

# Child Health Digital Ecosystem

Elizabeth Siderius

GGD IJsselmeer, Wedericklaan 56, 8265 DD Kampen, The Netherlands

Rare Care World

[e.siderius@kpnplanet.nl](mailto:e.siderius@kpnplanet.nl)



David Neubauer

Department of Child, Adolescent & Developmental Neurology

University Children's Hospital, Bohoriceva 20, 1525 Ljubljana, Slovenia

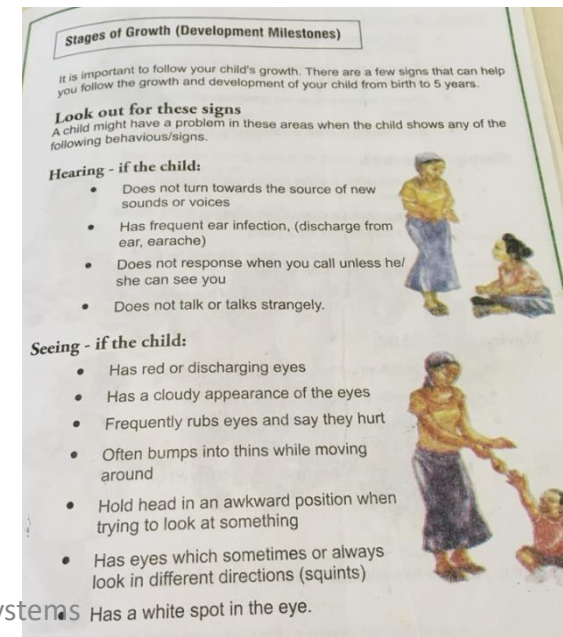
# Introduction

The **United Nations Sustainable Development Goals (SDG)** “ensure healthy lives and promote well-being for all ages”.

To meet the challenge of sustainable healthcare, communication and cooperation between **all involved in child health** should be supported.






Information systems, providing interoperability between public health, medical- and social care, **generate data to improve health.**

To establish a semantic interoperable child health record, efficient management and **publishing of terminologies** is a precondition.





World Health Organization

| Codification  | Meaning  |
|---|--|
| ICD & Orpha code  | International Code of <b>Diseases</b> / Orphanet code  |
| ICF (-CY)<br>  | The <i>International Classification of Functioning, <b>Disability</b> 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<br>      | A universal code system for <b>tests</b> , measurements, and observations.   |
| ICPC<br>      | 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<br>      | The purpose of the ATC/DDD system is to serve as a tool for <b>drug</b> utilization research in order to improve quality of drug use.  |
| ISO 9999<br> | ISO 9999:2011 establishes a classification of <b>assistive products</b> , especially produced or generally available, for persons with disability.   |



# Methods

The European Academy of Paediatrics has established a working group on ethics and rare disease to ensure the input of pediatricians in the universal right of the disabled child.

One global e- health model has been designed to harmonize interoperable child health by a Dutch patient group:

**A core set of terminologies was identified:**

**1) to identify 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**

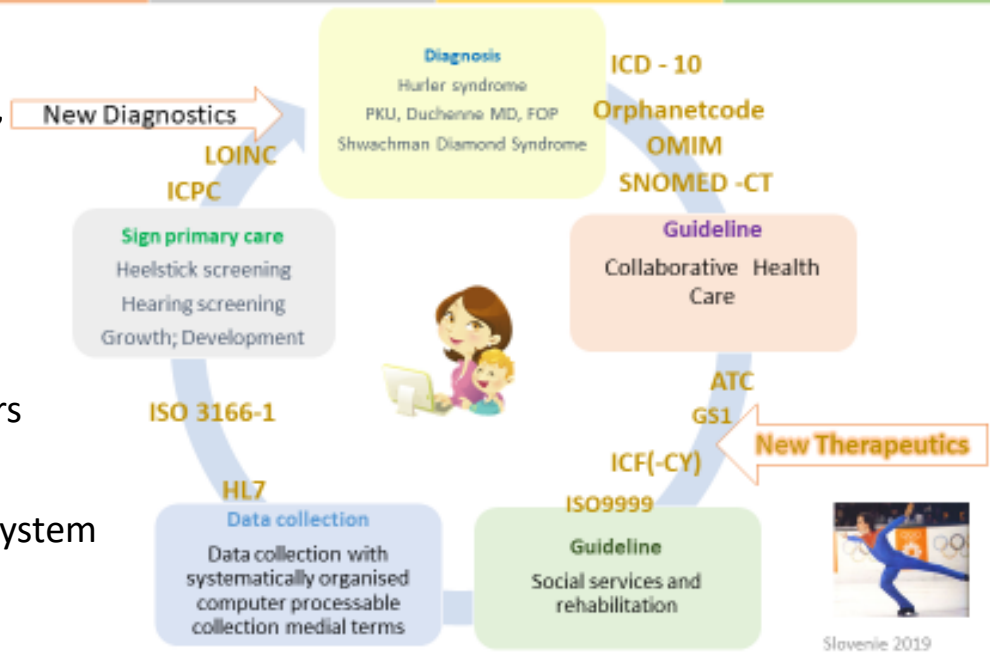
- ATC Anatomical Therapeutic Chemical Classification System
- SNOMED Clinical health terminology

