

European Networking for Rare: Equality, Equity or Justice?

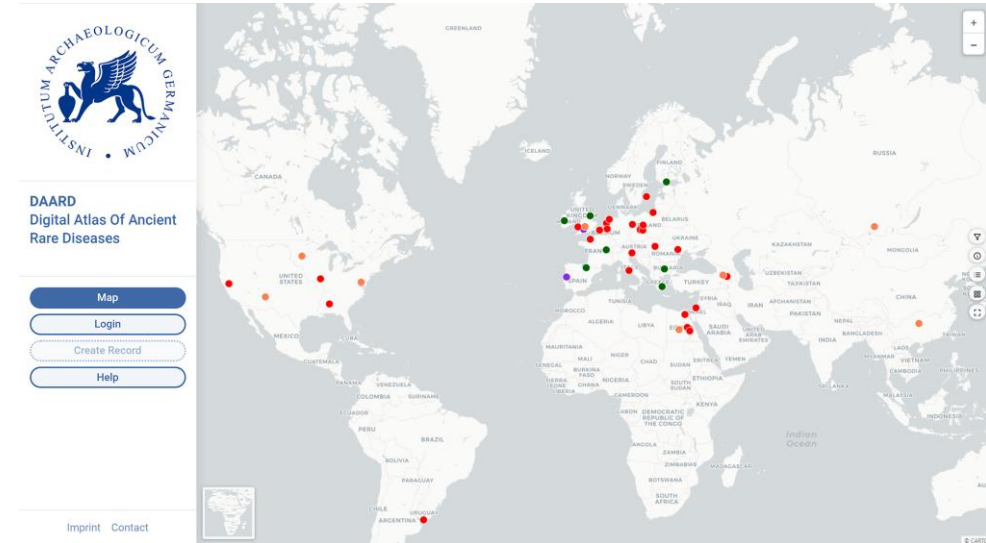
Violetta Christophidou Anastasiadou MD, PhD
Chair of the National Committee of Rare Diseases
Consultant Paediatrician, Clinical Geneticist



Rare Diseases In Antiquity

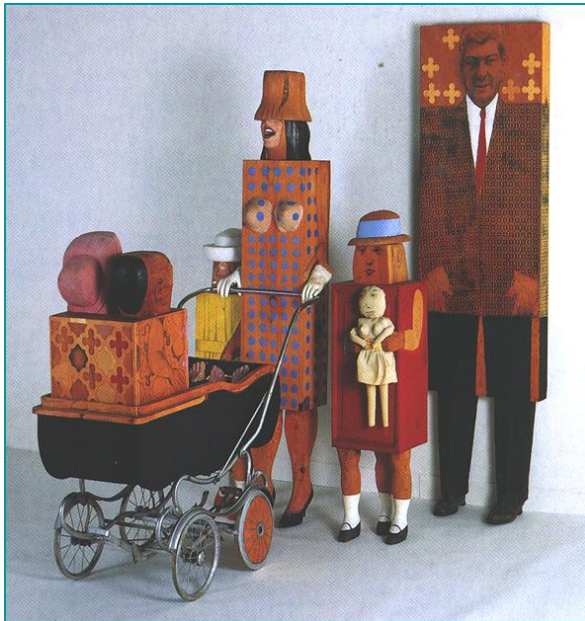


- Without microscopes or modern pathology, ancient physicians attributed **hidden or rare diseases** to a mix of humoral imbalances, environmental "miasma" (bad air), and the supernatural. In classical antiquity, illnesses whose causes were not visually apparent were frequently described as **apora** (puzzling or impenetrable objects of inquiry)



