Universal Health Coverage

Digital Health : Leave no child behind

Liesbeth Siderius
Rare Care World Foundation
Shwachman Syndrome Support Holland Foundation
The Netherlands

Patient Informatiom	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

ATC
New Therapeutics
ICF
ISO9999

ISO 3166-1
HL7/ FHIR

Registry
Data collection with systematically organised computer processable collection medial terms

Guideline
Social services and rehabilitation

International terminologies as a tool for interoperability in child health
Towards a Global Integrated Digital Preventive Child Health Model

One code = One meaning

CPC: International Classification of Primary Care

- HPO: Human Phenotype Ontology

LOINC: Standard for identifying health measurements, observations, and documents

- ICD: International Classification of Diseases

SNOMED CT: Systematized Nomenclature of Medicine Clinical Terms

- ICF: International Classification of function

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

REGULATION (EU) 2016/679 OF THE EUROPEAN PARLIAMENT AND OF THE COUNCIL

of 27 April 2016

on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation)

General Data Protection Regulation **2016**

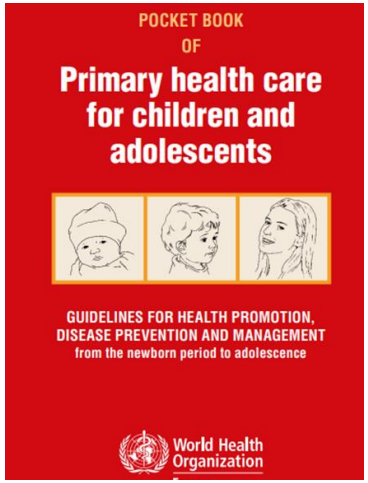
Art. 20 GDPR

Right to data portability

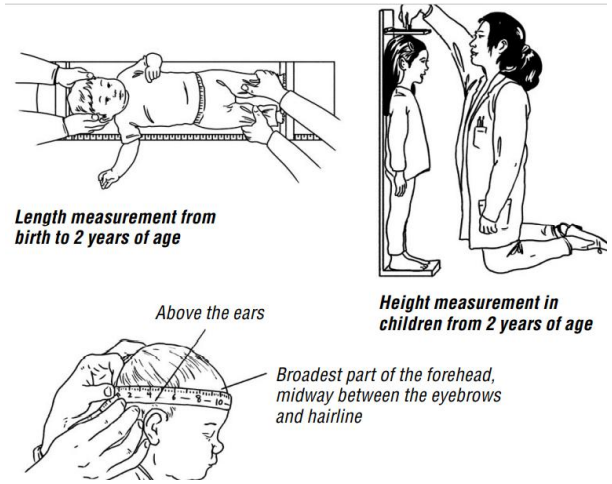
The data subject shall have the right to receive the personal data concerning him or her, which he or she has been provided to a controller, in a **structured, commonly used and machine-readable format** and have the right to transmit those data to another controller without hindrance from the controller to which personal data have been provide….

World Health Organization

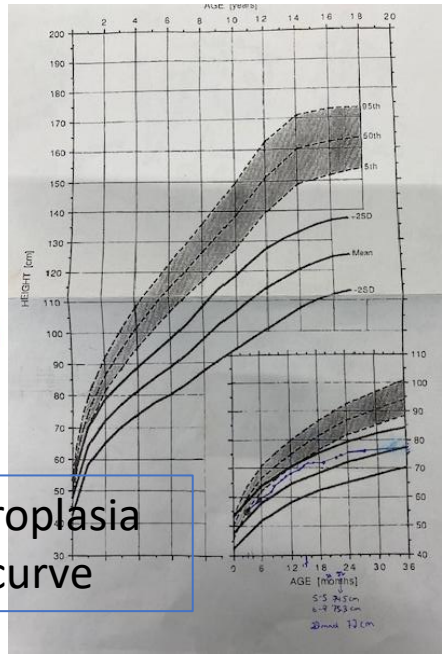
Digital Modelling of Primary Child Health
From home to each health system



https://www.who.int/europe/publications/i/item/9789289057622

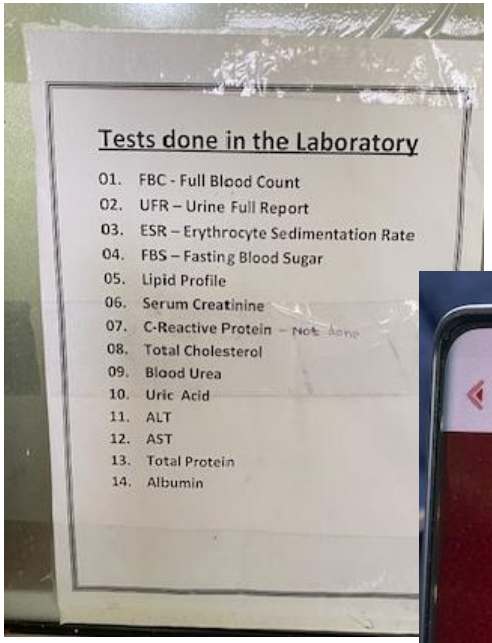


The Concept	Different Terms Used in Different Geographical Areas	Single Code Understand Globally
	English: “Body Height” Dutch: "Lichaamslengte" French: "Taille du corps" Spanish: "Altura del cuerpo" German: "Körpergröße" Italian: "Altezza del corpo" Portuguese: "Altura do corpo" Russian: "Рост тела" (Rost tela) Japanese: "身長" (Shinchō) Chinese (Simplified): "身高" (Shēn gāo) Arabic: ") "طول الجسمToul al jism)	**LOINC 8302-2 (Body height)**

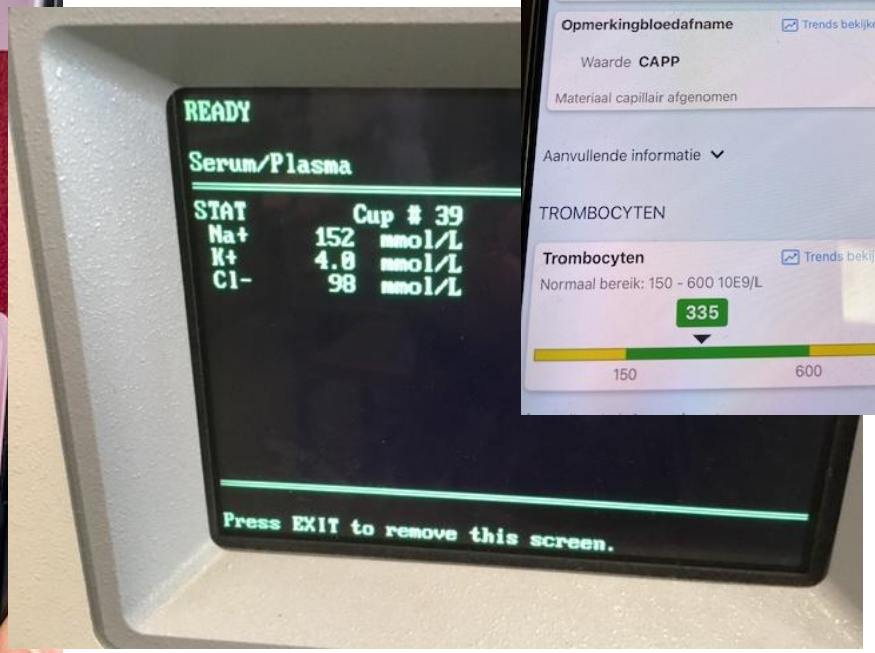


Achondroplasia Growthcurve


How do you say glucose?
Glucose 葡萄糖 Glükoos Glucose Glucose Glucose Glukose
LOINC
the lingua franca of clinical observation exchange
Γλυκόζη Glucosio 포도당 Glicose Glucosa Glucosa Glucosa


