Universal Health Coverage: leave no child behind

Towards a global digital child health record with

FHIR: get involved!

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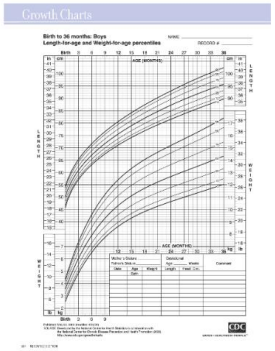
ce-chair Rare Disease Working Group (EAP/UEMS)
Rare Care World Foundation

Global Child Health

13. Recognize that primary health care brings people into first contact with the health system and is the most inclusive, effective and efficient approach to enhance people's physical and mental health, as well as social well-being, and that primary health care is the cornerstone of a sustainable health system for universal health coverage and health-related Sustainable Development Goals, as was declared in the Declaration of Alma-Ata and reaffirmed by the Declaration of Astana;

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

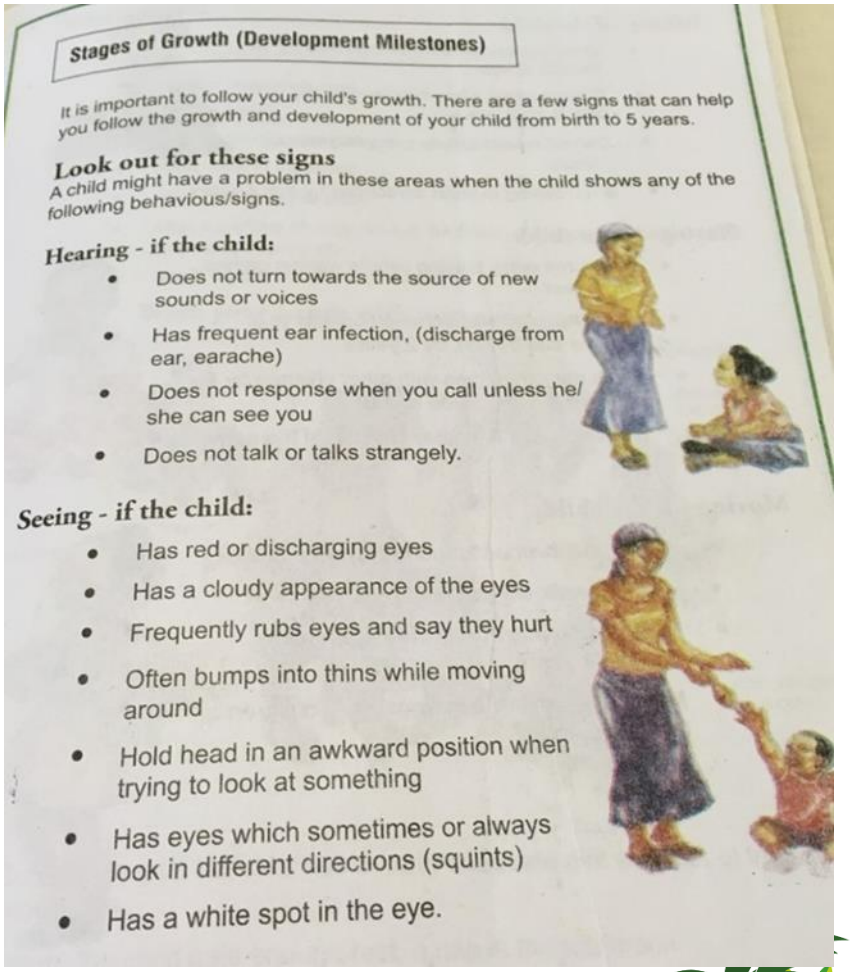
Preventive child health



Growth
Development
Physical examination
Vaccination

Global Child Health

World Health Organization



Ghana

Global Child Health Schemes

Collaborate on a interoperable global set for measurements, observations and surveillances

cademy of Pediatrics
E HEALTH OF ALL CHILDREN®

2015 Recommendations for Preventive Pediatric

Bright Futures/American Academy of Pediatric

amily is unique; therefore, these Recommendations for Preventive Pediatric Health Care are care of children who are receiving competent parenting, have no manifestations of any problems, and are growing and developing in satisfactory fashion. Additional visits may ıry if circumstances suggest variations from normal.

al, psychosocial, and chronic disease issues for children and adolescents may require ıg and treatment visits separate from preventive care visits.

