


Global Consensus in CHILD HEALTH

Distinct Features of Children with a Rare Disease in Preventive Child Health Care



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Life is simple,
but we insist on making it complicated.



孔夫子
Kǒng
fū
zǐ

It does not matter how slowly you
go as long as you do not stop.

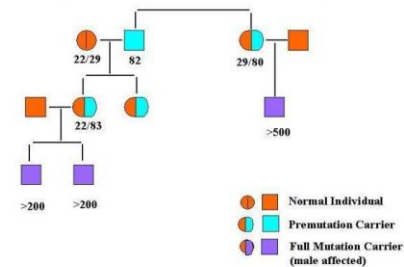
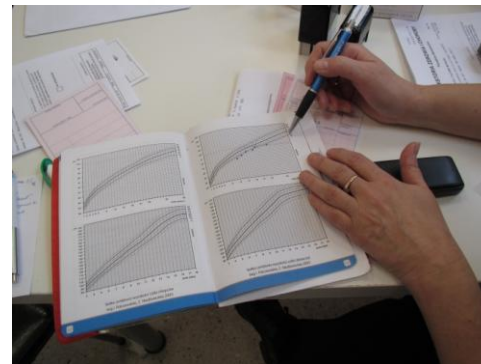
A female doctor with blonde hair, wearing a white lab coat and a stethoscope, is examining a baby's chest. The baby is sitting up, wearing a white diaper, and holding the doctor's hand. The doctor is looking down at the baby with a gentle expression. The background is a bright, out-of-focus indoor setting.



World map showing the percentage of the population aged 65 and over in 2009. The map is color-coded by region:

- <50% (4 countries or 2%)
- 50-79% (23 countries or 17%)
- 80-89% (25 countries or 18%)
- ≥90% (122 countries or 83%)
- Not available
- Not applicable

