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EURORDIS VISIT

Leslie Matalonga, Clinical Genomics Manager at CNAG

Barcelona, June 2025

AGENDA

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centro nacional de análisis genómico

09.30h. Group arrival

- **09.35h – 10.45h. Presentation of CNAG and RD projects**
- 09.35h. Welcome
- 09.40h. Presentation of CNAG and RD projects
- 10.30h. Open question time

10.45h - 11.00h. Break

- 11.00h - 12.00. Visit to the labs and CPD
- 11.00h - 11.25h. Group 1 visits to the labs and Group 2 visits to the CPD
- 11.25h - 11.50h. Group 1 visits to the CPD and Group 2 visits to the labs

- 11.50 - 12.00h. Questions and closing

CNAG presentation

1 CNAG

2 Activity in 2024

3 Research Projects

CNAG

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centre nacional d'anàlisi genòmica
centro nacional de análisis genómico

MAIN MILESTONES

- Created in 2009.
- Founded by Ministerio de Ciencia e Innovación and Generalitat de Catalunya.
- Additional sources of funding come from competitive projects and research services.
- More than 125 professionals, directed by Ivo Gut.
- We are part of OmicsTech (distributed ICTS for omic analysis, recognized by the Spanish Ministry of Science).

MISION

To carry out projects in genome analysis that will lead to significant improvements in people's health and quality of life, in collaboration with the Catalan, Spanish, European and International research and clinical community.

VISION

To be a high quality sequence analysis center and to be a world reference center for genomic analysis.


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Parc Científic de Barcelona
Clúster II - Torres R+D+I

CNAG UNITS

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SEQUENCING UNIT

MARTA GUT
Head of the Sequencing Unit

LÍDIA ÁGUEDA
Biorepository Lab Manager

JULIE BLANC
Sample Preparation Lab Manager

LAURA AGUILERA
Long-Read Sequencing Team Manager

KATJA KAHLEM
Sequencing Production Lab Manager

BIOINFORMATICS UNIT

SERGI BELTRAN
Head of the Bioinformatics Unit

MATTHEW INGHAM
Production Bioinformatics Manager

RAÚL TONDA
Lead Data Analyst of Variant Calling and Analysis Group

DAVIDE PISCIA
Lead Software Engineer of Data Platforms and Tools Development

QUALITY

LIDIA SEVILLA
Quality Manager

OUR TECHNOLOGIES

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SEQUENCING CAPACITY

> 20,000 Gbases/day
= 200 human
genomes/day at 30x

COMPUTING CAPACITY

10,000 cores
14PB disk +
8 PB tape

SEQUENCING INSTRUMENTS

4 Illumina sequencers
(1 NovaSeq X Plus, 2 NovaSeq6000, 1 MiSeq)

1 PacBio sequencer (Revio)

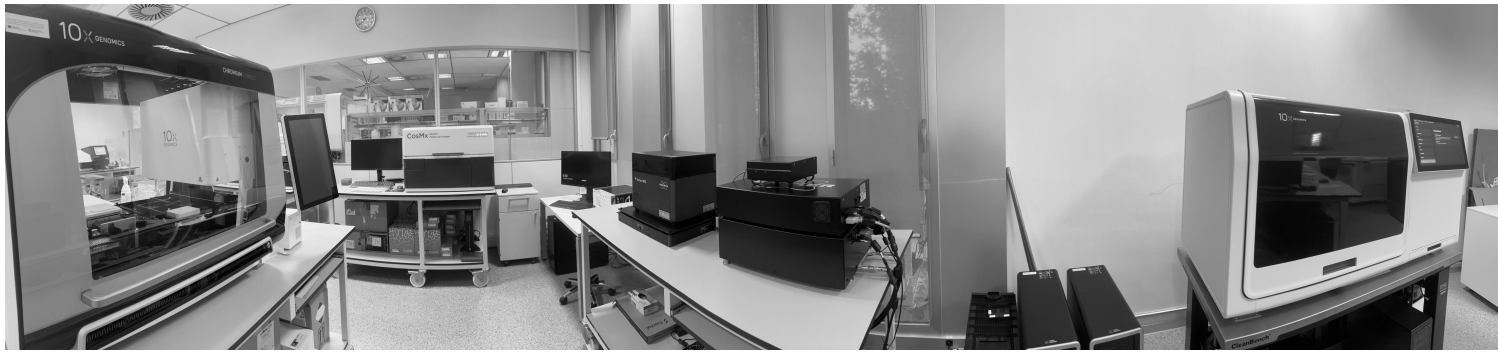
7 Oxford Nanopore Technologies sequencers
(1 GridIon, 1 Promethion, 5 MinION)

SINGLE CELL GENOMICS

10X Chromium X
10X Chromium Connect

SPATIAL GENOMICS

1 Bruker Vutara microscope
1 Nanostring CosMX
1 10X Genomics Xenium



QUALITY

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SGC Certification
ISO 9001: 2015.

ENAC ISO 17025: 2005 Accreditation.

SGS Certification ISO/IEC 27001:2022

BBMRI-ERIC
Expert Centre.

Oxford Nanopore Technologies Certified Service Provider

Dovetail Genomics Certified Service Provider

Coordination of an interlaboratory comparison program for Whole Genome Sequencing (Proficiency testing. ISO/IEC 70243)

Preparation of standarized guidelines for the International Organization of Standarization (ISO)

Future plans:
Esquema Nacional de Seguridad (ENS).

