UNIUNEA EUROPEANĂ
Proiect cofinanțat din Fondul Social European prin Programul Operațional CAPITAL UMAN 2014 – 2020

GENETICA în lumea BOLILOR RARE

lansarea proiectului PROGEN
20 aprilie 2018
Hotel Radisson Blu – București

INSTITUTUL FUNDENI

PROGEN

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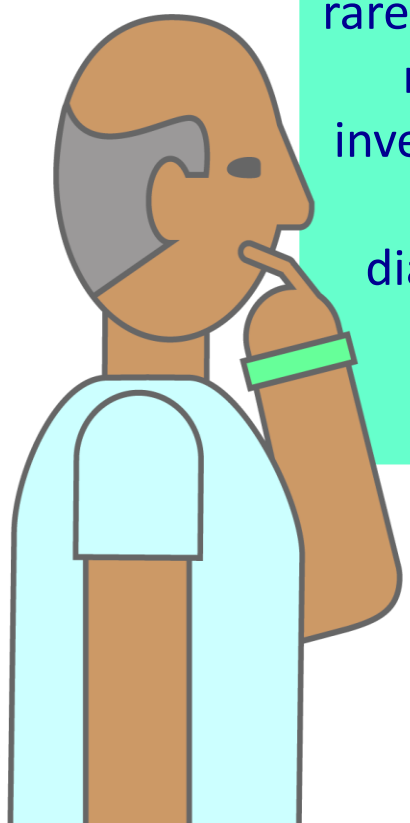
UMF- Carol Davila, București

BOLILE RARE : mit sau realitate ?



MIT

Bolile rare sunt atât de rare încât nu merită (este neprofitabil) să se investească niciun fel de efort pentru a le diagnostica, trata sau preveni.

REALITATE

Numărul de BR este estimat ~ 8000 de tipuri diferite; ca grup, diagnosticarea și managementul reprezintă o provocare medicală, plus lipsa stimulentelor financiare pentru dezv. și comercializarea medicamentelor orfane. PROBLEMA MAJORĂ DE SĂNĂTATE PUBLICĂ ÎN ÎNTREGA LUME!

http://www.thetgmi.org/genetics/how-many-rare-diseases-are-genetic/

CE SUNT BOLILE RARE ?

BR reprezintă o amenințare pentru sănătatea cetățenilor UE în măsura în care ele:

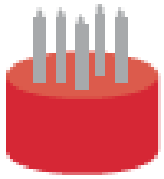
- sunt boli care pun în pericol viața sau
- provoacă o invaliditate cronică,
- având o prevalență redusă și un grad ridicat de complexitate.

În ciuda raritații acestora, există atât de multe tipuri diferite de boli rare, încât milioane de oameni sunt afectați.

BOLILE RARE : CIFRE și DATE

”Individually are rare, collectively are common”

- It is estimated that **350 million people worldwide** suffer from rare diseases



- About **30% of children** with these debilitating diseases **will not live** to see their 5th birthday

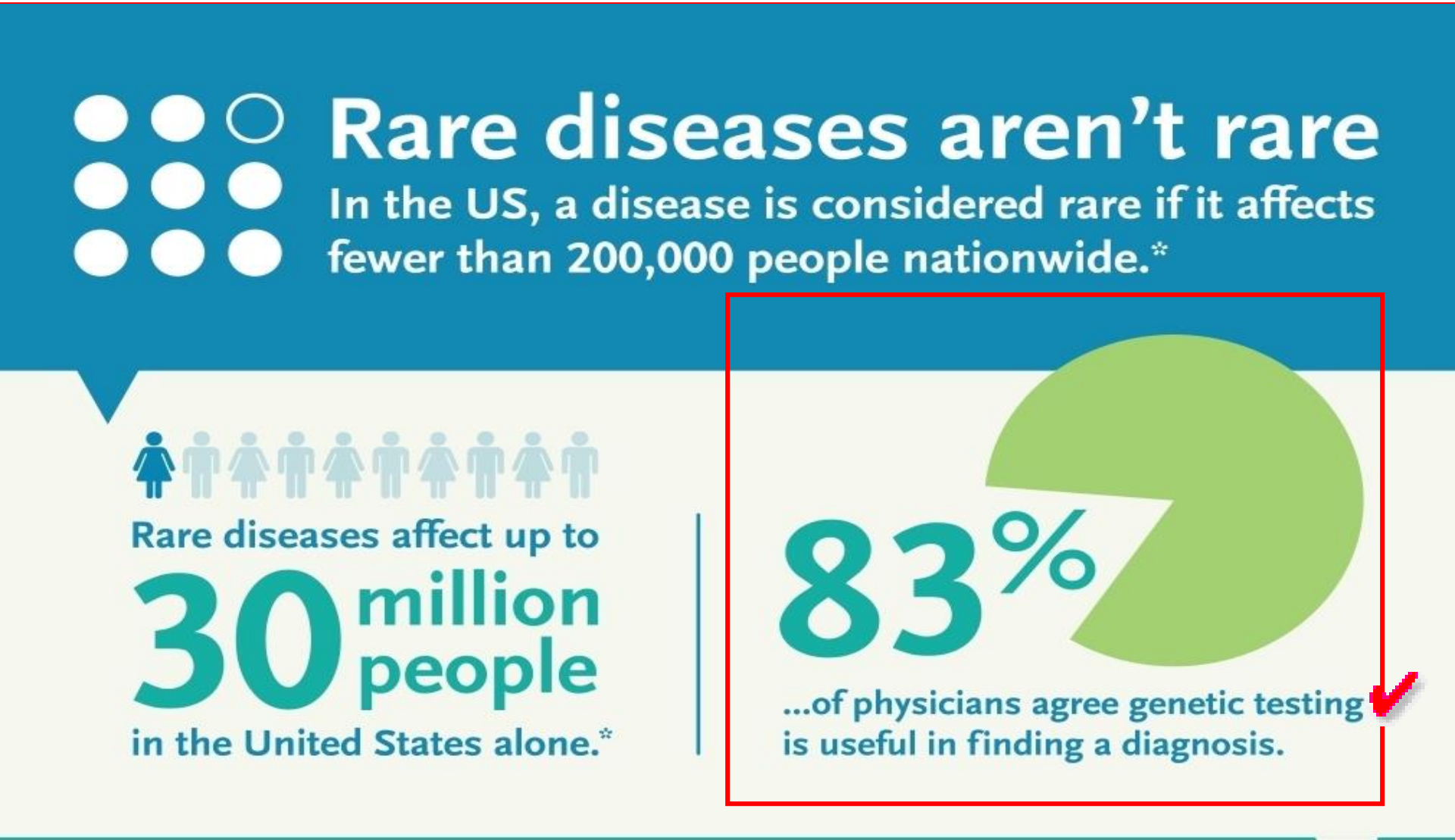
- About **80%** of rare diseases are genetic

