ORPHANET CONTRIBUTION TO IMROVING RARE DISEASE DIAGNOSTIC PATH

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THE RARE DISEASES DIAGNOSIS PYRAMID

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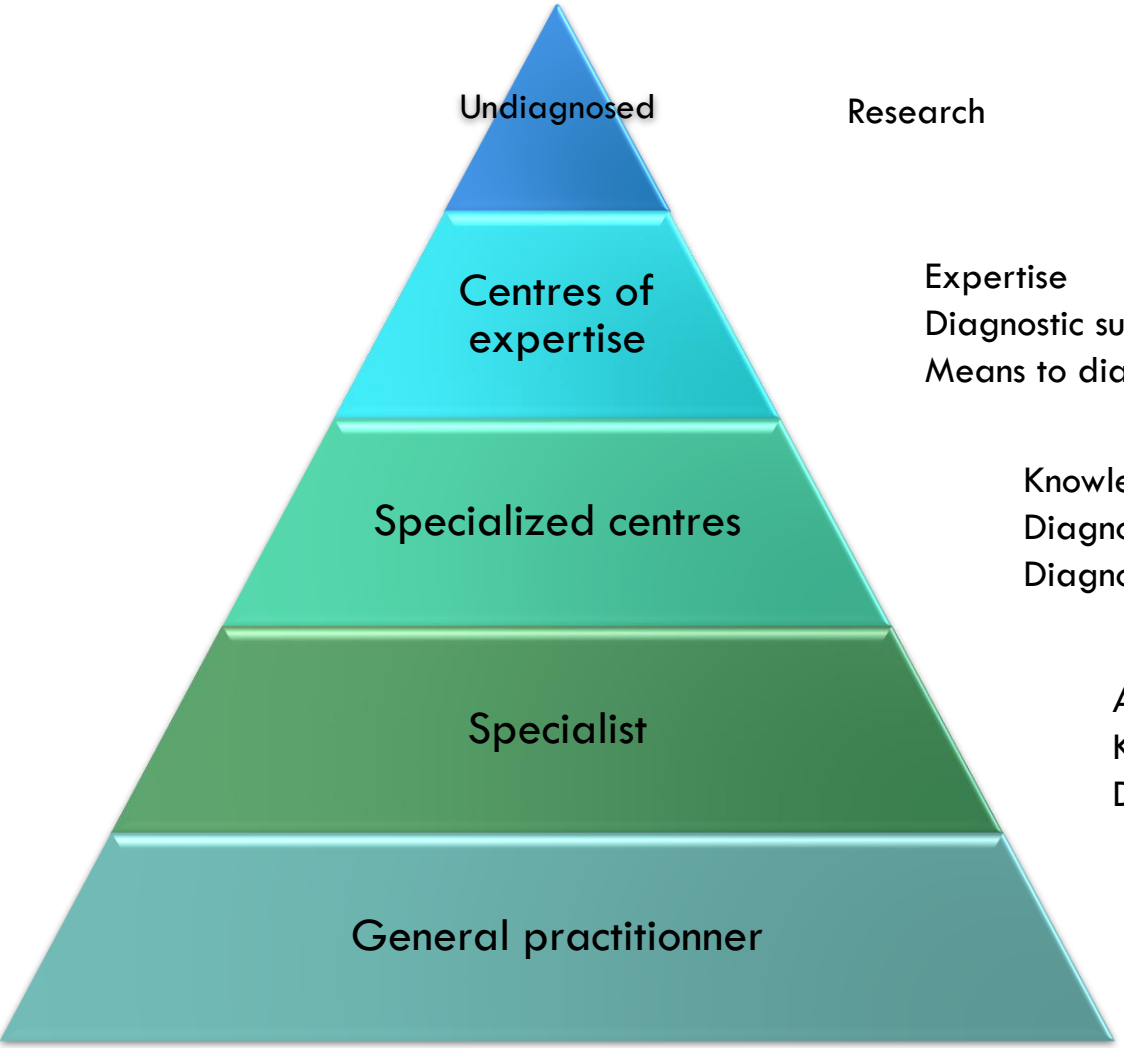

Research

Expertise
Diagnostic suspicion
Means to diagnose: NGS…

Knowledge
Diagnostic suspicion
Diagnosis

Awareness
Knowledge
Diagnostic suspicion?