Rarity Today

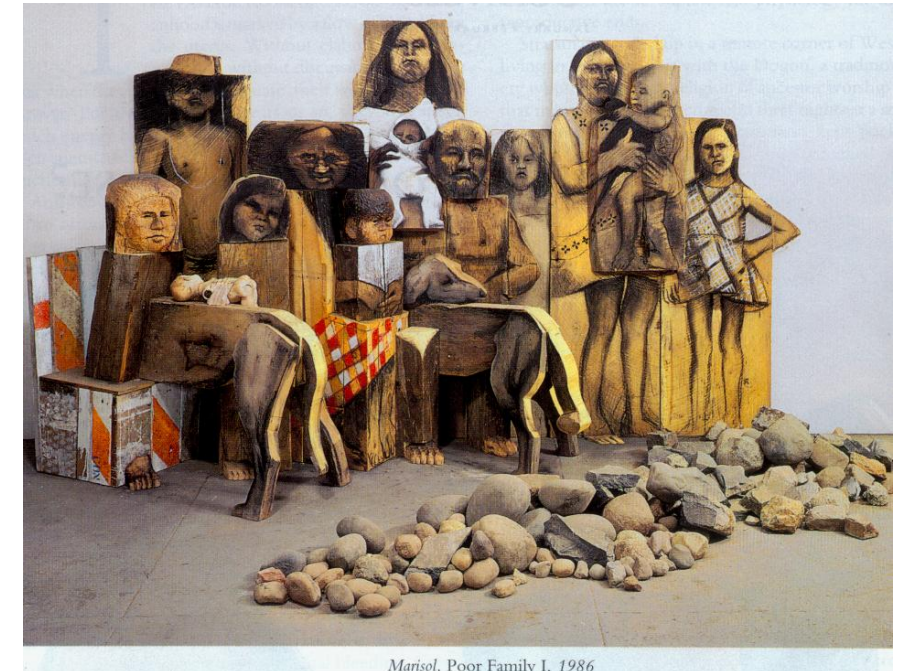
Family dimension



Diagnosis & Grief

- Shock-anger-denial
- Questioning-sadness
- Acceptance-mourning
- Health and Disease reappraised
- Care pathways in search
- Quality of life uncertainty
- Financial and professional challenges
- Family life balance endangered
- The “ordinary” sibling's burden
- Societal response
- **The STIGMA**

Family – societal disruption



Marisol, Poor Family I, 1986

Recognising Rare Within “Ordinary”

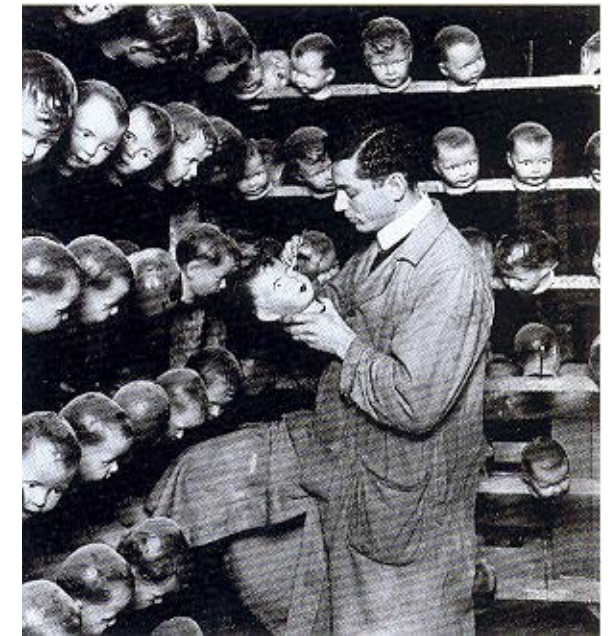
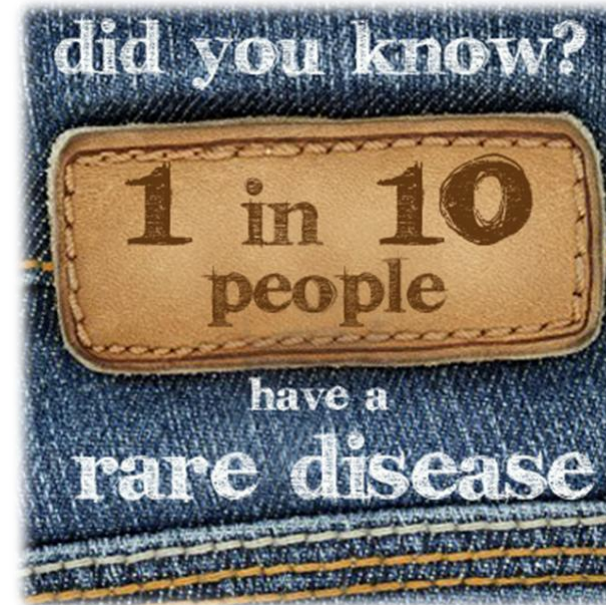
Rare diseases: a ‘common’ problem for physicians

Collectively more common than diabetes!
Almost as common as asthma!

