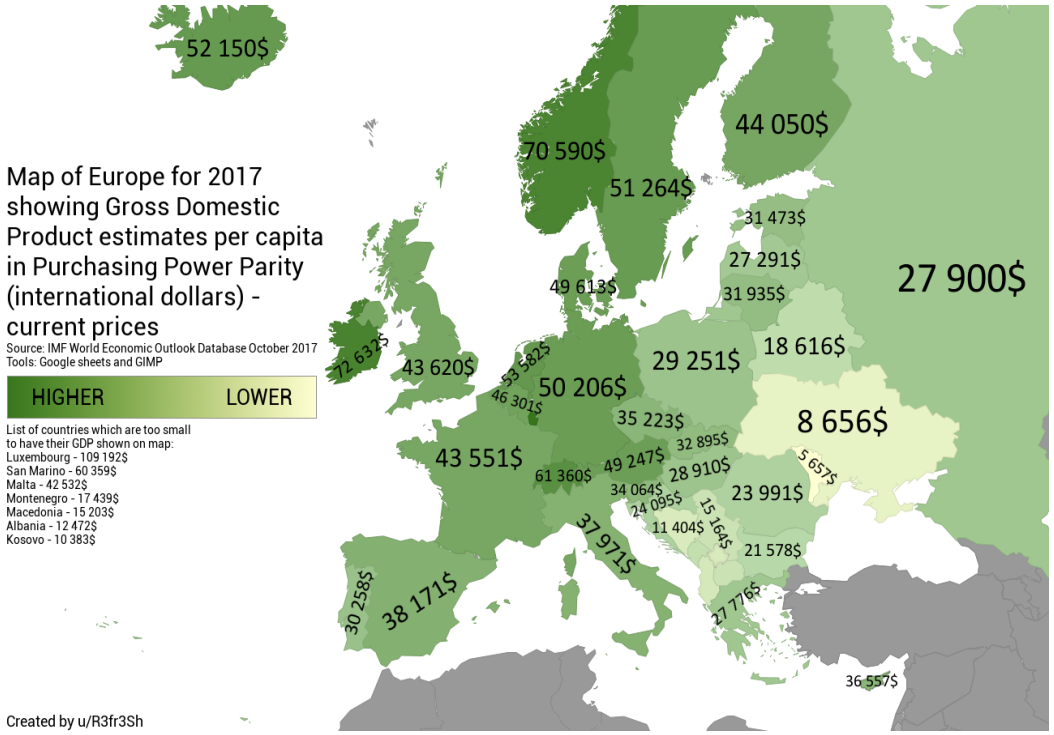
Rare Diseases in low economy countries - challenges and opportunities

Meeting of families with Alport Syndrome
OHRID, Macedonia
August 31th, 2018

Gross Domestic Product per Capita in Purchasing Power Parity



Macedonia $15,203

In 2017, the **average GDP per capita (PPP) of all of the countries of the world is USD $17,300.**
Using a PPP basis is comparing generalized differences in living standards between nations because PPP takes into account the relative cost of living.

Low quality healthcare is increasing the burden of illness and health costs globally

5 July 2018 News Release Geneva

Poor quality health services are holding back progress on improving health in countries at all income levels, according to a new joint report by the OECD, World Health Organization (WHO) and the World Bank.

Today, inaccurate diagnosis, medication errors, inappropriate or unnecessary treatment, inadequate or unsafe clinical facilities or practices, or providers who lack adequate training and expertise prevail in all countries.

Global Gathering for Rare Diseases

The inauguration of the Non Govermental Organization Committee for Rare Diseases took place 11 November 2016 the United Nations Headquarters in New York

Objectives

The NGO Committee for Rare Diseases is a multi-stakeholder, inclusive, global ecosystem focused on rare diseases, which aims:

To **increase visibility** of rare diseases at the global level

To **extend and share knowledge** about rare diseases and their unmet needs

To **connect** NGOs interested in rare diseases and their partners within a global platform

