Universal Health Coverage: leave no child behind

Rare Diseases in frame of Universal Health Coverage

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

Universal Health Coverage

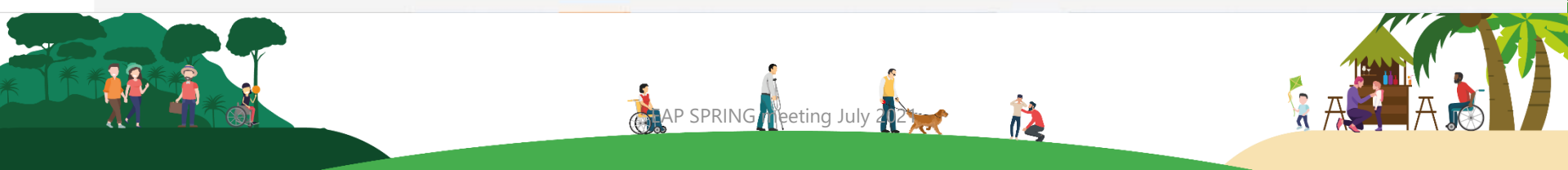
23/09/2019

UNITED NATIONS

MOVING TOGETHER
TO BUILD A HEALTHIER WORLD

UN High-Level Meeting on Universal Health Coverage, 23 September 2019, New York

On 23 September 2019, the United Nations General Assembly held a high-level meeting on universal health coverage. This meeting, held under the theme "**Universal Health Coverage: Moving Together to Build a Healthier World,**" aimed to accelerate progress toward universal health coverage (UHC), including financial risk protection, access to quality essential health-care services and access to safe, effective, quality and affordable essential medicines and vaccines for all.



UNITED NATIONS

UNIVERSAL HEALTH COVERAGE DAY

Political Declaration
of the High-level Meeting on Universal Health Coverage
"Universal health coverage: moving together to build a healthier world"

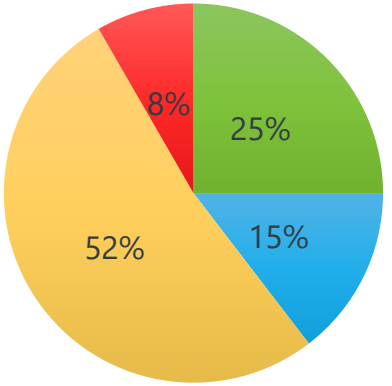
We, Heads of State and Government and representatives of States and Governments, assembled at the United Nations on 23 September 2019, with a dedicated focus for the first time on universal health coverage, reaffirm that health is a precondition for and an outcome and indicator of the social, economic and environmental dimensions of sustainable development and the implementation of the 2030 Agenda for Sustainable Development, and strongly recommit to achieve universal health coverage by 2030, with a view to scaling up the global effort to build a healthier world for all, and in this regard we:

1. Reaffirm the right of every human being, without distinction of any kind, to the enjoyment of the highest attainable standard of physical and mental health;

2. Reaffirm General Assembly Resolution 70/1 of September 2015, entitled "Transforming our world: the 2030 Agenda for Sustainable Development", stressing the need for a comprehensive and people-centered approach, with a view to leaving no one behind, reaching the furthest behind first, and the importance of health across all the goals and targets of the 2030 Agenda for Sustainable

Universal Health Coverage, leave no child behind

Respondents working in:

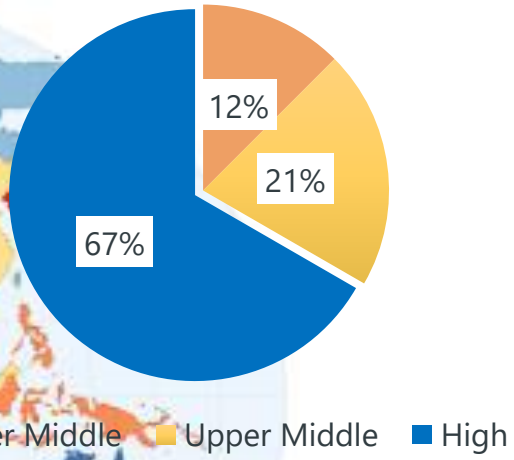


European Pediatric Network Rare Diseases Multiple Choice Questionnaire
October 2019- June 2020