- Rare diseases **impact more people** than AIDS or cancer combined

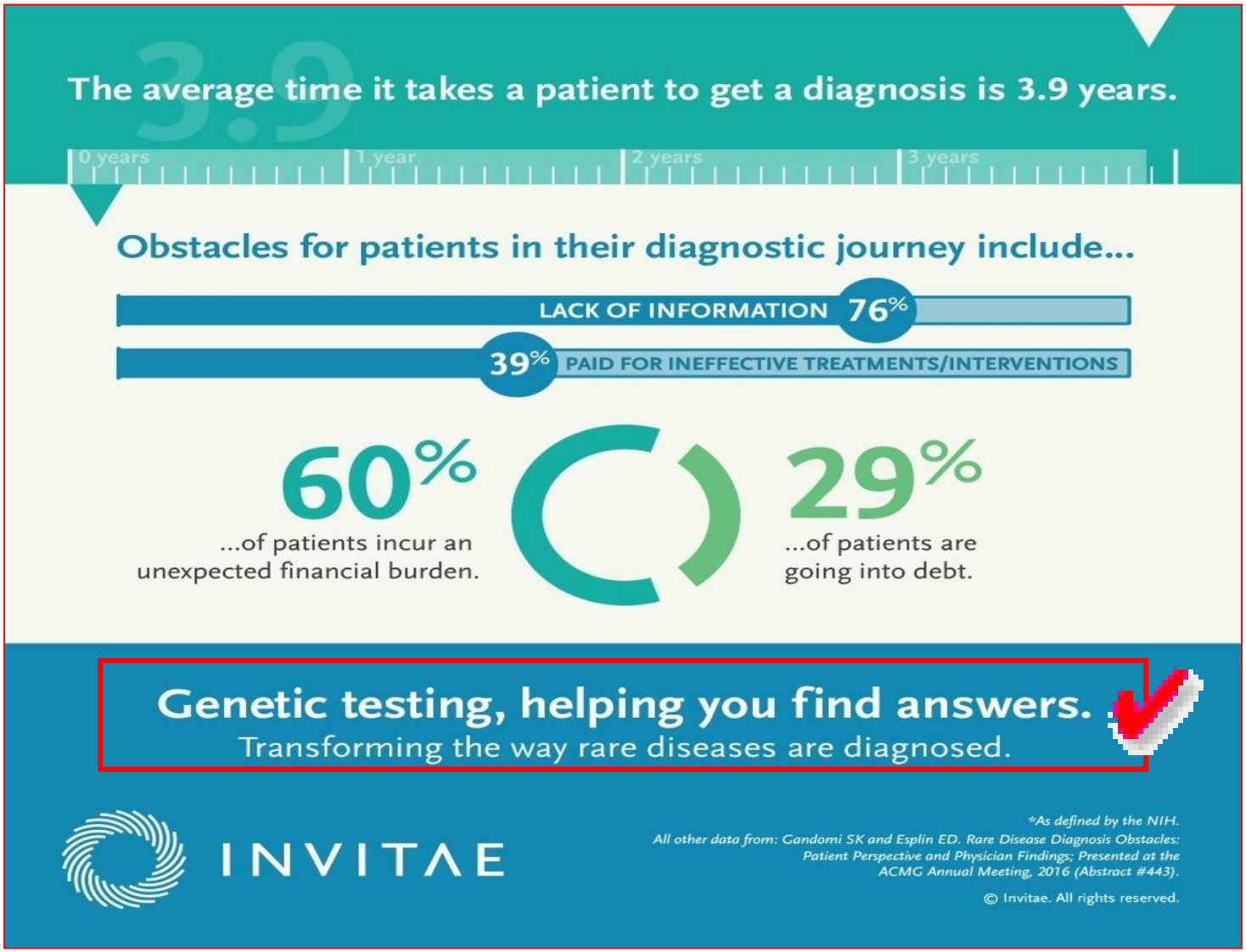
- **95% of rare diseases** do not have a single FDA-approved drug treatment