- Patients with rare diseases in every medical office
- **Neglected in medical education**
- Neglected in research
- **Specialized clinics are under-resourced**
 - Lack sustainable funding
 - Located in tertiary hospitals

HEALTH CARE SYSTEMS tremendous stress
alike “Women on the Verge of a Nervous
Breakdown” (1988) Pedro Almodovar

CHALLENGES



Advancement of Treatments FOR RARE DISEASES

When a Rare Disease is part of the Normality *the “ordinary Stigma”*



Hemoglobin
international journal for hemoglobin research

ISSN: (Print) (Online) Journal homepage: <https://www.tandfonline.com/loi/them20>

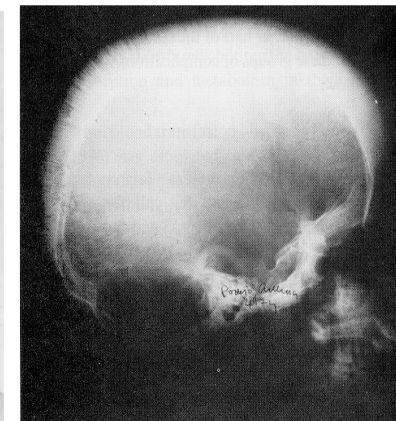
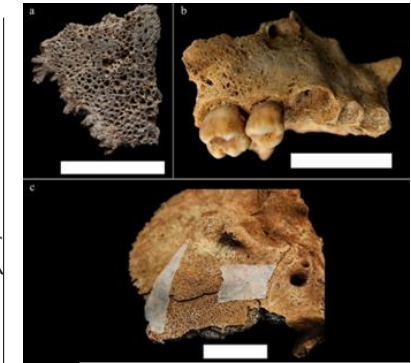
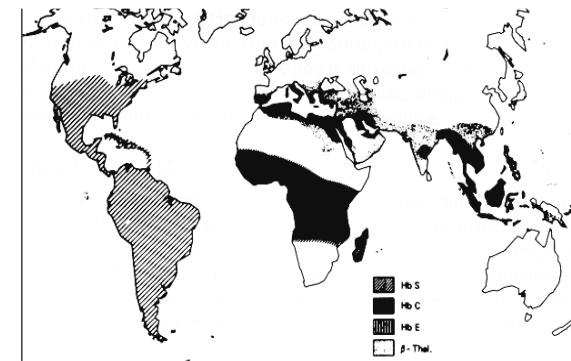


**The Prevention of Thalassemia Revisited:
A Historical and Ethical Perspective by the
Thalassemia International Federation**

Michael Angastiniotis , Mary Petrou , Dimitrios Loukopoulos , Bernadette
Modell , Dimitrios Farmakis , Panos Englezos & Androulla Eleftheriou



Yayoi Kusama, *Repetitive Vision*, 1996



The Cyprus Thalassaemia Association

Organisers:



The conference is funded by the
European Union (project no 101297283)

Advancement of Treatments FOR RARE DISEASES

An International Networking Example *worth following*



NIH National Library of Medicine
National Center for Biotechnology Information

Bookshelf Books Browse Titles Advanced Search Help Disclaim

Guidelines for the Management of α -Thalassaemia [Internet].
 Amir A, Lal A, Coates TD, et al., editors.
 Nicosia (Cyprus): Thalassaemia International Federation; 2023.

Chapter 13 **NOVEL AND EMERGING THERAPIES FOR α -THALASSAEMIA**
 Antonia Kattamis.
 • Author Information and Affiliations

Disease modifying therapies Go to:

Different pharmacological agents that may improve anaemia in α -thalassaemia by acting in different stages of the thalassaemia pathogenesis are being evaluated. These agents are not expected to have any benefit in patients with haemoglobin Bart's hydrops foetalis who do not have the ability to produce functional haemoglobin. The patients that will benefit the most from these agents are patients with moderate to severe forms of haemoglobin H (HH) disease, who require red blood cell transfusions and/or exhibit thalassaemia-related morbidities. These patients exhibit a high degree of haemolysis and/or ineffective erythropoiesis, and they usually carry non-deletional α -thalassaemia mutations.

World Blood Donor Day 2023 | One Drop of Humanity. Give Blood. Save Lives.

THALASSAEMIA INTERNATIONAL FEDERATION
 Treatment, prevention, final cure

A worldwide organisation for thalassaemia
 Treatment, prevention, final cure

SUPPORT US!

