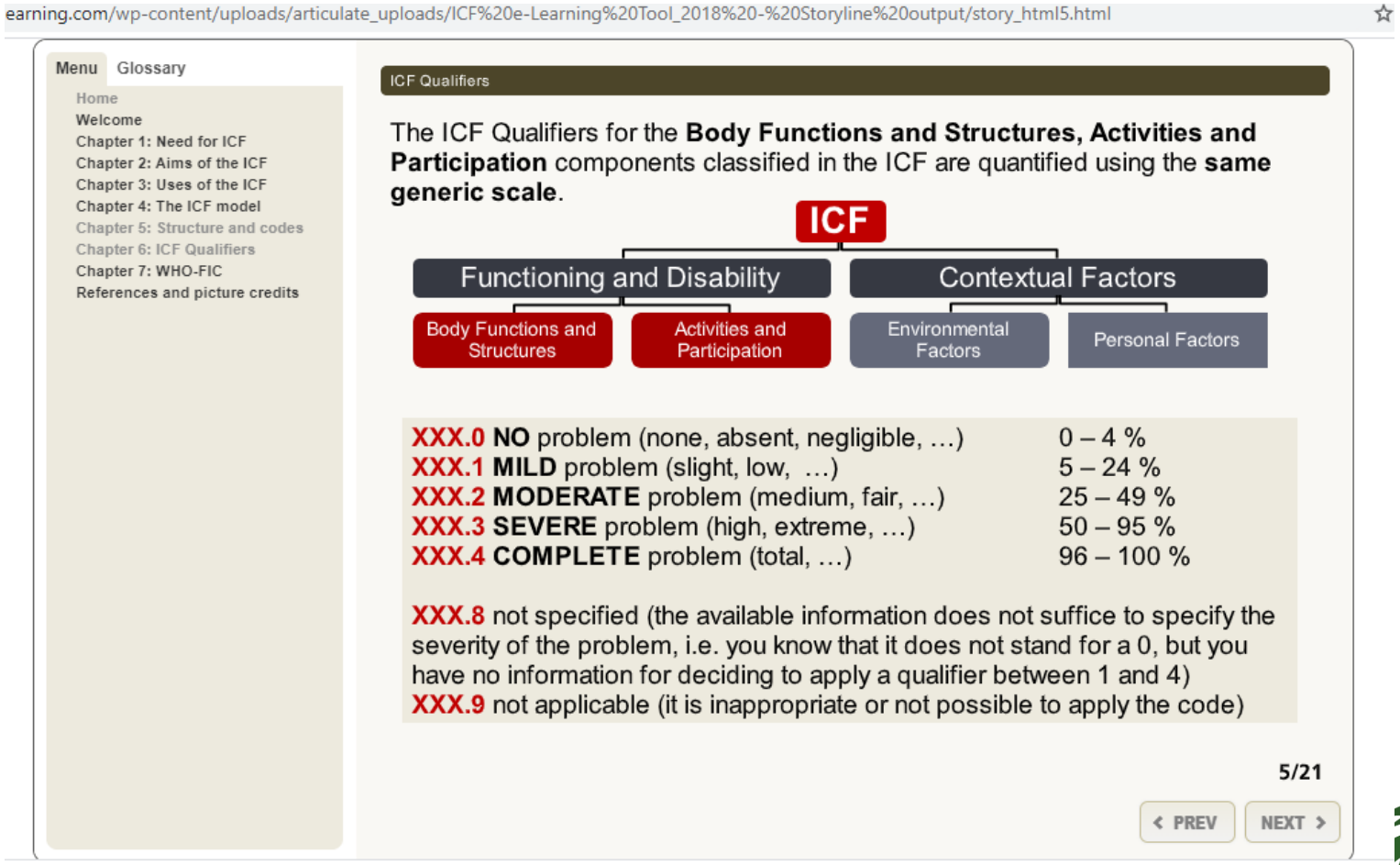
- d910
- d920 Recreation and leisure
- d930
- d940
- d950
- d998
- d999

Engaging in any form of play, recreational or leisure activity, such as informal or organized play and sports, programs of physical fitness, relaxation, amusement or diversion, going to art galleries, museums, cinemas or theatres; engaging in crafts or hobbies, reading for enjoyment, playing musical instruments; sightseeing, tourism and traveling for pleasure.

28/35

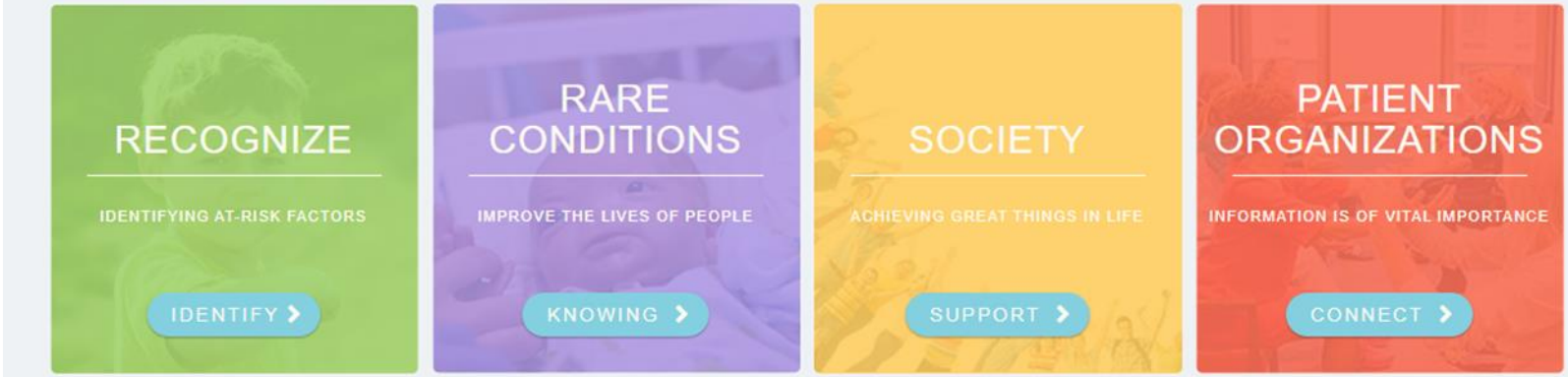
< PREV NEXT >

https://www.icf-elearning.com



Global Child Health Scheme Primary Care

- Physical examination
  - Microtia
    - ICPC: H80
    - ICD 10: Q17.2
    - HPO: 0008551
  - Facial asymmetrie
    - HPO: 0000324
  - Oculo-Auriculo-Vertebral Spectrum/Goldenhar Syndrome
    - ORPHA:141132 Oculo-auriculo-vertebral spectrum
    - OMIM # 164210 HEMIFACIAL MICROSOMIA; HFM
    - Loss of hearing
      - ICF b 230
    - Listening
      - ICF d 115
- Vaccination
  - Diphteria Hemophilus Influenzae B- Pertussis- Poliomyelitis- Tetanus- Hepatitis B
    - ATC J07CA09



rarecare.world

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

Thank

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- People with a rare condition and their families

Just normal people

Advocaters

Goldenhar syndrome

Thalassemia

Chromosome abnormality

Shwachman Diamond Syndrome

Thank You
For Your Attention