Source: Global Genes

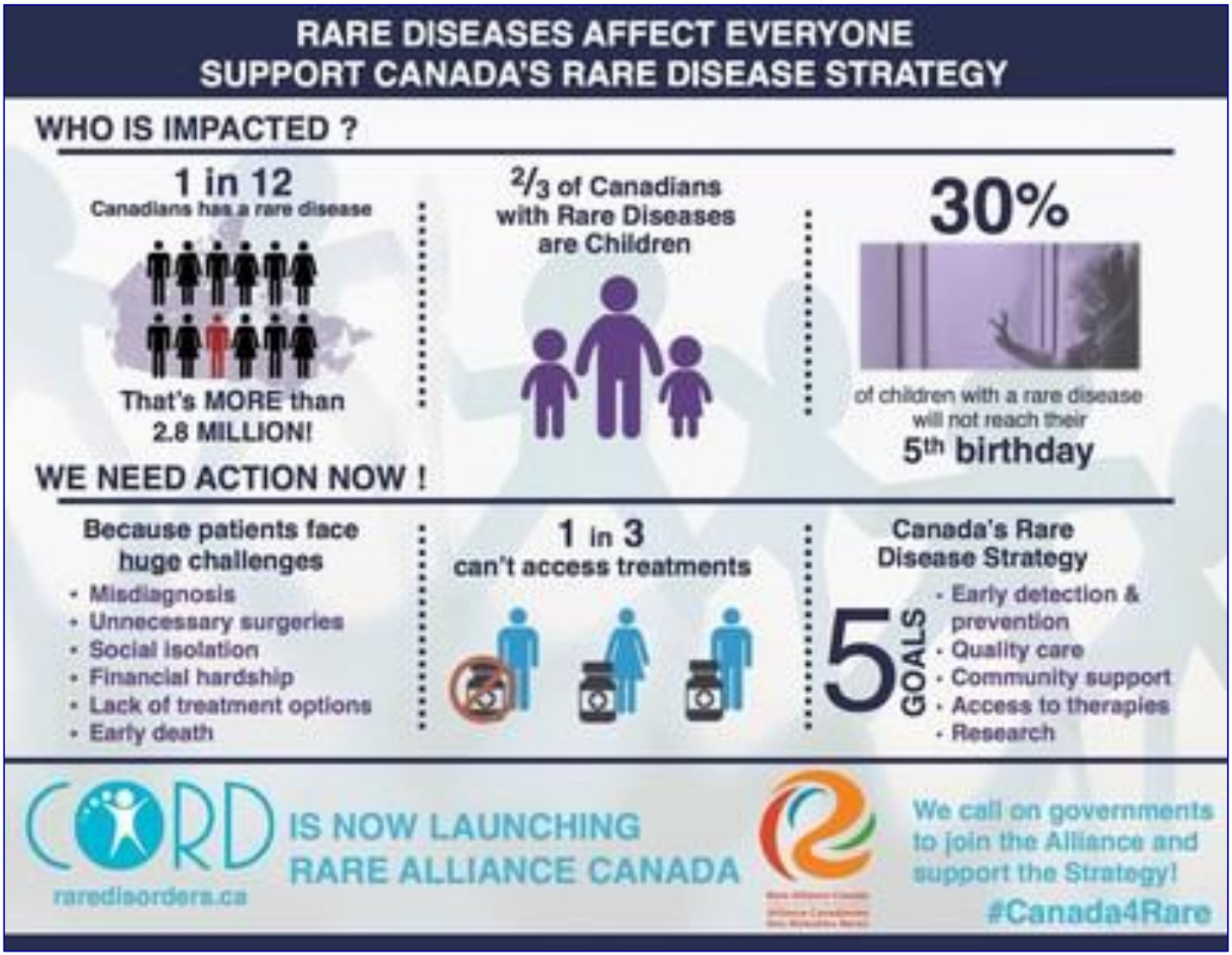
BOLILE RARE : CIFRE și DATE



BOLILE RARE : CIFRE și DATE



BOLILE RARE în CANADA



SINGAPORE JAPAN AUSTRALIA THE EU TAIWAN SOUTH KOREA

THESE COUNTRIES IMPLEMENTED ORPHAN DRUG LEGISLATION AFTER THE US PASSED THE ORPHAN DRUG ACT

cadth.ca/drugs-rare-diseases-evolving-trends-regulatory-and-health-technology-assessment-perspectives