These guidelines represent a consensus by the American Academy of Pediatri Bright Futures. The AAP continues to emphasize the great importance of conti comprehensive health supervision and the need to avoid fragmentation of care

Refer to the specific guidance by age as listed in Bright Futures guidelines Shaw JS, Duncan PM, eds. *Bright Futures Guidelines for Health Supervision o and Adolescents*. 3rd ed. Elk Grove Village, IL: American Academy of Pediatric

	INFANCY								EARLY CHILDHOOD							MIDD		
AGE[1]	Prenatal[4]	Newborn[4]	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	6 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		●[8]	★	★	★	★	★	★	★	★	★	★						
TAL/BEHAVIORAL ASSESSMENT																		
Developmental Screening[9]								●			●							
Autism Screening[10]											●	●						
Developmental Surveillance		●	●	●	●	●	●		●	●		●						
sychosocial/Behavioral Assessment		●	●	●	●	●	●	●	●	●	●	●						

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

Diagnosis

Hurler syndrome

PKU, Duchenne MD, FOP

Shwachman Diamond Syndrome

ICD - 10

Orphacode

OMIM

SNOMED -CT

DCOM

HPO

LOINC

ICPC

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

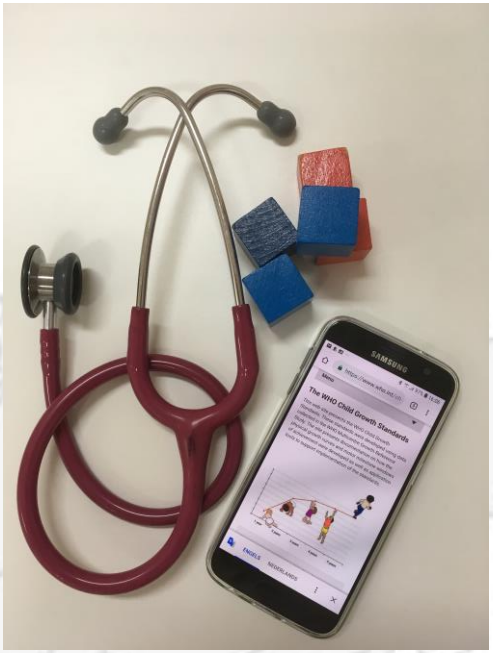
Registry

Data collection with systematically organised computer processable collection medial terms

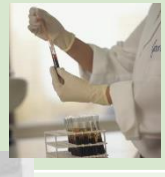
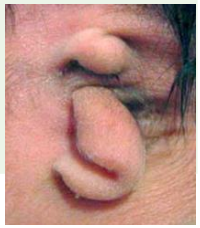
Guideline

Social services and rehabilitation

Data exchange by HL7



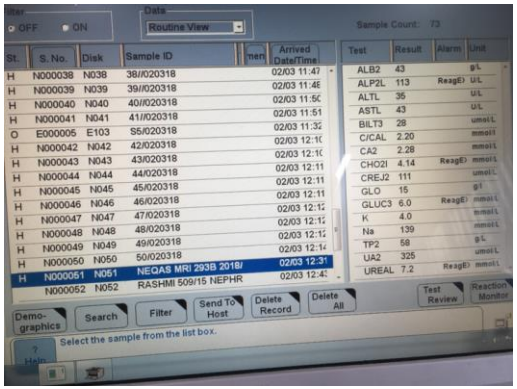
The Health Level Seven (HL7) standards are a set of international guidelines for transferring and sharing data between healthcare providers.

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 versio the *International Classification of Functioning, Disability a Health* (ICF, WHO, 2001) designed to record characteristic 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

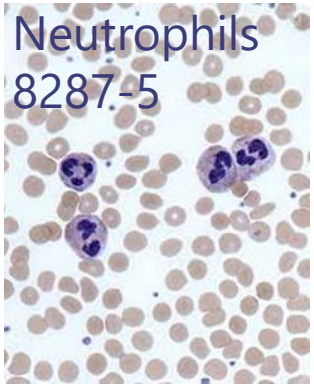
One global e- health model to harmonize interoperable child health
A core set of terminologies was identified:

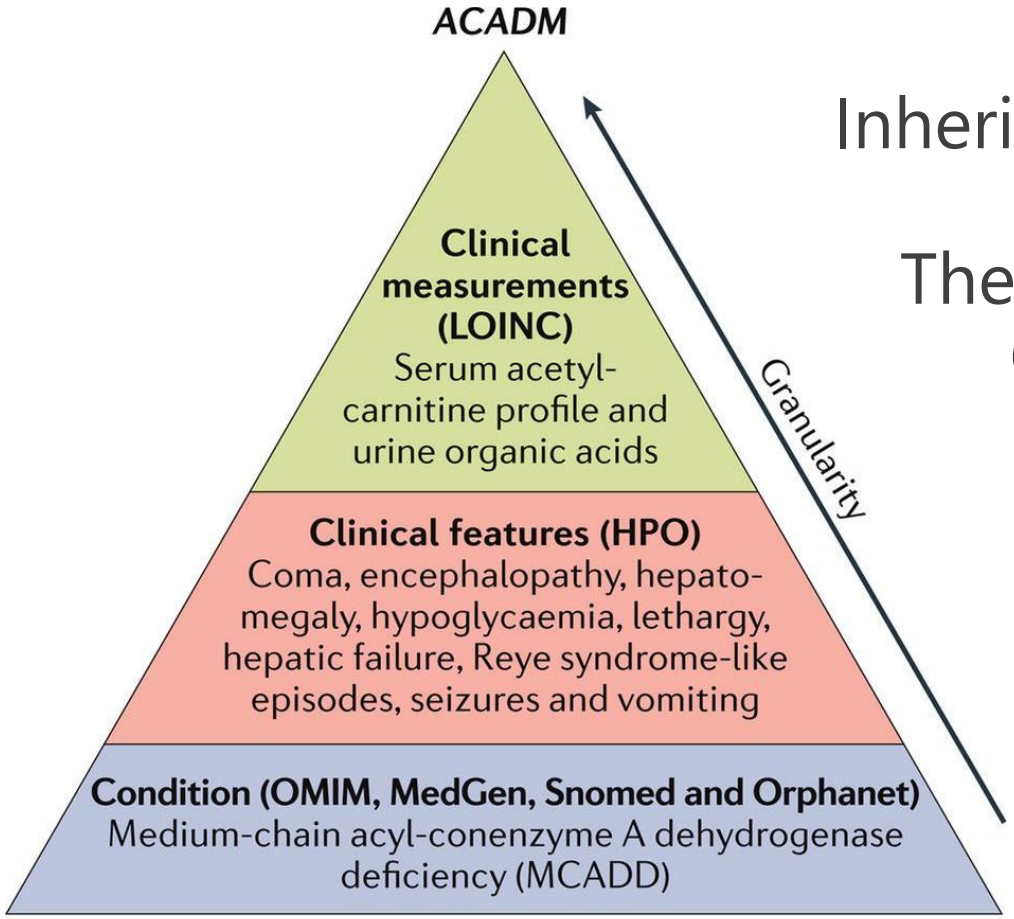
1**) to identity the child in primary care**
-ICPC International Classification of Primary Care
-HPO Human Phenotype Ontology
-LOINC Standard for identifying health measurements, observations, and documents
2**) to classify the disease**
-ICD International Classification of Diseases
-ORPHAnet Classification of rare diseases
-OMIM Catalog of Human Genes and Genetic Disorders
3) **for follow up and treatment**
- LOINC Standard for identifying health measurements
- ATC Anatomical Therapeutic Chemical Classification System
- SNOMED Clinical health terminology
4) **for social support**
-ICF International Classification of Functioning, Disability and Health
Three different rare conditions were selected to compare the e-health services.



46. Expand the delivery of and prioritize primary health care as a cornerstone of a sustainable people-centred, community-based and integrated health systems and the foundation for achieving universal health coverage, while strengthening effective referral system between primary and other levels of care, recognizing that community-based services constitute a strong platform for primary health care;

Strenghtening effective referaal systems, between primary and other leves of care





Online Mendelian Inheritance in Man (OMIM)

The Human Phenotype Ontology (HPO)