Tests done in the Laboratory
01. FBC - Full Blood Count
02. UFR – Urine Full Report
03. ESR – Erythrocyte Sedimentation Rate
04. FBS – Fasting Blood Sugar
05. Lipid Profile
06. Serum Creatinine
07. C-Reactive Protein – Not done
08. Total Cholesterol
09. Blood Urea
10. Uric Acid
11. ALT
12. AST
13. Total Protein
14. Albumin


Blood Sugar
मधुमेह (Blood Sugar)
80
PREV
NEXT

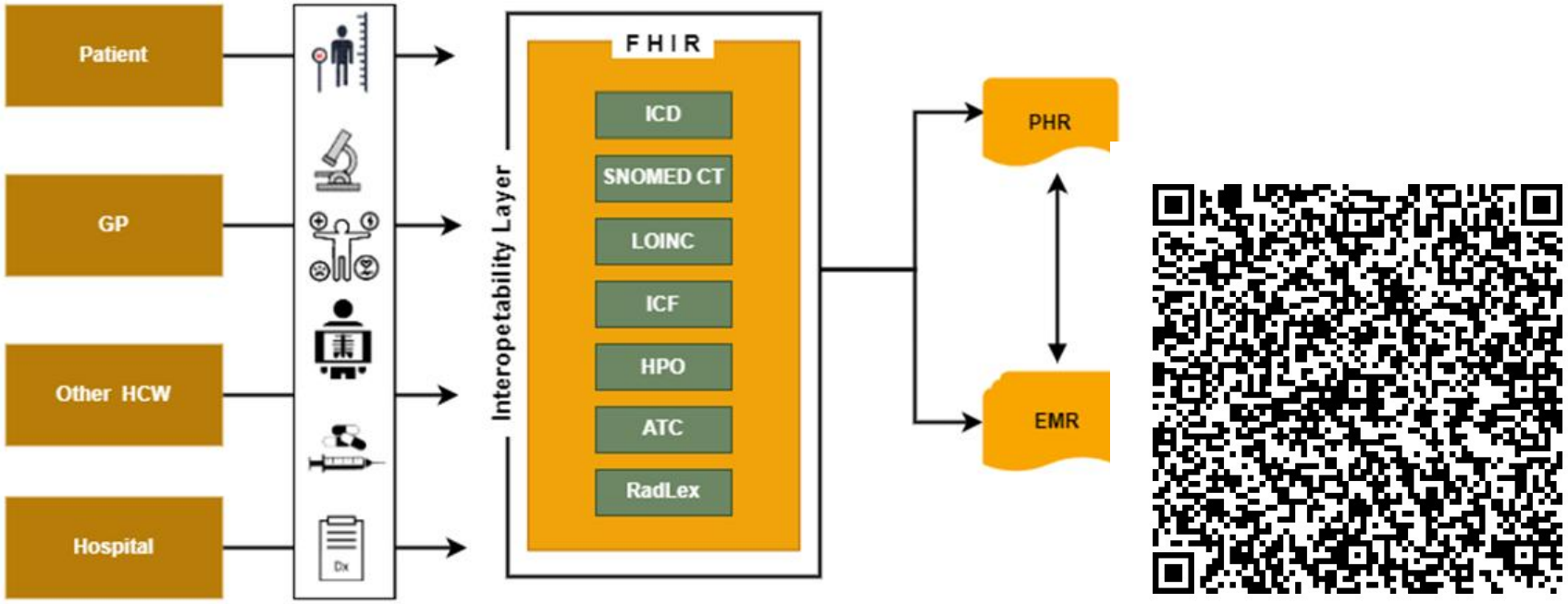

READY
Serum/Plasma
STAT Cup # 39
Na+ 152 mmol/L
K+ 4.0 mmol/L
Cl- 98 mmol/L
Press EXIT to remove this screen.

Terug
Onderzoeksdetails
Sluiten
Normaal bereik: 4,0 - 16,0 10E9/L
15,3
4
16
#Neutrofielen
Trends bekijken
Normaal bereik: 1,0 - 9,0 10E9/L
3,81
1
9
Opmerkingbloedafname
Trends bekijken
Waarde CAPP
Materiaal capillair afgenomen
Aanvullende informatie
TROMBOCYTEN
Trombocyten
Trends bekijken
Normaal bereik: 150 - 600 10E9/L
335
150
600

“Digital child health: opportunities and obstacles”,

by Liesbeth Siderius, Sahan Damsiri Perera, Lars Gelander, Lina Jankauskaite, Manuel Katz, Arunas Valiulis, Adamos A. Hadjipanayis, Laura Reali and Zachi Grossman

Front. Pediatr., 22 December 2023
Sec. Children and Health
Volume 11 - 2023 | https://doi.org/10.3389/fped.2023.1264829



Author Figure S.D. Perera, University Colombo, Srilanka

Chromosome 22 Q Deletion Syndrome

Autism

= ICD10 F84.0

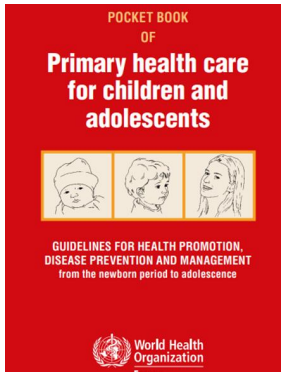
ICF

d 820 School education

d 210 Undertaking single task

d 610 Acquiring a place to live

Social participation



Problems during pregnancy

Diagnostic Short Intrauterine Femur Length US LOINC 11963-6

Body height Measured --at birth

LOINC 89269-5

Achondroplasia

=ICD 10 Q77.4

Managament Hydrocephalus risk

Head Occipital-frontal circumference by Tape measure LOINC 8287-5

Short Stature

Body Height LOINC 8302-2

Diagnostic test: Phosphate [Moles/volume] in Serum or PlasmaLOINC 14879-

Diagnosis: X-linked

hypophosphatemia (XLH)= OMIM 307800 = ORPHA:89936

new medicine

Therapy: Calcitriol ATC A11CC04 or

Failure to thrive

HPO: HP:0001531

LOINC:42819-3 Failure to thrive [CCC]

ICPC: T10

SNOMEDCT_US:36440009

Diagnostic test: Fatty Stool

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

Shwachman Diamond Syndrome

Diagnosis= Orphacode 811

Visual impairment

Infantile cataracts remain one of the **most treatable causes of lifelong visual impairment**.