Awareness
Knowledge

Integrated data for research

Diagnostic labs & tests

Best practices

Find an expert

Information

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Orphanet is 25!

ORPHANET'S MISSIONS

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Improve the **visibility** of RD by providing **a common language** across healthcare and research systems (ORPHAcodes)

Provide **high-quality information** and **expertise** on RD

Contribute to **generating knowledge**

→ piecing together the parts of the puzzle for better understanding of RD

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ORPHANET IS A NETWORK OF INSTITUTIONS ENDORSED BY MOH

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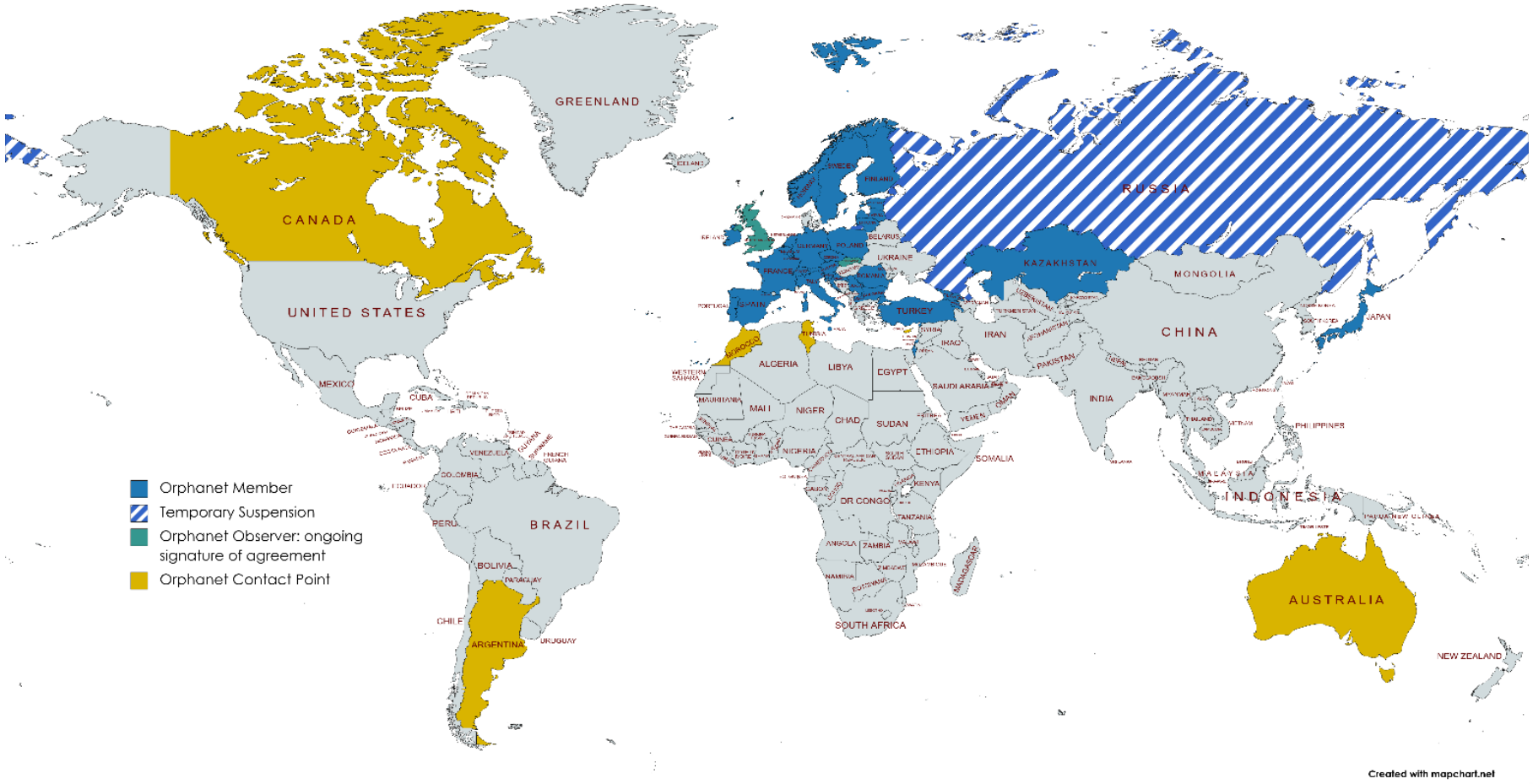

