

# Rare Diseases: From Gene to Society

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Primary care Paediatrician

Coordinator Rare Diseases Working Group

European Academy of Paediatrics

Shwachman Diamond Syndrome Patient representative

Sri Lanka, Kandy 21-09-2018

The Netherlands



# Diagnosis First



Cornelia de Lange Syndrome 1: 10.000 - 1: 30.000  
Severe developmental delay with characteristic features



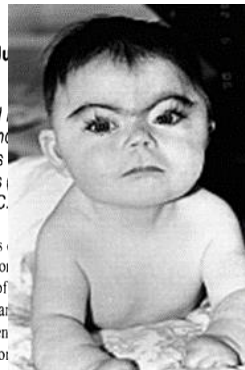
1933  
Clinical fetuare  
Cornelia de Lange

1981  
Chromosome  
Abnormality  
??

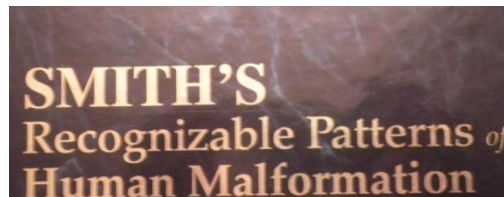
## Prometaphase Chromosomes in Five Patients With the Brachmann-de Lange Syndrome

Elizabeth J. Breslau, Christine Disteche, J. and Pat Cooper

Departments of Pathology, Pediatrics and  
ton and Children's Orthopedic Hospital and  
Washington (E.J.B., C.D., J.G.H.); Genetics  
Department of Social and Health Services  
ics Program, Walla Walla, Washington (P.C.



We analyzed the prometaphase chromosomes (of sibs) with the Brachmann-de Lange syndrome. Significant chromosome abnormality in any of tinct entities can be distinguished on clinical and the dup(3q) syndrome. We still recommend tients with BDLS and BDLS-like manifestation

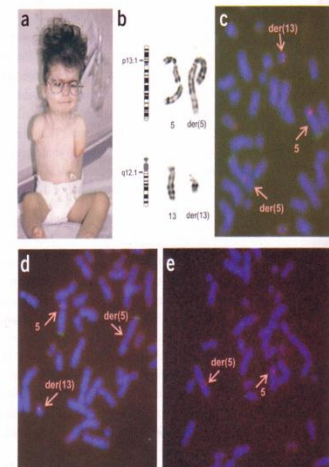


*NIPBL*, encoding a homolog of fungal Scc2-type sister chromatid cohesion proteins and fly Nipped-B, is mutated in Cornelia de Lange syndrome

2003 DNA mutation

Cornelia de Lange syndrome (CdLS) is a multiple malformation disorder characterized by dysmorphic facial features, mental retardation, growth delay and limb reduction defects<sup>1,2</sup>. We identified and characterized a new gene, *NIPBL*, that is mutated in individuals with CdLS and determined its structure and the structures of mouse, rat and zebrafish homologs. We named its protein product delangin. Vertebrate delangins have substantial homology to orthologs in flies, worms, plants and fungi, including Scc2-type sister chromatid cohesion proteins, and *D. melanogaster* Nipped-B. We propose that perturbed delangin function may inappropriately activate *DLX* genes, thereby contributing to the proximodistal limb patterning defects in CdLS. Genome analyses typically identify individual delangin or Nipped-B-like orthologs in diploid animal and plant genomes. The evolution of an ancestral sister chromatid cohesion protein to acquire an additional role in developmental gene regulation suggests that there are parallels between CdLS and Roberts syndrome.

The multisystem nature of the CdLS phenotype suggests that it is caused by a microdeletion or microduplication affecting several genes or by a single gene that regulates various target genes. A high-density BAC microarray comparative genome hybridization screen found no evidence for a consistent pattern of microdeletion or microduplication<sup>3</sup>. Because CdLS is rare and most cases are sporadic, genome-wide linkage screens are problematic. As an alternative, we analyzed chromosomal breakpoints associated with CdLS, focusing first on three classical cases with *de novo* balanced translocations, including the previously described translocations t(3;17)(q26.3;q23.1)<sup>4</sup> and t(14;21)(q32;q11)<sup>5</sup>. We first analyzed the 3q26.3 breakpoint because of