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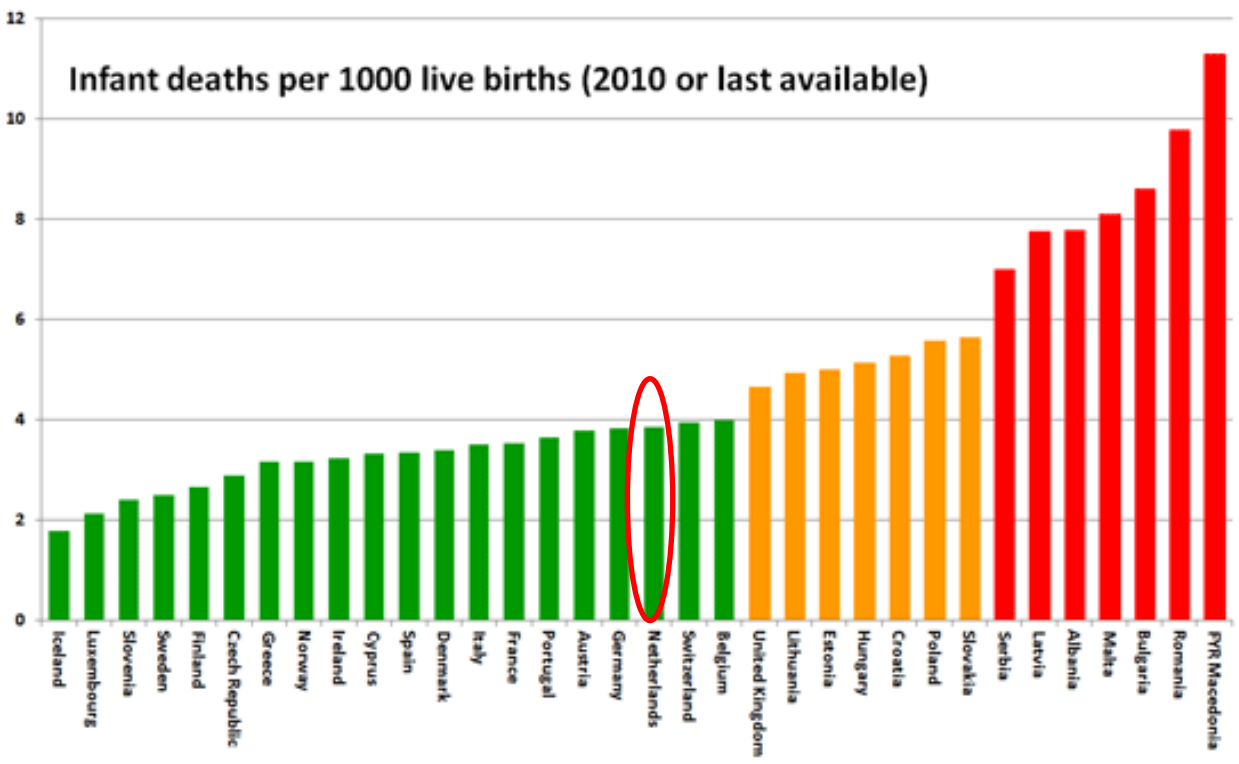
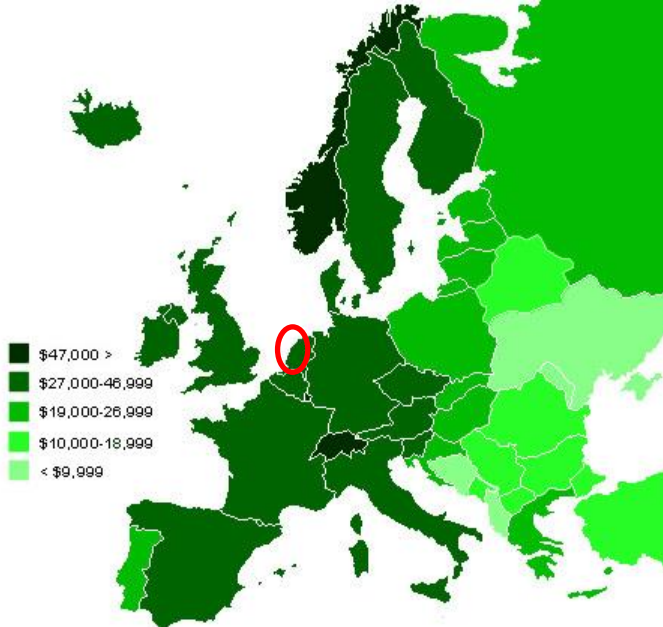


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Under 5 in mortality in Europe