50 YEARS OMIM
Human Genetics Knowledge for the World

Nature Reviews | Genetics

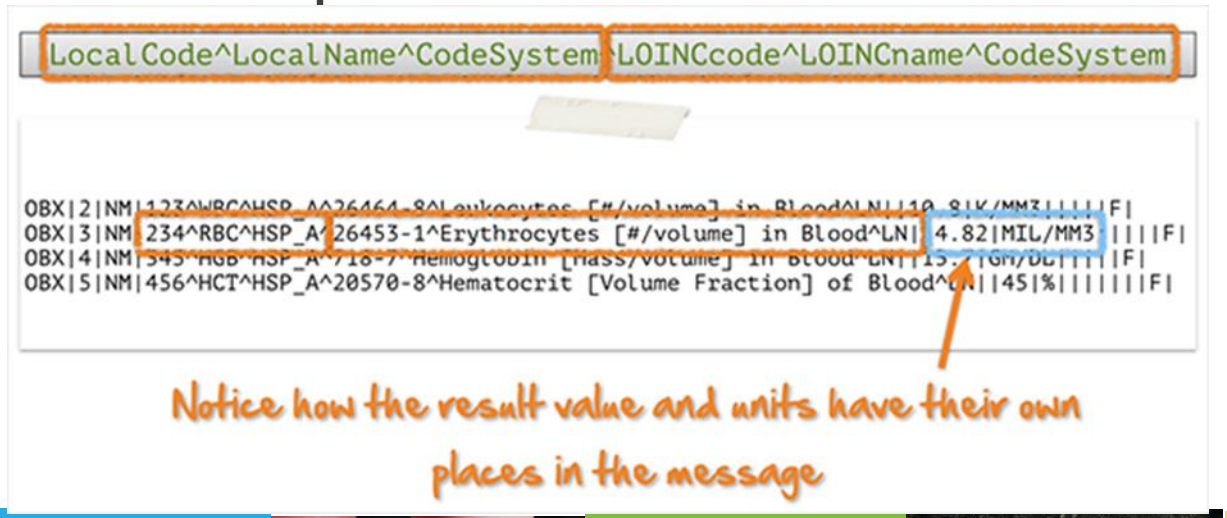
New variants found in Mendelian disease, what next? Review #bioinformatics scoring to prioritise 2017 https://www.nature.com/nrg/articles

twitter

LOINC

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

Interoperable codes in care



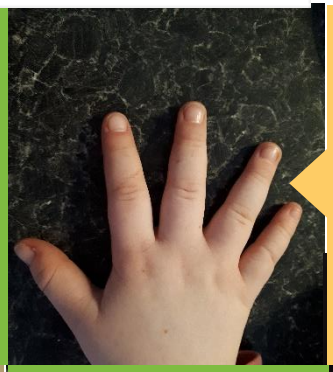
Goldenhar s
Abnormal ear

.

HP:0008551

FOP
Abnormal toe

HP:0010109

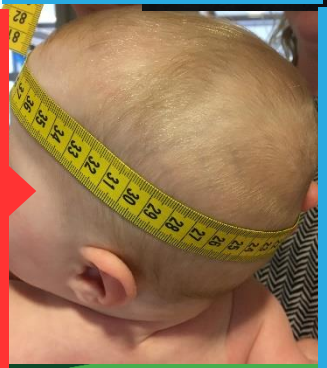


Coffin Lofry s
Tapered fingers

HP:0001182

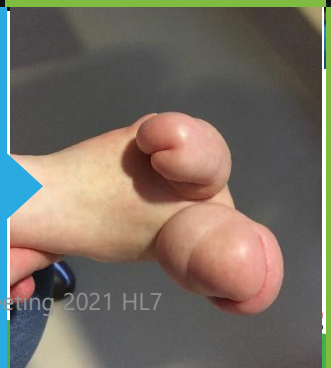
Measuring Head Circumference

LOINC
8287-5



Lower limb asymmetry

HP:0100559



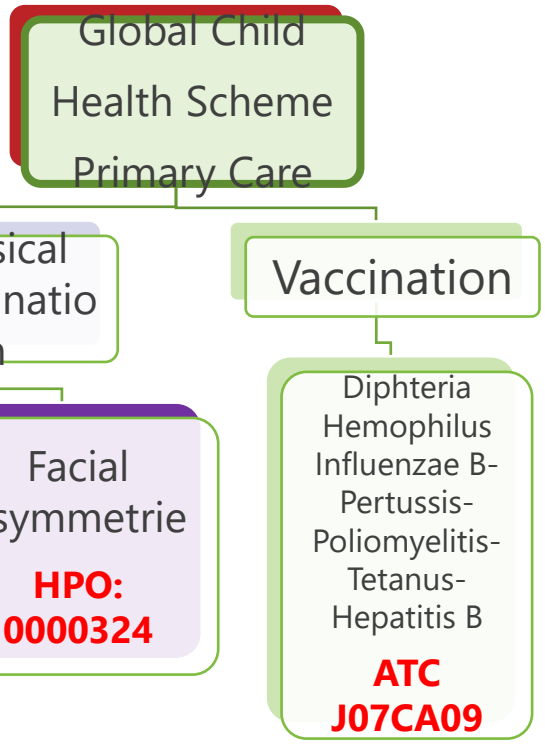
Shwachman DS

ATC

. A09AA02
Pancreatine

International classifications as a tool for interoperability in child health

Towards a Global Integrated Digital Preventive Child Health Model



Oculo-Auriculo-Vertebral Spectrum/Goldenhar Syndrome
ORPHA:141132 Oculo-auriculo-vertebral spectrum
OMIM # 164210 HEMIFACIAL MICROSOMIA; HFM