**Figure 1** FISH mapping of a 5p13 translocation breakpoint in an individual with classical CdLS. (a) Individual with classical CdLS with characteristic limb and facial abnormalities (including an upturned triangular nose, long philtrum, thin upper lip, downturned corners of the mouth; see fuller description for individual P46 in Table 1). (b) Giemsa chromosome banding showing a balanced *de novo* t(5;13)(p13.1;q12.1) translocation. (c-e) Metaphase chromosome FISH with the breakpoint-spanning BAC clone CTD-2653m23 (c) and overlapping fosmid clones G248P84262B4 (d) and G248P8840C10 (e), all labeled with Spectrum Red. Labeled in green is a chromosome 5q telomere-specific probe. Arrows indicate the normal chromosome 5 and the der(5) t(5;13)(p13.1;q12.1) and der(13) t(5;13)(p13.1;q12.1) chromosomes. In occasional metaphases a weak G248P84262B4 signal can be detected on the der(5) chromosome as well as a strong signal on the der(13). The combined data suggest that the most likely location for the breakpoint is close to the proximal end of the region of overlap for inserts of G248P84262B4 and G248P8840C10 (Fig. 2a).

# Abandoned



2014 - 2018

Georgia has become one of the first ex-Soviet republics to abolish state orphanages in favour of foster care.

But disabled children continue to be marginalised and face the prospect of life-long isolation from society.

[www.bbc.com/news/world-europe-25575094](http://www.bbc.com/news/world-europe-25575094)



## Sickle cell disease

1.1% of couples worldwide are at risk for having children with a haemoglobin disorder

WHO: Africa majority of children with the most severe form of the disease **die before the age of 5, usually from an infection or severe blood loss**



World Health Organization

# WHO, 7 December 2010

Fact sheet N°172

- A **collaborative management approach at primary health care** level with patients, their families and other health care actors is a must to effectively prevent many major contributors to the burden of disease.

## Essential elements for action

- **Support a paradigm shift towards integrated, preventive health care**
- **Promote financing systems and policies that support prevention in health care**
- **Equip patients with needed information, motivation, and skills in prevention and self-management**
- **Make prevention an element of every health care interaction**

Sri Lanka, Kandy 21-09-2018

# Rare Diseases Global Action

EURORDIS, Vienna, May 2018



European Conference on Rare Diseases & Orphan Products

- RD as a Collective Health and Social issue
- RD Universal Health Coverage
- Access to Diagnostics
- Access to Medicines / fair pricing
- Inclusion in the Non- Communicable Disease agenda
- RD in the Sustainable Development Goals



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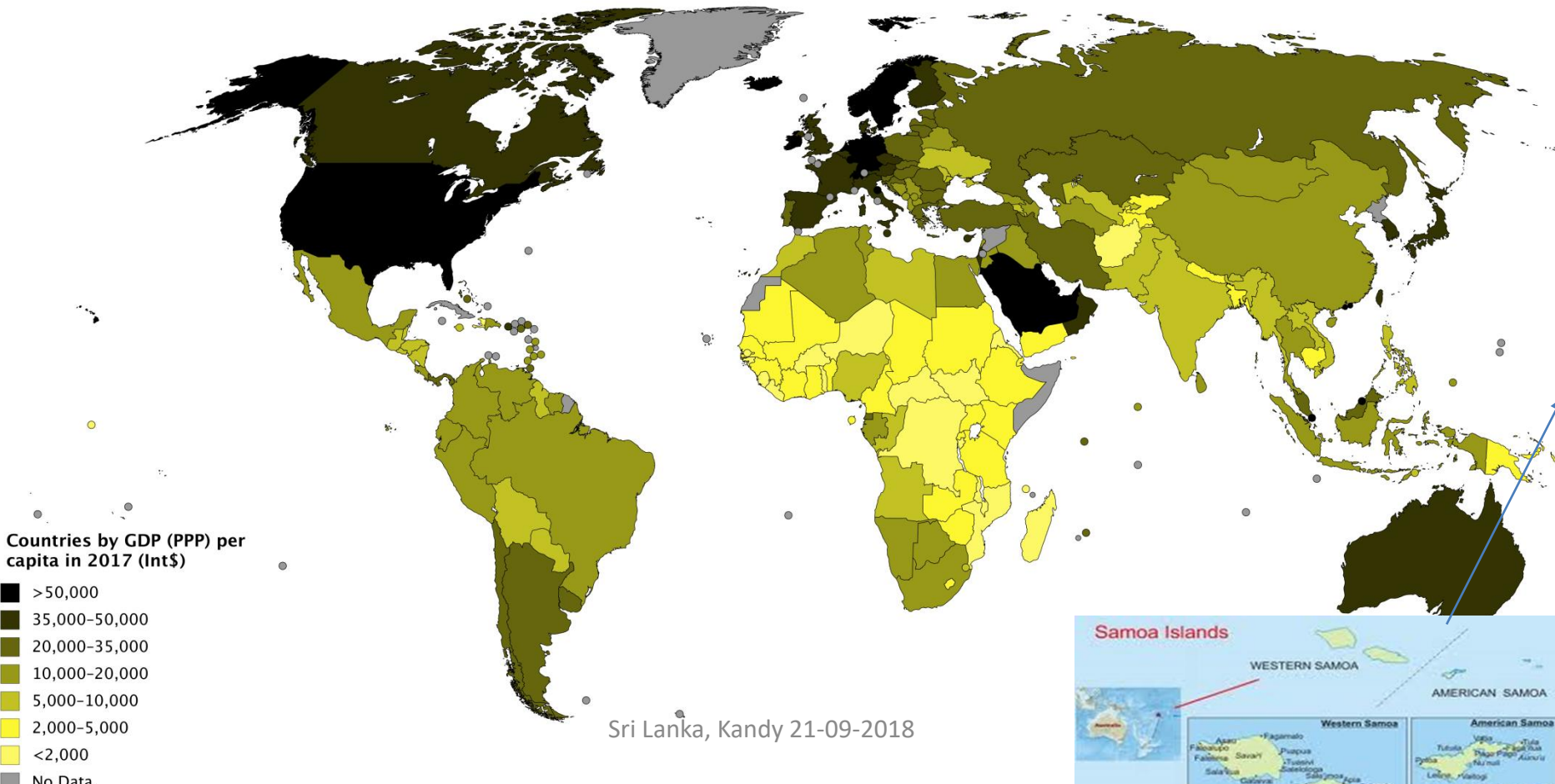