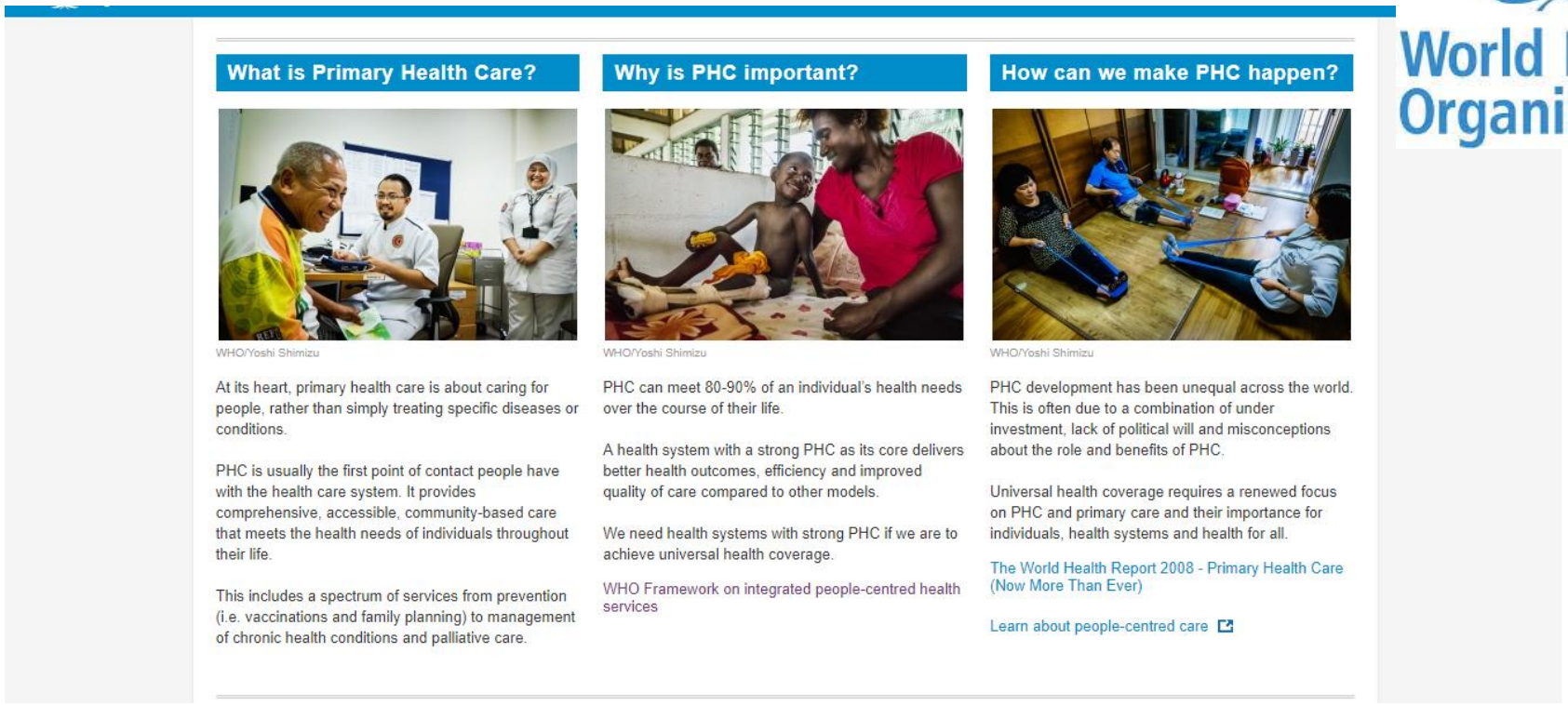
To **promote international, multi-stakeholder collaboration and actions** for rare diseases

To **align** rare diseases as a global priority in public health, research and medical and social care policies

Primary Health Care can meet 80-90% of an individual´s needs cover over the course of their life

http://www.who.int/primary-health/en/

World Health Organization



What is Primary Health Care?

WHO/Yoshi Shimizu

At its heart, primary health care is about caring for people, rather than simply treating specific diseases or conditions.

PHC is usually the first point of contact people have with the health care system. It provides comprehensive, accessible, community-based care that meets the health needs of individuals throughout their life.

This includes a spectrum of services from prevention (i.e. vaccinations and family planning) to management of chronic health conditions and palliative care.

Why is PHC important?