While the chance of improving vision for children with infantile cataracts has never been better,

Significant global and socioeconomic disparities still exist in their early management.

Lenhart PD, Lambert SR. Current management of infantile cataracts. Surv Ophthalmol. 2022 Sep-Oct;67(5):1476-1505. doi: 10.1016/j.survophthal.2022.03.005. Epub 2022 Mar 17. PMID: 35307324; PMCID: PMC10199332.

A Two Month old at PCH

Juvenile Cataract

Seeing red in young children: the importance of the red reflex Ayad Shafiq British Journal of General Practice 2015; 65 (633): 209-210. **DOI:** https://doi.org/10.3399/bjgp15X684625

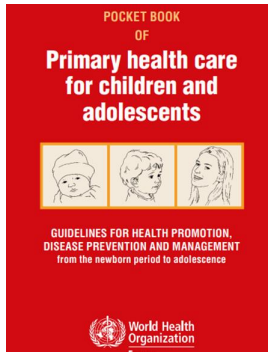
Primary Child Health

Child comes for a regular screening at PCH

Physical exam

Red eye reflex

Referal to ophtalmologist



Observation:

Study observation Left optic lens Slit lamp biomicroscopy Ophtalmol >

Diagnosis : Infantile cataract

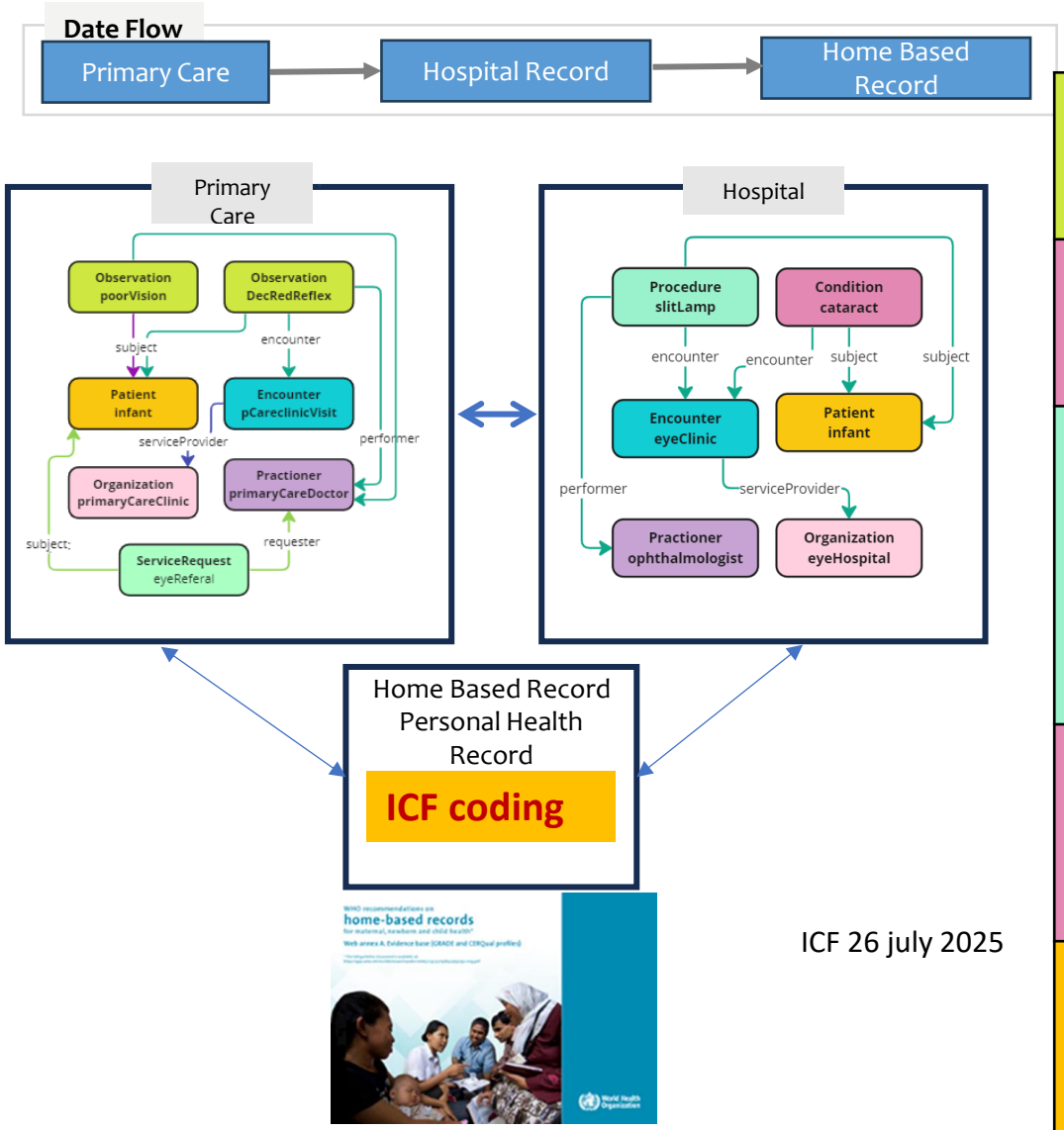
Cloudy lens or absent red reflex

A lens opacity (grey-white clouding of the lens) or absence of the red reflex, during the red reflex examination (p. 119), can be a sign of both congenital cataract (p. 459) and early retinoblastoma (p. 459).

- Refer newborns with an absent red reflex or a cloudy lens immediately to an eye specialist. Early detection and treatment are essential.

ICF 26 july 2025

Juvenile Cataract



ICF 26 july 2025

Terminologies

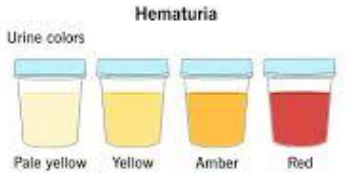
Red reflex absent	SNOMED CT	247079003
Abnormal vision	SNOMED CT	7973008
Study observation Left optic lens Slit lamp biomicroscopy	LOINC	79866-0
Infantile, juvenile and presenile cataract	ICD 10	H26.0
School education	ICF	d820