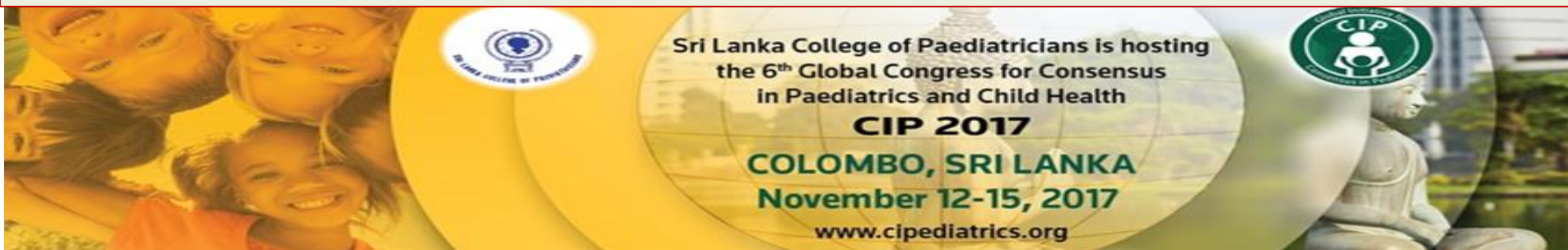
Bruck Syndrome 1: 1.000.000 world wide; bone fractures, limited movement, high incidence Samoa Islands

# Gross Domestic Product per Capita in Purchasing Power Parity per capita

estimated by IMF for year 2017



# Brugada syndrome 5-50 : 10.000; higher in south east Asia, recognizable through ECG abnormality



PP53:

**Revesz Syndrome: Rare Variant of Dyskeratosis Congenita. A Case Report**  
Imalke Kankanaratchchi<sup>1</sup>, Nuwan Wickramasinghe<sup>2</sup>, Sujeewa Am  
Sadani Vithana<sup>4</sup>, Thilina Madushanka<sup>4</sup>

<sup>1</sup>Paediatrics, Faculty Of Medicine University of Ruhuna, Sri Lanka

<sup>2</sup>University Paediatric Unit, Teaching Hospital Karapitiya, Sri Lanka

PP54:

PP78:

**Oculo-Auricular- Vertebral (Oav) Spectrum Disorders: Experience From Sri  
WMDAS Wanninayake<sup>1</sup>, Romesh Gunasekera<sup>2</sup>, Saman Yasawarden  
Basnayake<sup>4</sup>, Malka Jayathilake<sup>2</sup>, DDC Nilini<sup>6</sup>, Deepthi de Silva<sup>7</sup>**