Responce 48
29 countries

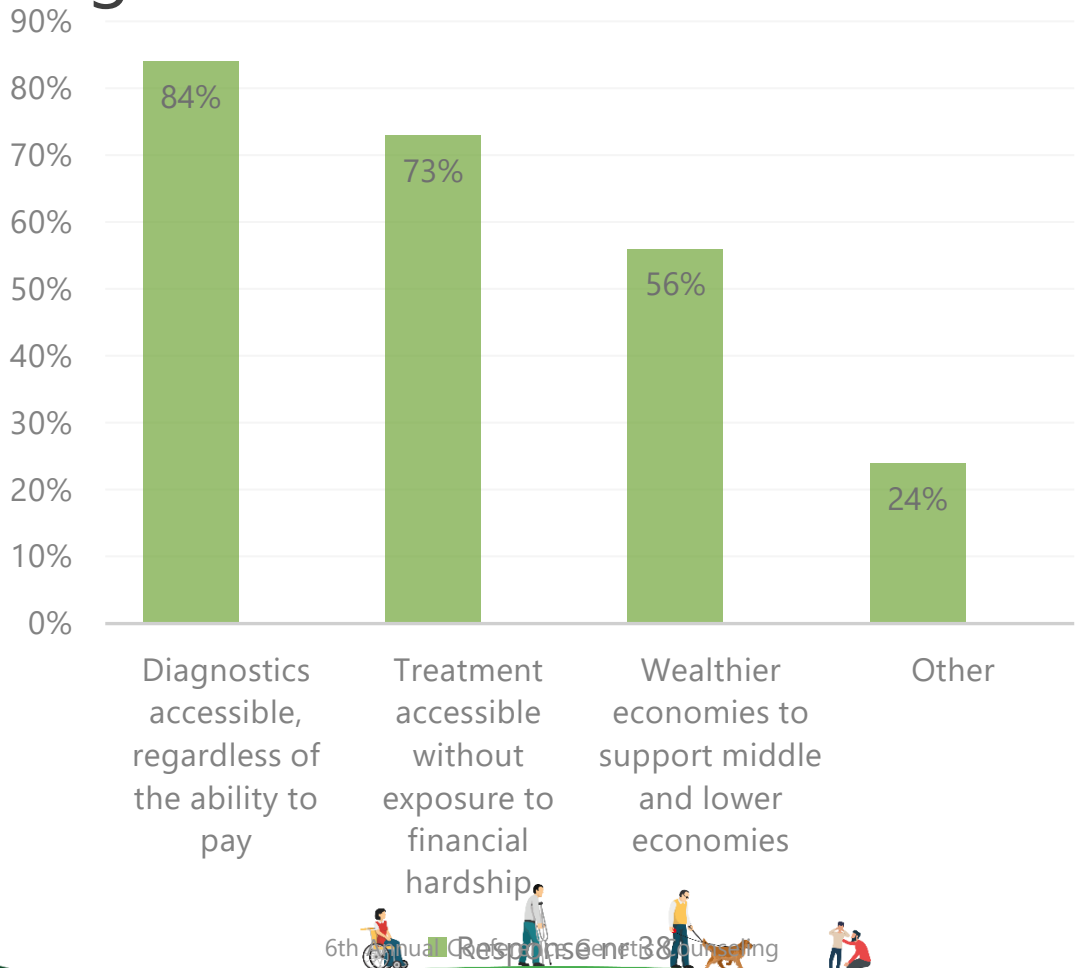
Respondents by Country Income

Respondents (48) working in:

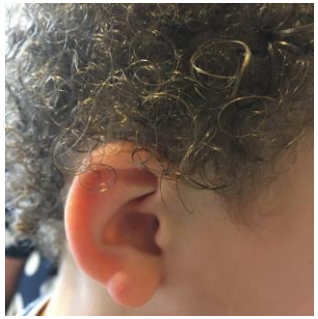


34. Also strengthen efforts to address eye health conditions and oral health, as well as rare diseases and neglected tropical diseases, as part of universal health coverage;

What global action would be necessary?



Recognize



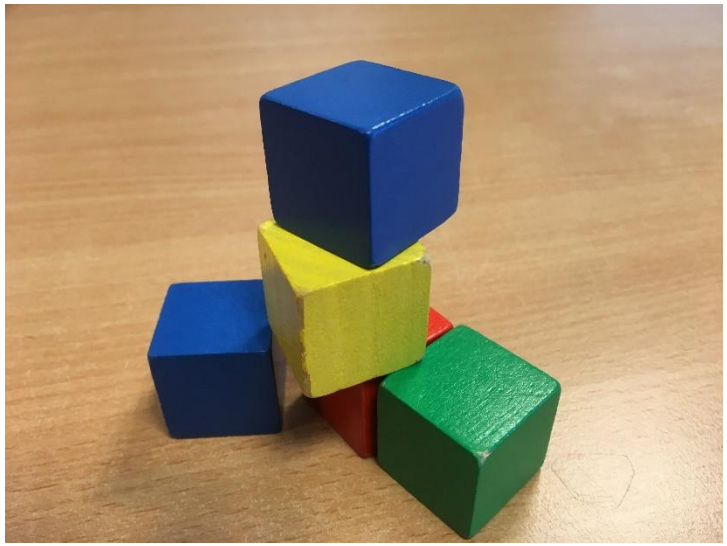
Children living with the diagnosis

- autism,
- developmental delay,
- prolonged neonatal icterus,
- abnormal features,
- failure to thrive