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2024 Annual Report

STRATEGIC AREAS

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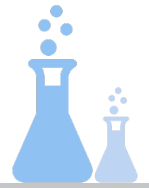
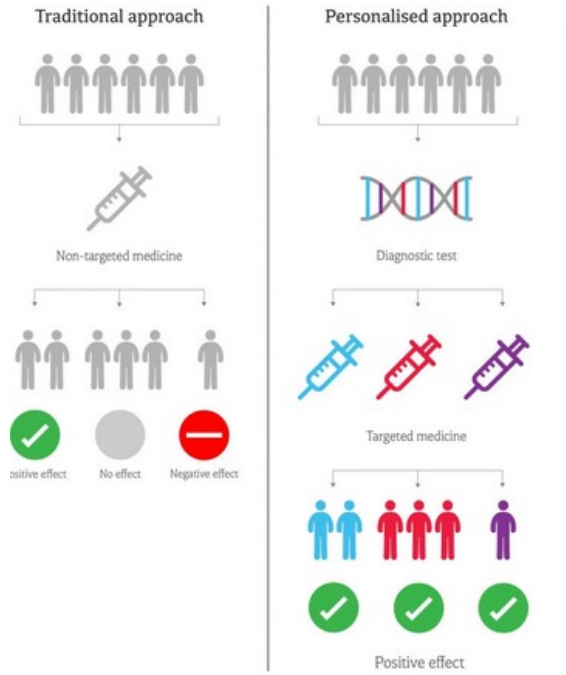
- Personalized medicine
- Rare diseases
- Cancer
- Single-Cells Analysis
- Genome in action
- Biodiversity



CNAG strategic pilars

centre nacional d'anàlisi genòmica
centro nacional de análisis genómico

Personalised medicine | **Rare diseases** | **Cancer** | **Single-Cell Analysis** | **Genome in action** | **Biodiversity**



Cambridge Design Partnership Ltd 2024

CNAG strategic pilars

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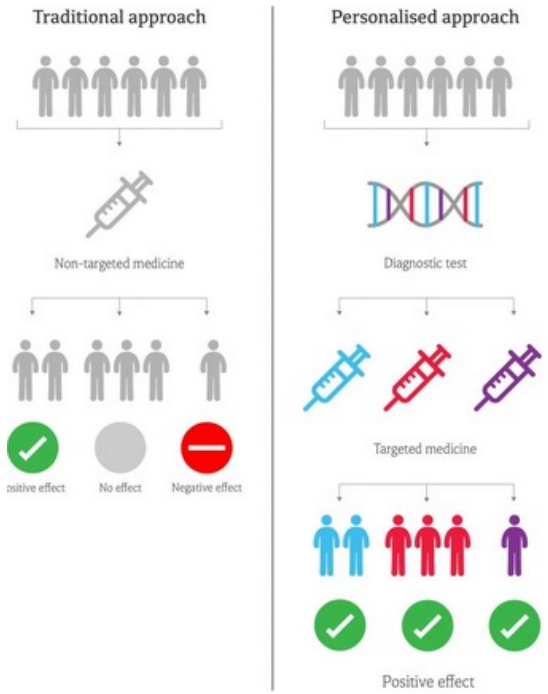
Personalised medicine	Rare diseases	Cancer	Single-Cell Analysis	Genome in action	Biodiversity



Cambridge Design Partnership Ltd 2024

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Rare disease and personalized medicine projects

Rare diseases and genomics

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Complexity of rare diseases

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- Rare disorders - lack of medical knowledge
- Multisystemic disorders-requires strong expertise from different hospital units
- High phenotypic and genetic heterogeneity
- Progressive disorders: complex monitoring and follow-up

