



L'action d'Orphanet, base de données internationale des maladies rares

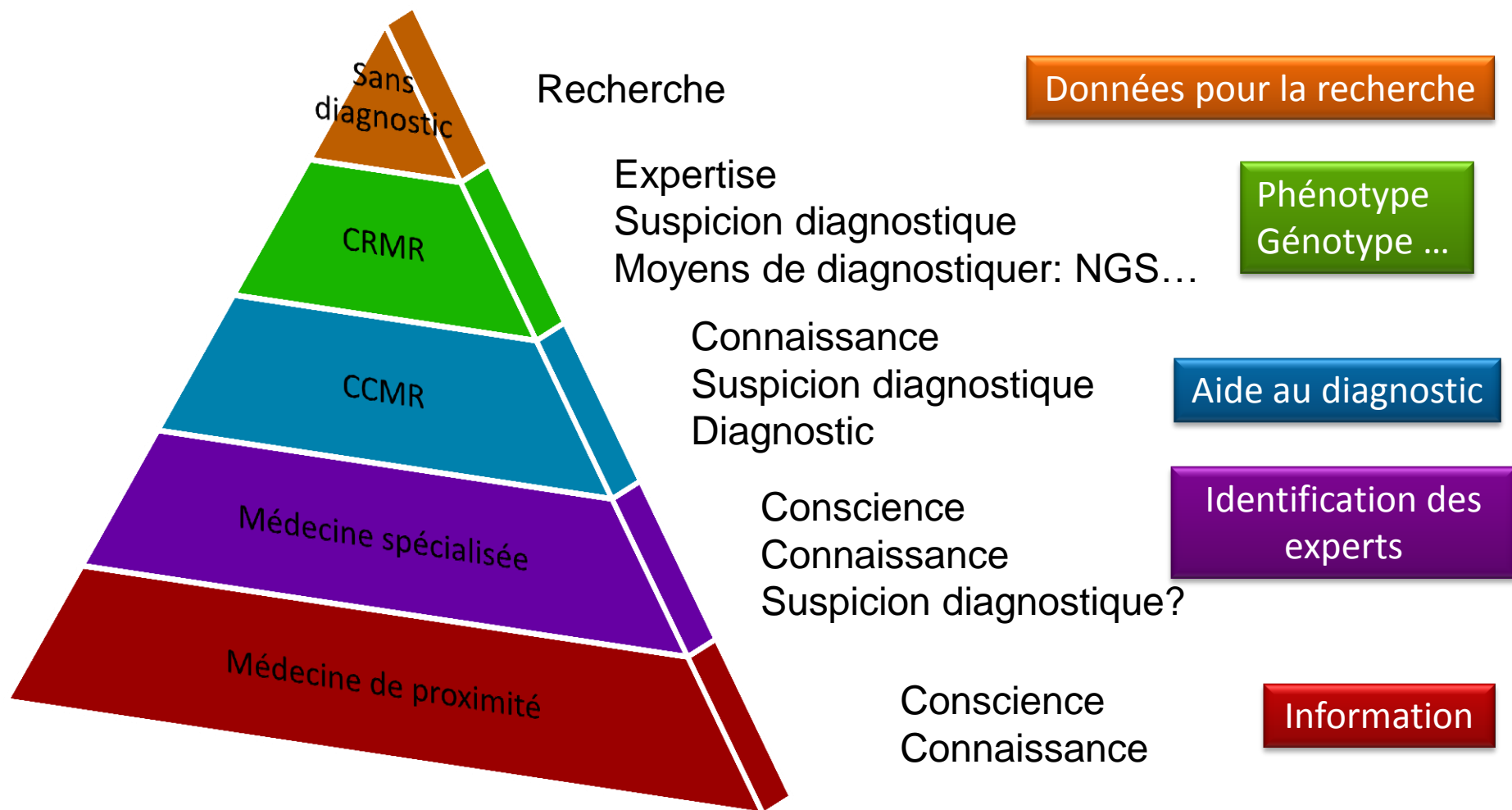
Ana Rath

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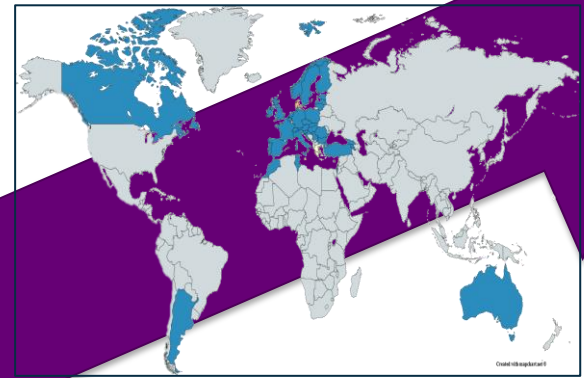
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Journée des anomalies du développement pour les personnes
Sans diagnostic et uniques