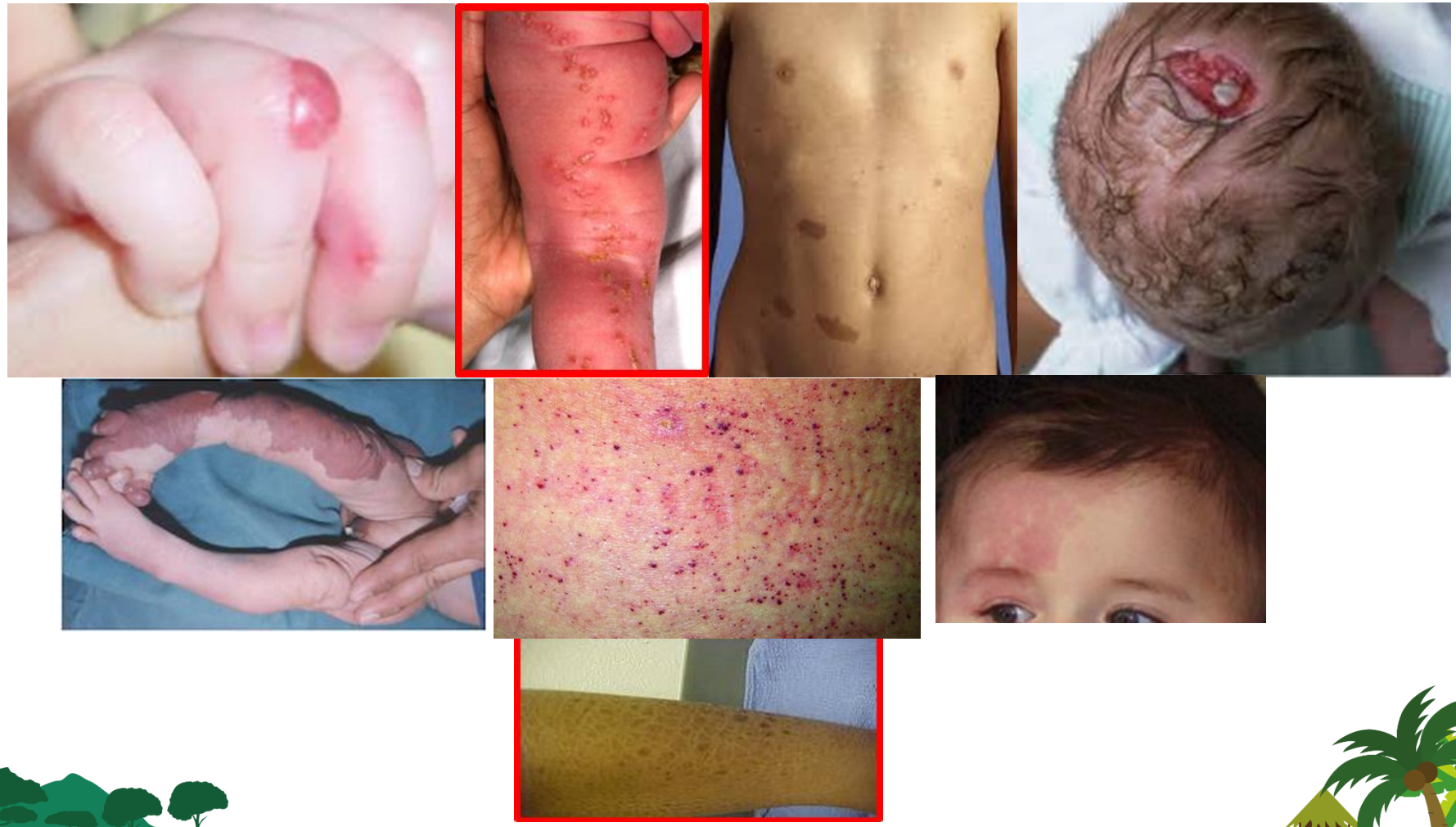
may very well have a

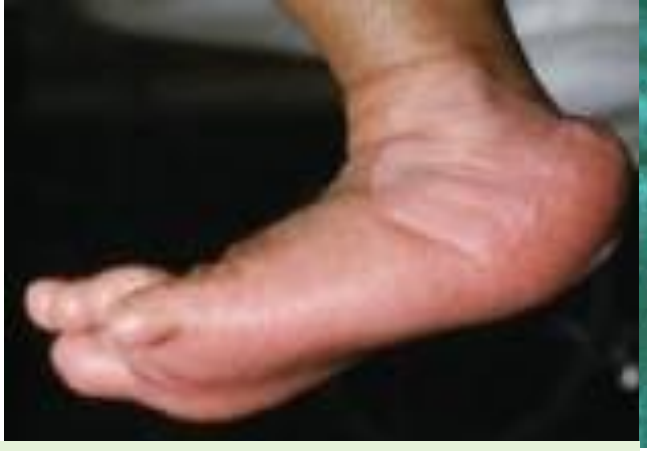
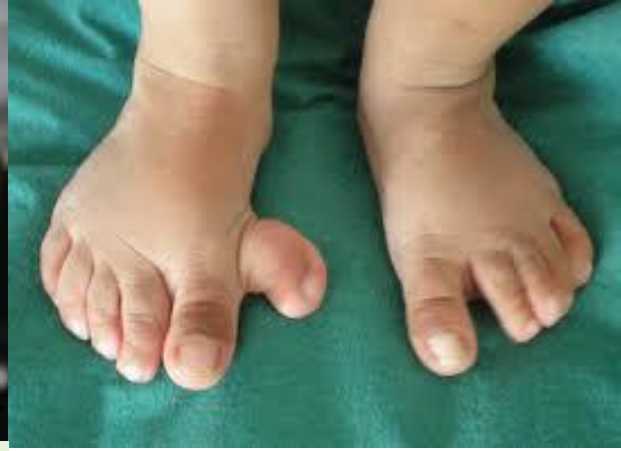
rare condition with

➢ specific health risks and treatment.