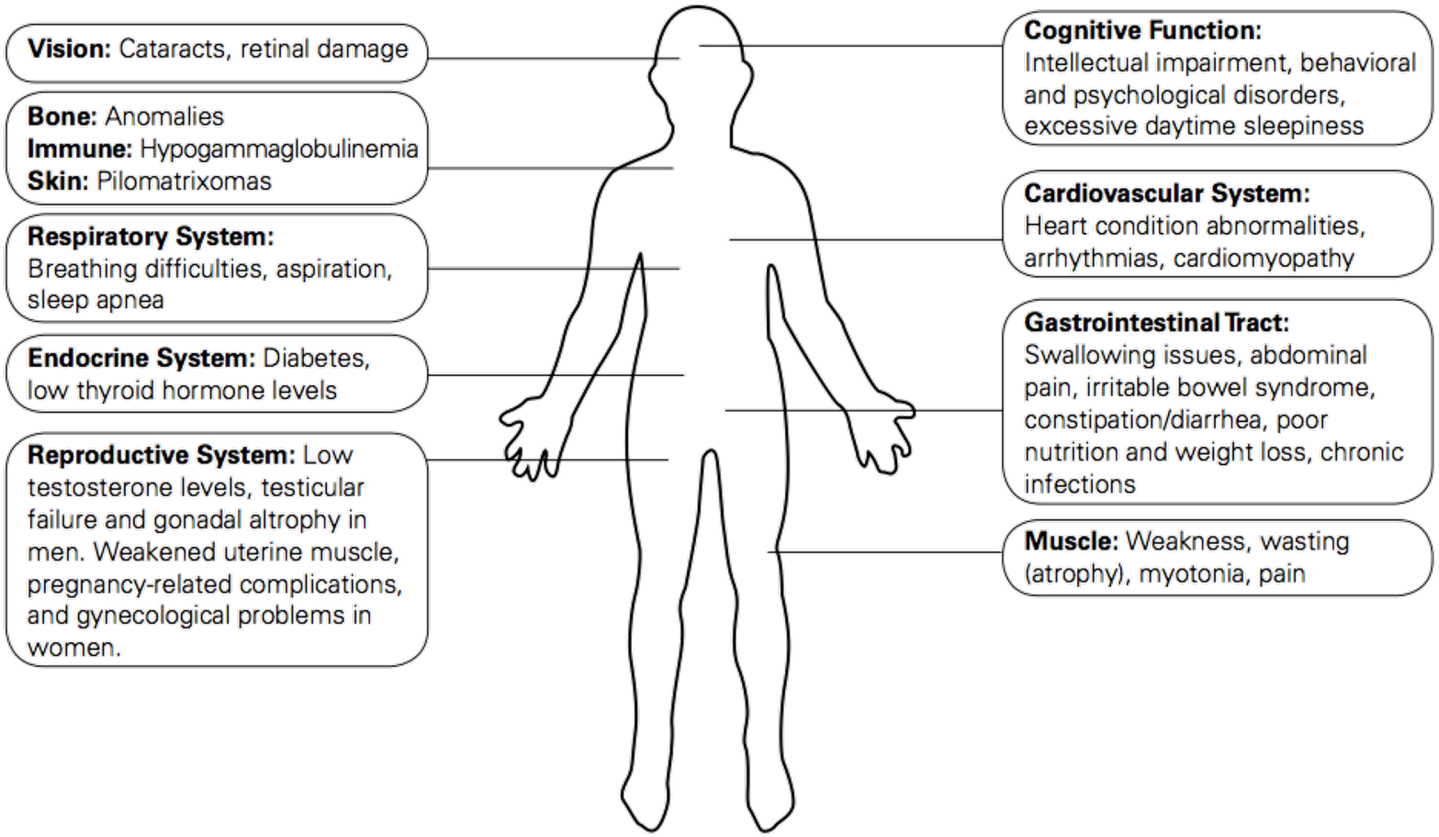


Image from Myotonic Dystrophy Foundation

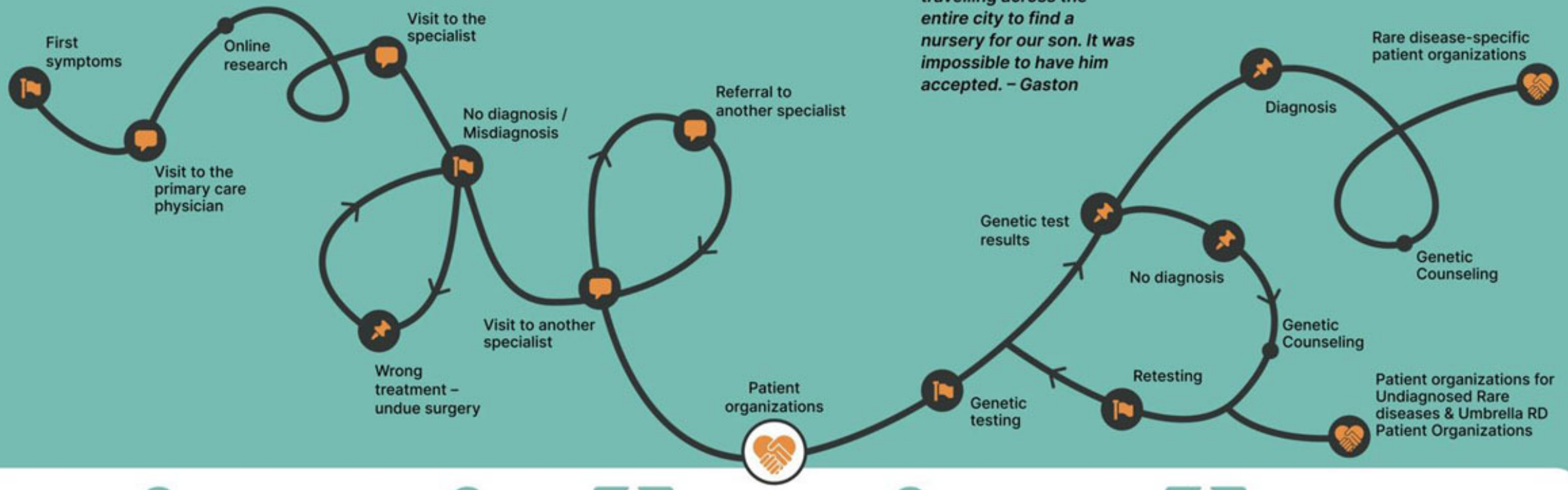
Patient Journey through diagnosis

> *"It's a waiting game, but you tell a mum to wait when she's waited 15 years. It's difficult. – Nuria*

> *"People began to ask which side of the family it came from...It was a difficult time for us as parents. – Alexa*

> *"A diagnosis may be bad news, it may be very bad news or it may be no news. But all of that's OK and there's help and support for whatever spectrum you end up on. – Peter*

> *"We went around, travelling across the entire city to find a nursery for our son. It was impossible to have him accepted. – Gaston*



Information and training for patient organizations

Helplines

Information on secondary findings

Next Steps Toolkit

EUROGENTEST Guidelines

Experience Based Co-Design

RareConnect .org

ENSERIO study. Time to Diagnosis

Undiagnosed Photo Project

Protocol to support ultra-rare diagnosis

Training for professionals

EURORDIS

SolveRD

CHEO

European Patient Advocacy Group

EuroGentest

GLOBAL COMMISSION

UNIAMO

WILHELM FOUNDATION

feder

SWAN UK

UDNI

Patient Journey through diagnosis

"It's a waiting game, but you tell a mum to wait

"People began to ask which side of the family it came from...It was a difficult time for us as parents. – Alexa

"A diagnosis may be bad news, it may be very bad news or it may be no news. But all of that's OK and support ctrum you r