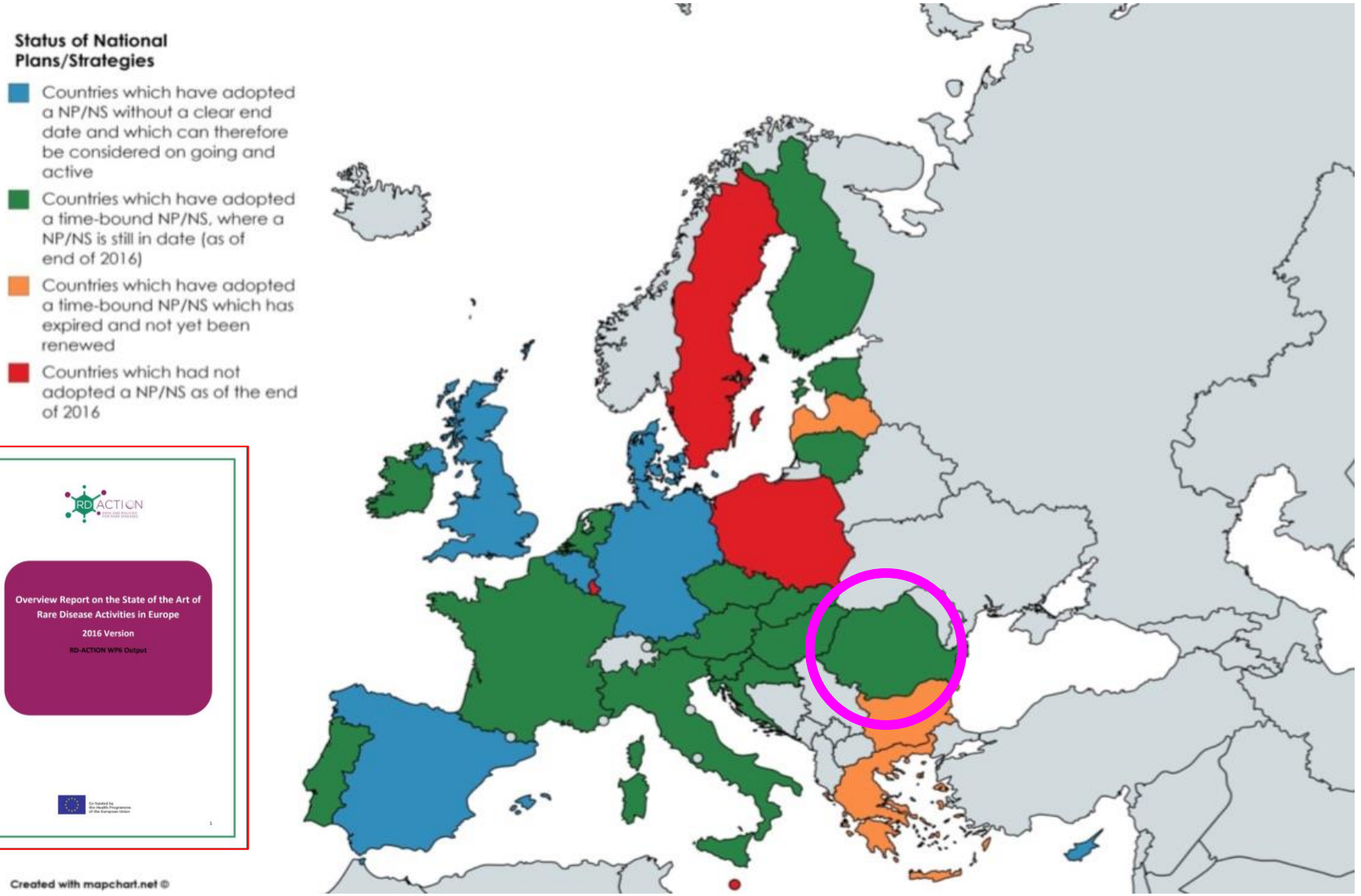
DE CE SUNT SPECIFICE BOLILE RARE ?

- BR sunt o problemă de sănătate publică pretutindeni în lume (una dintre cele mai mari comunități de pacienți din lume);
- Persoanele cu BR au NEVOI SPECIALE;
- NECESITĂ îngrijiri medicale ultraspecializate (de înaltă calitate și sigure), servicii sociale și sprijin specific;

BR sunt o PRIORITATE

în strategiile de sănătate naționale dar și o STRATEGIE GLOBALĂ bazată pe cooperare, colaborare și reglementare la nivel mondial (reunirea expertizei și utilizarea eficientă a resurselor disponibile).

BOLILE RARE în EUROPA



POLITICA de sănătate europeană pentru bolile rare

SCOP:

1. reducerea morbidităţii semnificative şi evitarea unei mortalităţi premature;
2. îmbunătăţirea calităţii vieţii pacienţilor cu BR;
3. îmbunătăţirea potenţialului socio-economic al persoanelor afectate.

POLITICA de sănătate europeană pentru bolile rare

ACTIVITĂȚI PRIORITARE:

1. DEFINIREA, CODIFICAREA ȘI INVENTARIEREA ADECVATE BOLILOR RARE
2. PLANURI ȘI STRATEGII NATIONALE ÎN DOMENIUL BOLILOR RARE
3. CERCETAREA ȘTIINȚIFICĂ ÎN DOMENIUL BOLILE RARE

POLITICA de sănătate europeană pentru bolile rare

ACTIVITĂȚI PRIORITARE:

4.	REUNIREA EXPERTIZEI ÎN DOMENIUL BOLILOR RARE LA NIVEL EUROPEAN
5.	RESPONSABILIZAREA ORGANIZAȚIILOR DE PACIENȚI
6.	CENTRE DE EXPERTIZĂ ȘI REȚELE EUROPENE DE REFERINȚĂ PENTRU BOLILE RARE
7.	DURABILITATE

INTEGRATED CARE FOR RARE DISEASES:
BRIDGING THE GAP TO IMPROVE THE LIVES OF 30 MILLION PEOPLE IN EUROPE

RARE DISEASES

Rare, complex, chronic, disabling

30 million people affected in Europe

Fewer than **1 in 2000** people affected by each rare disease

No cure for the vast majority and few treatments available

Expertise and knowledge on their consequences are scarce and difficult to access

PEOPLE LIVING WITH A RARE DISEASE AND THEIR CARERS FACE SERIOUS CARE BURDEN[1]

65% have to visit different health, social and local services in a short period of time

67% feel that these services communicate badly between each other

7in10 do not feel well informed about their rights

7in10 find that organising care is time-consuming; 6 in 10 find it hard to manage

INTEGRATED HEALTH AND SOCIAL CARE IS ESSENTIAL TO ENABLE PEOPLE LIVING WITH A RARE DISEASE TO

hope. it's in our genes.™

The technological push toward reforming a healthcare system to adopt the ethos of personalized medicine—that the right drug be given to the right patient at the right time.