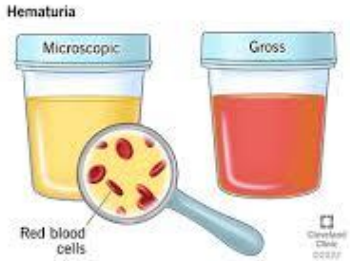
Hearing Loss

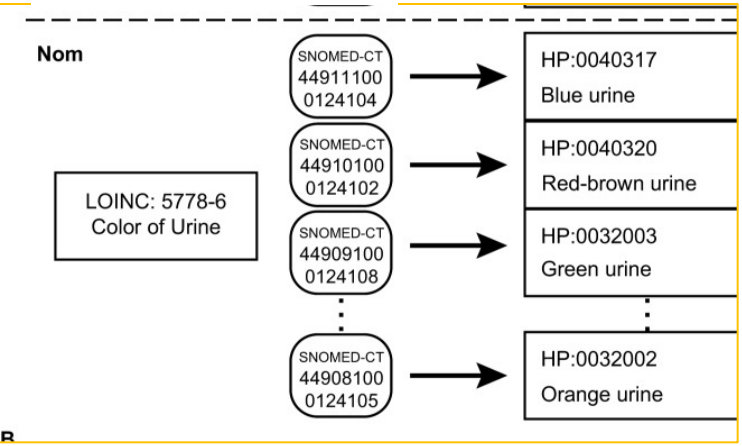
The impacts of hearing loss are broad and can be profound. They include a loss of the ability to communicate with others **delayed language development** in children, which can lead to social isolation, loneliness and frustration, particularly among older people with hearing loss.

Many areas lack sufficient accommodations for hearing loss, which effect academic performance and **options for employment**. Children with hearing loss and deafness in developing countries rarely receive any **schooling**.

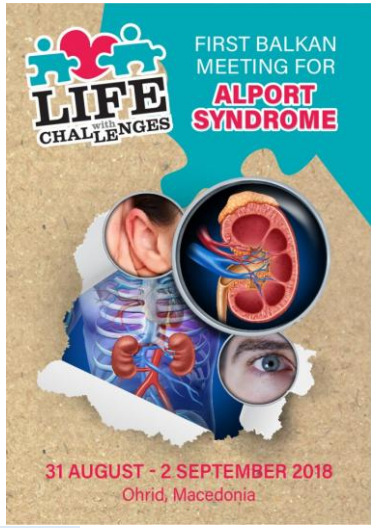
Alport syndrome







Zhang XA, Yates A, Vasilevsky N, et al. Semantic integration of clinical laboratory tests from electronic health records for deep phenotyping and biomarker discovery. *NPJ Digit Med*. 2019;2:32. doi:10.1038/s41746-019-0110-4



LOINC 33051-4 Erythrocytes [Presence] in Urine

LOINC 2888-6 Protein [Mass/volume] in Urine

ICD-10-CM Diagnosis Code N04.9 **Steroid-resistant nephrotic syndrome**

LOINC 53853-8 COL4A5 gene targeted mutation analysis

OMIM # 301050
ALPORT SYNDROME 1, X-LINKED
ORPHA:63 Alport syndrome

LOINC LL6016-1 Hearing Loss - Degree

ICF b 230 Hearing Function

ICF d 820 School education

NEN-EN-ISO 21388:2021
Hearing aid

ICF d 840 Work and Employment

https://datafirst-ailater.health/home

Rare diseases: ethical challenges in the era of digital health

POLICY AND PRACTICE REVIEWS article
Front. Digit. Health, 09 July 2025
Sec. Human Factors and Digital Health
Volume 7 - 2025

Ayushman Bharat
Digital Mission
Building Digital Health Ecosystem

- Empower citizens with digital access to healthcare services.
- Ensure interoperability across public and private health ecosystems.
- Promote real-time data sharing of health records with patient consent.
- Improve the efficiency, quality, and affordability of healthcare.
- Facilitate evidence-based planning and policy-making through anonymized health data.

By digitizing healthcare, ABDM seeks to create a secure, trustworthy, and user-friendly ecosystem that benefits both service providers and patients.

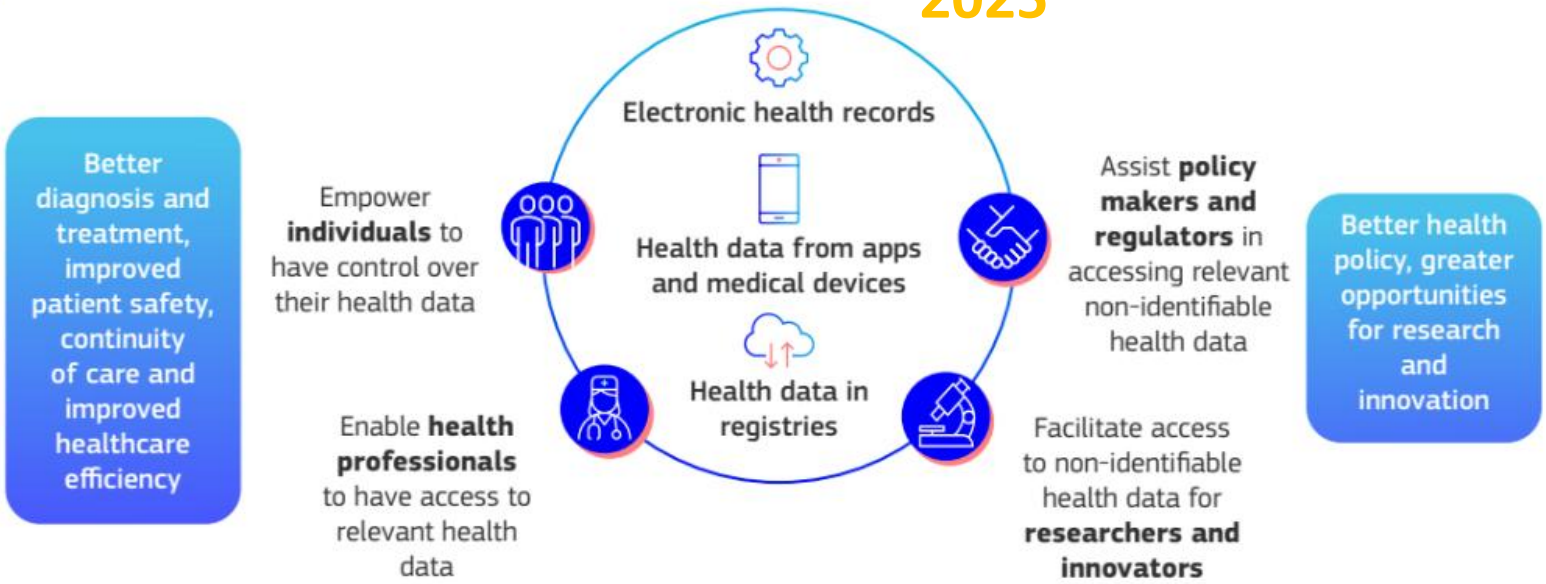
https://www.cheggindia.com/general-knowledge/ayushman-bharat-digital-mission/