La pyramide des maladies rares



20 years Orphanet



Inserm



orphanet



Inserm

www.orpha.net

Information

www.orpha.net



The portal on rare diseases and orphan drugs

« Rare diseases are rare, but rare disease patients are numerous »

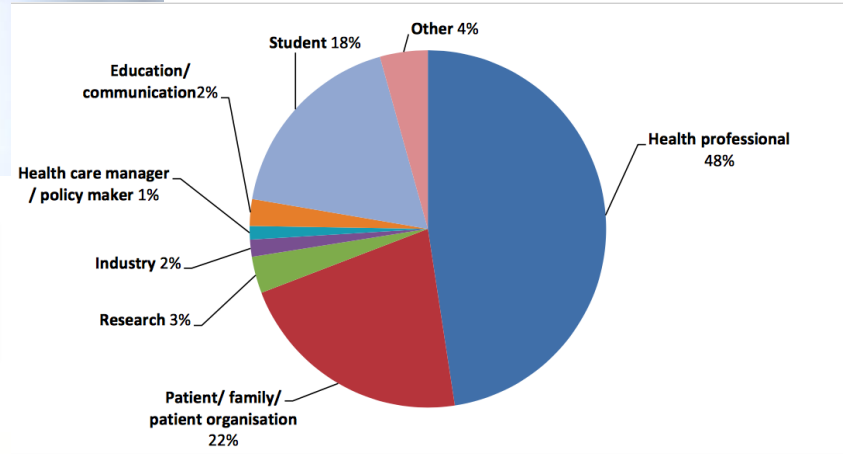
Access our Services

Inventory, classification and encyclopaedia of rare diseases, with genes involved	Inventory of orphan drugs	Directory of patient organisations	Directory of professionals and institutions
Directory of expert centres	Directory of medical laboratories providing diagnostic tests	Directory of ongoing research projects, clinical trials, registries and biobanks	Collection of thematic reports: Orphanet Reports Series

Orphanet website in numbers

- 47,5 million pages viewed
- More than 12 million PDFs downloaded
- Over 9 million visitors from 232 countries

Portail web international et multilingues
Pour un public varié



Donner un nom à chaque maladie: nomenclature ORPHA



View classifications by disease or by group of diseases

Rare metabolic disease
Metabolic disease involving complex molecules
Peroxisomal disease
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

Rare neurologic disease
Neurometabolic disease
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

Rare neurologic disease
Rare epilepsy
Metabolic diseases with epilepsy
Peroxisomal disease
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

Rare neurologic disease
Leukodystrophy
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

Rare endocrine disease
Rare adrenal disease
Primary adrenal insufficiency
Chronic primary adrenal insufficiency
Genetic chronic primary adrenal insufficiency
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form