Gross Domestic Product Europe
Languages Europe



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Under 5 mortality in China

Unicef, 2013

Source: National Bureau of Statistics, NPA Monitoring Statistics, 2014

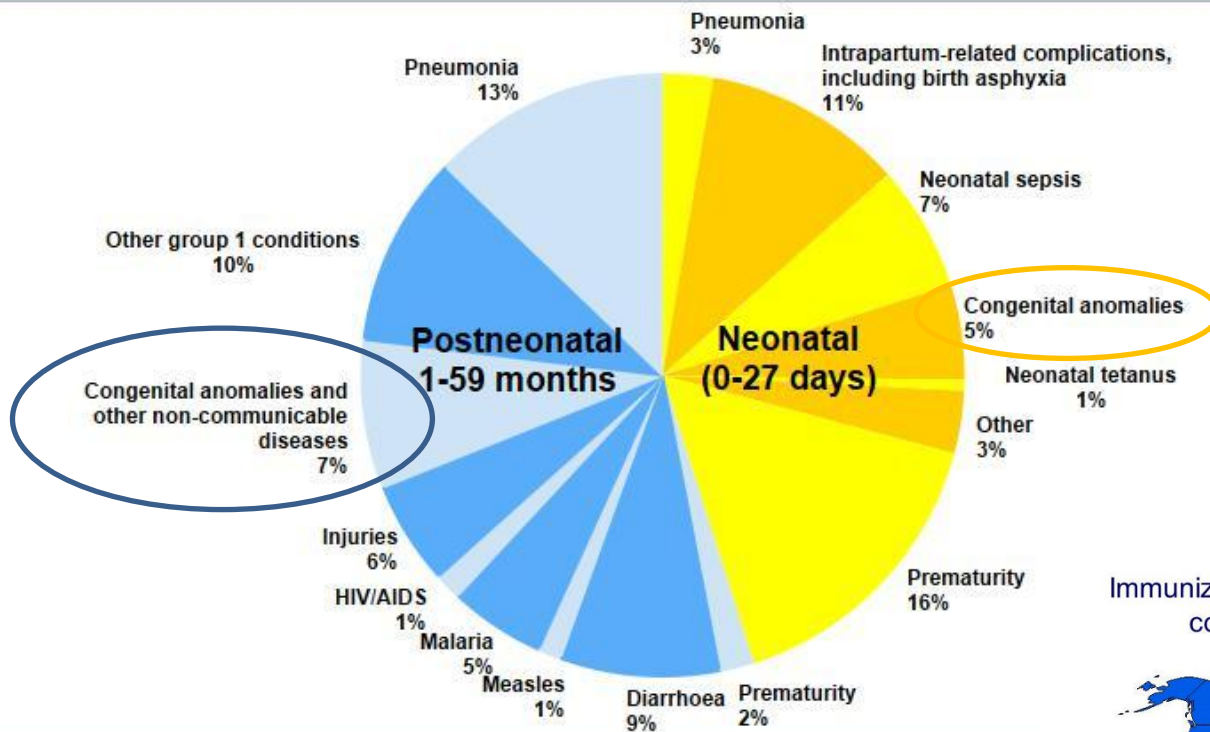


Great disparities exist in the infant mortality rates of different provinces. The infant mortality rate is generally highest in western provinces and lowest in eastern provinces, while central provinces mainly fall in between. The infant mortality rate ranges from **2.3 per thousand live births in Beijing** to around **20 per thousand live births in Xinjiang and Tibet**.

Yan, March 2016

WHO indicators under 5 mortality

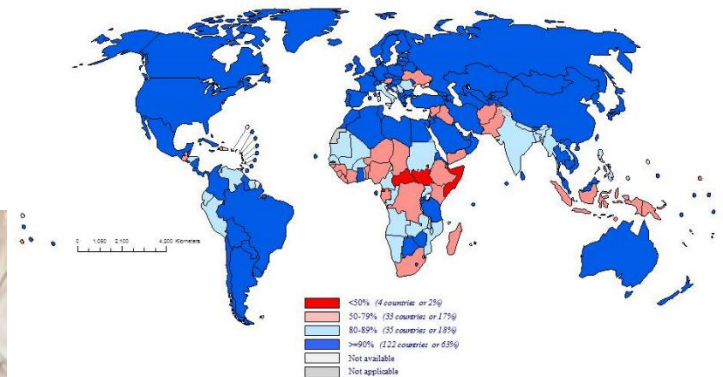
Causes of deaths among children under 5 years, 2015



Source: WHO-MCEE methods and data sources for child causes of death 2000-2015 (Global Health Estimates Technical Paper WHO/HIS/IER/GHE/2016.1)

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Immunization coverage with 1st dose of measles containing vaccines in infants, 2014

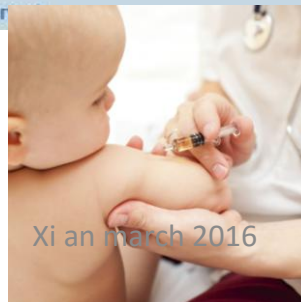


Source: WHO/UNICEF coverage estimates 2014 revision, July 2015. Map production: Immunization Vaccines and Biologicals (IVB), World Health Organization. Date of slide: 16 July 2015

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Rare is common in pediatrics