WHO/Yoshi Shimizu

PHC can meet 80-90% of an individual's health needs over the course of their life.

A health system with a strong PHC as its core delivers better health outcomes, efficiency and improved quality of care compared to other models.

We need health systems with strong PHC if we are to achieve universal health coverage.

WHO Framework on integrated people-centred health services

How can we make PHC happen?

WHO/Yoshi Shimizu

PHC development has been unequal across the world. This is often due to a combination of under investment, lack of political will and misconceptions about the role and benefits of PHC.

Universal health coverage requires a renewed focus on PHC and primary care and their importance for individuals, health systems and health for all.

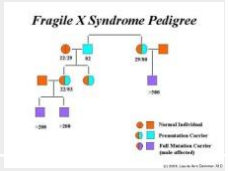
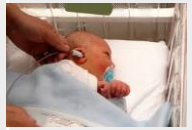
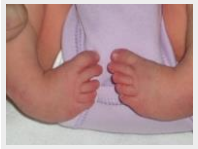
The World Health Report 2008 - Primary Health Care (Now More Than Ever)

Learn about people-centred care

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**
- Over 50 % manifests in childhood, almost each chronic condition in childhood,
- < 5 y 2-3 % have a rare disease

Identifying children with a disability through primary care, child health surveillance, and vaccination programs

Preventive Child Health	age	feature	Rare Disease
Family history	neonatal	X linked and autosomal recessive & dominant 	• Alport syndrome • Neurofibromatosis
Heel prick screening	neonatal	National program	• Sickle cell disease • Phenyl Keton Uria
Hearing screening *Otoacoustic emission* *Automated Auditory Brainstem Response*	neonatal 	Hearing deficit 0,2% > 76% cause identified 39% - 60% genetic , 30% aquered (cytomegalovirus) Other causes	• Usher syndrome 50% hereditary forms combining deafness and blindness prevalence of 3 to 6.2 per 100,000
Congenital anomaly	neonatal 	Club feet 1/700-1/1000 of liveborns	20% associated with • distal arthrogryposis, • congenital myotonic dystrophy,or other genetic syndromes

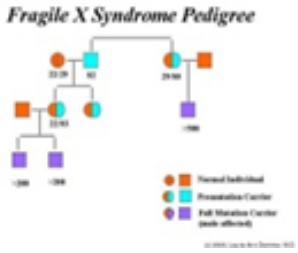
Recognize Alport syndrome

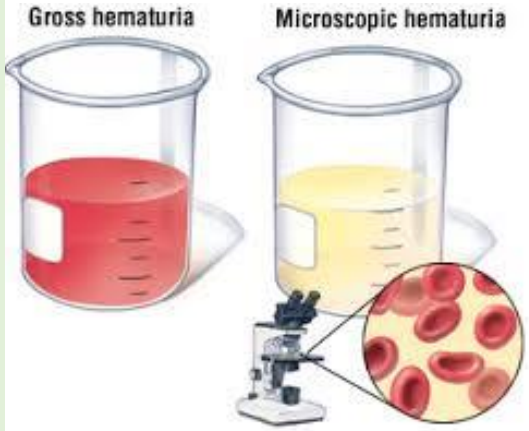
Primary Care

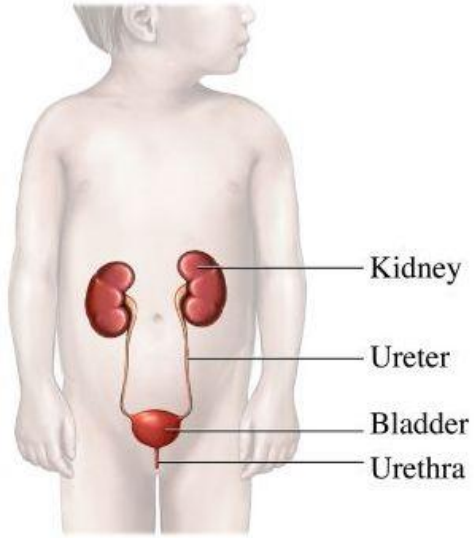
- Family history kidney disease

Tests or exams make a diagnosis:

- Urine test: protein and blood in urine.
- Blood test: A blood test levels of protein, and wastes in your blood.
- Ultrasound kidney
- Hearing test: A hearing test will be done to see if your hearing has been affected.
- Vision test: A vision test will be done to see if you vision has been affected.