First symptoms

Online research

Visit to the primary care physician

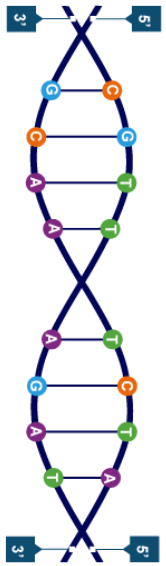
Rare disease-specific patient organizations

Genetic Counseling

testing

Patient organizations for Undiagnosed Rare diseases & Umbrella RD Patient Organizations

REACHING A MOLECULAR DIAGNOSIS IS CRUCIAL FOR PATIENTS



- Treatment
- Patient management
- Disease prognosis
- Genetic counselling
- Reduce anxiety of uncertainty

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Genetic origin

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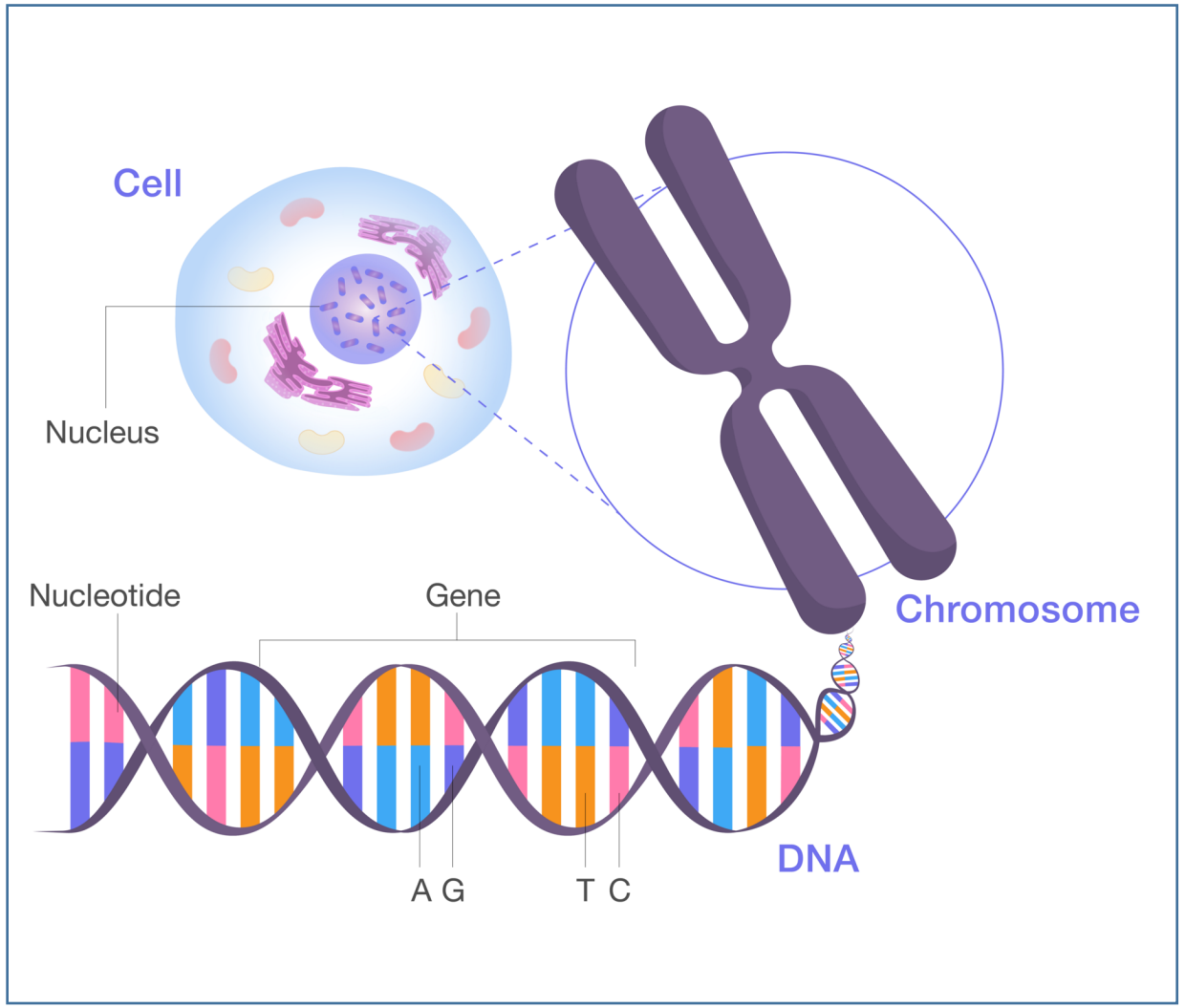

46 chromosomes (books)
>20,000 genes (chapters)
3,200 million bases (letters)

>3-7M variants! (letters comparison)

Is it possible to sequence our genome?

- **2000:** 1 genome >10y, aprox $3,000 millions
- **2024:** 1 genome = 1 day, <1000 euros