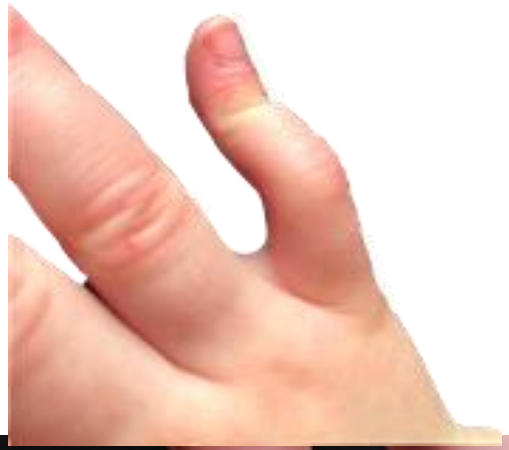


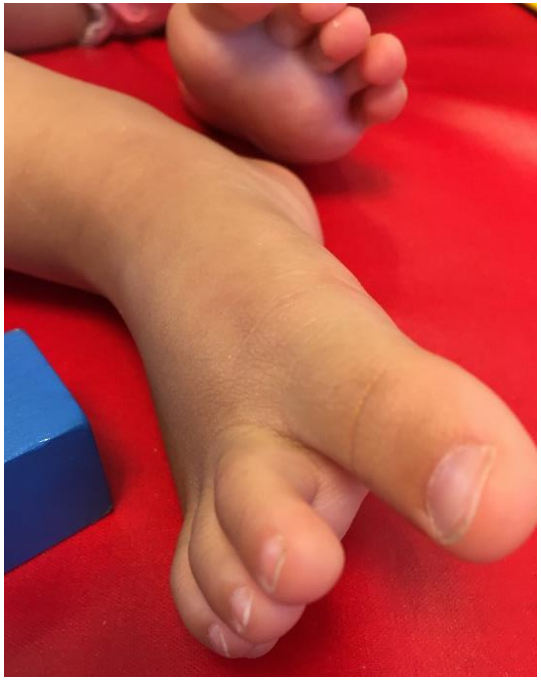
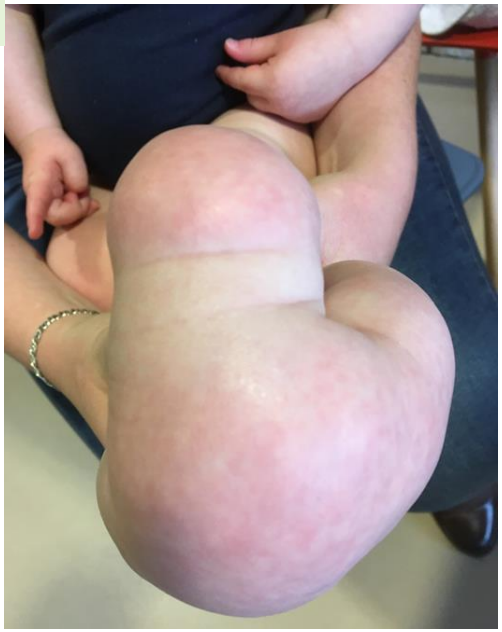
Global Child Health



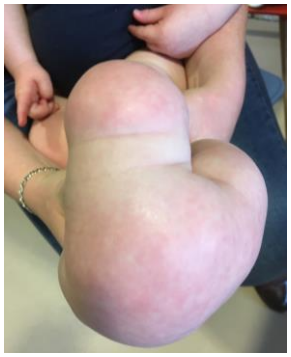
Access to Diagnostics



Girl, 11 month old
Diagnosis PIK3CA mutation

What is the clinical diagnosis?



PIK3CA is responsible for Proteus syndrome and PIK3CA-Related Overgrowth Spectrum, respectively

Mosaic Disorders of the PI3K/PTEN/AKT/TSC/mTORC1 signaling pathway

Neera Nathan, MD[1], **Kim M. Keppler-Noreuil, MD**[2], **Leslie G. Biesecker, MD**[2], **Joel Moss, MD, PhD**[3], and **Thomas N. Darling, MD, PhD**[1]

[1]Department of Dermatology, Uniformed Services University of the Health Sciences, Bethesda, MD, USA

[2]Medical Genomics and Metabolic Genetics Branch, National Human Genome Research, Institute, National Institutes of Health, Bethesda, MD, USA

[3]Cardiovascular and Pulmonary Branch, National Heart, Lung, and Blood Institute, National Institutes of Health, Bethesda, MD, USA

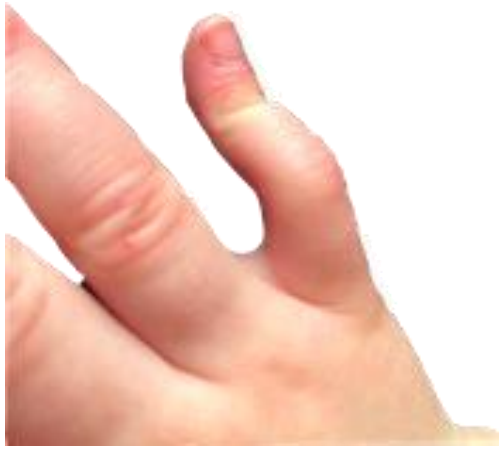
Abstract

Mosaicism is the presence of two or more genetically distinct cell lineages originating from a single zygote. The skin frequently marks mosaic conditions through migration patterns of a population of mutant cells during embryogenesis. Somatic mutations in genes of the PI3K/PTEN/AKT/TSC/mTORC1 signaling pathway can result in segmental overgrowth, hamartomas, and malignant tumors, given the crucial role of this axis in cell growth. Mosaicism for activating mutations in *AKT1* and *PIK3CA* is responsible for Proteus syndrome and PIK3CA-Related Overgrowth Spectrum, respectively. These frequently exemplify Happle's patterns of cutaneous

Accessible diagnostics

Chromosome study

12 months old girl with reduced growth in length and weight and minor dysmorphic features >
Array CGH > Materials: EDTA blood 2x 3-6 ml >
result term: 5 weeks
Resulted in diagnosing a **moziac duplicate chromosome 12q12-q13.2**



DNA panels

2 year old girl with a developmental delay >
Exome panel developmental delay (1338 genes) >
Materials: EDTA Blood, isolated DNA >
Result term regulair: 3 months / rapid: 15 workdays :
Resulted in diagnosing **a mutation in NAA15**