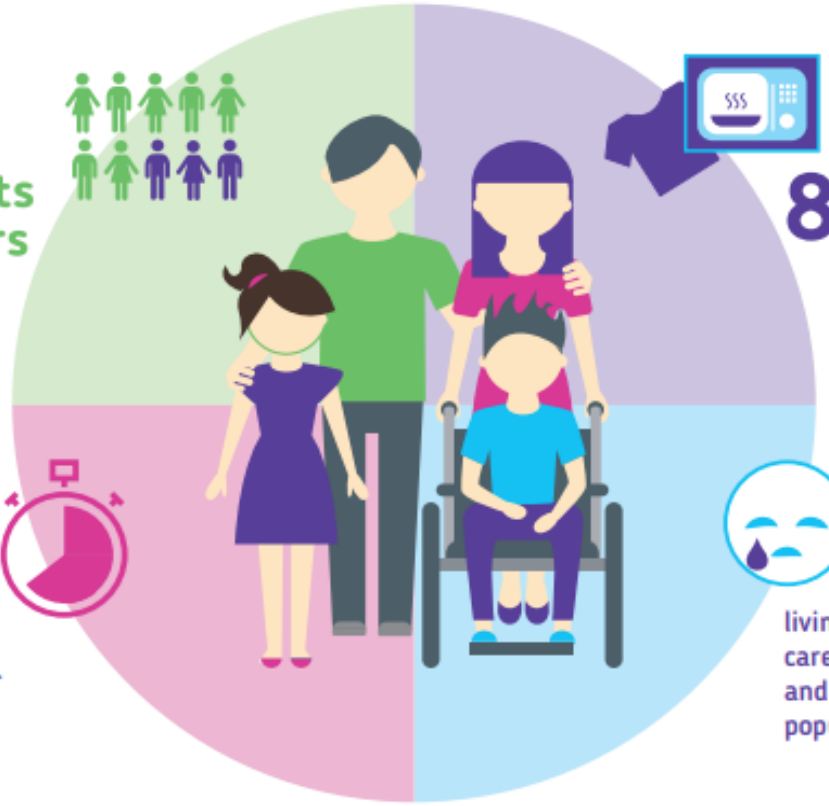






- Glomerular filtration rate (GFR): A blood test will be done to know how well your kidneys are filtering the wastes from your body.
- Kidney biopsy: looked at under a microscope.
- Genetic test: This can help confirm the diagnosis and determine the genetic type of Alport syndrome you may have.

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

A EURORDIS INITIATIVE

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.