**4) for social support**

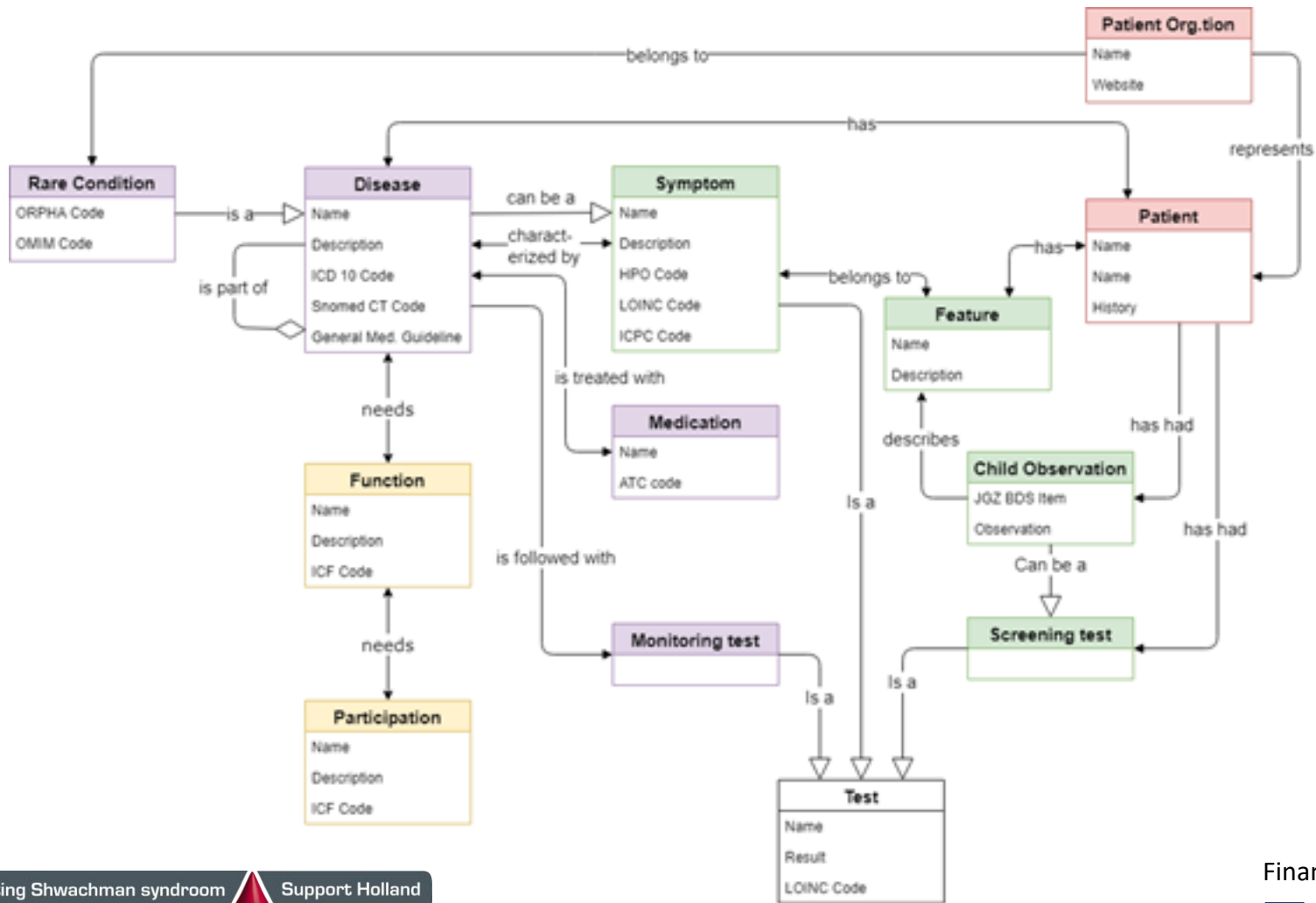
- ICF International Classification of Functioning, Disability and Health

Three different rare conditions were selected to compare the e-health services.