Genetic origin

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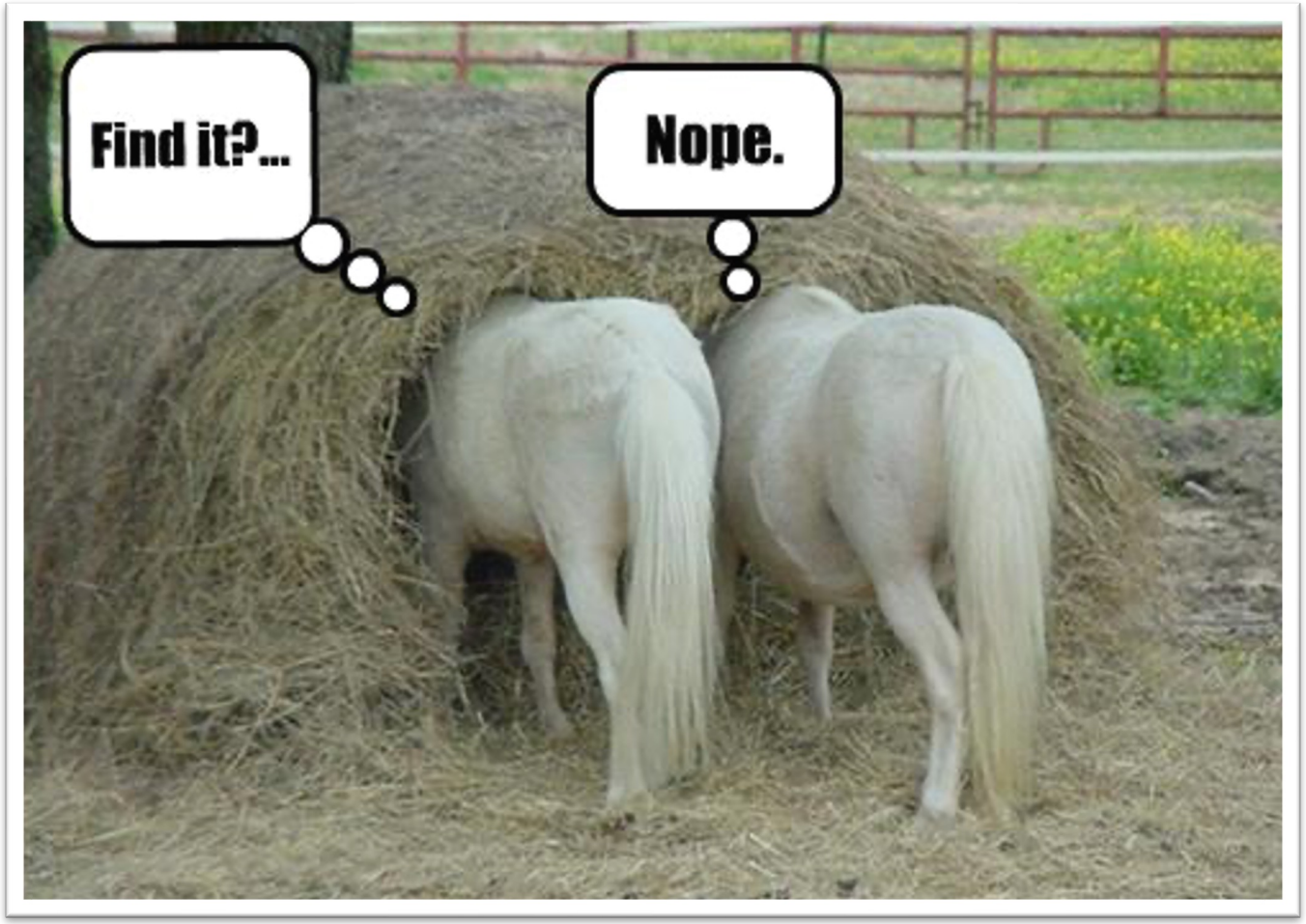
How do I find the pathogenic variant within more than 3,000,000?



Rare diseases and genomics at CNAG

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1. Sequencing – lab visit with Marta Gut
2. Data handling and processing – CPD visit with Simon
3. Bioinformatics Unit
    1. Data analysis -group led by Raul tonda
    2. Infrastructure – GPAP analysis platform – group led by Davide Piscia
    3. Results interpretation -group led by Sergi Beltran

Unmet needs and bottlenecks in rare diseases

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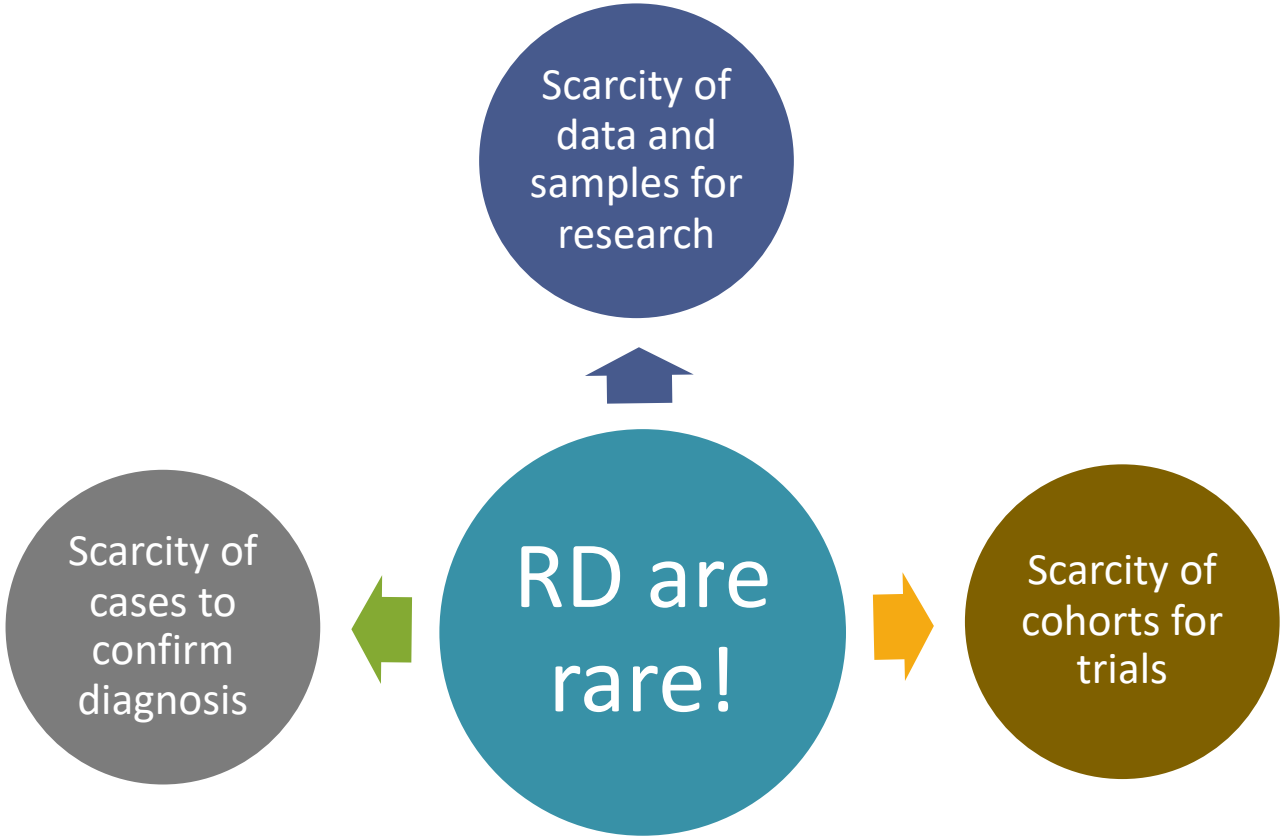
Lack of diagnosis