<sup>1</sup>Demonstrator, Department of Physiology, Faculty of Medicine, Un  
Kelaniya, Sri Lanka, <sup>4</sup>Consultant Plastic Surgeon, Lady Ridgway Hos  
Colombo, Sri Lanka

<sup>3</sup>Consultant ENT Surgeon, Lady Ridgway Hospital, Colombo, Sri Lanka

<sup>4</sup>Lady Ridgway Hospital, Colombo, Consultant Orthodontist, Sri Lanka

<sup>5</sup>Speech and Language Therapist, Lady Ridgway Hospital, Colombo, Sri Lanka

<sup>6</sup>Research Assistant, Department of Chemistry, Faculty of Science, University of Kelaniya, Sri Lanka

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<sup>32</sup>Research Assistant, Department of Chemistry, Faculty of Science, University of Kelaniya, Sri Lanka

PP81:

**Partial 1Q Trisomy Syndrome Due Maternal T(1;4) Balance Translocation; A Case Report**  
R.J.M.K.A Jayasundara<sup>1</sup>, M.D.C.J.P. Jayamanne<sup>1</sup>, G.B.A.M. Rathnasiri<sup>1</sup>, S.  
Mayoorathy<sup>1</sup>, Ranmali Rodrigo<sup>2</sup>

<sup>1</sup>Colombo North Teaching Hospital, Registrar in Paediatrics, Sri Lanka

<sup>2</sup>Faculty of Medicine, University of Kelaniya, Consultant Neonatologist, Sri Lanka

PP82:

**Pallister Killian Syndrome: Experience of Clinical Diagnosis and Molecular Confirmation in Sri Lankan Patients**

Nirman Ratnayake<sup>2</sup>, DDC Nilini<sup>1</sup>, Samantha Waidyanatha<sup>3</sup>, Kumudu

Weerasekera<sup>4</sup>, Nicole de Leeuw<sup>5</sup>, Deepthi de Silva<sup>6</sup>

<sup>1</sup>Department of Chemistry, Faculty of Science, University of Colombo, Research Assistant, Sri Lanka, <sup>2</sup>Department of Physiology, Faculty of Medicine, University of Kelaniya, Demonstrator, Sri Lanka, <sup>3</sup>Lady Ridgway Hospital, Consultant Paediatrician, Sri Lanka

<sup>4</sup>Department of Human Genetics, Radboud University Medical centre, Nijmegen, the Netherlands, Clinical Laboratory Geneticist, Netherlands

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<sup>28</sup>Department of Physiology, Faculty of Medicine, University of Kelaniya, Demonstrator, Sri Lanka

Teaching Hospital Karapitiya, Medical Officer, Sri Lanka

PP105:

**Oculocerebrorenal Syndrome of Lowe - A Case Report**

Sadani Vithana<sup>1</sup>, Imalke Kankanaratchchi<sup>1</sup>, Thilina Madusha

Wickramasinghe<sup>1</sup>, Sujeewa Amarasekera<sup>1</sup>, Harshini Dharmawansa

Rukshani

<sup>1</sup>professorial unit, Paediatrics, Teaching Hospital Karapitiya, Sri Lanka

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<sup>3</sup>paediatric Nephrology, Teaching Hospital Karapitiya, Sri Lanka

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<sup>39</sup>paediatric Nephrology, Teaching Hospital Karapitiya, Sri Lanka

180 Posters  
36 Rare Diseases  
24 from Sri Lanka

PP50:

**A Paediatric Case of Brugada Syndrome**

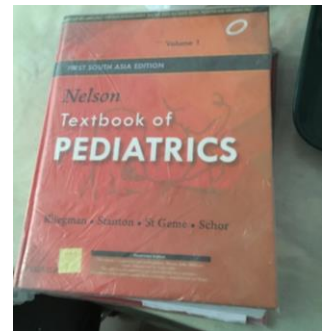
Navabalasooriyar Pratheep, Kankanaratchchi Imalke, Kitulwatte Nalin

Medical Intensive Care Unit, Lady Ridgeway Hospital for Children, Sri Lanka



# Rare Diseases Forum

- Well trained and **experienced paediatricians**
- Families have their own data and are **co-managing**
- Little access to diagnostic tests
- Little access to internet
- No facilities for electronic data collections



**With limited resources :  
accurate diagnosis !**



# Rare Diseases Paediatric Global Action

What can paediatricians do to improve health and wellbeing of children with a rare and disabling condition and their families?