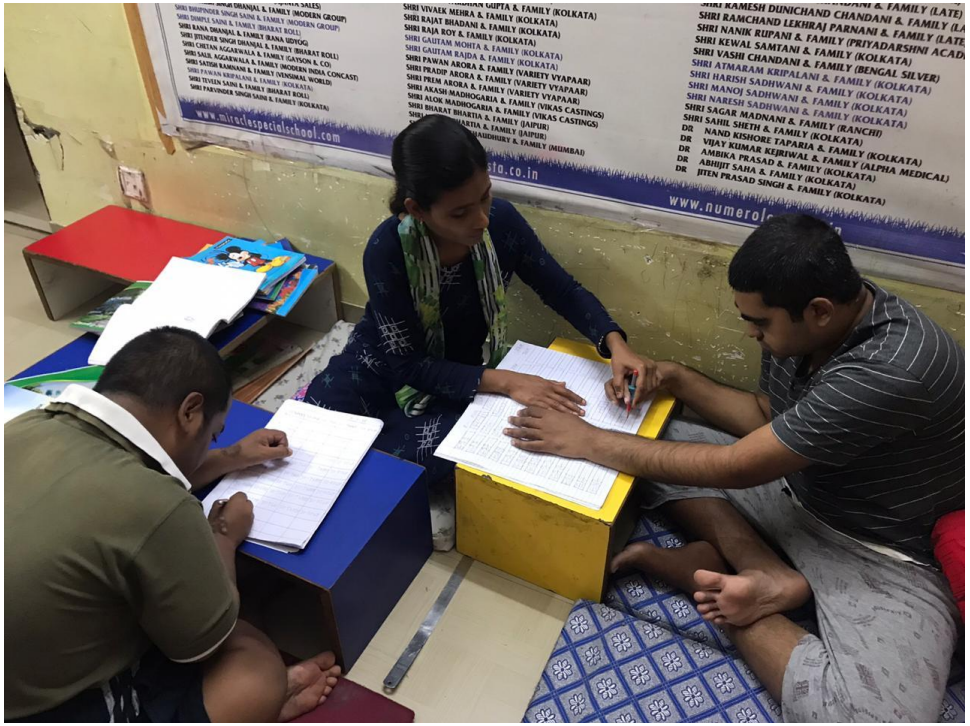
Digital Health Europe

ABOUT NEWS & EVENTS TWINNINGS RESULTS & PUBLICATIONS SUPPORT SERVICES RESOURCES Participate

DigitalHealthEurope recommendations on the European Health Data Space

March 18th 2025





The life experiences of individuals living with a condition are underrepresented in the digitalising world.

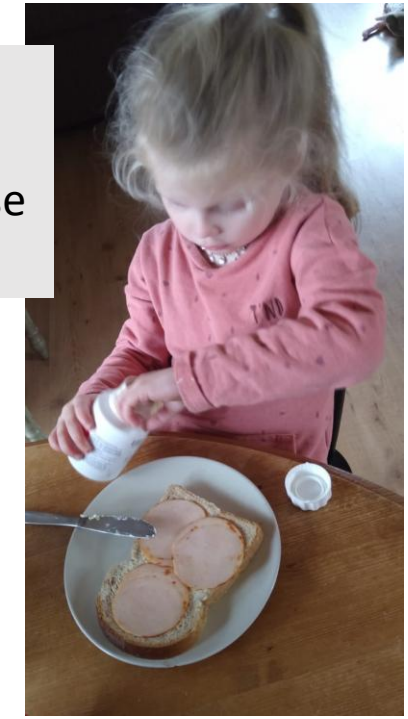
These gaps result in deficiencies in health and social, integrated, holistic, real-life digital data.

Rare diseases: ethical challenges in the era of digital health

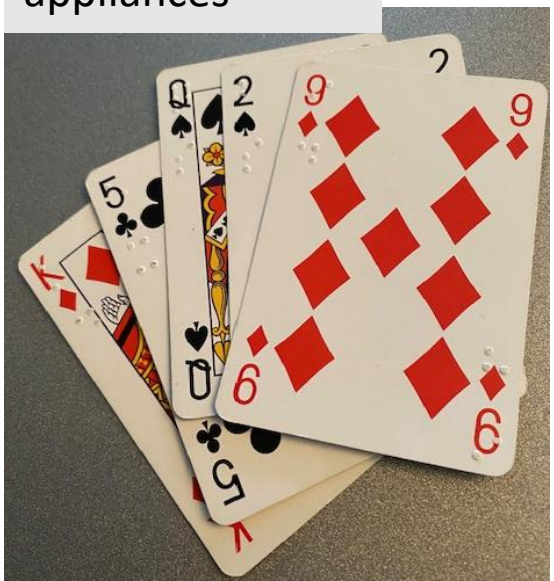
Siderius L, Perera SD, Jankauskaite L, Bhattacharya A, Gonçalves P. Rare diseases: ethical challenges in the era of digital health. Front Digit Health. 2025 Jul 9;7:1539841. doi: 10.3389/fdgth.2025.1539841. PMID: 40704368; PMCID: PMC12283590.

- Rare diseases as an identity (1).
- Public Policies: (2).
- Technical considerations: digital health opportunities in relation to interoperable medical data (3).
- A case study (4).
- AI data comparison: (5).
- Ethical challenge: European law brought concerning Kant's notion of noumena (6).

A09AA02 - **Multienzymes** (Lipase, Protease Etc.)



ISO 17049:2013 Accessible design — Application of **braille** on signage, equipment and appliances



- Public Policies: (2).

The 78th World Health Assembly (May 2025) approved the extension and renewal of the Global Strategy on Digital Health.

January 2025, the Indian Ministry of Health and Family Affairs of India posted: From Data to Diagnosis Transforming Healthcare through Digitalisation. Indicating India's Ayushman Bharat Digital Mission and the Digital Health Incentive Scheme can set a global benchmark for digital healthcare transformation.

On Mar 26, 2025, the European Health Data Space entered force.

However, many existing public health policies do not adequately account for the unique needs of rare disease populations, particularly in **integrating patient-reported outcomes into digital frameworks.**

From Feature to Medical Guideline
- A case study (4)

Feature

- Fatty Stool
- Failure to thrive
- Common infections

Shwachman Diamond Syndrome- *Management*

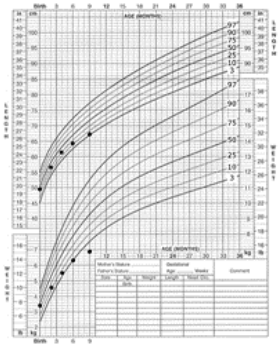
- Pancreas insufficiency
- Neutropenia
- Skeletal Dysplasia
- Autisme like

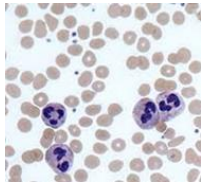
ICF d 820 School education

Failure to thrive
- HP:0001531
- LOINC:42819-3 Failure to thrive

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

Pancreas insufficientie
- ICD -10 K86.81





Ann. N.Y. Acad. Sci. ISSN 0077-8923

ANNALS OF THE NEW YORK ACADEMY OF SCIENCES
Issue: Annals *Meeting Reports*

Draft consensus guidelines for diagnosis and treatment of Shwachman-Diamond syndrome