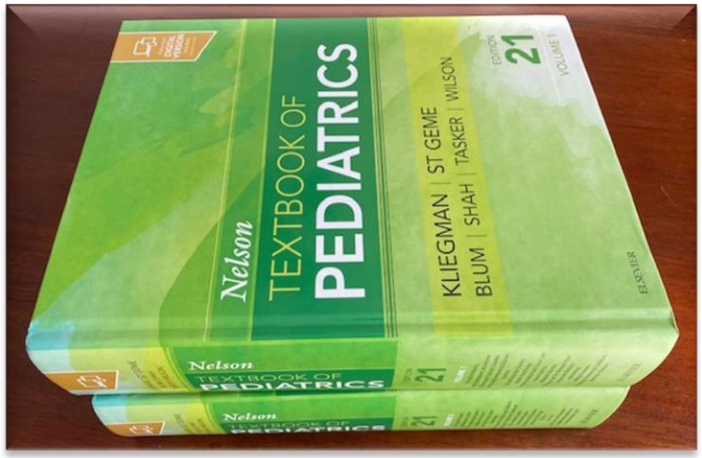
Information

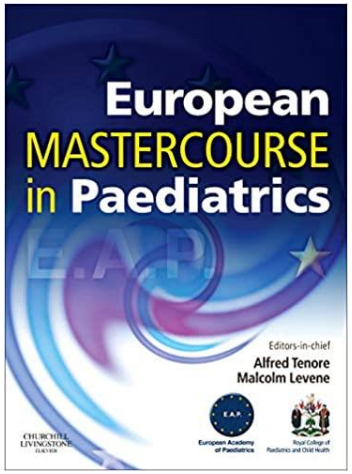
Truncating Variants in *NAA15* Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies

Hanyin Cheng,[1,49] Avinash V. Dharmadhikari,[1,49] Sylvia Varland,[2,45] Ning Ma,[3,4,5] I
Robert Kleyner,[7] Alan F. Rope,[8] Margaret Yoon,[7] Asbjørg Stray-Pedersen,[9,10] Jennifer
Sarah R. Crews,[11] Mohammad K. Eldomery,[9] Zeynep Coban Akdemir,[9] Andrea M. I
Vernon R. Sutton,[9] Jill A. Rosenfeld,[9] Erin Conboy,[13] Katherine Agre,[13] Fan Xia,[1,9]
Magdalena Walkiewicz,[1,9,14] Mauro Longoni,[15,16] Frances A. High,[15,17,18] Marjon A.
Grazia M.S. Mancini,[19] Candice R. Finnila,[20] Arie van Haeringen,[21] Nicolette den H
Claudia Ruivenkamp,[21] Sakkubai Naidu,[22] Sonal Mahida,[22] Elizabeth E. Palmer,[23,24]

Table 1. Summary of Phenotypes

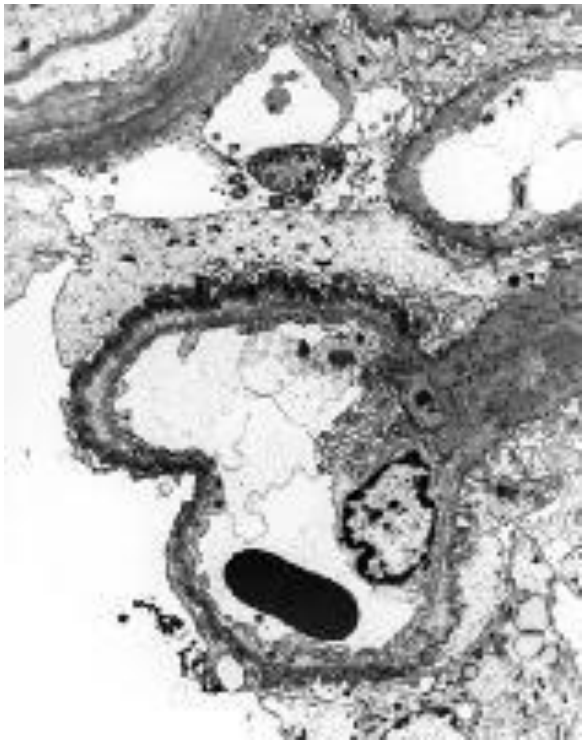
Phenotype	Number of individuals with phenotype	Number of individuals with relevant data	Percentage
Brain Structure and Function			
Intellectual disability (ID)[a]	23	23	100
ASD, ADHD, or behavioral issues	30	33	91
Abnormal brain MRI	2	11	18
Speech delay	32	33	97
Seizures	6	26	23
Motor Impairments			
Motor delay and related abnormalities	31	32	97
Muscle tone issues	7	18	39
Feeding difficulties	8	14	57
Cardiovascular			
Congenital cardiac defects	4	19	21
Major vessel anomalies	2	19	11
Arrhythmias	1	19	5
Hypertension	1	19	5
Other			
Mild dysmorphism	18	28	64
Skeletal or connective-tissue defects	8	20	40





Diagnostic criteria for Alport syndrome

Velibor Tagic (North Macedonia)



*Family history for haematuria, with progression to End Stage Kidney Disease especially in males