PROACTIVE VERSUS REACTIVE MEDICINE

"Genomics and Personalized Medicine Empower People to Save Their Own Lives"

OMIM

OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh. Its official home is omim.org.

January 7th,2017

4,919 confirmed Genetic disorders

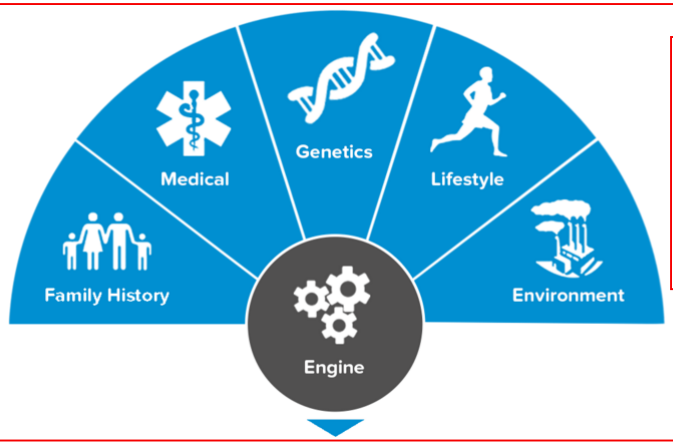
(phenotype and known gene)

Heterogeneous disorders are broken down into subtypes for each different gene. Growing focus on genetic subtypes, because many new therapies are gene-specific, or even mutation specific.

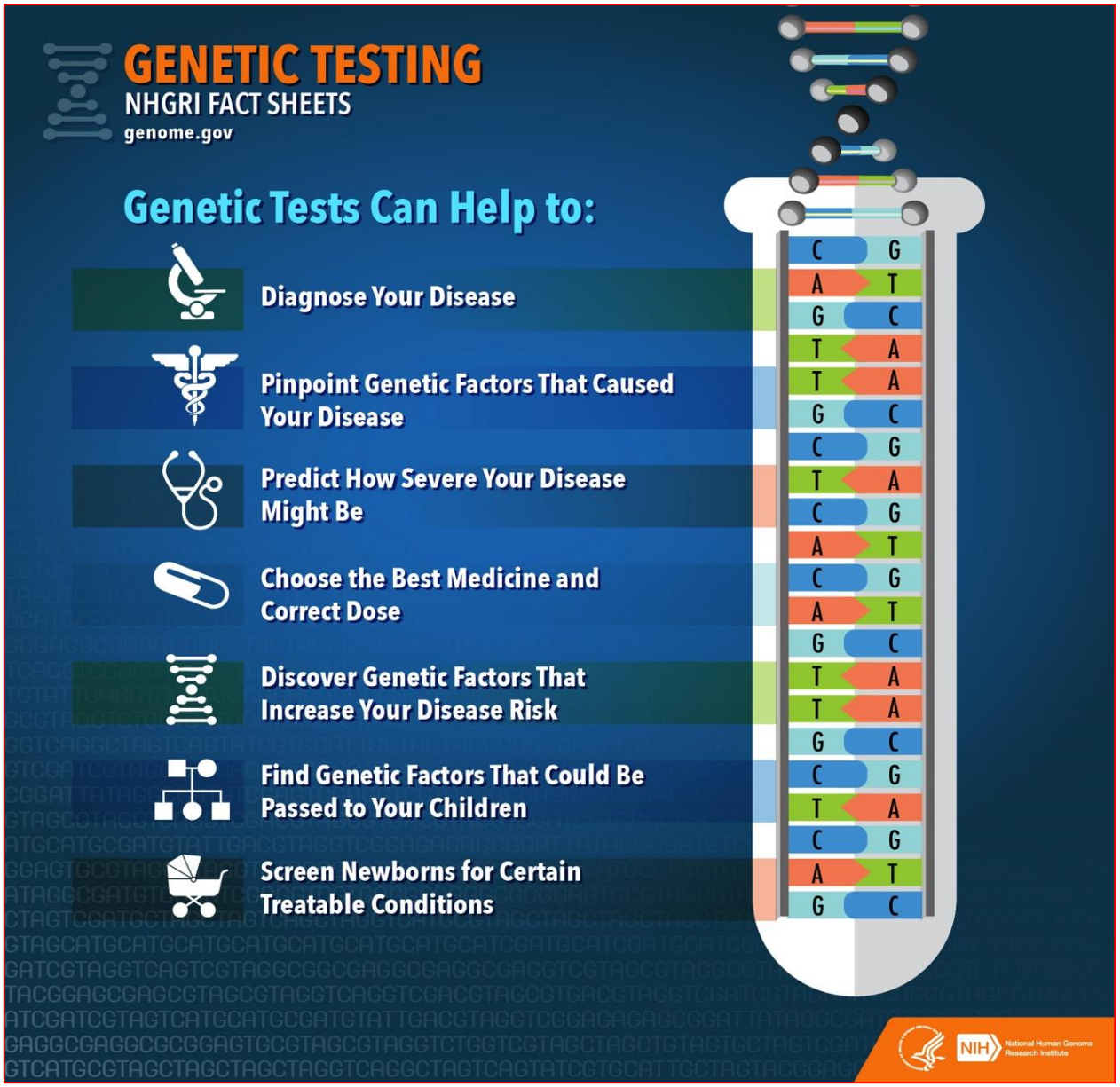
"individual's genetic profile to guide decisions made in regard to the prevention, diagnosis, and treatment of disease."