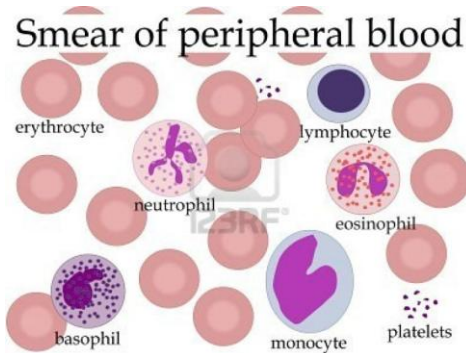
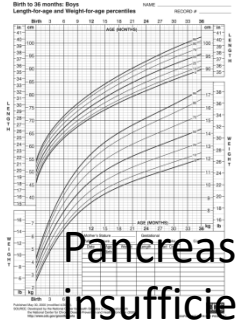


- A disease or disorder is defined as rare in Europe when it affects less than **1 in 2000**.
- There are between **6000** and 8000 rare diseases: **5-8%** population
- 80% of rare diseases are of genetic origin, and are often **chronic** and **life-threatening**.

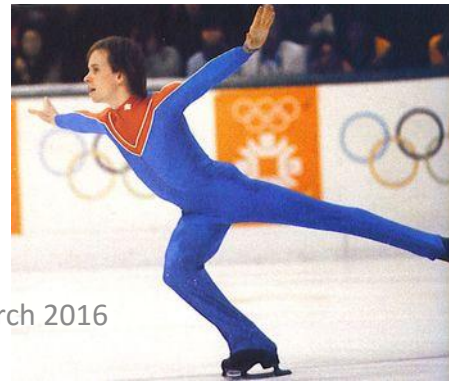


Shwachman Diamond Syndrome (SDS)

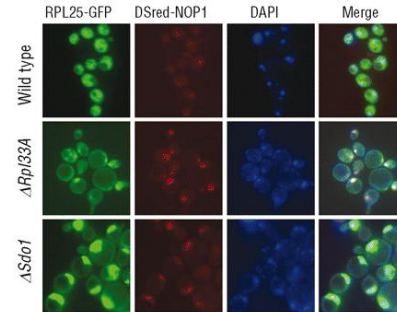
Growth Charts



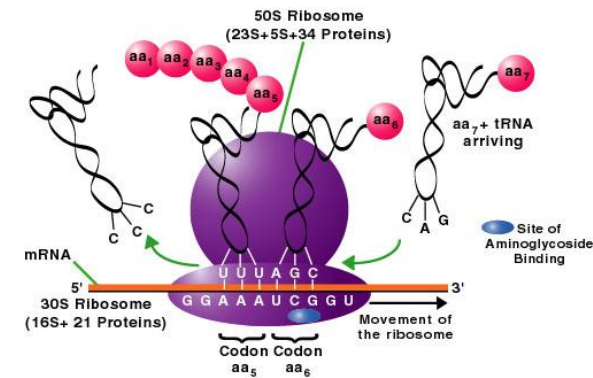
Neutropenia



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Ribosomopathy



EURORDIS
Rare Diseases Europe

Stichting Shwachman syndroom Support Holland



Listen to the Voices of Patients with Rare Diseases



EURODIS
Rare Diseases Europe



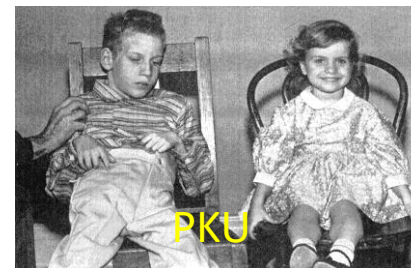
Duchenne



Fibrodysplasia
Ossificans
Progressiva



Down Syndrome



PKU



Osteogenesis Imperfecta

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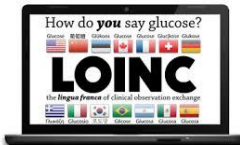





Fibula
Hypoplasia



Tuberous scelosis

After first symptoms,
battle for diagnosis,
which can last for years.
After the diagnosis,
faced with
inadequate health and **social care**.