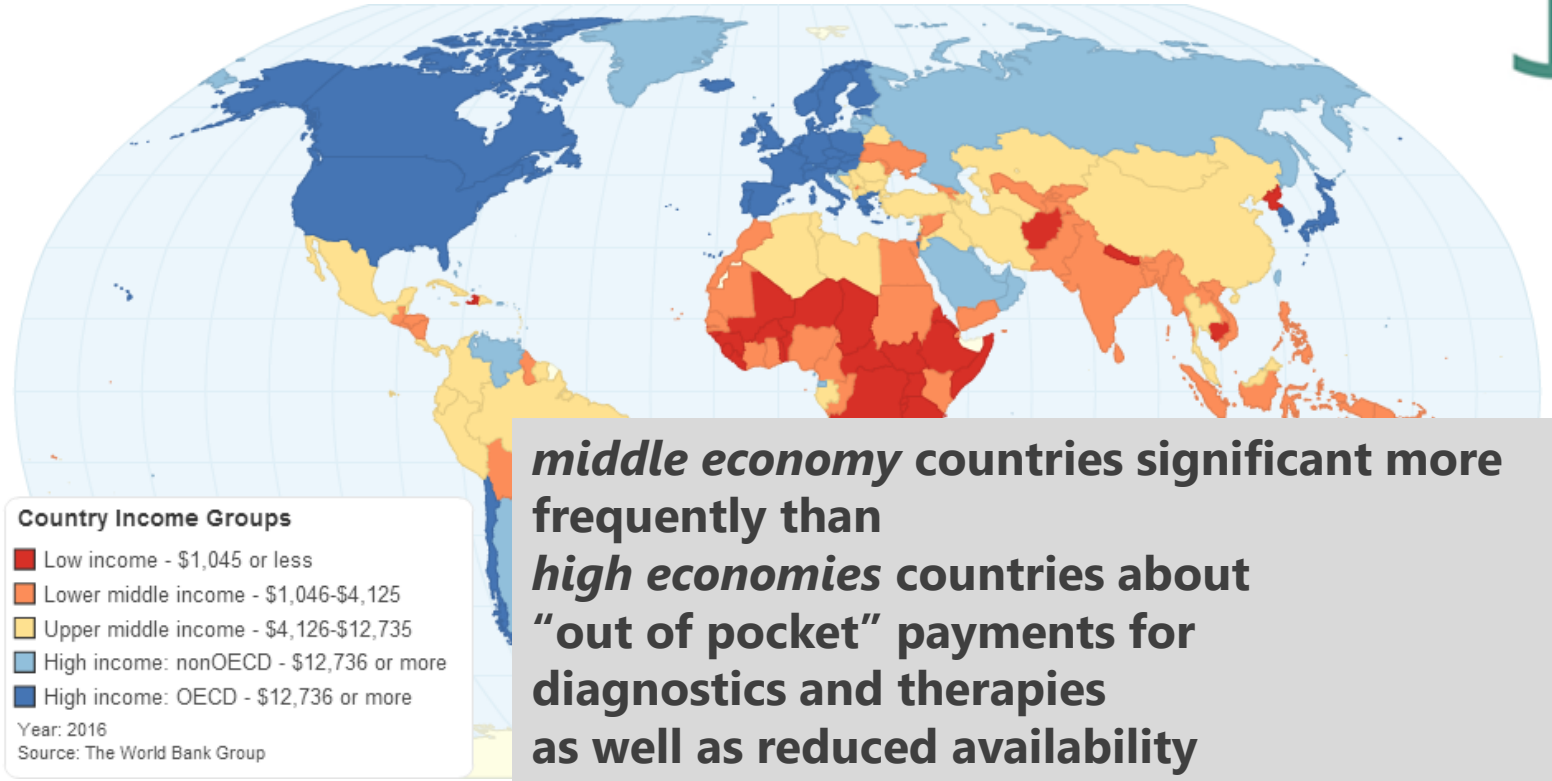
*Thickening of the glomerular basal membrane on Electronic Microscope

*Progressive, sensorineural deafness

*Anterior lenticonus and peri-macular flecks

50. Improve availability, affordability and efficiency of health products by increasing transparency of prices of medicines, vaccines, medical devices, diagnostics, assistive products, cell- and gene-based therapies, and other health technologies across the value chain, including through improved regulations and building constructive engagement and a stronger partnership with relevant stakeholders, including industries, private sector and civil society, in accordance with national and

UNIVERSAL HEALTH COVERAGE DAY



middle economy **countries significant more frequently than** ***high economies*** **countries about "out of pocket" payments for diagnostics and therapies as well as reduced availability**

Spinal Muscular Atrophy

Kyiv Post

Tech Opinion Lifestyle Video Ukraine Digest Investigations Tip us off More Employment

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Ukrainian family in battle to raise $2.3 million to save their son

By Daria Shulzhenko. Published April 8. Updated April 8 at 6:59 pm

ELSEVIER

Available online at www.sciencedirect.com

ScienceDirect

Neuromuscular Disorders 28 (2018) 103–115

www.elsevier.com/locate/nmd

Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care

Eugenio Mercuri [a,b,1,*], Richard S. Finkel [c,1], Francesco Muntoni [d], Brunhilde Wirth [e],

incidence around 1 in 10,000 live births have been estimated with SMA type I

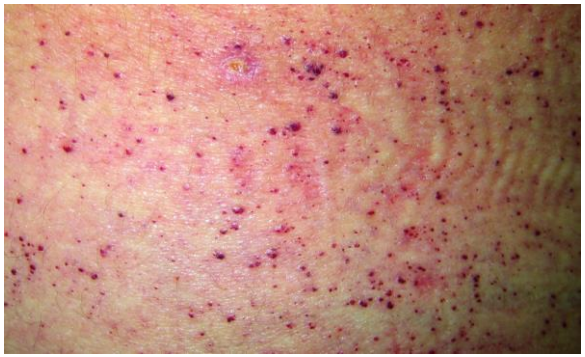
12 mg/5 mL
Spinraza intrathecal solution

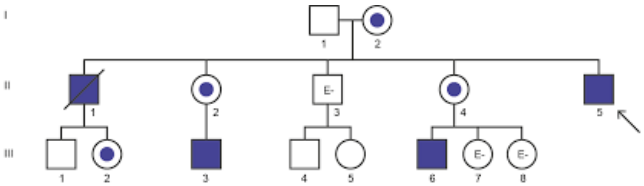
from $133,119.50
for 5 milliliters

Quantity	Per unit	Price
5 milliliters	$26,623.90	$133,119.50

Important: When there is a range of pricing, consumers should normally expect to pay the lower price. However, due to stock shortages and other unknown variables we cannot provide any guarantee.