Who we are | What we do | Haemoglobin Disorders | Positions & Policies | Publications | Media Centre | Contact us

TIF e-Academy

e-Academy is TIF's newly developed project, within the context of TIF 2.0 Era. Using a variety of interactive tools and allowing users a deep learning experience, TIF e-Academy looks to increase the knowledge of patients, parents and healthcare professionals on thalassaemia and sickle cell disease (SCD) facts, past and current advances.

Visit e-Academy

An Introduction to the TIF e-Academy
 Thalassaemia TIF

WELCOME TO TIF'S E-ACADEMY!

Watch on YouTube

For Patients

Brief summary and recommendations
 References

In TIF e-Academy, Thalassaemia patients (and parents) can take TIF's Thal e-Course to learn more about the disorder, its genetic cause, the blood component affected, new advances in treatment, lifestyle and many more. Sickle Cell Disease patients, their families and caregivers, can gain in-depth knowledge on all aspects of SCD clinical management through the SCD e-Course for Patients, with content developed by eminent international medical experts, with extensive



ERICA About Work Packages ERICs News Resources

Overview of ERICs

ERN EuroBloodNet
 European Reference Network for Hematological Diseases

Haematological diseases involve abnormalities of blood and bone marrow cells, lymphoid organs and coagulation factors, and almost all of them are rare. They can be subdivided into six categories: rare red blood cell defects; bone marrow failure; rare coagulation disorders; haemochromatosis and other rare genetic disorders of iron synthesis; myeloid malignancies; and lymphoid malignancies.

Diagnosis of rare haematological diseases (RHs) requires considerable clinical expertise and access to a broad range of laboratory services and imaging technologies. These tests allow precise disease classification according to WHO criteria using international scoring systems and, where possible, biomarkers.

Given these requirements and the fact that some RHs are very rare, diagnosis is frequently overlooked or delayed, especially in elderly patients. Treatment is also often difficult due to the specialised infrastructures and teams required and the difficulty accessing specific treatments such as allogeneic stem cell transplantation or coagulation factors.

Preventive programmes are in place in some countries for certain conditions, but there is an urgent need for harmonisation in the field of screening.

EuroBloodNet, with the experience gained thanks to the EU-funded European Network for Rare and Congenital Anaemias (ENERCA) and the European Haematology Association (EHA), will seek to improve access to healthcare for RH patients; to promote guidelines and best practice to improve training and knowledge sharing; to offer clinical advice where national expertise is scarce; and to increase the number of clinical trials in the field.

Contact information

- Website of Network
- LinkedIn
- Twitter
- Facebook

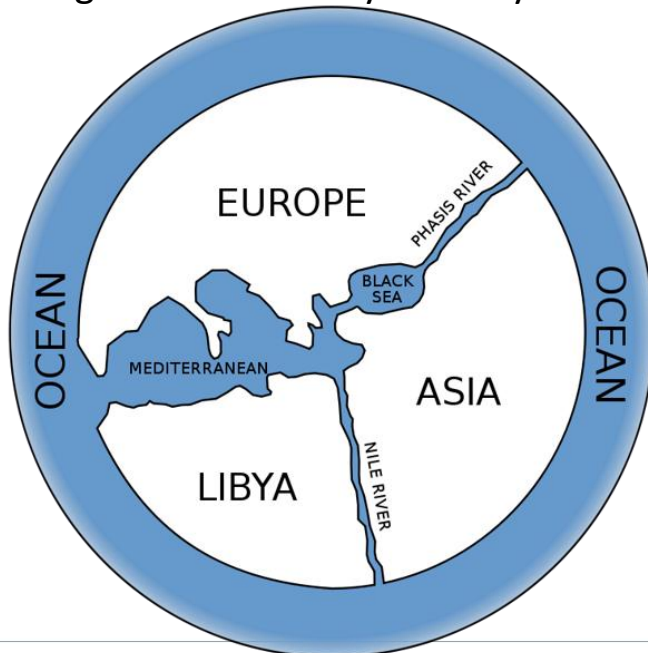
ebn
 ERN | EuroBloodNet

Advancement of Treatments FOR RARE DISEASES

The cornerstone of three continents...

The First Map of the world by Anaximandros
c. 610 – c. 546 BC

Anaximander was a pre-Socratic Greek philosopher who lived in Miletus, a city of Ionia, the Aegian coast of today's Turkey



A Snapshot of 70 centuries Cypriot History...