## RECOGNIZE

- strengthen primary paediatric care with sufficient knowledge
- advocate for access to proper diagnostics and treatment
- collect and share data
- support families

Turner syndrome : 50 per 100,000 liveborn females;  
coarctation of the aorta and bicuspid aortic valve reduce life expectancy

**Primary Health Care** can meet 80-90% of an individual's needs over the course of their life  
<http://www.who.int/primary-health/en>



World Health Organization

**@WHO\_Europe 7 sep.**  
**#PrimaryHealthCare** has a strong focus on quality of care.

Quality health services, build trust in patients and encourage individuals to engage in their health and the health of their families. We cannot have Health For All without quality care.

# Rare Diseases Paediatric Global Action

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# Next Generation Sequencing



## Welcome to NGS

The massively parallel sequencing technology known as next-generation sequencing (NGS) has revolutionised the biological sciences. With its ultra-high throughput, scalability, and speed, NGS enables researchers to perform a wide variety of applications and study biological systems at a level never before possible.

<https://www.illumina.com/science/technology/next-generation-sequencing.html>

.



Fabry 1 : 40 - 60,000 males Fabrazyme X linked , pain skin pigmentations,  
Heart disease, stroke

## Orphan Drugs: European Academy of Paediatrics position statement on the Paediatric Regulations and Rare diseases December 2017

Rare diseases should not be overlooked in discussing how to improve impact of Paediatric Regulation

Developers of medicines for rare paediatric diseases need to comply with both the Orphan and Paediatric Regulations. Decisions of the Orphan and Paediatric Committees should be aligned.

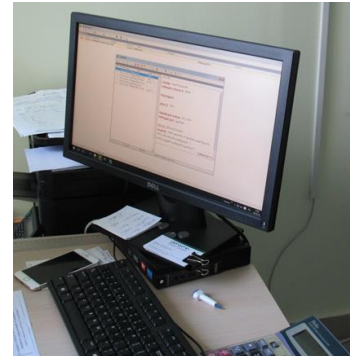
To improve the availability of high quality medicines for use in children, **transparency on development costs** and agreement on maximum price, is warranted.

The price should match a considerable and measurable clinical benefit.

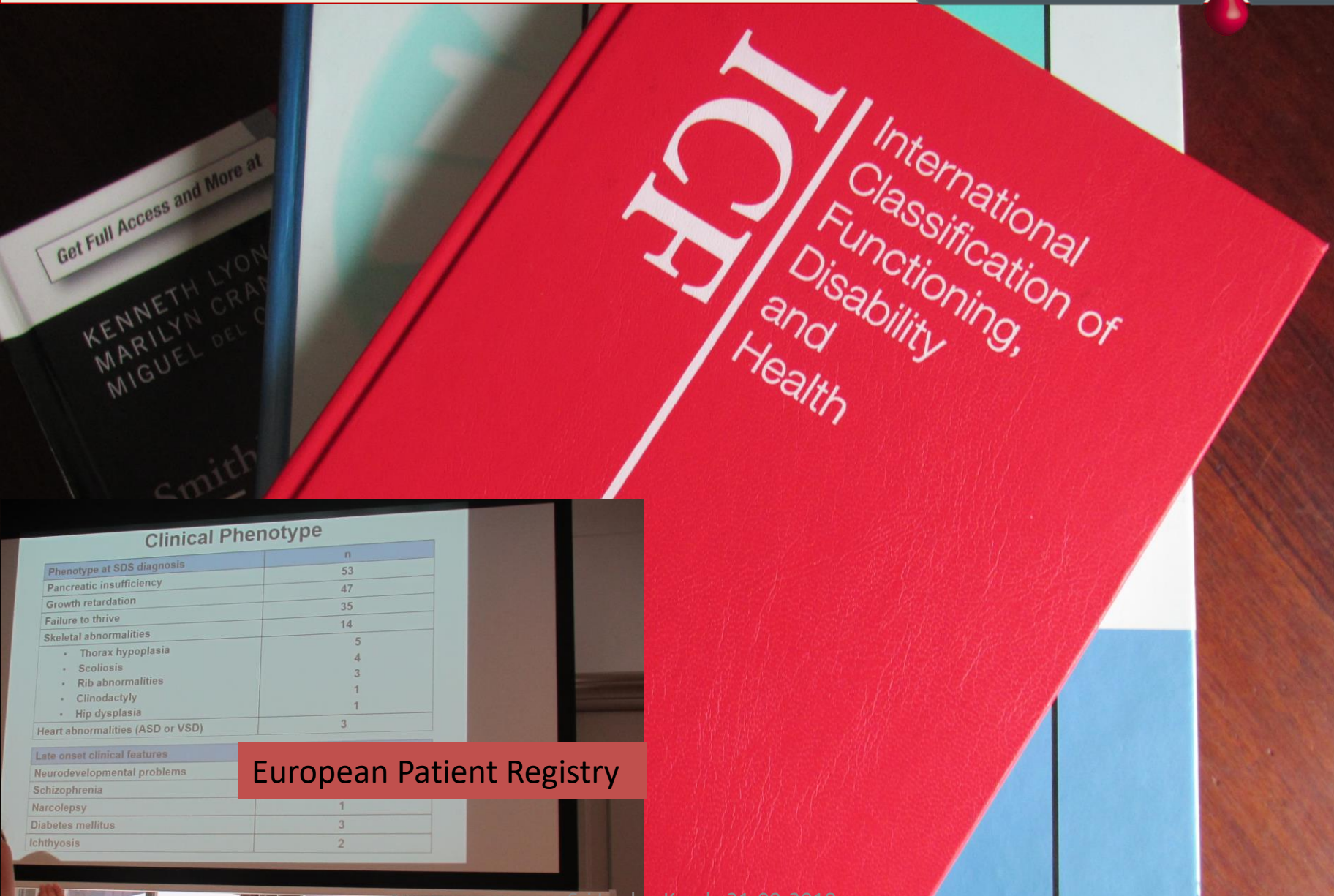
# Rare Diseases **Paediatric** Global Action