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



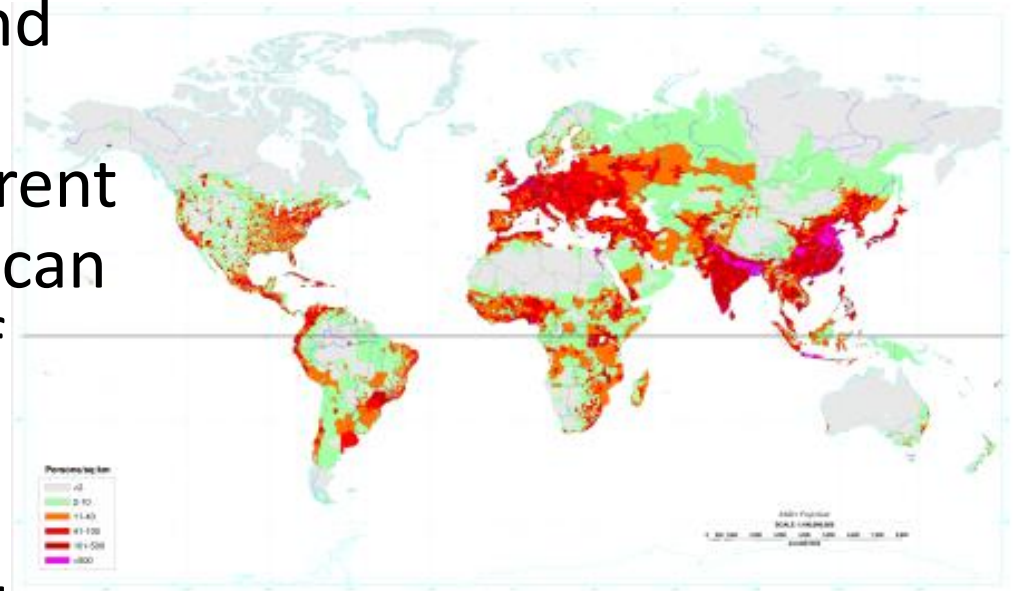
# Unified Modeling Language (UML)



# Results

Duchenne Muscular Dystrophy, neonatal cholangiostasis, and Shwachman Diamond Syndrome, completely different rare and complex disorders can all be affiliated to this set of international classifications.

Still harmonization of data exchanges within states and across borders seems to be the major hurdle in digital personalized health.



*Population density (people per km<sup>2</sup>) map of the world in 1994. In relation to the equator it is seen that the vast majority of the human population lives in the Northern Hemisphere, as 67% of the Earth's land area is there. (Wikipedia)*



# 310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD OMIM

H02AB07 prednisone ATC



# neonatal cholangiostasis

1975-2 Bilirubin.total [Mass/volume] in Serum or Plasma LOINC

HP: 0006579 Prolonged neonatal jaundice HPO



About Membership Ecosystems International Projects Discover News & Events



Slovenian ECHAlliance Ecosystem



Scottish Digital Health and Care Ecosystem



Digital Health Rheinland Ecosystem



Northern Ireland Connected Health Ecosystem



shutterstock.com • 163524896



NICE / PACA Provence



stem



Digital Health Malta Ecosystem



Digital Technologies and Health Ecosystems Ljubljana, 12 Sept 2019

SNOMEDCT/129104009 Developmental mental disorder (disorder)

J07CA09 diphtheria-hemophilus influenzae B-pertussis-poliomyelitis-tetanus-hepatitis B ATC



# 260400 SHWACHMAN-DIAMOND SYNDROME 1; SDS1 OMIM

16142-2 : Fat [Mass/time] in 24 hour Stool LOINC



# Building the Rare Disease

knowledge and information ecosystem

Thalassemia, 3500 patients have been identified in Sri Lanka.



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)

### OMIM

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

Hemoglobinopathies

Digital Technologies and Health Ecosystems

Ljubljana, 12 Sept 2019



# Discussion and conclusions

The World Health Organization states that a **collaborative management** approach at the primary health care level involving patients, their families and other health care providers may reduce the burden of disease for the child, family, and society. The **family-centered chronic care management originated in pediatric care.**

A **general global set of interoperable classifications** can help to ensure awareness and optimal health for children with a rare and disabling condition. Knowledge of the diseases as well as e health is a precondition in designing efficient digital strategies. A **holistic semantic structure connects primary care, hospital care and integrated person centered care.** These structures are not in place and /or locked in vendor dependent IT systems.