Yigal Dror,[1] Jean Donadieu,[2] Jutta Koglmeier,[3] John Dodge,[4] Sanna Toiviainen-Salo,[5] Outi Makitie,[5] Elizabeth Kerr,[1] Cornelia Zeidler,[6] Akiko Shimamura,[7] Neil Shah,[3] Marco Cipolli,[8] Taco Kuijpers,[9] Peter Durie,[1] Johanna Rommens,[1] Liesbeth Siderius,[10] and Johnson M. Liu[11]

Shwachman Diamond S
ORPHA:811
OMIM# 260400

Cystic Fybrosis
ORPHA:586
OMIM # 21970

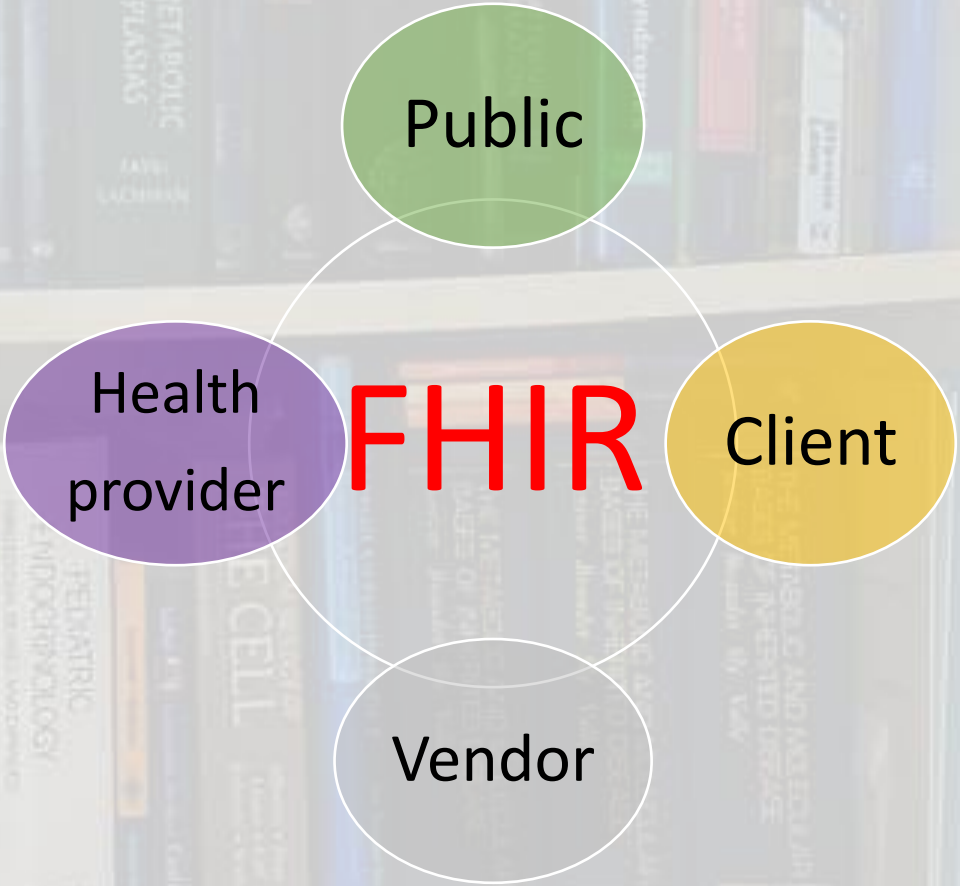
Open Access FHIR RESTfull API Library

Mother and Child Health
- Growth & Development
- Conditions

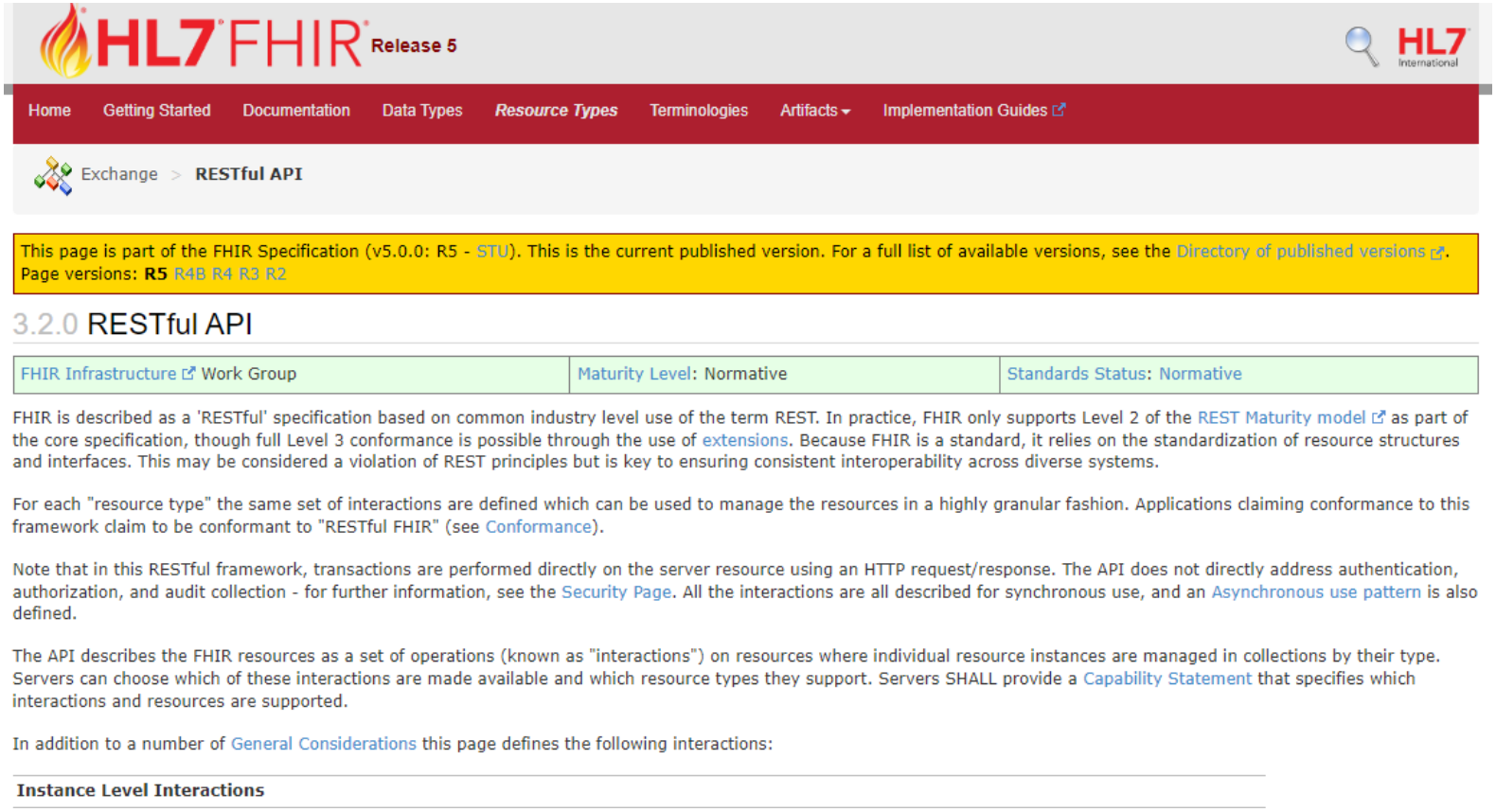
Computable clinical guidelines
- Thalassemia
- Shwachman Diamond Syndrome