Early History

- Cyprus Stone Age (5800-3000 BC)
- Cyprus Bronze Age (3000-2300 BC)
- The Achaeans in Cyprus (1225 BC)
- The Phoenicians in Kition (1100 BC)
- The Assyrians in Cyprus (709 BC)
- The Egyptians in Cyprus (560 BC)
- Persian Dominance in Cyprus (522-486 BC)

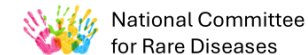
Local Indigenous Population

- Greeks
- Phoenicians
- Assyrians
- Egyptians
- Persians

- Cypriots form an allegiance with Alexander the Great (332 BC)
- The Ptolemies in Cyprus (294 – 58 BC)
- Romans and Christianity (58 BC)
- Byzantine Times (395 AD)

Recent

- Franks in Cyprus (1192-1489 AD)
- Venetian Period (1489-1571 AD)
- The years under the Ottoman rule (1570-1878 AD)
- The British rule in Cyprus (1878 – 1960 AD)
- Cyprus gains its independence (1960)



Organisers:



Funded by
the European Union

The conference is funded by the
European Union (project no 101297283)

Advancement of Treatments FOR RARE DISEASES

- **CYPRUS NATIONAL COMMITTEE FOR RARE DISEASES**
- First National Strategy for Rare Diseases (2010...today 2026 update)
- **VISION: To develop a comprehensive strategy that ensures fair access for all patients with Rare Diseases to a well-organized system of medical care and social welfare.**



2010, Εθνική Στρατηγική για τα Σπάνια

- Η ανάπτυξη ολοκληρωμένης στρατηγικής που να διασφαλίζει την ισότιμη πρόσβαση όλων των Ασθενών με Σπάνια Νοσήματα σε ένα άρτια οργανωμένο σύστημα ιατρικής φροντίδας και κοινωνικής μέριμνας
- 8000 σπάνιες ασθένειες αφορούν 8% πληθί
- Εξαιρετικά δύσκολη διάγνωση
- Ετερογενείς, σοβαρές έως ανίατες
- Ορφανά φάρμακα
- Πολυασπληριές-ειδικές ανάγκες
- Ατομικό-οικογενειακό-κοινωνικό κόστος
- Δημόσια υγεία



Health Care Systems and Professionals awareness / expertise in Rare Diseases PUBLIC and patient awareness

- By education:**
 - Limited awareness ?
 - Limited education and training ?
 - Scarce expertise ?
- By incentives:**
 - Limited inspiration ?
 - Limited identification by the NHC-systems ?
 - Health care economics ratification ?
- Consequences:**
 - Fear for families
 - Fear and reservation for professionals- development and financial safeguarding ?
 - More scarcity ?

- Public awareness limited
- Patients and families
 - Stigma
 - Everyday life obstacles



Activities-Public Awareness

ORPHANET Cyprus

- Network committed to maintain, update and develop an internet database for rare diseases and orphan drugs

Networking for Genetic Disorders

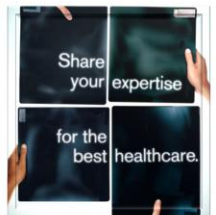
- American Embassy project

GeneNet Cyprus

- Biocommunal project
- Trilingual-leaflets and website

ERNs # CBHC building bridges-bridge the gaps

- ERNs TRIAD: Health Care, Academy and Research
- Enhancing Access/ Membership/ Affiliation to ERNs
 - Absolute importance for all MS, large to small
 - Reduce inequity
 - Develop knowledge and expertise
 - Establish a mentality of interaction, networking and collaboration
 - Capacity Building in national health systems
- Integrating ERNs in CBHC will guarantee CBHC sustainability and invest on patients' rights



Advancement of Treatments FOR RARE DISEASES



European
Reference
Networks

24 networks of excellence in RD

➤ Kick-off: 2017-03-09, Vilnius

> **1600 Centers of Excellence, ~1 000 000 patients/year**

- **Triangle of highly-specialized healthcare, research and education**
- **The largest EU platform for virtual and physical cross-border healthcare, clinical and translational research in rare and complex diseases**
- **Economies of speed, scale and scope for multiple tasks**
develop and implement RD clinical guidelines, collect cohorts and data, create a curriculum for RD education, perform an ultra-rare disease clinical trial, make RD monitoring for policy decisions, etc.
- **Multidisciplinarity, accessibility, quality assurance, tackling inequities**