Codification	Meaning	 World Health Organization
ICD & Orpha code	International Code of Diseases / Orphanet code	
ICF (-CY) 	The <i>International Classification of Functioning, Disability and Health for Children and Youth (ICF-CY)</i> is a derived version of the <i>International Classification of Functioning, Disability and Health</i> (ICF, WHO, 2001) designed to record characteristics of the developing child and the influence of environments surrounding the child .	
LOINC 	A universal code system for tests , measurements, and observations.	
ICPC 	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 	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.	



ICF-CY

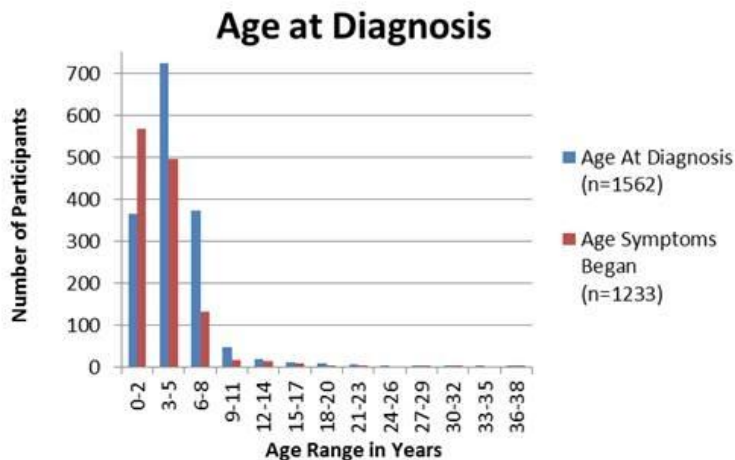
Duchenne Muscular Dystrophy

Diagnosis



LOINC
Lab code

Tests are needed before DMD can be diagnosed. The first step in making the diagnosis is a [blood test](#). This tests for **creatine kinase**. Children with DMD always have a very high level of creatine kinase (about 10-100 times normal). Therefore if a child's creatine kinase level is normal, then DMD is ruled out. If the creatine kinase level is high, further tests are needed to see whether this is due to DMD or to some other condition.