Co-funded by
the Health Programme
of the European Union

A global network improving **visibility**, **awareness**, **information** and **knowledge** in the field of rare diseases

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GIVING A NAME FOR ALL RARE DISEASES

ORPHA CODES

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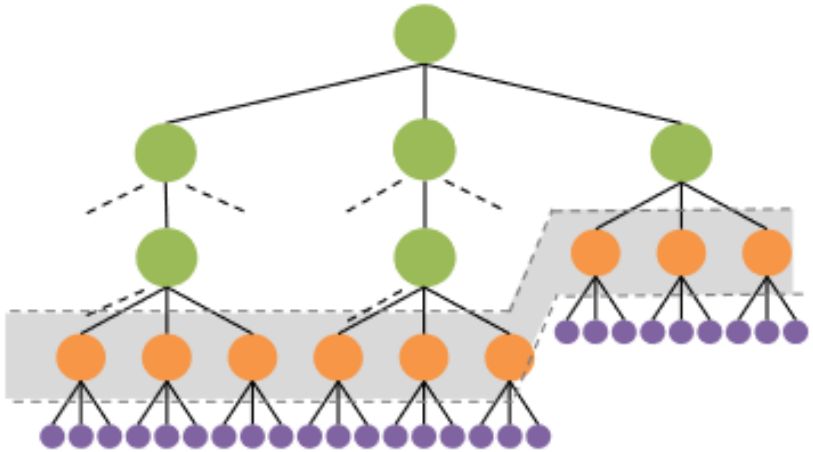
A MEDICAL TERMINOLOGY SPECIFIC TO RARE DISEASES (<1 in 2000 cases)

Clinical definition:

Disorders are clinically homogeneous entities described in at least two independent individuals, confirming that the clinical signs are not associated by fortuity.



Group

Disorder

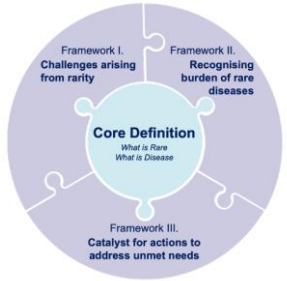
Subtype

9,339 clinical entities

2,097 groups

6,227 disorders

1,014 subtypes



"Classification level"

ENG FR SP DE IT NL PG PL RU CHN TUR

Comprehensive, standardized, evidence-based, interoperable, versioned, computable and free (CC-BY 4.0)

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. . . INCLUDING UNDIAGNOSED RD

Rare disorder without a determined diagnosis after full investigation

Suggest an update

Disease definition

A rare disorder for which all reasonable efforts have been done by rare diseases experts to determine a diagnosis according to the state of the art and available diagnostic capabilities, but did not enable to conclude on a clinically known concept. It is recommended to restrict the use of this entity for coding purposes to rare disease experts.

ORPHA:616874

Classification level: Disorder

Synonym(s):