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- Strengthen primary pediatric care with sufficient knowledge
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- Support families







Clinical Phenotype

Phenotype at SDS diagnosis	n
Pancreatic insufficiency	53
Growth retardation	47
Failure to thrive	35
Skeletal abnormalities	14
• Thorax hypoplasia	5
• Scoliosis	4
• Rib abnormalities	3
• Clinodactyly	1
• Hip dysplasia	1
Heart abnormalities (ASD or VSD)	3
Late onset clinical features	
Neurodevelopmental problems	
Schizophrenia	
Narcolepsy	1
Diabetes mellitus	3
Ichthyosis	2

European Patient Registry

# Building the Rare Disease

knowledge and information ecosystem

Thalassemia, 3500 patients  
have been identified in Sri Lanka.

## RareCare.World

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 ( $\alpha 2 \delta 2$ ). Another way of identifying people with **thalassemia** major is neonatal screening. Neonatal screening ...

#### Rare Condition

[Thalassemia major or Beta Thalassemia](#)

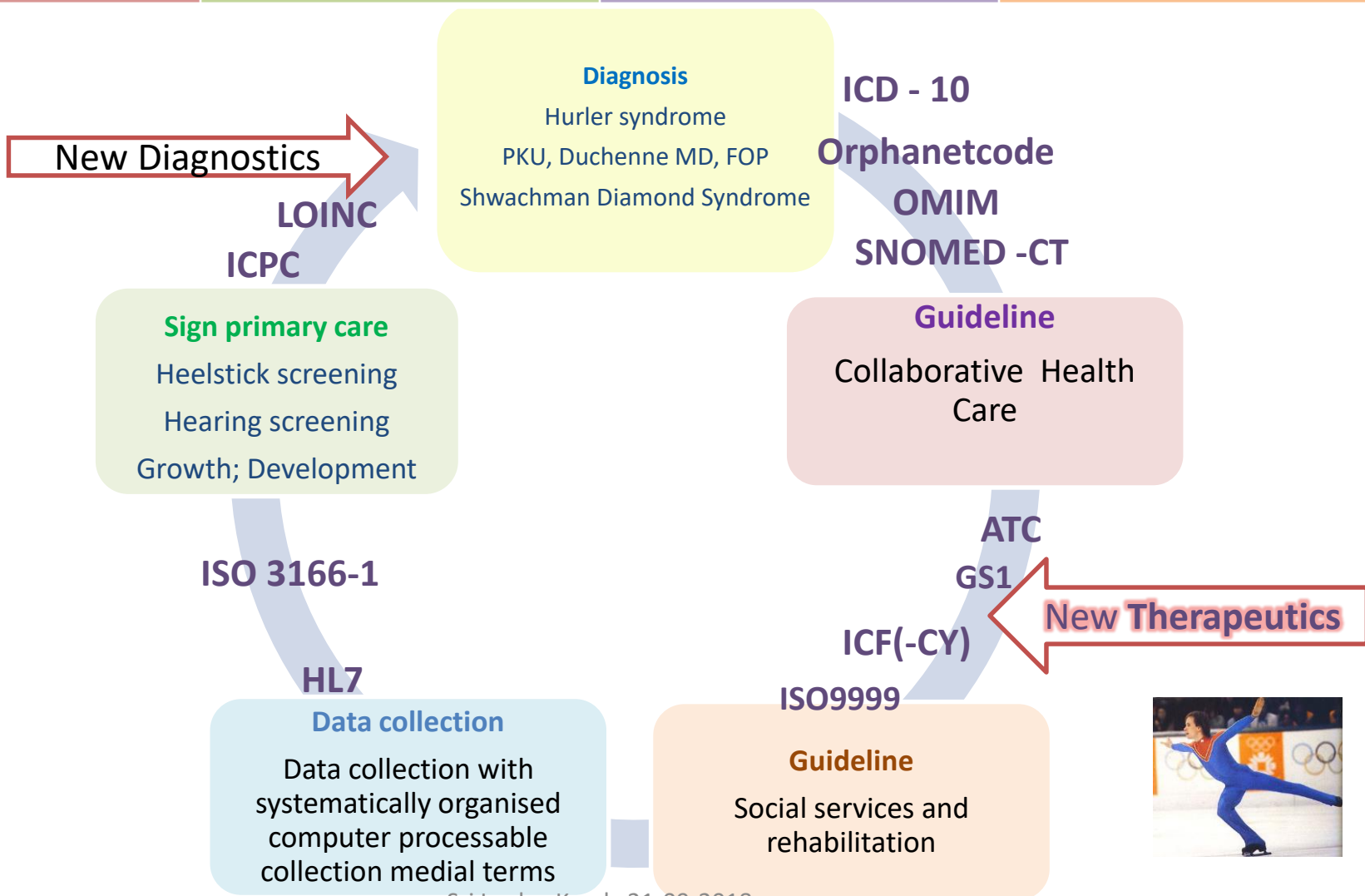
#### Disease

[Hemoglobinopathies](#)



Sri Lanka, Kandy 21-09-2018

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





# Rare Diseases Paediatric Global Action



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Anal atresia 1:5000, after operation parents daily wash the bowel, child will not easily be toilet trained if at all