Immunizations
- Vaccination schemes

Social Support
- ICF
- ISO 9999



Terminologies enable semantic interoperability in health information exchange standards systems using HL7 CDA and FHIR



HL7 FHIR Release 5

HL7 International

Home | Getting Started | Documentation | Data Types | *Resource Types* | Terminologies | Artifacts | Implementation Guides

Exchange > **RESTful API**

This page is part of the FHIR Specification (v5.0.0: R5 - STU). This is the current published version. For a full list of available versions, see the Directory of published versions.
Page versions: **R5** R4B R4 R3 R2

3.2.0 RESTful API

FHIR Infrastructure Work Group	Maturity Level: Normative	Standards Status: Normative

FHIR is described as a 'RESTful' specification based on common industry level use of the term REST. In practice, FHIR only supports Level 2 of the REST Maturity model as part of the core specification, though full Level 3 conformance is possible through the use of extensions. Because FHIR is a standard, it relies on the standardization of resource structures and interfaces. This may be considered a violation of REST principles but is key to ensuring consistent interoperability across diverse systems.

For each "resource type" the same set of interactions are defined which can be used to manage the resources in a highly granular fashion. Applications claiming conformance to this framework claim to be conformant to "RESTful FHIR" (see Conformance).

Note that in this RESTful framework, transactions are performed directly on the server resource using an HTTP request/response. The API does not directly address authentication, authorization, and audit collection - for further information, see the Security Page. All the interactions are all described for synchronous use, and an Asynchronous use pattern is also defined.

The API describes the FHIR resources as a set of operations (known as "interactions") on resources where individual resource instances are managed in collections by their type. Servers can choose which of these interactions are made available and which resource types they support. Servers SHALL provide a Capability Statement that specifies which interactions and resources are supported.

In addition to a number of General Considerations this page defines the following interactions:

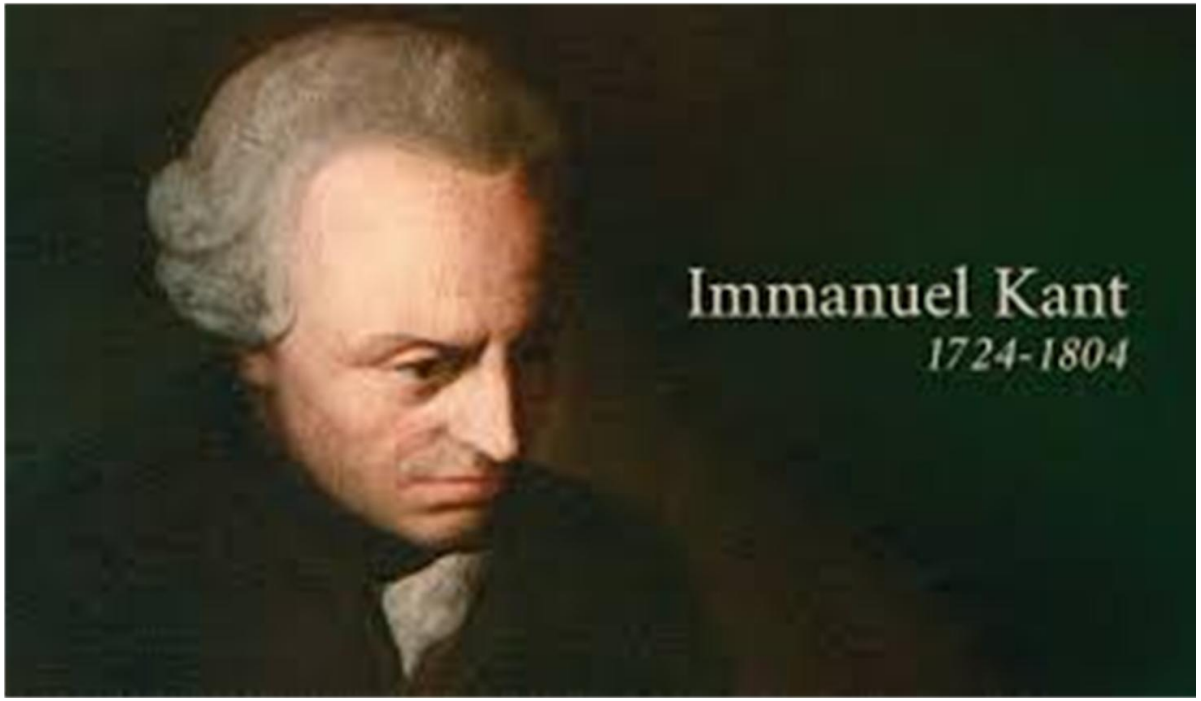
Instance Level Interactions

Harita Y, Kitanaka S, Isojima T, Ashida A, Hattori M. **Spectrum of LMX1B mutations: from nail-patella syndrome to isolated nephropathy.** Pediatr Nephrol. 2017 Oct;32(10):1845-1850. doi: 10.1007/s00467-016-3462-x. Epub 2016 Jul 23. PMID: 27450397.

Ethical challenge: European law brought concerning Kant's notion of noumena (6).

Rare Diseases Europe recently (2025) reported the impact of living with a rare disease. 8 / 10 people with rare diseases live with disabilities.

58% experienced **discrimination** related to the rare disease or disability in **healthcare, housing, employment and education**.



How we perceive the world is shaped by how we see and think.

We can not know the world as it is because we can only experience it through our senses and thoughts.

Growing up with Shwachman Diamond syndrome
International Classification of Function (ICF)

Materials & Methods
Representatives of the Dutch SDS patient organisation selected 12 categories from the domain activities and participation of the ICF core-set autism brief and included these items in a questionnaire.