Fully investigated rare disorder without a determined diagnosis

Prevalence: -

Inheritance: -

Age of onset: -

ICD-10: -

OMIM: -

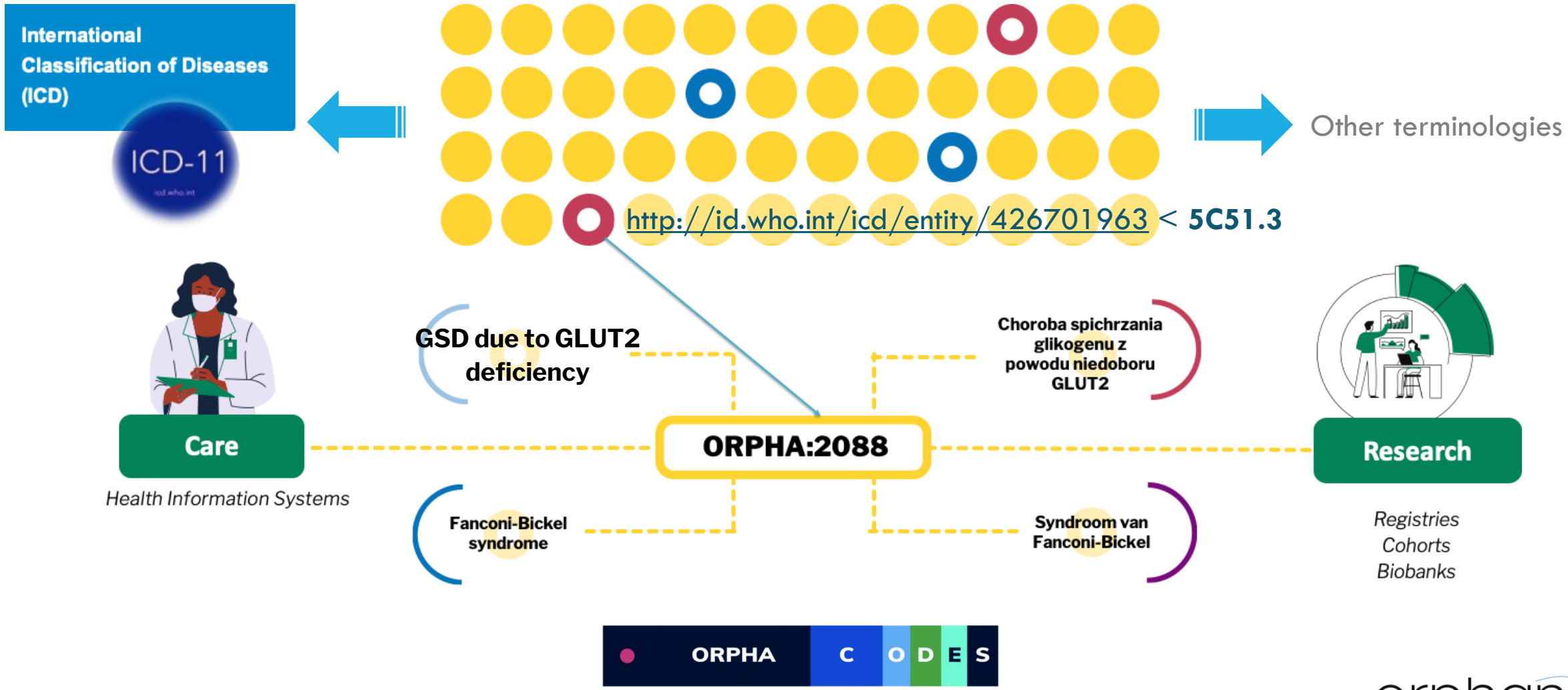
UMLS: -

MeSH: -

GARD: -

MedDRA: -

TOWARDS GENERAL SEMANTIC INTEROPERABILITY FOR RD



INFORMING ABOUT EACH RARE DISEASE

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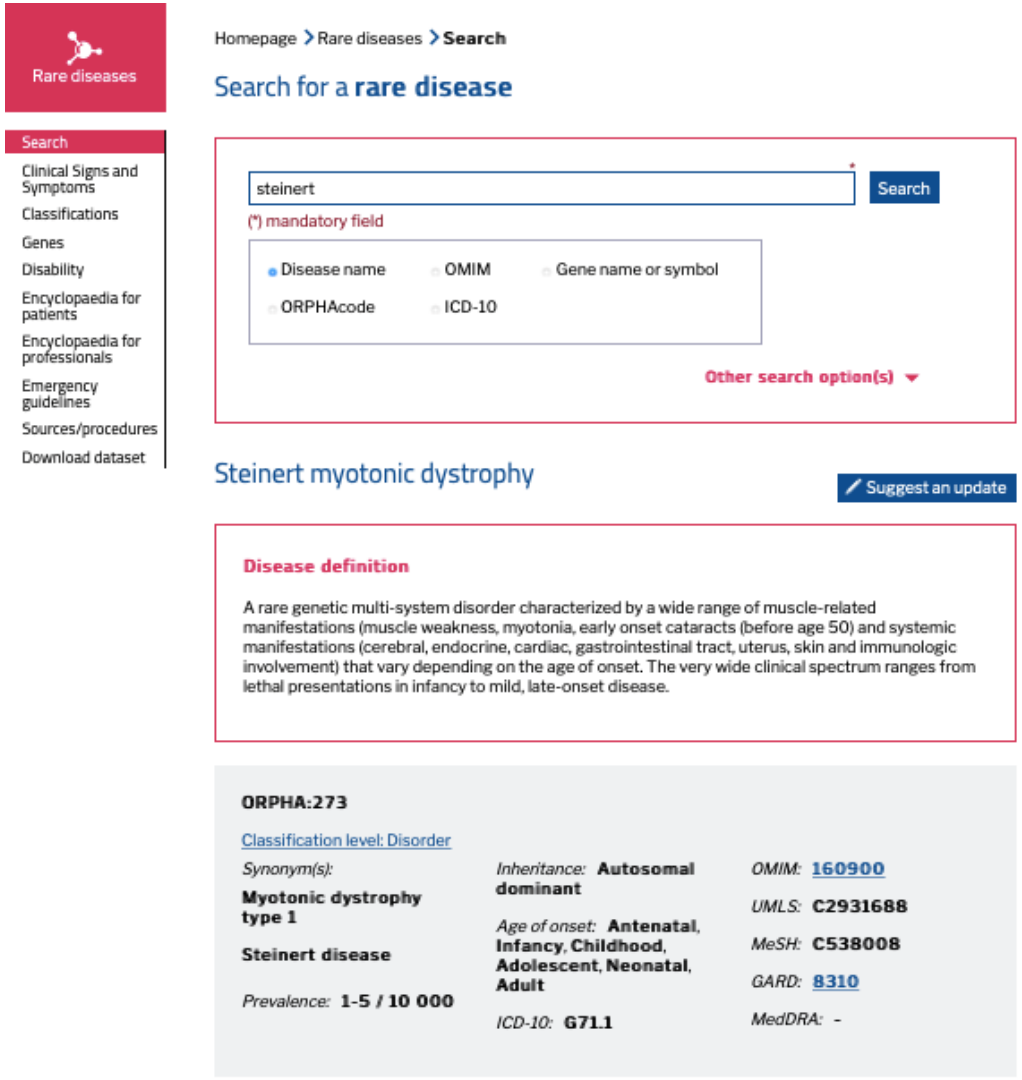

www.orpha.net

9 languages

Detailed information

Article for general public

Français (2006, pdf)
Svenska (2015)
Italiano (2009, pdf)
Español (2012, pdf)

Professionals

› Emergency guidelines
Español (2014, pdf)
Deutsch (2010, pdf)
Polski (2010, pdf)
Italiano (2010, pdf)
English (2010, pdf)
Français (2010, pdf)

› Anesthesia guidelines
Czech (2014)
Deutsch (2014)
English (2014)
Español (2014)

› Review article
Français (2007, pdf)

› Clinical practice guidelines
Español (2019)
Deutsch (2012)

› Guidance for genetic testing
Français (2016, pdf)

› Clinical genetics review
English (2021)

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Portál pro vzácná onemocnění a léčiva pro vzácná onemocnění

*" Jedním vzácným onemocněním trpí vždy jen hrstka lidí, ale **dohromady je pacientů se vzácným onemocněním mnoho** "*

Přístup k našim službám

- Seznam, klasifikace a encyklopedie vzácných onemocnění se souvisejícími geny
- Seznam léčivých přípravků pro vzácná onemocnění
- Adresář pacientských organizací
- Adresář odborníků a institucí
- Adresář specializovaných center
- Adresář lékařských laboratoří poskytujících diagnostické testy
- Adresář probíhajících výzkumných projektů, klinických hodnocení, registrů a biobank
- Sbírka tematických reportů: Série reportů Orphanet

Vyhledat onemocnění

Hledat

INFORMING ON WHERE EXPERTISE IS FOR EACH RARE DISEASE

Figure 9 Directory of expert services (January 2020)