Rare diseases seriously impact everyday life

**7 in 10** patients & carers

reduced or stopped professional activity due to their or their family member's rare disease.



**8 in 10** patients & carers

have difficulties completing daily tasks (household chores, preparing meals, shopping etc.)



**2/3** of carers

spend more than 2 hours a day on disease-related tasks.



**3 times** more people

living with a rare disease and carers report being unhappy and depressed than the general population\*



\* Rare Barometer Voices sample compared to International Social Survey Programme, 2011



Rare Barometer Voices is a EURORDIS-Rare Diseases Europe online survey initiative. It brings together over 6,000 patients, carers and family members to make the voice of the rare disease community stronger. Results are shared with policy decision makers to bring about change for people living with a rare disease.

**3,071**  
people responded to the survey.

The survey was conducted in  
**23** languages across  
**42** countries

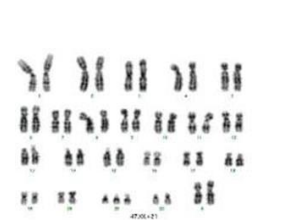


Thank you to all Rare Barometer Voices participants and partners!

For more information visit [eurordis.org/voices](http://eurordis.org/voices) or email [rare.barometer@eurordis.org](mailto:rare.barometer@eurordis.org)

[www.eurordis.org/content/contribute-rare-barometer-programme](http://www.eurordis.org/content/contribute-rare-barometer-programme)