Lack of therapies



Data sharing is essential...

RD-Connect Genome-Phenome Analysis Platform (GPAP)

https://platform.rd-connect.eu/

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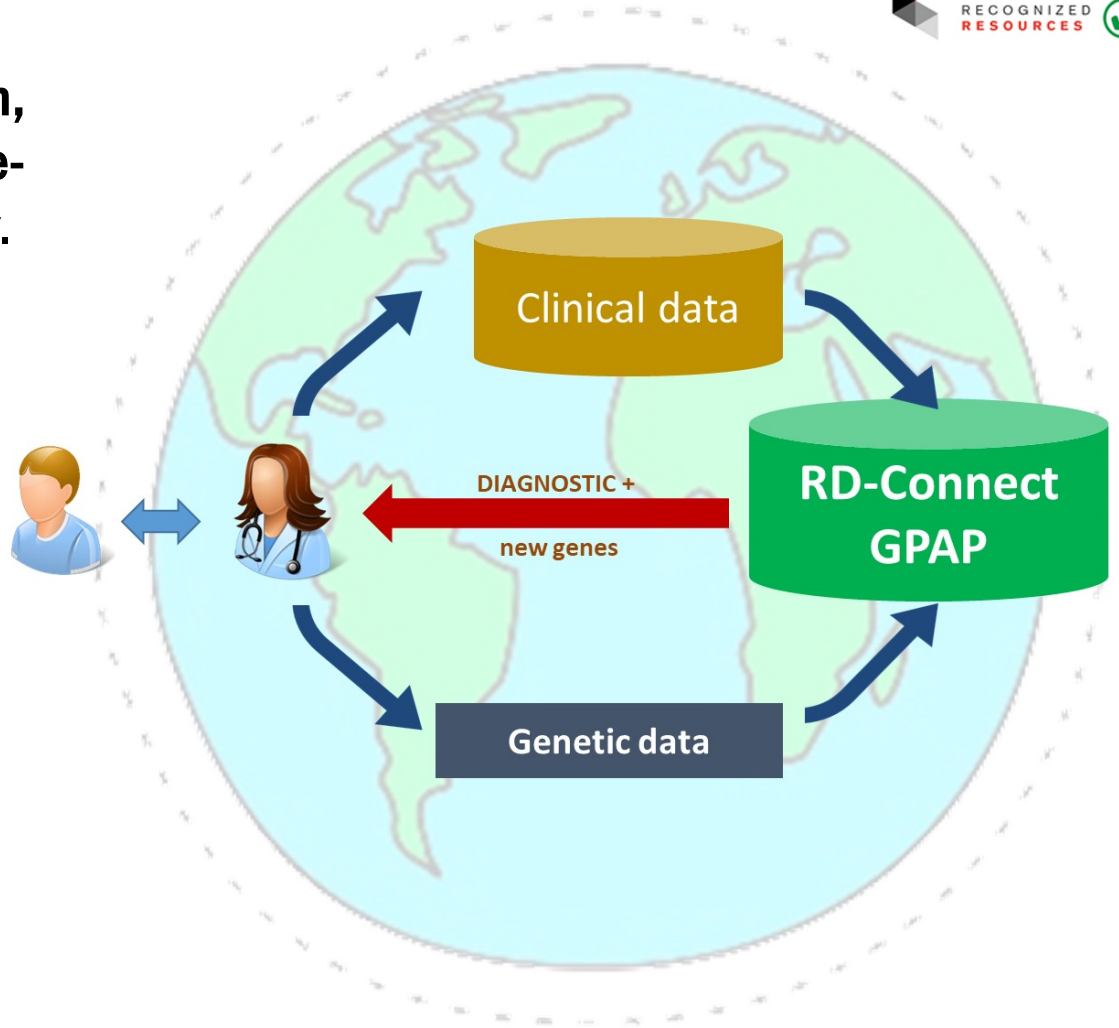
IRDiRC
RECOGNIZED RESOURCES

The RD-Connect GPAP is an online system that facilitates **collation, sharing, analysis and interpretation of integrated genome-phenome datasets** for Rare Disease diagnosis and gene discovery.

