Brussels, 6 December 2019

Accelerating Progress for Safe Patient-Centred Universal Health Coverage

Patient Solidarity Day

6 December 2019| #PSD2019 | www.patientsolidarityday.org

EAP
Rare Diseases

Sustainable Development Goals, Universal Health Coverage and Rare Diseases

Liesbeth Siderius, pediatrician

Introduction

Where you live in the world dramatically affects your access to essential health services
#HealthForAll
World Health Organization

- **Sustainable Development Goals, United Nations 2015**

Rare is common, in pediatrics

- **Universal Health Coverage, United Nations, 2019**

Pediatric Support

- **Disabled child**

Coordinated care

- **Digital Health for all**

Interoperability

Rare disease 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
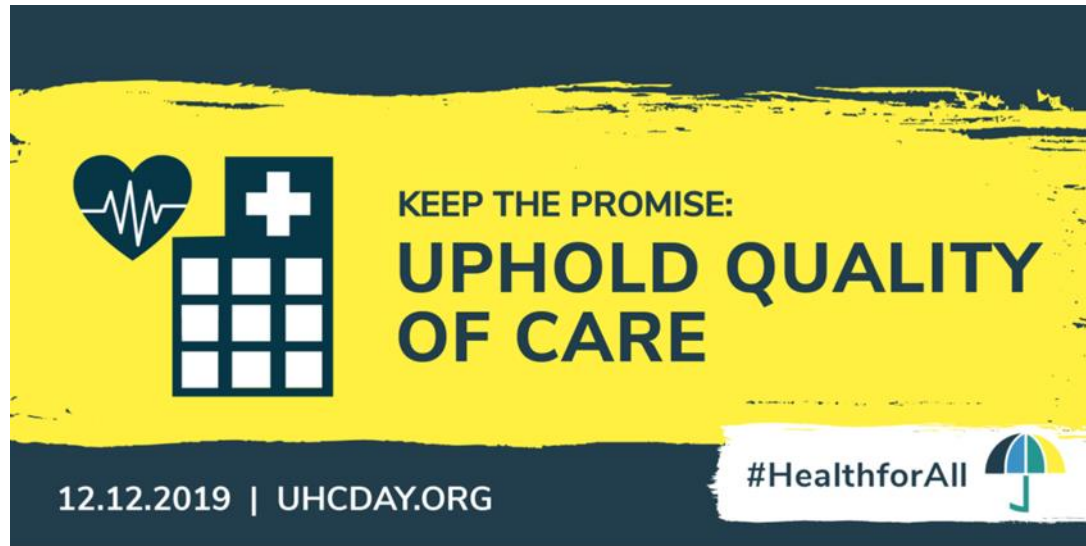- achieve **universal health coverage**

KEEP THE PROMISE:
LEAVE NO ONE BEHIND

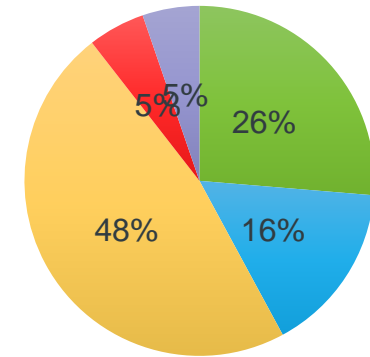#HealthforAll

12.12.2019 | UHCDAY.ORG

Share Knowledge

- Persons living with a rare disease remain a **marginalised and invisible** with **little information** available about their diseases

- They suffer **inequality** in accessing health care services and treatment, and in the prices they have to pay, due to their **social status** or their country of origin.

KEEP THE PROMISE:
UPHOLD QUALITY OF CARE

12.12.2019 | UHCDAY.ORG

#HealthforAll

Universal Health Coverage, leave no child behind

Pediatricians working in:

- ■ Primary care
- ■ Secundary care
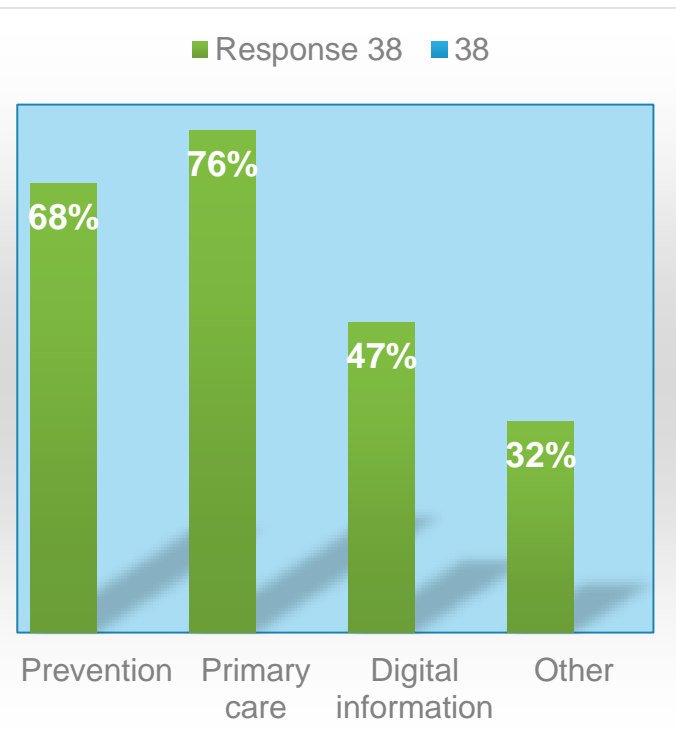- ■ Tertiary care
- ■ Trainee
- ■ Other

European Pediatric Network Rare Diseases

Questionnaire October- November 2019

Responce 38 ; 24 countries

Measures to reduce child mortality and increase access to quality health-care services
(UHC nr 29)

What would be necessary?

Prevention
Improve efficiency of preventive child healthcare schemes

Primary care
Promotion of pediatric primary care

Other
;

Digital information
Improved integration of digital information

Response 38 ◼ 38

- Prevention — 68%
- Primary care — 76%
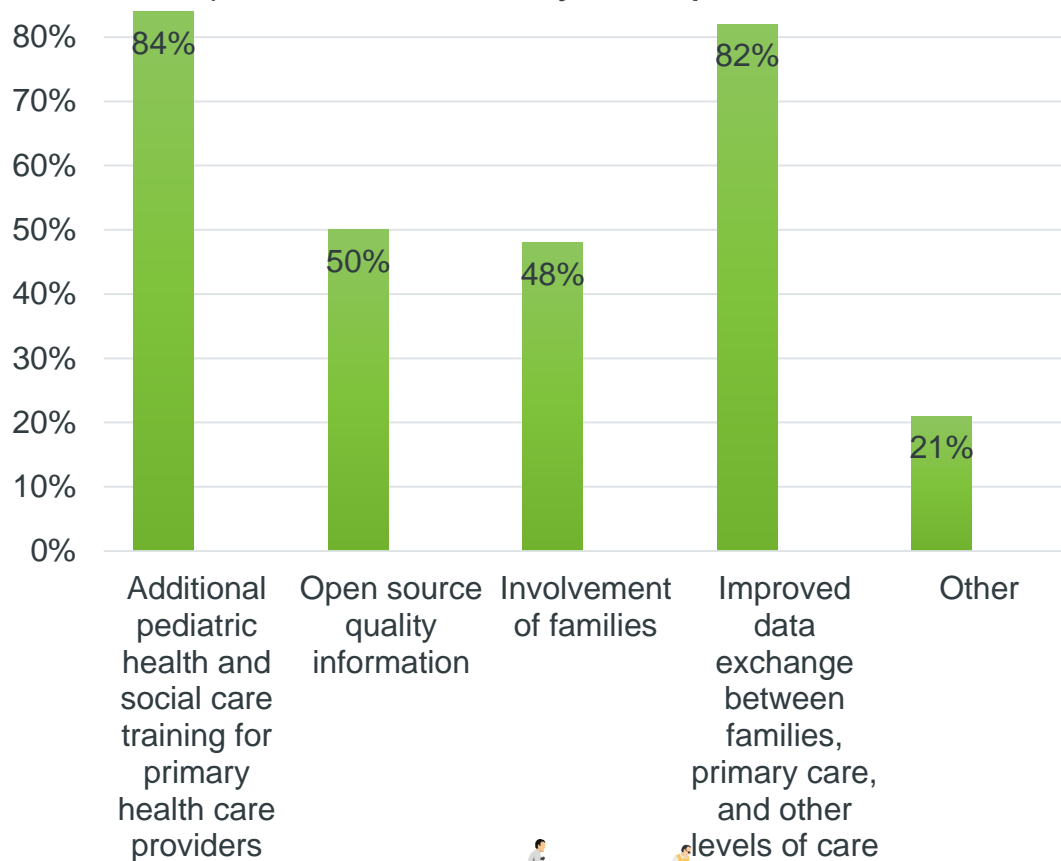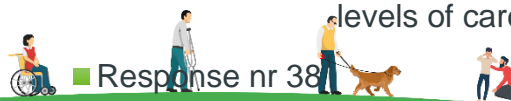- Digital information — 47%
- Other — 32%

Recognize that **primary health** care brings people info first contact with the health system to enhance people's physical and mental health as well as social well-being (~~UHC~~ nr 13)