Organisers:



THE CYPRUS INSTITUTE OF
NEUROLOGY & GENETICS



DEPUTY MINISTRY OF
RESEARCH, INNOVATION
AND DIGITAL POLICY
REPUBLIC OF CYPRUS



CY2026.EU
CYPRUS PRESIDENCY
OF THE COUNCIL OF THE EU



Funded by
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Advancement of Treatments FOR RARE DISEASES

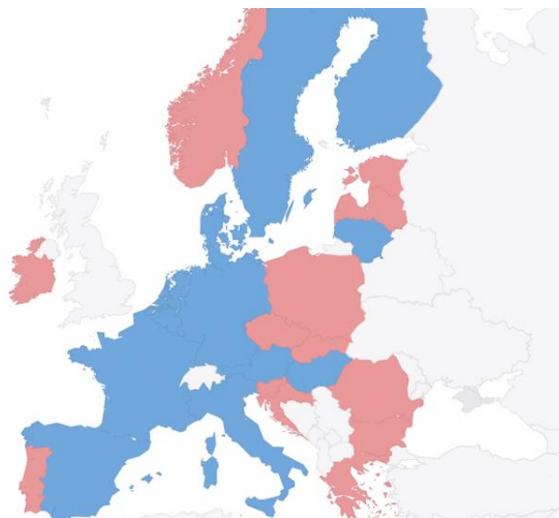
EUROPEAN NETWORKS

The ERNs today

- 24 ERNs
- 1605 Clinical teams
- 375 Hospitals
- 28 Countries

But...

- 14 Countries still lacking full coverage



OBJECTIVE

- Improve geographical coverage of ERN expertise across Member States and Norway
- Facilitate access to specialised care in countries or disease areas where full ERN membership is not yet established
- Support cross-border knowledge exchange and clinical collaboration
- Strengthen the integration of ERNs into national healthcare systems