- >28,000 genome-phenome datasets
- > 100 contributing groups
- > 600 registered users
- Data is pseudonymized
- User activity logged
- Platform security audited periodically by an external company



RD-Connect elixir INB EUROPEAN JOINT PROGRAMME RARE DISEASES SolveRD

GPAP – user interface

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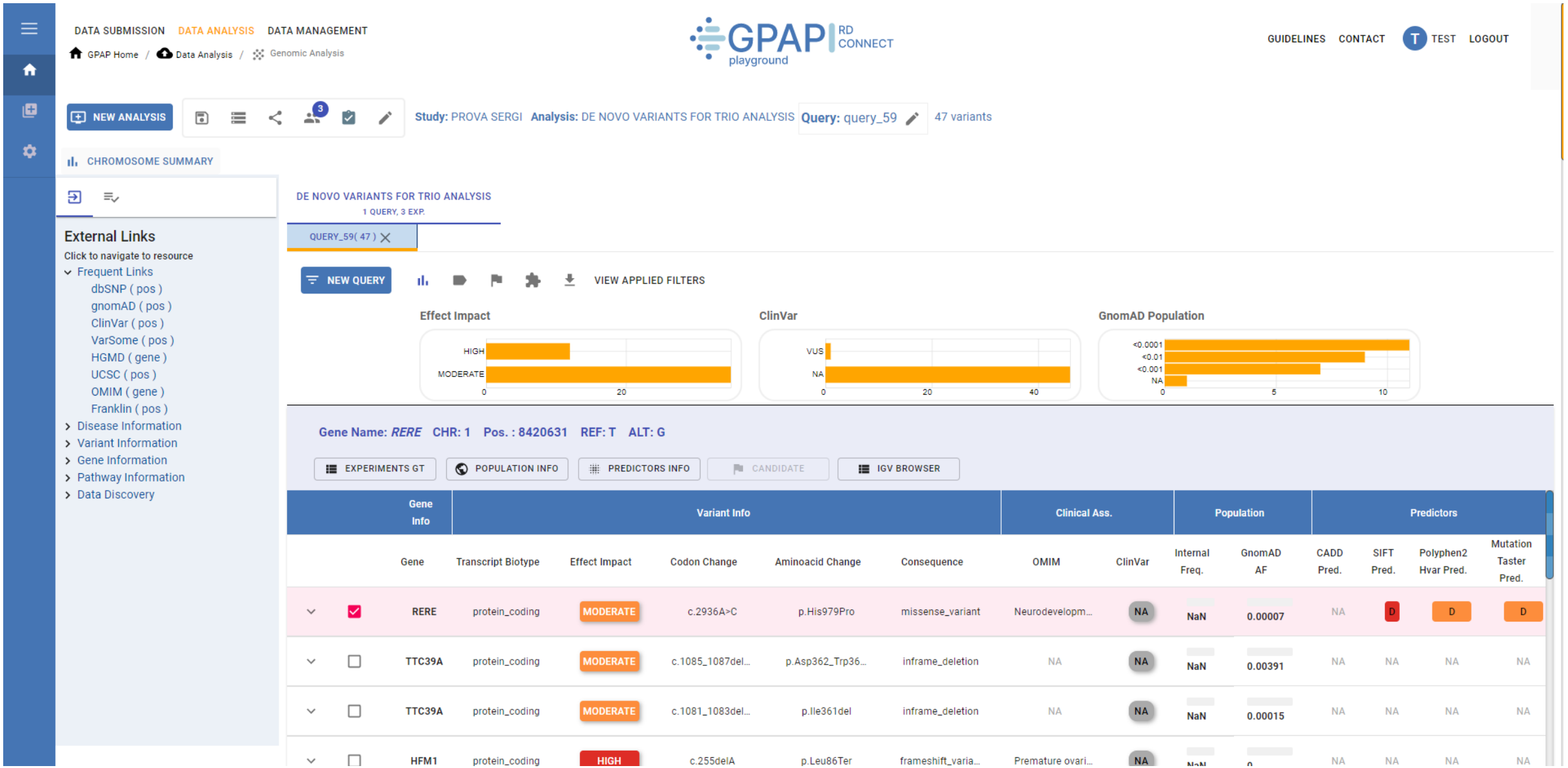

GPAP

RD CONNECT

genome phenome
analysis platform

A platform for rare diseases developed by cnag

GPAP instances

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GPAP
Genome - Phenome Analysis Platform

G — IRDiRC RECOGNIZED RESOURCES — RD-Connect
>30.000 WES/WGS
https://platform.rd-connect.eu/

G — NAVARRABIOMED CENTRO DE INVESTIGACIÓN BIOMÉDICA
>2900 WGS
https://navbiomed.cnag.gpap.eu/

G — URDCat
>1850 WES/ WGS
https://rdcat.gpap.cnag.eu/

S G — GPAP | CANCER
https://ca.gpap.cnag.eu/

G S — GPAP | CNAG
>2500 WES/WGS
https://portal.gpap.cnag.eu

SolveRD
European Reference Networks
Reanalysis
>7000 negative cases
>1000 solved

246 consanguineous cases
Diagnostic yield 86%
8 novel disease-causing genes

bbmri-lpc
317 RD cases
Diagnostic yield 37% (8-95%)
10 novel candidate genes

NAGEN PROYECTO GENOMA 1000 NAVARRA
NAGEN1000
NAGENPEDIATRICS
NAGENCOL
NAGENMX
REPRONAGEN
PHARMANAGEN
Current diagnostic yield 15-52%

PERIS 2016 2020
934 Neurological cases
Diagnostic yield 32.5%
Reanalysis 20.7%

La Marató 3 — ciberer Enfermedades Raras
844 Neurological cases
Reanalysis 15.57%

Instand NGS for Personalized Therapy
hereditary cancer
Tumor/Normal
RNA fusion
Tumor characterization

IMPaCT Genómica
136 hereditary cancer
941 rare diseases

GPAP instances

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GPAP
Genome - Phenome Analysis Platform