Fabry Disease





Neuropathic Pain (hand and feet)
Abdominal Pain
Dark red spots
Hearing loss tinnitus
Heart problems
Kidney disease
Strokes

ELSEVIER journal homepage: www.elsevier.com/locate/ymgme

Commentary

Fabry disease during the COVID-19 pandemic. Why and how treatment should be continued

Juan Politei*

Fundation for the Study of Neurometabolic Diseases, FESEN, Argentina

37. Increase access to health services for all persons with disabilities, remove physical, attitudinal, social, structural, and financial barriers, provide quality standard of care and scale up efforts for their empowerment and inclusion, noting that persons with disabilities, representing 15% of the global population, continue to experience unmet health needs;

1) Paediatricians providing quality information regarding **the cause of disabilities** 83%

2) Disabilities may be caused by an undiagnosed rare condition 71%

3) Families with a disabled child become **isolated from society** 50%

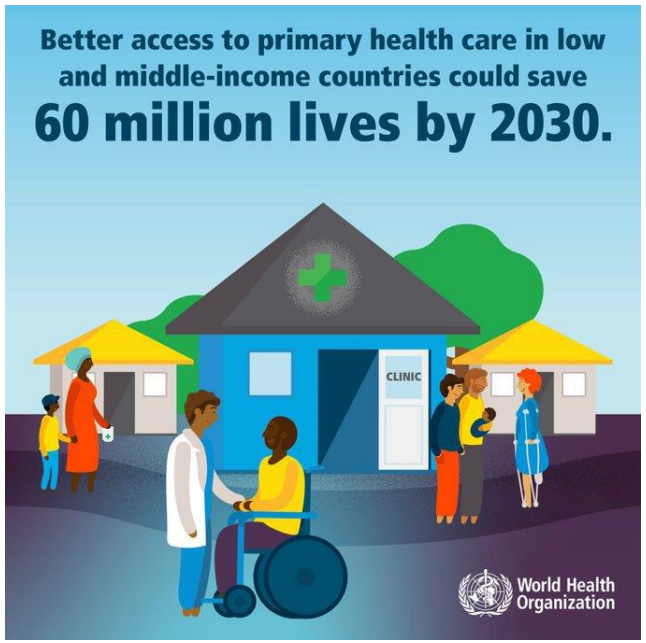
4) Disabled children have high quality care in my country/ state **35%**

5) Families are empowered and do not need (paediatric) support **2%**

Sri Lanka

29. Take measures to reduce maternal, neonatal, infant and child mortality and morbidity and increase access to quality health-care services for newborns, infants, children, as well as all women before, during and after pregnancy and childbirth;

Measures to reduce child mortality and increase access to quality health-care services; what would be necessary?



Primary care 81 %

Promotion of pediatric primary care

;

Prevention 75 %

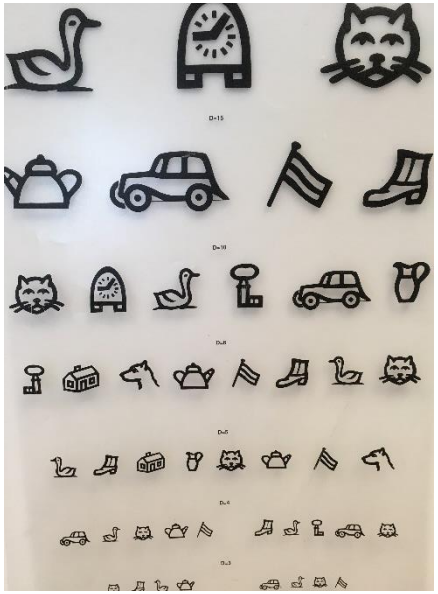
Improve efficiency of preventive child healthcare schemes

Digital information 50%

Improved integration of digital information

Primary Health Care

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;



Universal Health Coverage directive #13

- *"first contact with the health system*
- *most inclusive, effective and efficient approach*
- *enhance people's physical and mental health, as well as social well-being",*
- cornerstone of a sustainable health system for universal health coverage

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;

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;

Deliver **primary care as a cornerstone** by
44 respondents working in paediatric practice:
primary, secondary or tertiary care:

N=44	Primary (12)	Secondary (7)	Tertiary (25)
Training	10 (83%)	5 (71%)	21 (84%)
Information	5 (42%)	6 (86%)	10 (40%)
Family	5 (42%)	4 (57%)	14 (56%)
Data exchange	8 (67%)	6 (86%)	20 (80%)

EAP's 'Core Training' syllabus

GENETICS AND DYSMORPHOLOGY

EBP EXAM
EUROPEAN BOARD OF PAEDIATRICS EXAMINATION

Knowledge base

- Principles and molecular basis of Mendelian- and non-Mendelian inheritance
- Embryological basis of malformation and environmental factors in fetal development
- Principles of dysmorphology and syndrome identification
- Basis of genetic and molecular techniques
- Ethical and social implications of genetic testing
- Indications and limitations of prenatal diagnosis
- Rationale of newborn screening