Genomic Sequencing: A laboratory method that is used to determine the entire genetic makeup of a specific organism or cell type. This method can be used to find changes in areas of the genome that may be important in the development of specific diseases, such as cancer.

Personalized Medicine: Sometimes referred to as precision or individualized medicine, personalized medicine, is an emerging field of medicine that uses diagnostic tools to identify specific biological markers, often genetic, to help assess which medical treatments and procedures will be best for each patient.



Approaches will be effective based on GENETIC, environmental, and lifestyle factors.



National Human Genome Research Institute

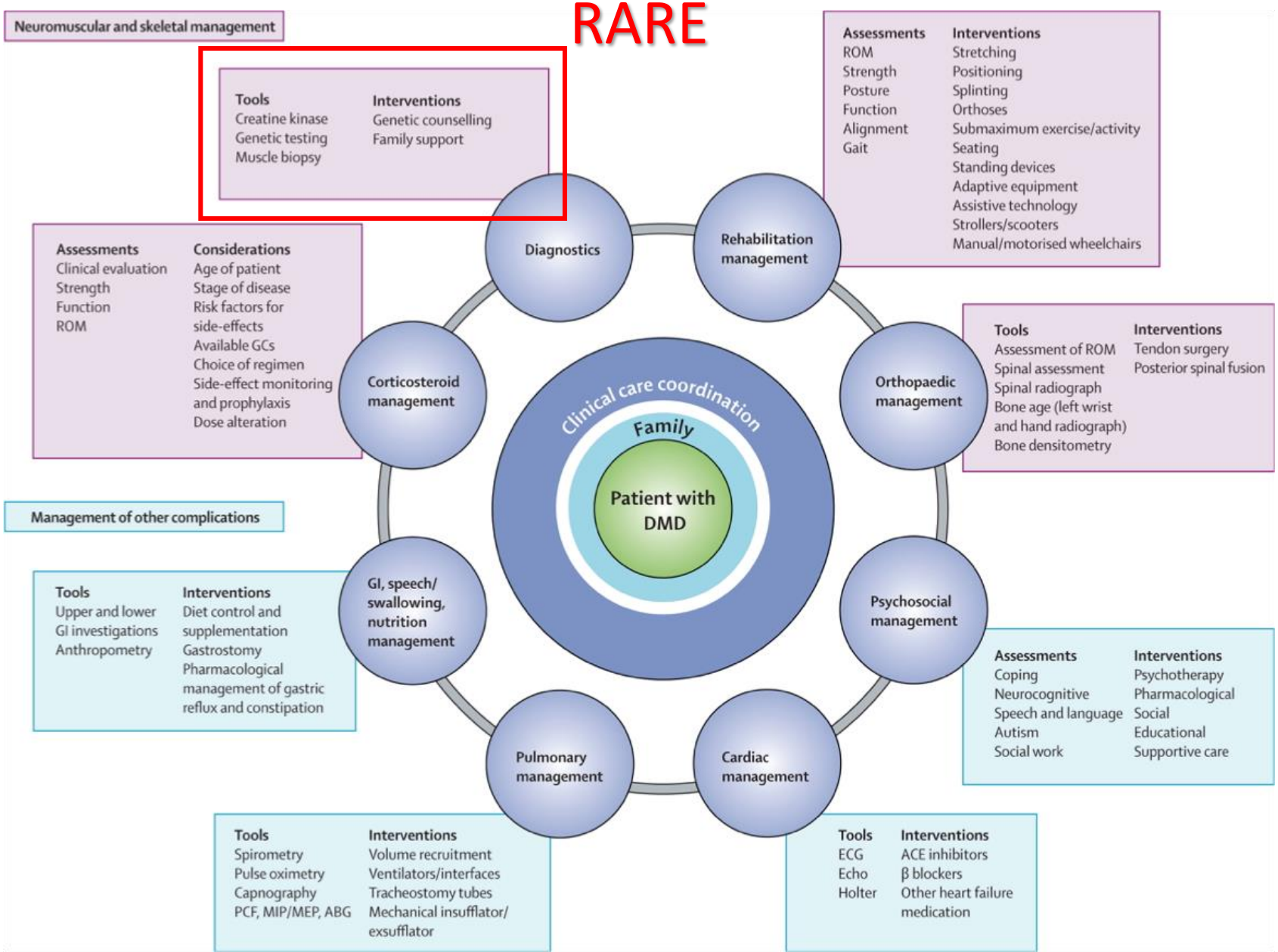
7 REASONS TO USE GENETIC TESTING:

1. Diagnostic
2. Identificarea cu precizie a factorilor genetici care cauzează o boală
3. Predicția severității bolii
4. Alegerea corectă a medicamentului și dozei
5. Detectarea factorilor genetici care cresc riscul de îmbolnăvire
6. Factorii genetici de risc pentru descendenți
7. Screening-ul NN pentru afecțiuni tratabile.

Choosing the right DNA test for your needs

Discuss your DNA questions with your healthcare provider he/she may be able to guide you in your kit considerations, and those surrounding chronic disease. Consider the financial cost, benefits, limits. Be mindful of the significance and impact the results will have on you and others.