www.orphadata.org



Orphanet Rare Disease Ontology



Identification
des experts

Catalogue de services

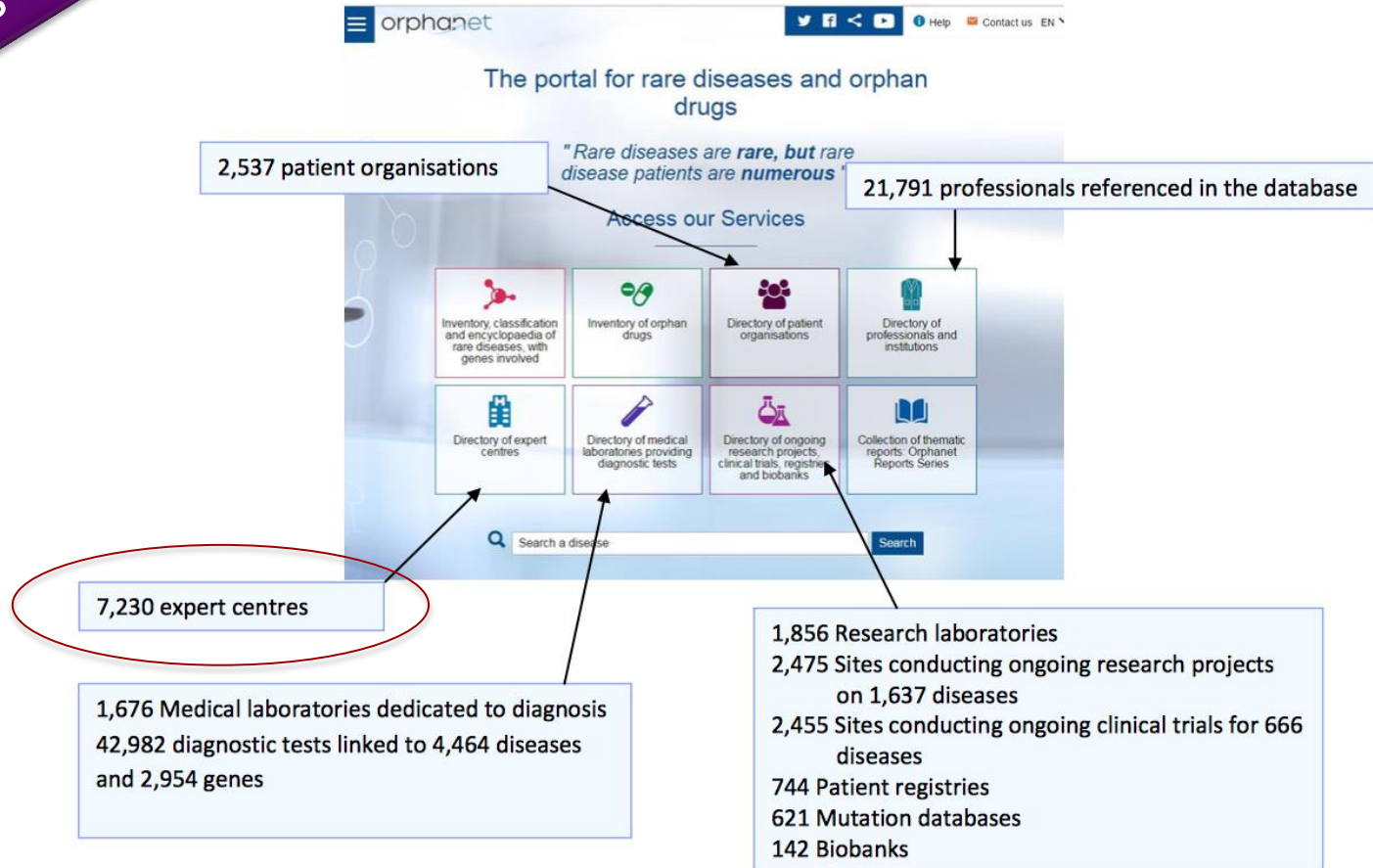


Figure 12 Directory of expert services⁴

Aide au diagnostic



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Le portail des maladies rares et des médicaments orphelins

Menu. Support the Orphanizer. Help. The Orphanizer

Features. Diseases. **Ontology.** Patient's Features.

HPO.	Feature.	Modifier.	Num diseases.
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News

This is Orphanizer

- Orphanizer and used data updated on April 18th, 2017
- This tool is maintained and developed by [Sebastian Köhler](#), Peter Robinson, and the [Orphanet team](#)
- This tool is funded by the [HIPBI-RD](#) project
- The algorithm used is [BOQA](#).

- ORGAN ABNORMALITY
 - Abnormal cellular phenotype
 - Abnormality of blood and blood-forming tissues
 - Abnormal bleeding
 - Abnormal onset of bleeding
 - Abnormal umbilical stump bleed
 - Bleeding requiring red cell transfusion
 - Bleeding with minor or no trauma
 - Cephalohematoma
 - Epistaxis
 - Excessive bleeding after a venipuncture
 - Excessive bleeding from superficial cuts
 - Gingival bleeding
 - Internal hemorrhage
 - Menorrhagia
 - Oral cavity bleeding
 - Persistent bleeding after trauma
 - Prolonged bleeding following procedure
 - Prolonged bleeding time
 - Subcutaneous hemorrhage
 - Abnormal thrombosis
 - Abnormality of bone marrow cell morphology
 - Abnormality of coagulation
 - Abnormality of leukocytes
 - Abnormality of thrombocytes
 - Extramedullary hematopoiesis
 - Hematological neoplasm
- Abnormality of connective tissue
- Abnormality of head or neck
- Abnormality of limbs
- Abnormality of metabolism/homeostasis
- Abnormality of prenatal development or birth
- Abnormality of the breast
- Abnormality of the cardiovascular system
- Abnormality of the digestive system
- Abnormality of the ear
- Abnormality of the endocrine system

Clear. Get diagnosis.