European Reference Networks

PROVIDING AN ADDED-VALUE KNOWLEDGE BASE FOR HEALTH AND RESEARCH

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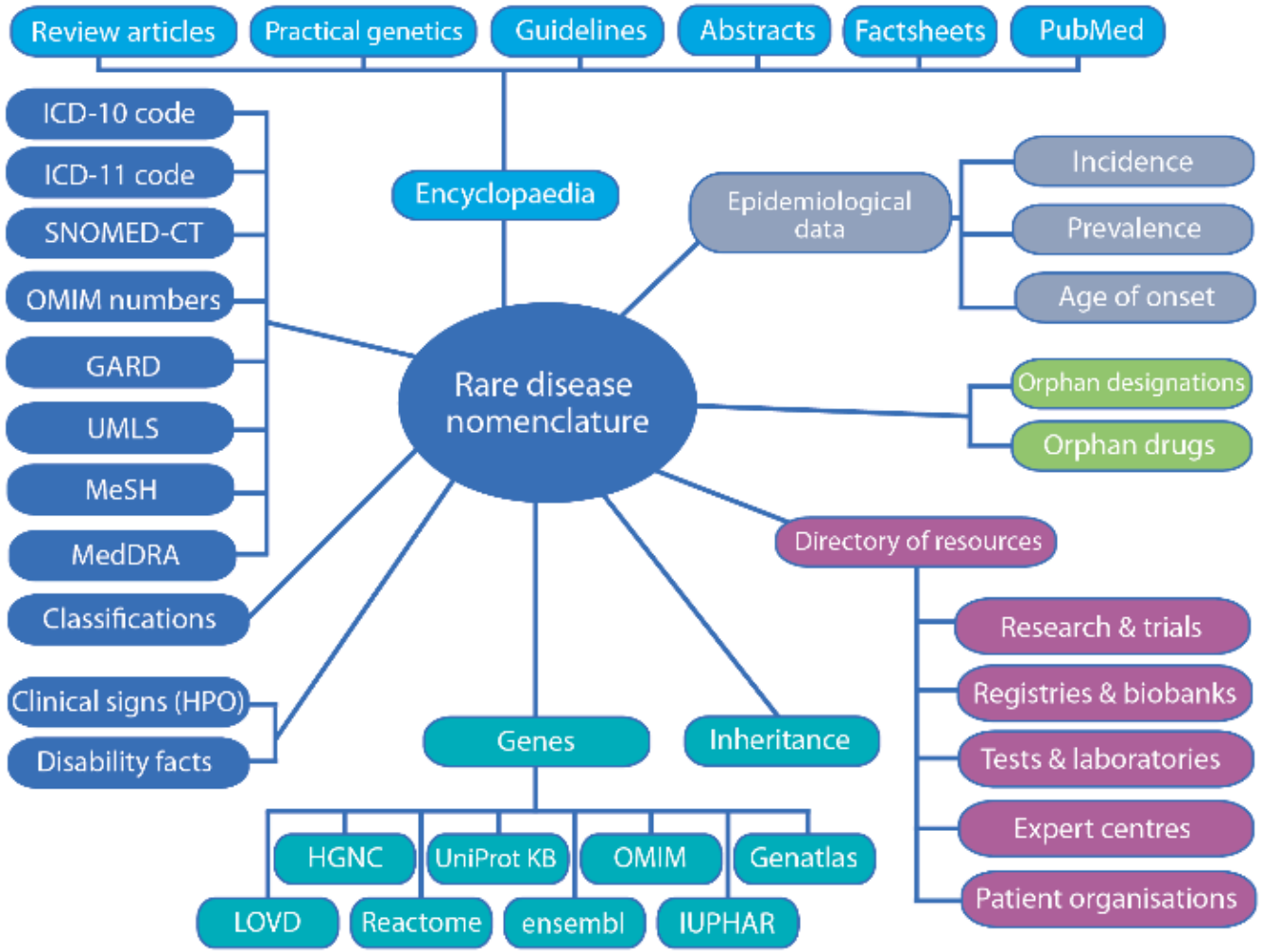