ABORDAREA MULTIDISCIPLINARĂ A BOLILOR RARE



Patients with rare genetic diseases are among the first beneficiaries of the NGS revolution

Translation of next-generation sequencing into clinical diagnostics for patients with rare diseases

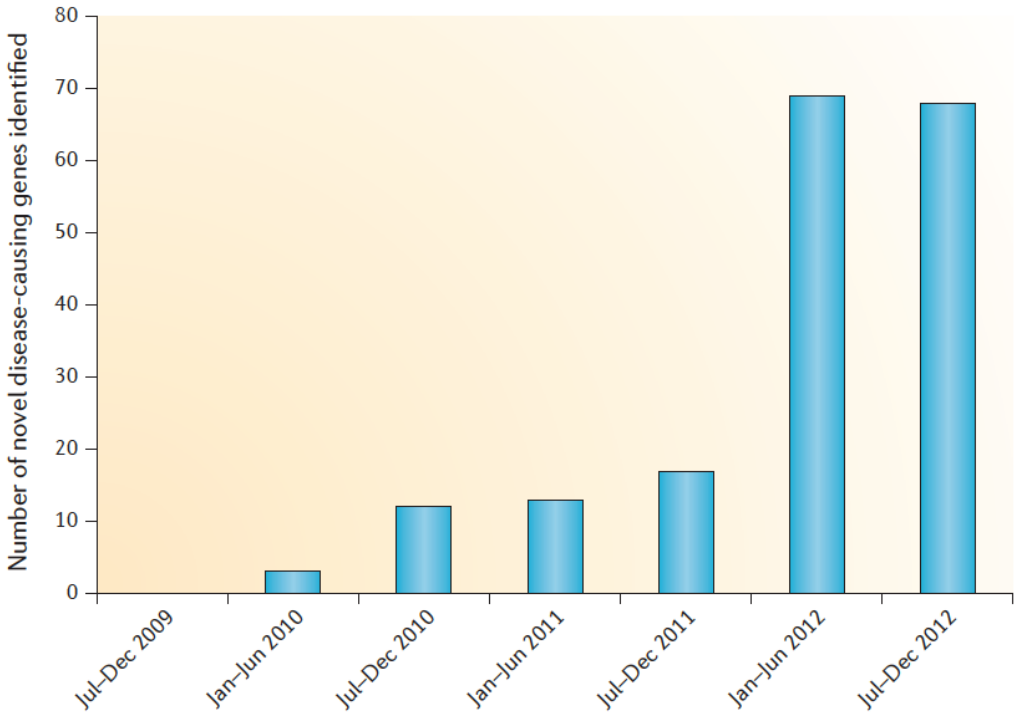


Figure 1 | **Pace of discovery of novel rare-disease-causing genes using whole-exome sequencing.** Since the first whole-exome sequencing (WES) proof-of-concept experiment[11], the discovery of disease-causing genes using WES has increased rapidly, with a marked jump from 2011 to 2012 and a stabilization of this rate in the latter half of 2012. More than 180 novel genes have been discovered in this manner. As next-generation sequencing technology becomes less costly and more widely used, we anticipate another jump in the rate of discovery, provided that the infrastructure for the large-scale sharing of deep phenotypic and genetic data sets emerges.

K.M.Boycott et al. ,2013

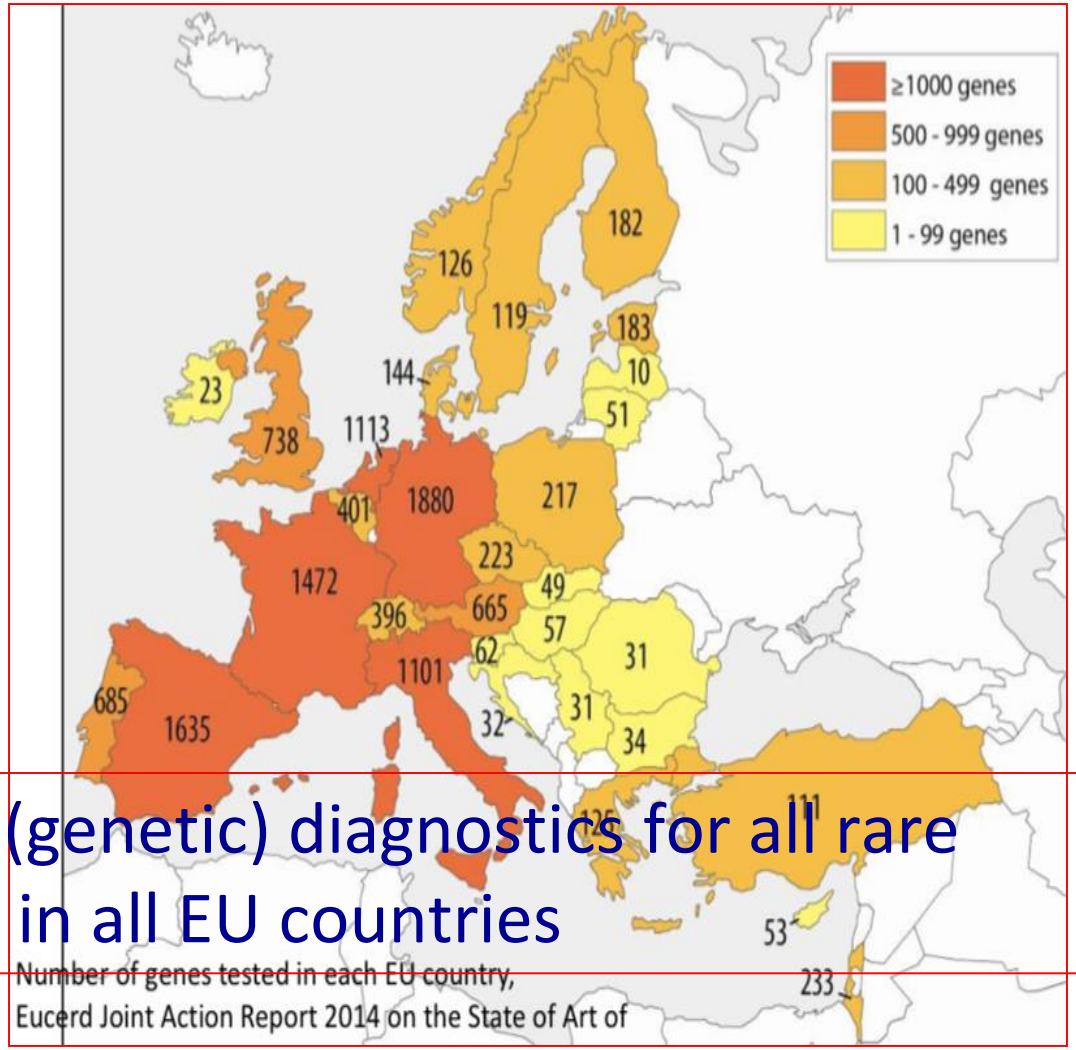
CROSS BORDER GENETIC TESTING OF RARE DISEASES