Décrire chaque maladie avec ses phénotypes

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environ 6,000 maladies rares

2787 MR

57,635
annotations

HPO phénotypes

5008 HPO

Fréquences

Obligatoire (100%)

Très fréquent (99-80%)

Fréquent (79-30%)

Occasionnel (29-5%)

Très rare (1-4%)

Absent 0%

Critères diagnostiques

Pathognomonique

Critère diagnostique

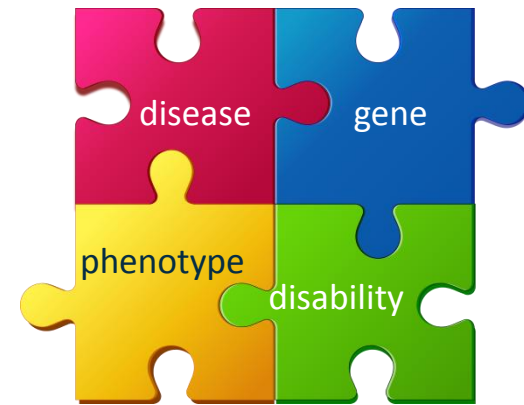
Annotations
faites par
Orphanet

Relayées par
Orphanet et
HPO

Phénotype
Génotype ...

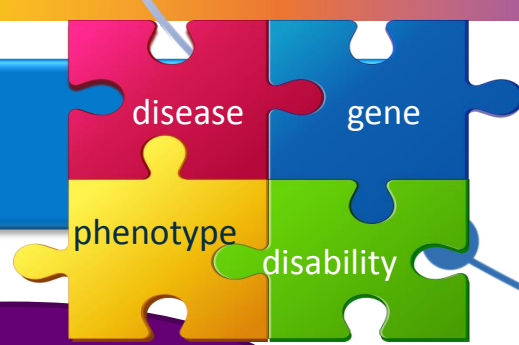
Base de connaissances

- Mettre ensemble les pièces du puzzle
 - Autour de la classification Orphanet des MR
- Fournir des données intégrées et réutilisables
 - Orphanet Rare Disease Ontology (ORDO)
- Promouvoir l'interopérabilité
 - Passerelle entre le soin et la recherche

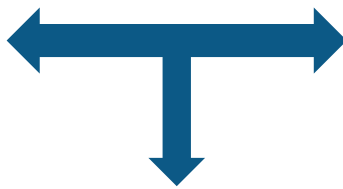


Données pour la recherche

Aider à résoudre le puzzle



Care
Health Information
System (EHRs)