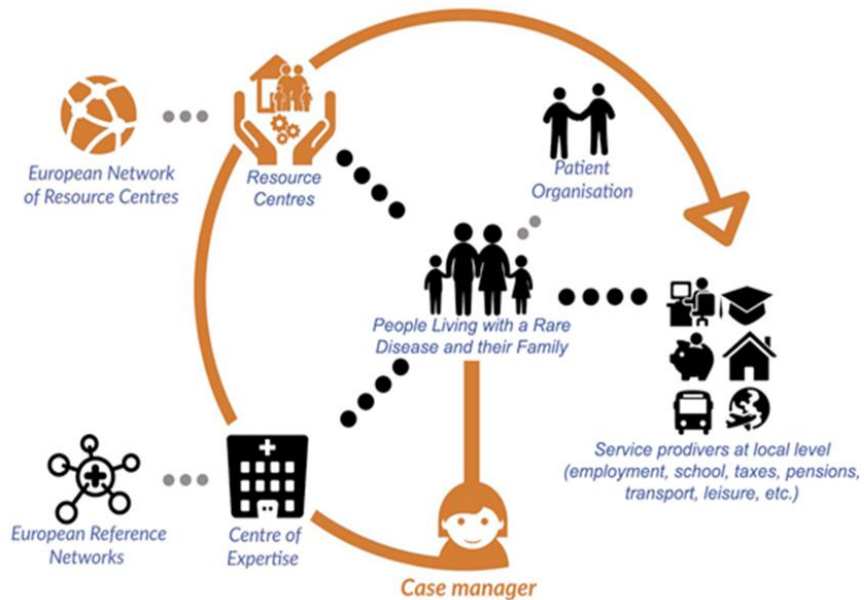
WHY THIS CALL: ERNs COVERAGE

ERN Centres	AT	BE	BG	CY	CZ	DE	DK	EE	ES	FI	FR	GR	HR	HU	IE	IT	LT	LU	LV	MT	NL	NO	PL	PT	RO	SE	SI	SK	TOTAL	Mbrs	APs	MS not covered	
BOND	1	2			1	6	3		4	1	5			1	1	10	2	1		1	4	1		2		1	1	1	48	42	6	3	
CRANIO	1	3			1	2	2		3	1	3			2	1	7	1	1	1	1	4	2	1			3	1	1	42	35	7	7	
Endo-ERN	2	7	2	1	2	13	3	1	5	1	9	3	2	1	1	20	1	1	2	1	8	2	4	3	4	2	1	2	104	92	12	0	
EpiCARE	2	2			1	2	4	2	2	3	2	6		2	1	7	2	1	1	1	1	1	1	3	1	1	1	1	50	38	12	4	
ERKNet	2	3			2	11	3	2	5	1	7	1		1	1	15	1	1	2	1	6	1	3	1	1	1	1	1	73	64	9	4	
ERN-EYE	3	3			1	8	2	2	7	1	6		1	1	1	9	1	1	1	1	4		1	1	1	1	1	59	51	8	4		
ERNICA	4	3			1	6	2	1	3	1	6		1	1	1	6	2	1	1	1	5	1	2			3	1	52	39	13	7		
ERN-LUNG	3	6			1	4	11	2	1	4	1	5		1	2	1	23	2	1	1	1	7	1	2	3	2	2	1	87	78	9	3	
ERN-RND	1	4	1	1	4	9	2	1	8	1	5	1	1	3	1	9	1	1	1	1	6		2			2	1	67	63	4	4		
ERN-SKIN	2	3			4	8	2		1	1	6		1	3	1	12	1	1	1	1	5				1	1	1	56	52	4	8		
EURACAN	1	6			1	4	8	1	2	6	2	12	1	2	1	1	26	2	1	1	1	8	1	3	3	1	3	1	99	90	9	2	
EuroBloodNet	2	6	2	1	3	10	2	1	4	1	11	4		1	1	32	1	1	1	1	5	2	2	2	1	1	1	96	89	7	4		
eUROGEN	1	3			2	11	2		7	1	2		1	1	1	7	1	1	1	1	6	2	1		4	1	1	56	51	5	8		
EURO-NMD	1	5	1	1	2	10	2	1	7	2	9	1		1	1	20	1	1	1	1	6	1	1	1	2	1	1	80	73	7	3		
GENTURIS	2	4			1	2	6	2	1	2	2	1		1	1	9	1	1	1	1	5	1	1	3	1	1	1	51	44	7	5		
GUARD-HEART	2	3			2	2	2	1	7	1	5		1	1	1	9	2	1		1	4		1	3	1	1	1	53	44	9	5		
ITHACA	1	5		1	1	9	3	1	3	2	7		1	2	1	13	1	1	1	1	6	1	3	2	1	1	1	70	65	5	2		
MetabERN	4	6	1		1	11	1	1	7	1	10	1	1	1	1	18	1	1	1	1	6	2	3	6	1	2	1	91	85	6	1		
PaedCAN	1	4	1	1	2	19	2	2	6	4	7	1	3	2	1	12	1	1	1	1	2	1	6	2	2	3	1	90	79	11	0		
RARE-LIVER	1	4			1	10	2	1	3	1	5	1	2	2		13	1	1	2	1	3	1	2	2	2	2	1	62	52	10	5		
ReCONNET	1	3			1	4	1	1	3	1	7	1	1	1	1	20	1	1	2	1	6		1	3	1	1	1	63	54	9	5		
RITA	3	2			2	11	2	1	4	1	5	2	1	1	1	14	2	1	1	1	5	1	3	2	2	1	1	70	61	9	3		
TRANSPLANTCHILD	1	2			2	2	1	6	1	2	1	1	1	1	1	5	1	1	1	1	3	1	1	3	3	3	3	40	33	7	7		
VASCERN	2	5		1		5	3		2	1	4			1	1	7	1	1		1	6	1			1	2	1	46	40	6	9		
Full Members	9	94	7	4	45	196	46	5	110	32	146	18	7	27	18	323	17	1	6	0	121	14	44	48	13	45	13	5	1414				
Affiliated Partners	35	0	1	7	0	0	4	19	0	0	0	0	16	6	0	0	14	23	18	24	0	6	1	0	2	0	10	5	1605				
Current situation	44	94	8	11	45	196	50	24	110	32	146	18	23	33	18	323	31	24	24	24	121	20	45	48	15	45	23	10	1605				
Areas not covered	0	0	0	0	13	2	0	0	5	0	0	0	12	7	0	6	0	0	0	4	0	0	7	3	3	13	0	1	1				

Advancement of Treatments FOR RARE DISEASES

ERNs integration in NHS in every MS

Knowledge, expertise, research, networking and sharing- transfer into practice



Is there a distinguish between patients and the "others"?
• We all are conditional patients