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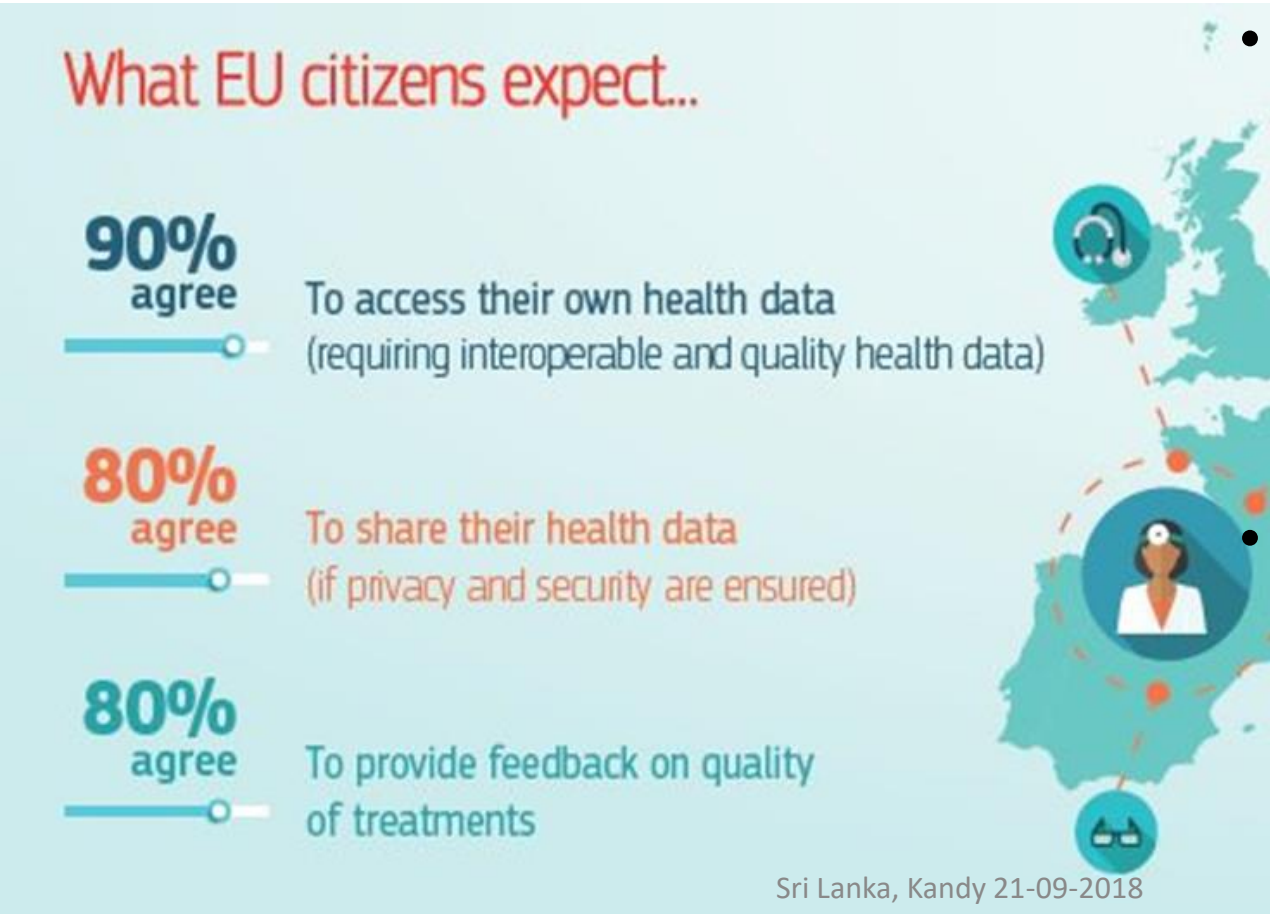


# @EU\_Health

## 25 April 2018



- EU\_Health
- Citizens' expectations on digital health: access to their own data, privacy & security and the ability **to give feedback on treatment.**
- Data in the EU: Commission steps up efforts to increase availability and boost **health care data sharing**





Familial Hypercholesterolaemia 1:250, diet and medicine will prolong life expectancy, patient organisation developed and app  
<https://fheurope.org/about-fh/familial-hypercholesterolaemia/>




# @WHO\_Europe 30 August 2018

## Q: What is health literacy?

communities. #HealthForAll

Tweet vertalen




What is health literacy ?

The ability

*to gain access to, understand and use information*

to promote and maintain good health



Sri Lanka, Kandy 21-09-2018

07:09 - 30 aug. 2018

# Rare becomes Common Patient-Centered Universal health coverage

Quality health care is *people-centred*.  
This means that decisions about your care are  
tailored to your needs and preferences and you  
are treated with respect and compassion.



THE "UNIVERSAL" IN  
UNIVERSAL HEALTH COVERAGE  
MEANS:  
OUT-OF-POCKET PAYMENTS  
DO NOT PUSH PEOPLE INTO  
POVERTY