One code = One meaning

ICPC: International Classification of Primary Care

HPO: Human Phenotype Ontology

LOINC Standard for identifying health measurements, observations, and documents

ICD: International Classification of Diseases

ATC: Anatomical Therapeutic Chemical Classification System

ORPHA: Classification of rare diseases

OMIM: Catalog of Human Genes and Genetic Disorders

Use of terminologies enables semantic interoperability between systems using HL7 CDA and FHIR

Building the Rare Disease

knowledge and information ecosystem

rarecare.world

Application Programming Interface

SEARCH

Find and share knowledge about

Rare diseases all over the world

Sri Lanka

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)

… causes bone marrow expansion and cortical thinning. **Thalassemia** major, which not properly t… 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 … 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

Ministerie van Volksgezondheid, Welzijn en Sport

FHIR/ HL7 Get involved

RarecareFHIRIG - Local Development build (v0.1.0). See the Directory of published versions

1 Home

This repository contains the FHIR resources for the "Een PGO voor iedereen" ("A personal healthcare environment for everyone") project.

Note: *All example content is example only!* It is based on Shwachman Diamond Syndrome (SDS) data from rarecare.world, but for brevity it is much shorter than the actual data would be.

1.1 MedMij and the "PGO for everyone" project

The Netherlands has a national effort, MedMij, to provide all Dutch citizens with a personal healthcare environment, web or mobile. However, persons with rare diseases have trouble seeing their condition properly represented. Due to the rare nature of their condition, vendors are hesitant to invest in small populations. Having a machine-readable Rare Condition profile would enable vendors to simply read the necessary data to provide those persons with customized dashboards, graphs and questionnaires to address their conditions properly. Moreover, healthcare professionals, patient organizations and researchers could all benefit from the structured collection of data.

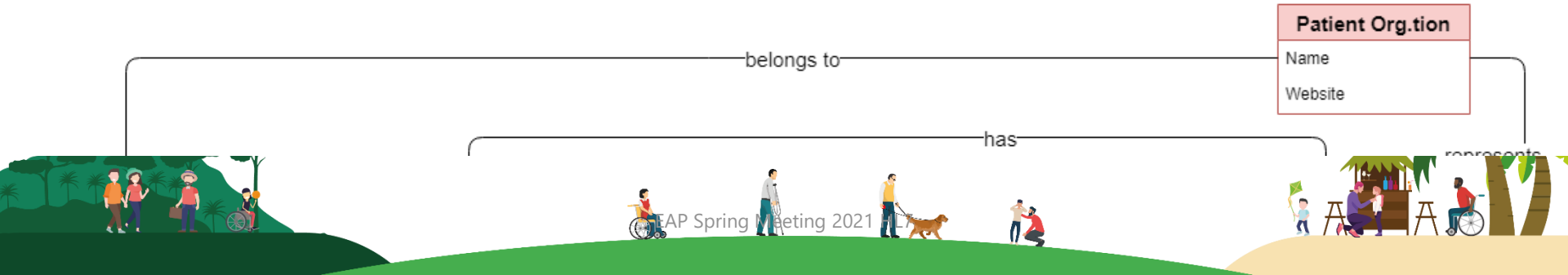
The "PGO for everyone" project aims to provide such a machine-readable API for PGO's. The definitions for specific rare conditions are published as FHIR resources. PGO's can pull those in with an API, and use the definitions to provide disease-specific dashboards and questionnaires for those rare conditions.

1.2 The RareCare Data Model

The Rare Care models are maintained at https://rarecare.world

and (only partially complete yet) https://decor.nictiz.nl/art-decor/decor-datasets-zaz-

From those resources FHIR profiles are generated. The basis is a Data Model of Rare Conditions:



Thank

- European Pediatric Rare Disease Network

John Dodge, U.K.
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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, Kolkata, India
Marc de Graauw, IT Expert, Netherlands
Martin Postma, IT Expert, Netherlands

- People with a rare condition and their families