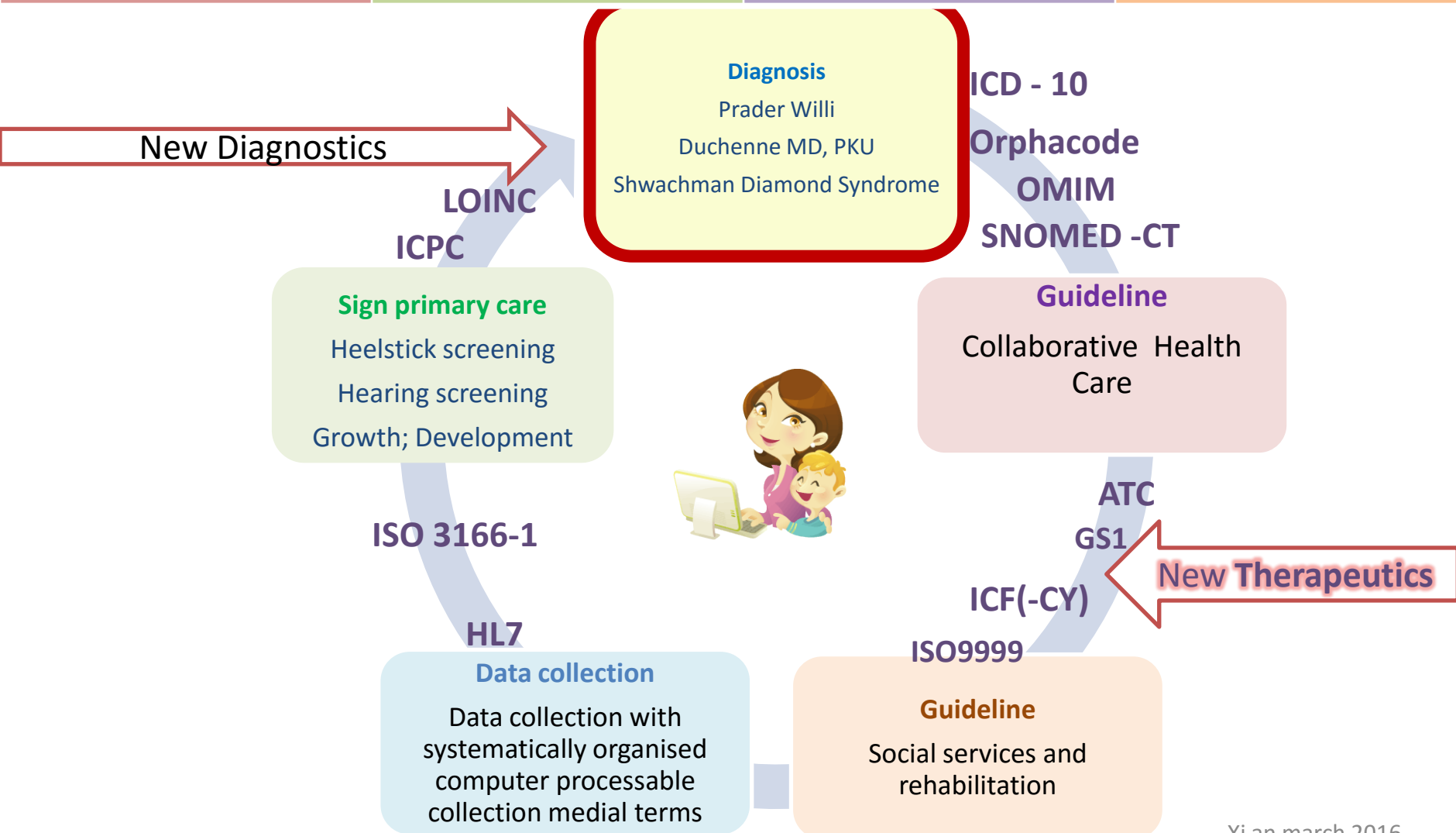


DuchenneConnect is targeted toward participants in the US

<http://www.patient.co.uk/health/duchenne-muscular-dystrophy-leaflet>

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Patient Information	Primary Care	Diagnosis Collaborative care	Social Services
www.shwachman.nl (ORPHA811) (ICD 10 : D61.0)	Growth retardation Recurrent infections (LOINC)	Guideline SDS (Orphanetcode; SNOMED, ATC e.a.)	Recurrent illness Fatigue, Short (ICF-CY; ISO 9999)



10 Rare Diseases	Primary Feature	Open information	Code
Anal Atresia	Con		ICD
Craniosynostosis			Code Dutch child health
Treacher Collins			
Cystic Fibrosis			ICD Orphacode
Duchenne Muscular Dystrophy			ICF - CY, LOINC
Fibrodysplasia Ossificans Progressiva			Code Dutch child health
Glaucoma			ICD
Hurler Syndrome			
Neurofibromatosis			Code Dutch child health
Rett syndrome			ICF- CY cm
Shwachman Diamond Syndrome	Granulocyte neutrophils		Kg, cm, LOINC
Sickle Cell Disease	Heel Stick Sample	 	ICD, LOINC, orphacode

A serene landscape featuring a body of water, likely a lake or a wide river, with tall reeds in the foreground. A wooden pier or dock extends into the water on the left side. The sky is filled with soft, white clouds, and the overall atmosphere is calm and peaceful.

The Convention of the Rights of the Child and the *Convention of the Rights of Persons with a disability* highlight how children with a disability have the same rights as other children.

Children with disabilities and their family have ordinary needs and must have access to **mainstream programs** and services.

We thank



FOP Stichting Nederland

Stichting Shwachman syndroom



Support Holland

And you for
your
attention

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