Expand the delivery of and prioritize primary health care as a cornerstone of **sustainable people-centred**, community-based and integrated health systems, (~~UHC~~ nr 46) What would to your opinion be necessary:
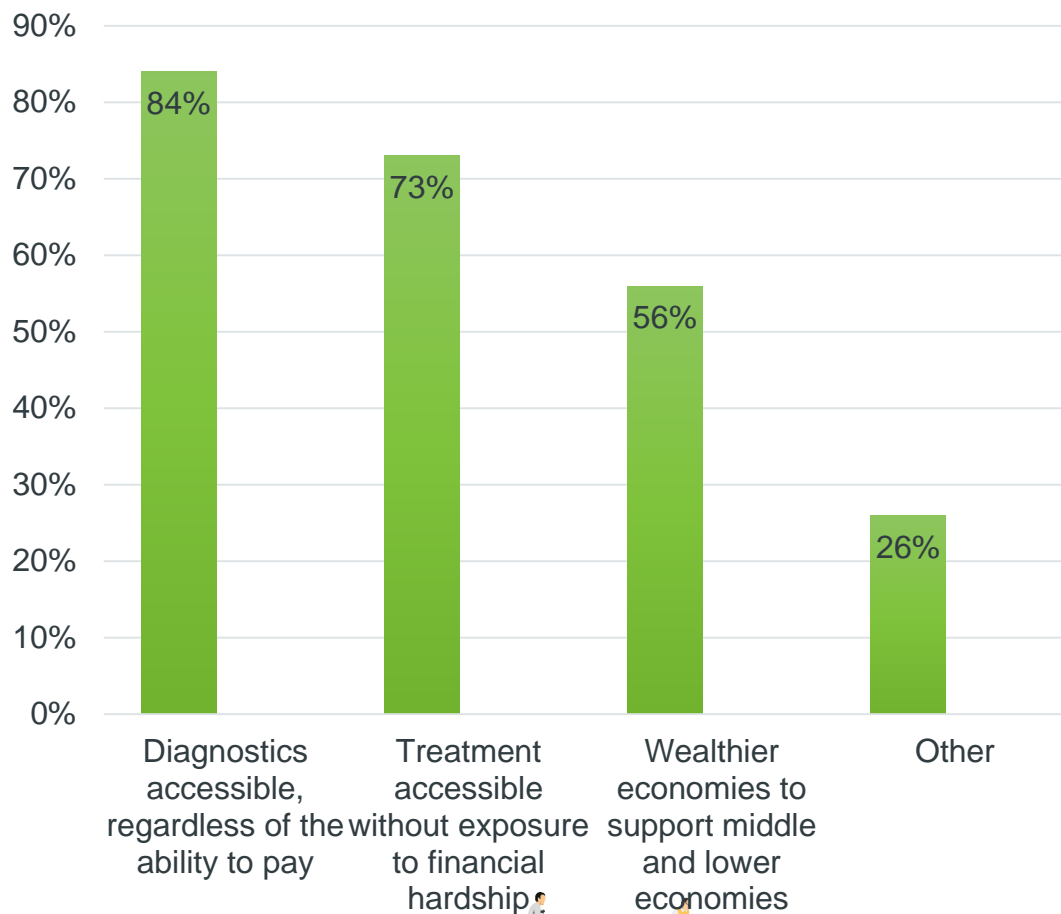

Chart with vertical axis from 0% to 80%:
- Additional pediatric health and social care training for primary health care providers: 84%
- Open source quality information: 50%
- Involvement of families: 48%
- Improved data exchange between families, primary care, and other levels of care: 82%
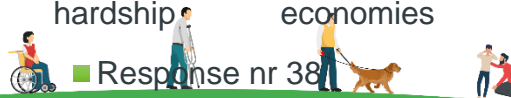- Other: 21%

Response nr 38

Strengthen efforts to address… **rare diseases** …as part of Universal Health Coverage (UHC nr 34)

What global action would be necessary?

Increase **access to health** services to all persons with disabilities and scale-up efforts for their empowerment and inclusion (UHC nr 37)

What would be necessary:

- Pediatricians should take an active role in **providing quality information** on the cause of disabilities   86% (30)

- Disabilities may be caused by an undiagnosed rare condition   66% (23)

- Disabled children have high quality care in my country/ state   43% (15)

- Families with a disabled child become isolated from society   43% (15)

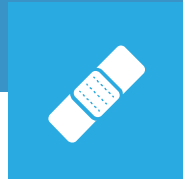- Families are empowered and do not need (pediatric) support   3%   (1)

At least half of the world's people do not have access to essential health services.