Research
Registries/Cohorts

Orphanet central
nomenclature

Genes

- HGNC
- OMIM
- UniProt
- Reactome
- Ensembl
- Genatlas
- IUPHAR

Phenotypes



Disabilities



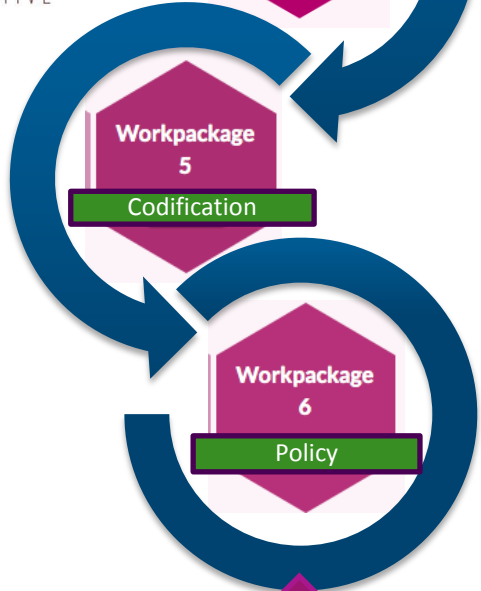
Terminologies

- OMIM
- ICD10/11
- UMLS
- MedDRA
- SNOMED

Côté santé



Guides et outils
Codification
& exploitation



Implémentation
Politiques EU
Recos CEGRD
Guides



www.rd-action.eu

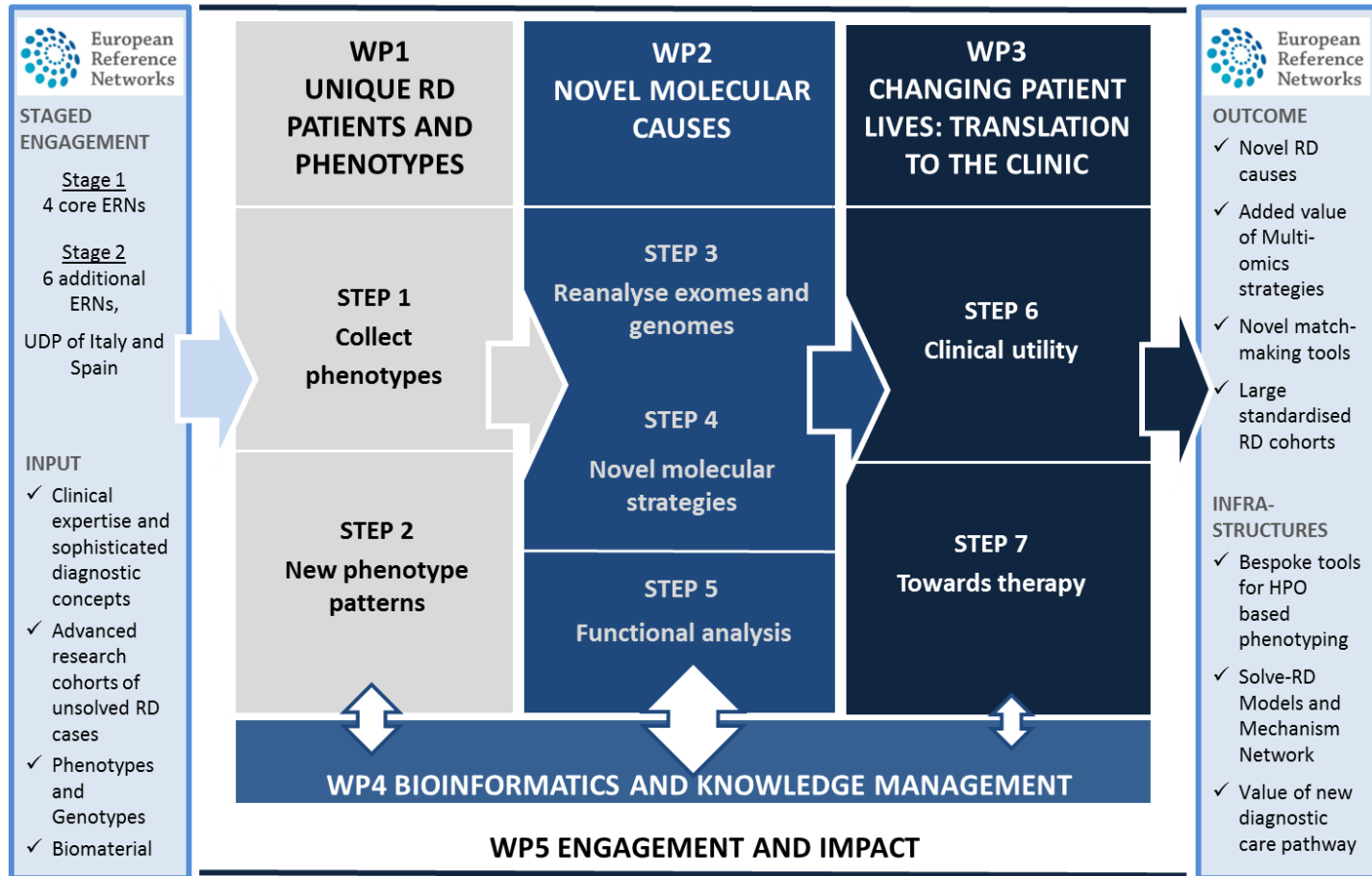


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Côté recherche

SolveRD



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MERCI DE VOTRE ÉCOUTE