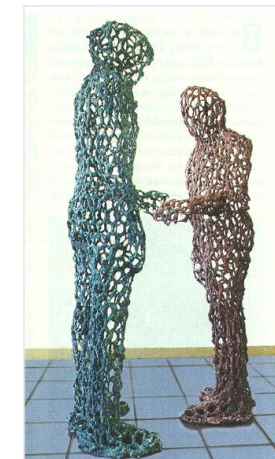
Most families need to address both common and rare or complicated health challenges

Health care economics : one single pie

Health care systems : one pie-connected jars

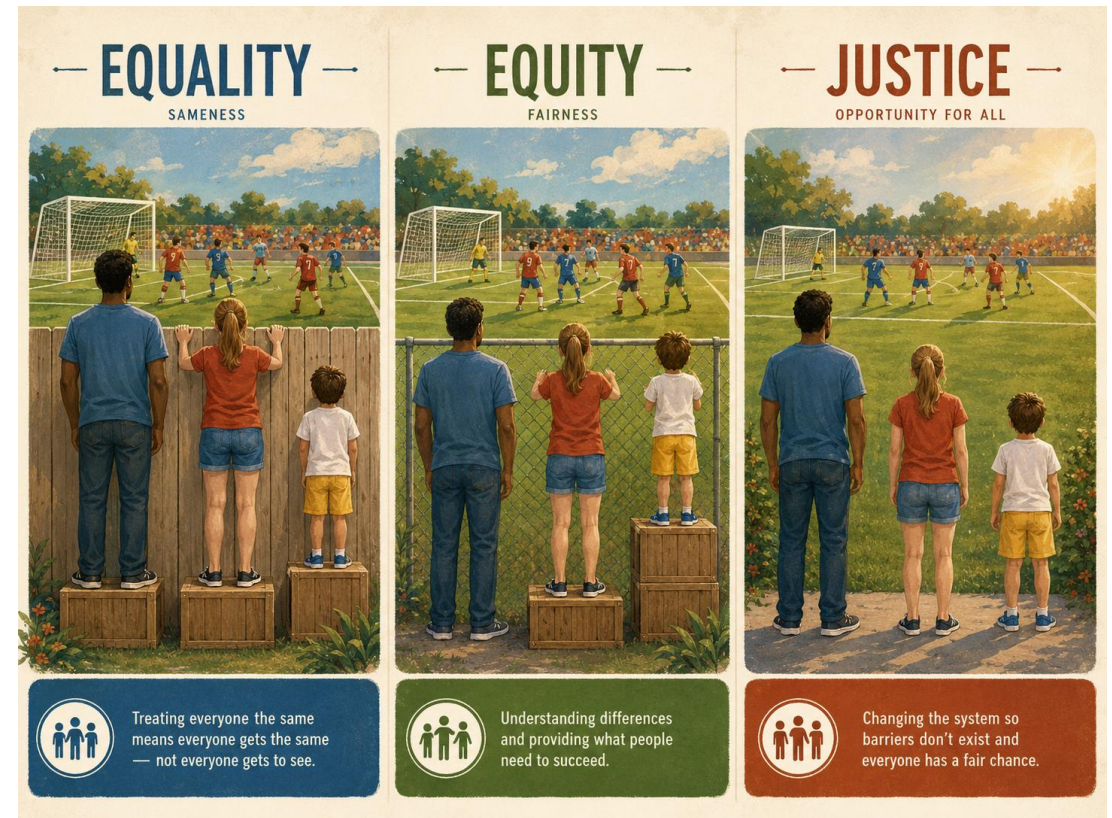
Equity: access to health care is unequal

Justice : access to health care needs be addressed with justice



EQUALITY, EQUITY AND JUSTICE

- Equality, equity, and justice
 - are foundational pillars of the European Union,
 - enshrined in Article 2 of the Treaty on European Union (TEU) and the [Charter of Fundamental Rights](#).
 - These values ensure a society defined by pluralism, non-discrimination, tolerance, and solidarity.



Equality, Equity or Justice? Is it time to reconsider?



- Rare diseases are ultra rare in underpopulated regions
- Expertise is more threatened in small countries and remote regions
- Unmet needs are larger in underpopulated regions
- Infrastructure is more restraint in small size health care systems
- Health economy is of smaller scale and more endangered
- Research faces larger and multiple obstacles



Advancement of Treatments FOR RARE DISEASES

THANK YOU!