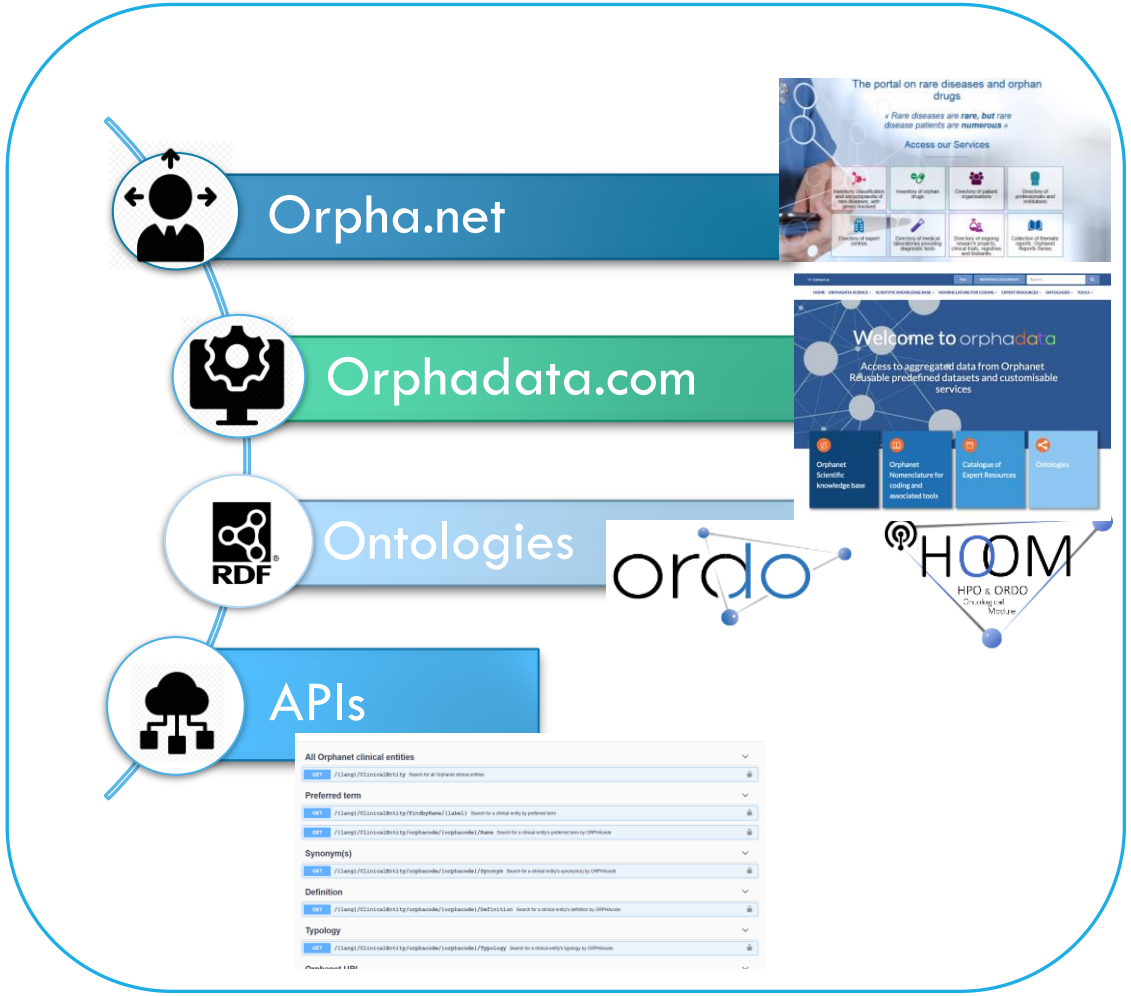

DATA FOR HELPING SOLVING THE UNSOLVED

SolveRD Cases
(Phenopackets)

{JSON}

XML

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hpo

Orphanet Data
with HPO
annotations

Similarity
computations
(8 algorithms)

Rare Diseases Cases
Ontology
(RDCO)

blazegraph

Cystoscape
Clustering

Cases-**ORPHA** Networks

GENTURIS RND
ITHACA RND-NMD
ORPHA

cnag

CRG

Cases-Cases Networks

WHAT IS FORTHCOMING

- Orphanet is preparing a dedicated webpage to gather published evidence
- On NBS for RD
- For which a NBS programme exists in European countries
- Based on a continuous literature survey

The need to document and identify good practice in existing national NBS programmes so that these lessons may be promoted and adopted more widely.