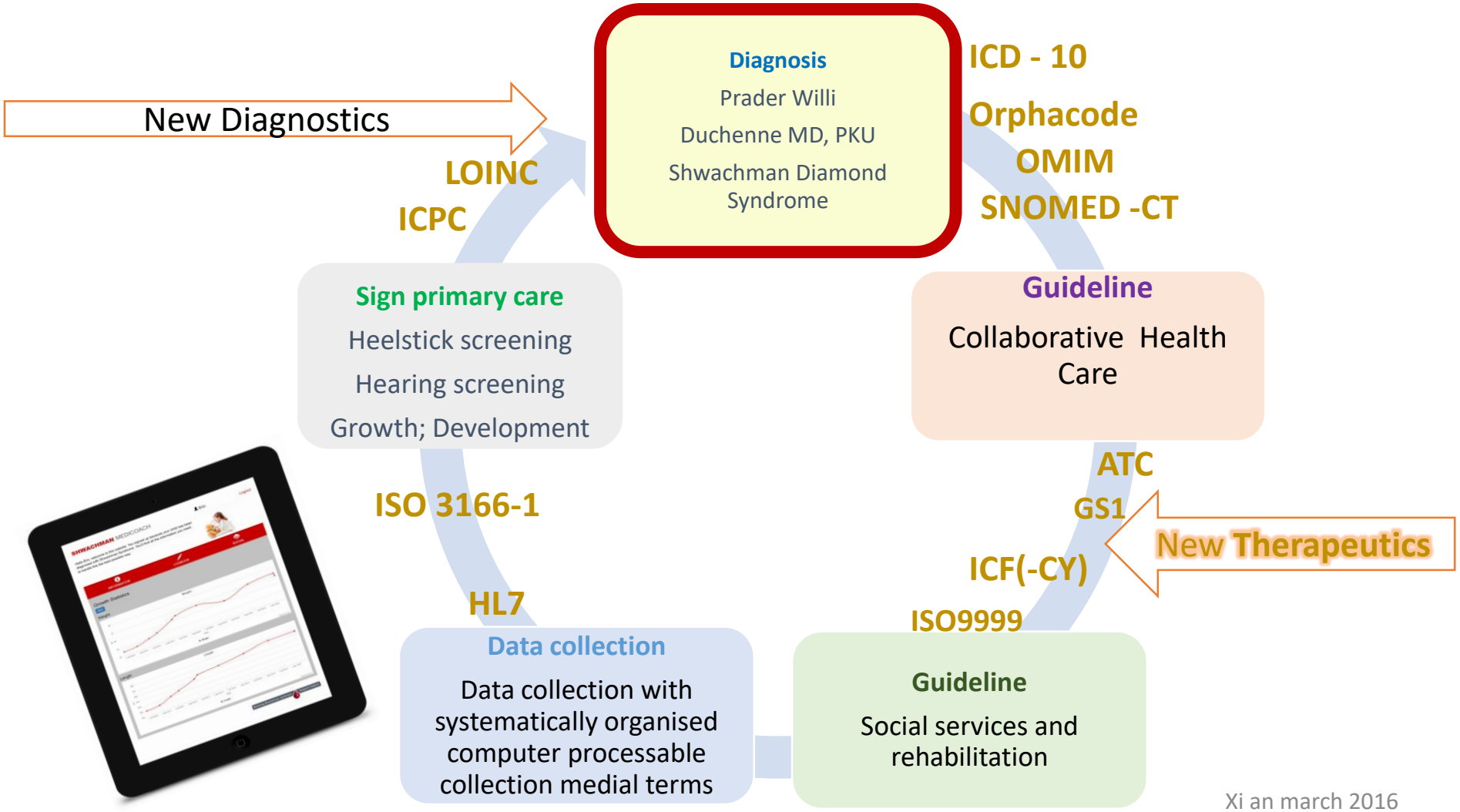
The survey was conducted in **23** languages across **42** countries

Thank you to all Rare Barometer Voices participants and partners!

www.eurordis.org/content/contribute-rare-barometer-programme

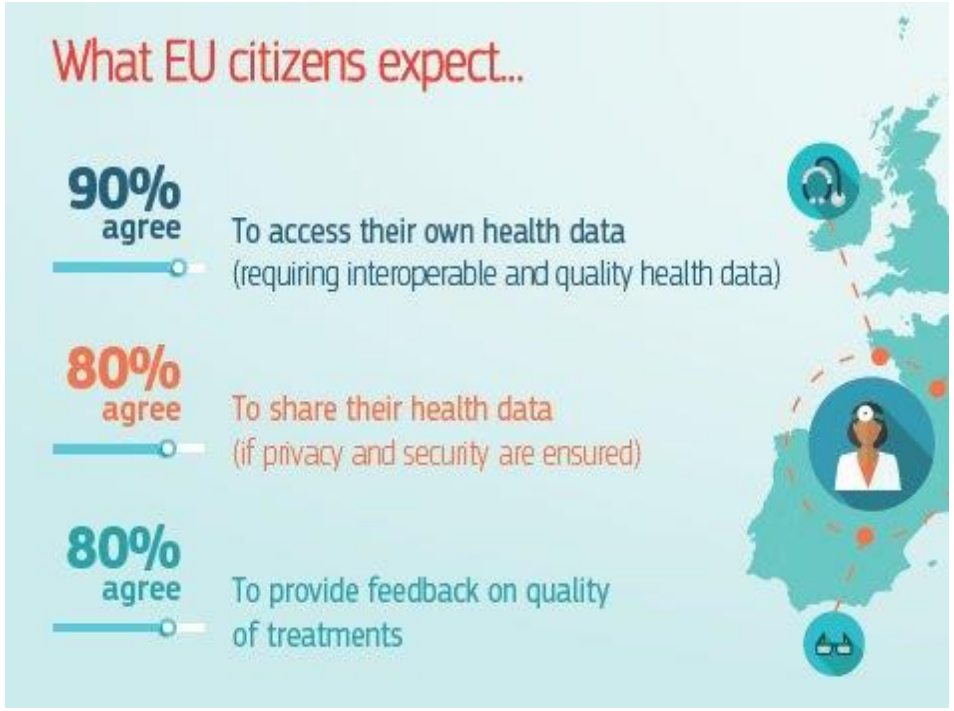
For more information visit **eurordis.org/voices** or email **rare.barometer@eurordis.org**