Commission Expert Group on Rare Diseases

Recommendation on

CROSS BORDER GENETIC TESTING OF RARE DISEASES IN THE EUROPEAN UNION

13 November 2015



Number of genes tested in each EU country, Eucerd Joint Action Report 2014 on the State of Art of

To improve access to (genetic) diagnostics for all rare diseases in all EU countries

CONCLUSIONS

R.A.R.E.

RARE DISEASE. AWARENESS. RESEARCH. EDUCATON.

A DISEASE MAY BE RARE
BUT HOPE SHOULD NOT BE

=

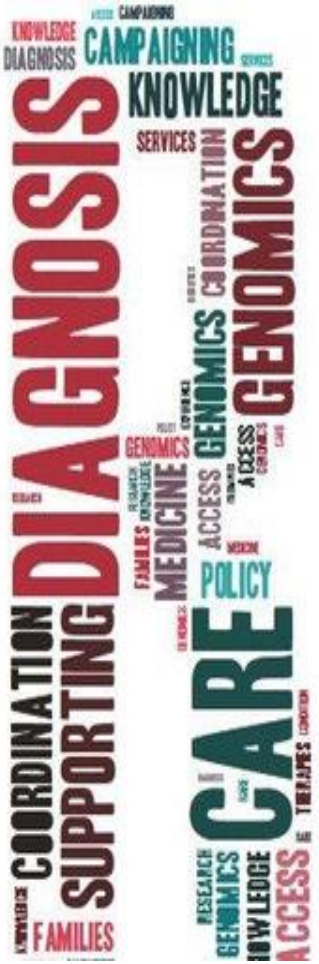

EURORDIS
Rare Diseases Europe
10 000 thanks!!!

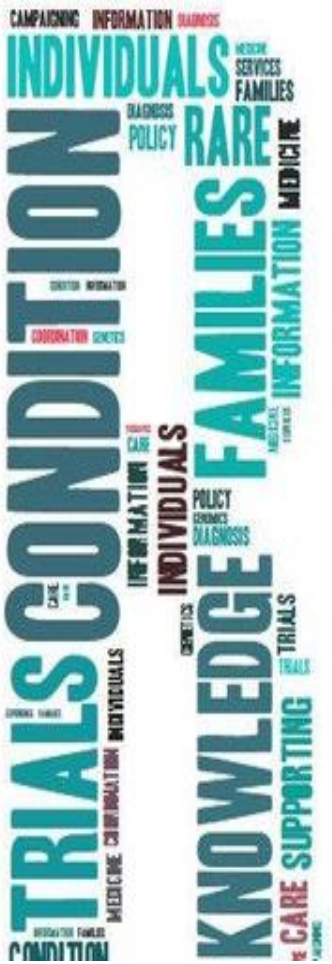
PROGEN

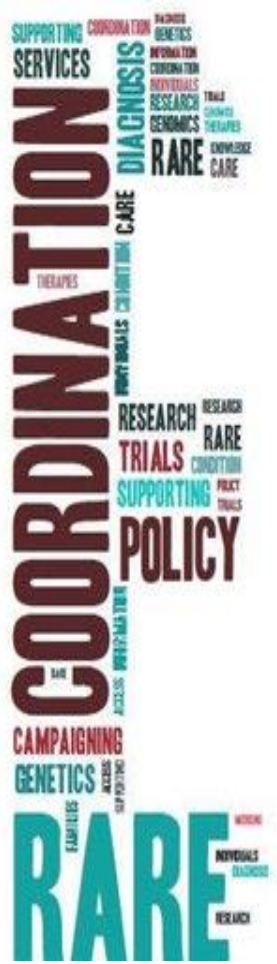
UNIUNEA EUROPEANĂ

Instrumente Structurale 2014-2020

Proiect cofinanțat din Fondul Social European prin Programul Operațional CAPITAL UMAN 2014 – 2020







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