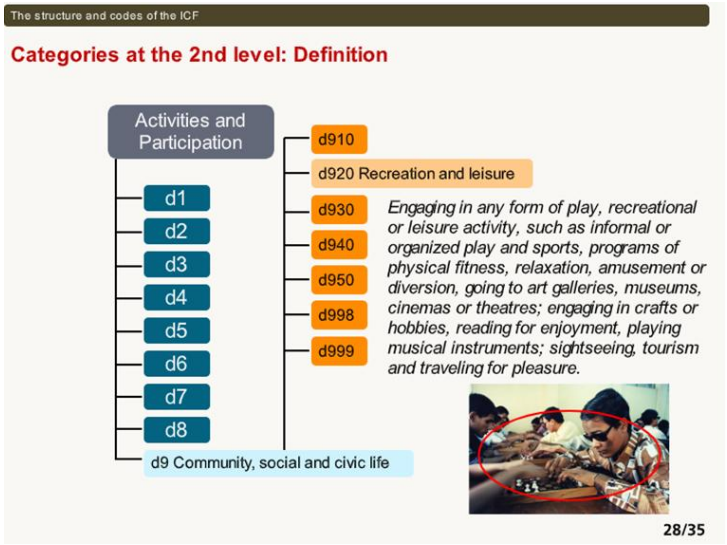
Results
The table shows ICF ≥6 quotations from ≥ 3 respondents; the most frequent are on top. Not only activities and participation categories were used frequently, but also functions and environmental factors

Activities and participation	Functions
d920 Recreation & leisure	b152 Emotional functions
d240 Handling stress and other psychological demands	b126 Temperament and personality functions
d850 Remunerative employment	b455 Exercise tolerance functions
d570 Looking after one's health	b125 Dispositions and intra-personal functions
d475 Driving	
d310 Understand spoken messages	**Environmental factors**
d720 Complex interpersonal interactions	e310 Immediate family
d610 Acquiring a place to live	e330 Peoples in positions of authority
d750 Informal social relationships	e355 Health professionals
d640 Doing housework	e360 Other professionals
d710 Basic interpersonal interactions	e120 Transportation
d230 Carrying out daily routine	
d210 Undertaking a single task	

Conclusion
Understanding the positive / neutral and negative aspects of living with a rare condition may help parents and communities to support growing up towards a fulfilled life.
Incorporating the **ICF in personal digital health records promotes health and well-being at all ages (Sustainable Development Goal #3, United Nations**)

ICF d 920.0

Recreation and leisure



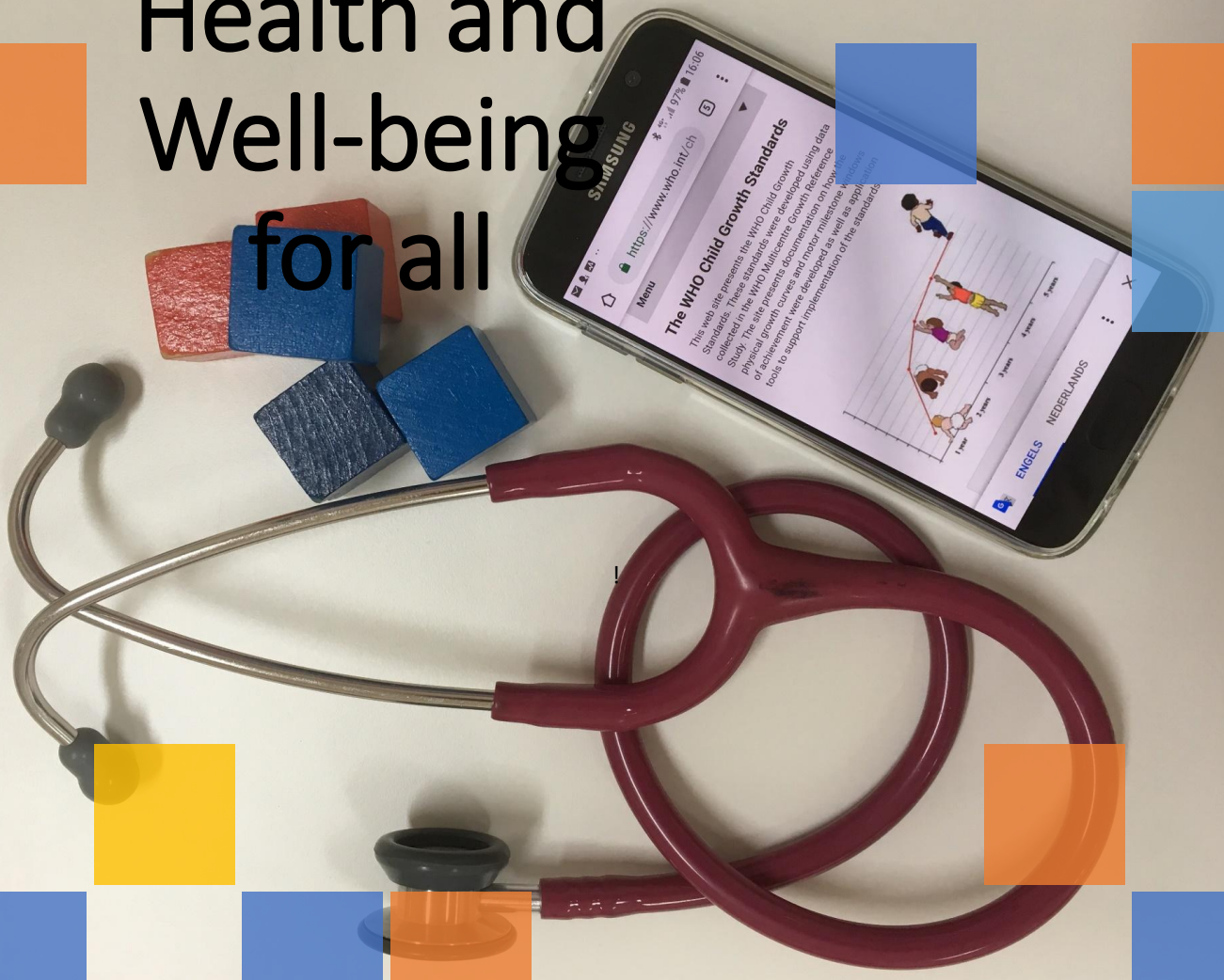
Indian Mother and Childcare
Kolkata, 2020

Rare diseases: ethical challenges in the era of digital health

Ethical challenge

- Patient-reported outcomes and lived experiences are crucial for developing accurate, person-centred digital health frameworks. Digital data on social participation are vital to ensure person-centred health services, particularly for poorly understood conditions.
- Individuals with rare chronic disabling conditions provide valuable insights based on their perception of reality as the effect of biology and experience varies in time and place. They should be actively involved in all decisions about their care and data exchange, either as groups or individuals.
- Research on digital literacy and training programs for people with a rare condition is a precondition.
- To drive innovation, it is essential to incorporate the human perspectives on how digitalisation can **enhance health and wellbeing**.

Health and Well-being for all



Thank

- European Pediatric Rare Disease Network

John Dodge, U.K.
Lali Margvelashvili, Georgia
Velibor Tasic, N- Macedonia
David Neubauer, Slovenia
Arunas Valiulis, Lithuania
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- Consensus in Pediatrics and Child Health

Manual Katz, Israel

- Forum Rare Diseases, Sri Lankan Pediatric Society

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