Patient Informatiom	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)



Xi an march 2016

@EU_Health
25 april 2018



EU_Health

Citizens' expectations on #digitalhealth: access to their own data, privacy & security and the ability to give feedback on treatments.

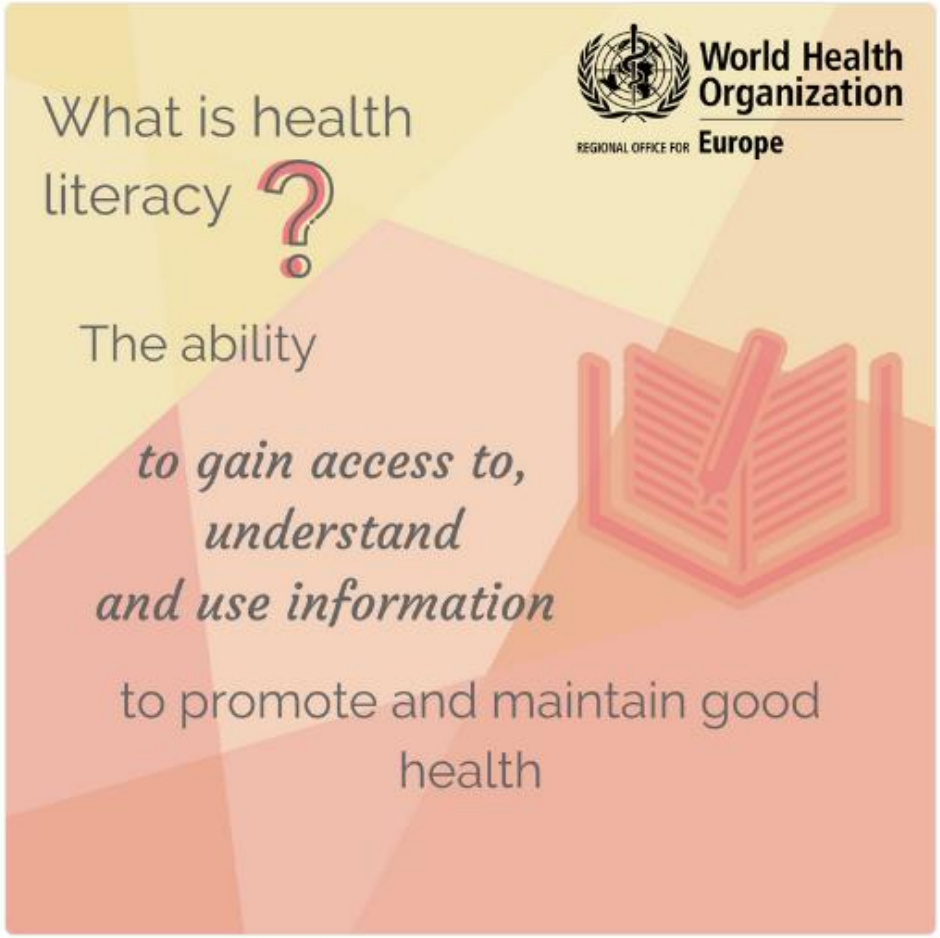
Data in the EU: Commission steps up efforts to increase availability and boost healthcare data sharing

Twitter WHO 30 August 2018

World Health Organization

communities. #HealthForAll

Tweet vertalen



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