A EUROPEAN TOOL FOR ALL STAKEHOLDERS

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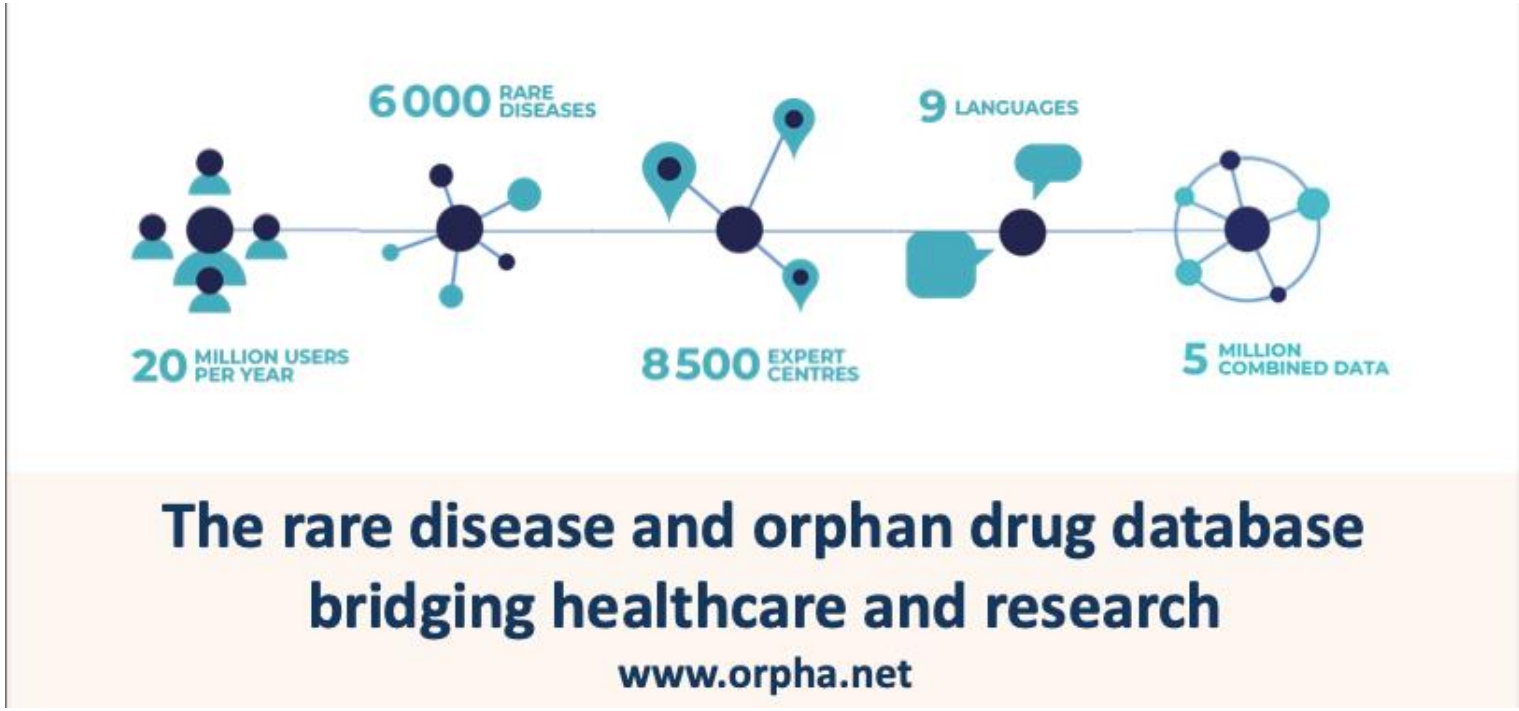

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THANK YOU!

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KNOW THE RARE

FOR BETTER CARE