UHC ensures that

NO ONE IS LEFT BEHIND.

12.12.18
UHC

Accessible Diagnostics

Quality Information

Training Primary Care

Data exchange

World Health Organization

WHO recommendations on
home-based records
for maternal, newborn and child health*

Web annex A. Evidence base (GRADE and CERQual profiles)

*The full guideline document is available at:
http://apps.who.int/iris/bitstream/handle/10665/274327/9789241550352-eng.pdf

World Health Organization

Stages of Growth (Development Milestones)

It is important to follow your child's growth. There are a few signs that can help you follow the growth and development of your child from birth to 5 years.

Look out for these signs
A child might have a problem in these areas when the child shows any of the following behavious/signs.

Hearing - if the child:
- Does not turn towards the source of new sounds or voices
- Has frequent ear infection, (discharge from ear, earache)
- Does not response when you call unless he/she can see you
- Does not talk or talks strangely.

Seeing - if the child:
- Has red or discharging eyes
- Has a cloudy appearance of the eyes
- Frequently rubs eyes and say they hurt
- Often bumps into thins while moving around
- Hold head in an awkward position when trying to look at something
- Has eyes which sometimes or always look in different directions (squints)
- Has a white spot in the eye.

Ghana

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

Diagnosis
Hurler syndrome
PKU, Duchenne MD, FOP
Shwachman Diamond Syndrome

HPO
LOINC
ICPC

ICD - 10
Orphacode
OMIM

SNOMED -CT
DCOM

Sign primary care
Heelstick screening
Hearing screening
Growth; Development

Guideline
Collaborative Health Care
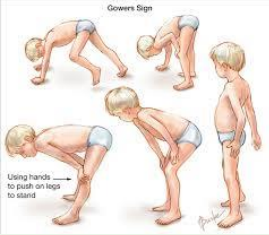
Interoperable data model

ISO 3166-1

ATC

New Therapeutics

ICF

HL7/ FHIR

ISO9999

Registry
Data collection with systematically organised computer processable collection medial terms

Guideline
Social services and rehabilitation

UNIVERSAL HEALTH COVERAGE DAY

©SDSS Holland

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 characterist[...] 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


```
LocalCode^LocalName^CodeSystem   LOINCcode^LOINCname^CodeSystem
```

```
OBX|2|NM|123^WBC^HSP_A^26464-8^Leukocytes [#/volume] in Blood^LN||10.8|K/MM3|||||F|
OBX|3|NM|234^RBC^HSP_A^26453-1^Erythrocytes [#/volume] in Blood^LN||4.82|MIL/MM3|||||F|
OBX|4|NM|345^HGB^HSP_A^718-7^Hemoglobin [Mass/volume] in Blood^LN||15.7|GM/DL|||||F|
OBX|5|NM|456^HCT^HSP_A^20570-8^Hematocrit [Volume Fraction] of Blood^LN||45|%|||||||F|
```

Notice how the result value and units have their own places in the message

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
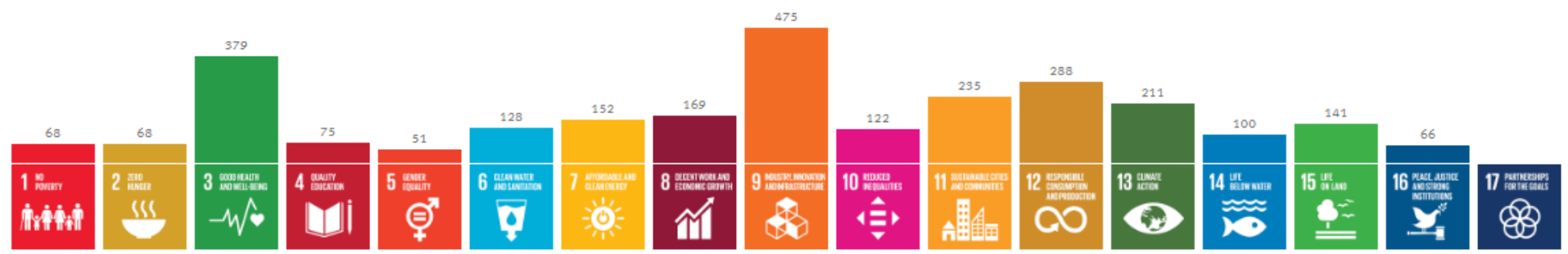
ATC

. A09AA02
Pancreatine

ISO= International Standards globally recognized guidelines and frameworks

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.

KEEP THE PROMISE: **MOVE TOGETHER**

12.12.2019 | UHCDAY.ORG

#HealthforAll

Doing nothing is not an option
Support the UHC is the least paediatricians can do

Thank You For Your Attention