Skills

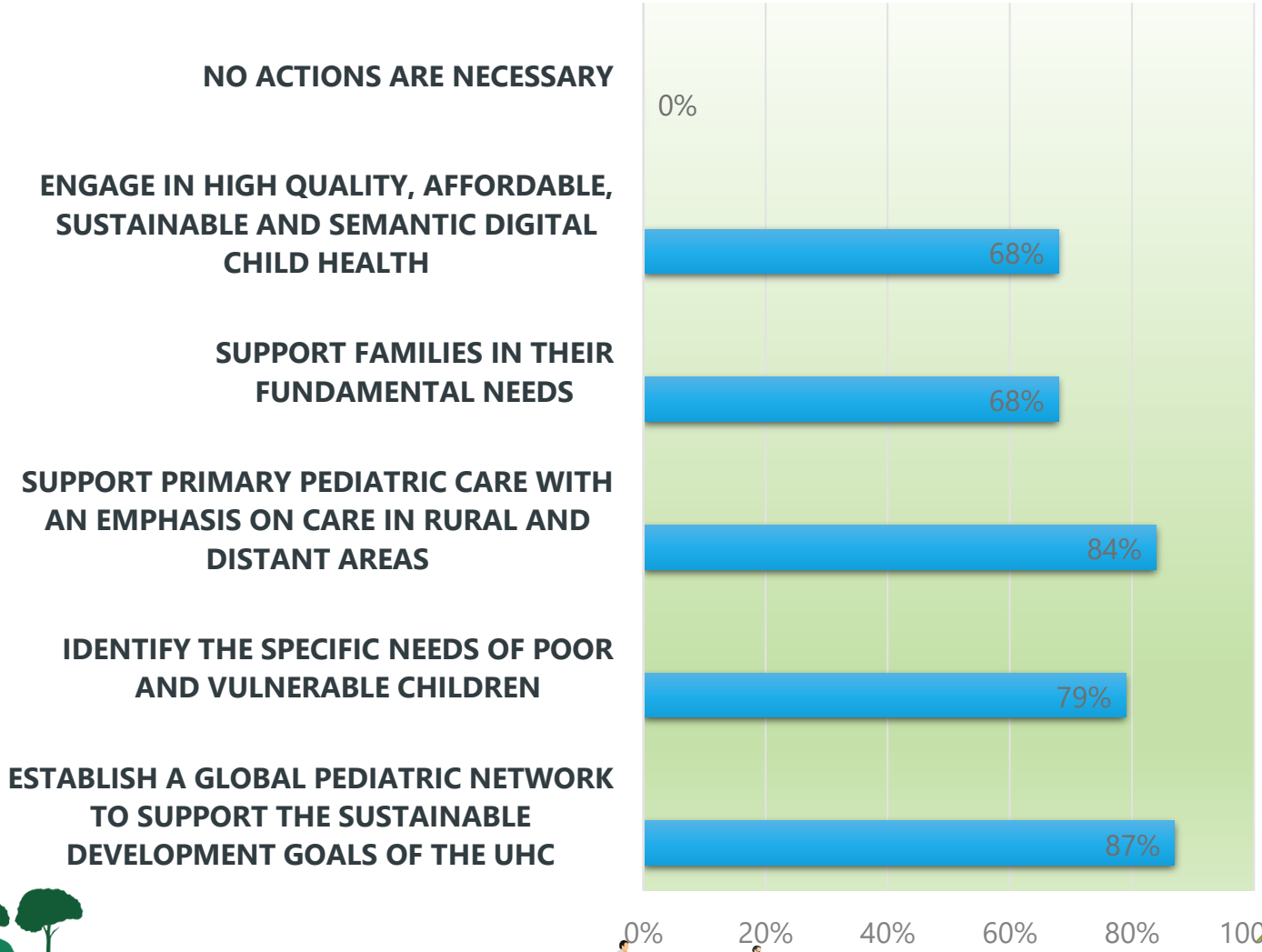
- Construction and interpretation of a family pedigree
- Recognition of common genetic, chromosomal and dysmorphic syndromes
- Genetic counselling related to common conditions
- Ability to access genetic databases

Training in pediatric health and social care

- As most rare disease present in childhood;
- Paediatricians, at the **hospital as well as in primary care in the community**, to recognize and take are of children with rare and disabling conditions;
- The **EAP's 'Core Training' syllabus** resonates with **Universal Health Coverage**, the rights of the child and the rights of the disabled;
- The first European Board of Paediatrics Exam took place in 2020, and two further examinations are planned in 2021.

EBPEXAM
EUROPEAN BOARD OF PAEDIATRICS EXAMINATION

Increase **global awareness**, international solidarity, international collaboration and action towards the achievement of the universal health coverage (UHC nr78)



Conclusion

Due to the lack of affordability and availability of diagnostic facilities and therapies in **middle - let alone lower - economy countries**, chronically ill and disabled children are deprived of receiving accurate diagnosis or proper therapy.

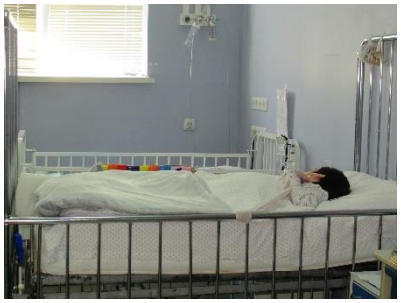
Unless deliberate efforts are made to reach these children, they **face preventable morbidity and death** without having a chance to profit from advances in medical and digital science.

The **implementation of a sustainable digital child health** system, in line with the **WHO recommendations on digital health** as well as the UHC, requires support of policy makers.

Further research should include identification of more specific actions: **"to support primary pediatric care with an emphasis on care in rural and distant areas and support the specific needs of chronically ill and disabled children".**

Leave no one behind

‘What would be the least **YOU** could do to contribute to the UHC?’



The Netherlands

Day Centre
Coffin Lowrey Syndrome

India

NIEPID
To be diagnosed

Georgia

Abandoned
Undiagnosed

Peru

Living with family
In rural village
‘ Birth trauma’

Thank

- European Pediatric Rare Disease Network

John Dodge, U.K.
Lali Margvelashvili, Georgia
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, India

- People with a rare condition and their families