G

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IMPaCT Genómica

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NAGEN: precision medicine

https://www.navarrabiomed.es/es/nagen

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PROYECTO GENOMA 1000 NAVARRA

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Current diagnostic yield 15-52%

- Whole genome sequencing
- >2250 datasets reported
- CNAG participates in sequencing, analysing and interpreting the data

WGS for rapid clinical management: NAGENPEDIATRICS

Information kindly provided by Josune Hualde and Sara Ciria

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NAVARRABIOMED
CENTRO DE INVESTIGACIÓN BIOMÉDICA

259 patients
(751 WGS)

- Recruitment age (average) = 3,8 years
- Turn around time (average) = 18,28 days (12-25)

- Interpretation:
  - 1628 variants interperted (in 629 genes) and TAGed in GPAP
  - 259 diagnostic reports issued
  - 39,2% diagnostic rate in ICU cohort
  - Impact in clinical management: 53,8%

Grupo	Nº Familias	Variantes pertinentes	Tasa diagnóstica
UCI	79	31	**39,2%**
TEA	179	26	**14,85%**
Cribado	5	3	**60%**
Tumores sólidos	7	6	-

NAGEN: precision medicine

https://www.navarrabiomed.es/es/nagen

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EXAMPLES Clinical Impact UCI

	Age	Clinical manifesta-tions	Molecular result	Variant analysis	Results	Clinical impact	OMIM
NP003	3m	Seizure	SCN1A c.2995A>C p.Asn999His (het)	Candidata strong	19d	Antiepileptic drug adjustments and inclusion in chronic illness program	Developmental and epileptic encephalopathy (OMIM #182389)
NP062	7d	Encephalopathy, feeding disorder, peculiar phenotype	ALG1 c.605A>G p.Asp202Gly (hom)	Patogénica	19d	Inclusion in palliative care program. Exitus. Genetic counselling offered to the family (ongoing pregnancy)	Congenital disorder of glycosylation, type Ik (OMIM #608540)
NP165	Newborn	Microcephaly, small for gestational age, metabolic alteration, seizure	HIBCH c.1127T>G p.Phe376Cys; c.439-2A>G (compound het)	Patogénica	18d	Personalised diet (low in valine)	3-hydroxyisobutryl-CoA hydrolase deficiency (congenital metabolic disorder, OMIM #250620)
NP265	13y	Intellectual disability and catatonia	SHANK3 c.3727dupG p.Ala1243Glyfs Ter69 (de novo)	Patogénica	16d	Drug adjustments	PHELAN-MCDERMID SYNDROME (OMIM #606232)

GPAP instances

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GPAP
Genome - Phenome Analysis Platform

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941 rare diseases

IMPaCT Genómica

https://genomica-impact.es/

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IMPaCT-GENóMICA is a collaborative infrastructure, which establishes the networks and flows necessary to contribute to the diagnosis of rare diseases and other genetic diseases, beyond usual clinical practice, equitably throughout the territory.

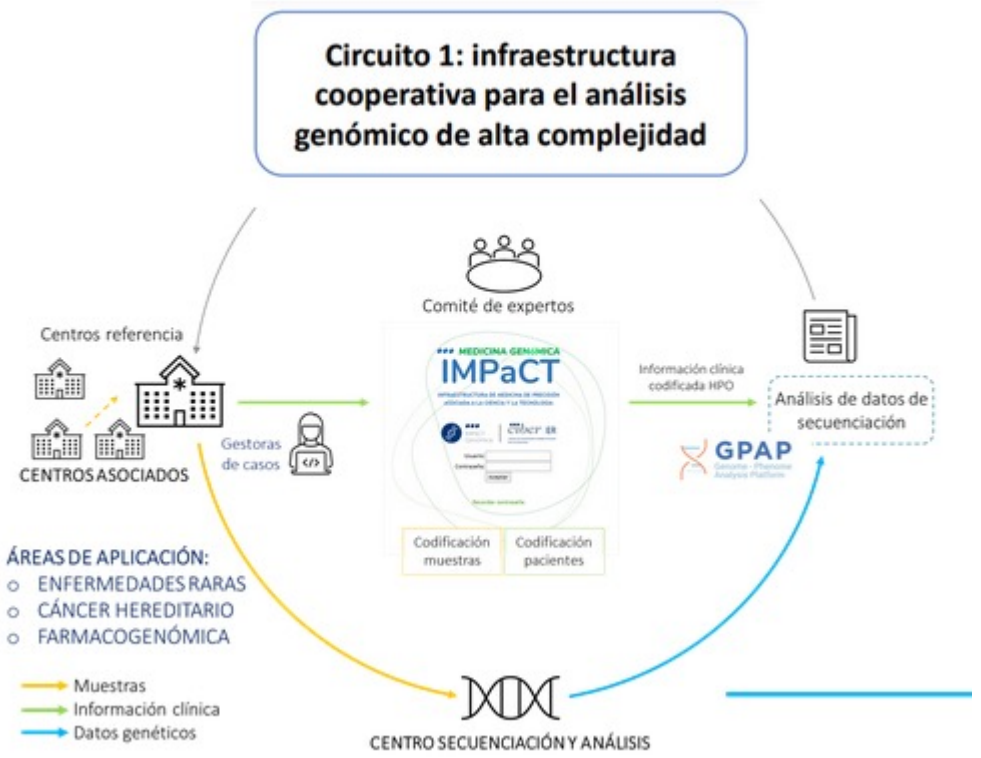
17
Comunidades Autónomas

37
Entidades Asociadas

+100
Hospitales

3
Centros de secuenciación

+300
Colaboradores directos



Rare disease projects

1. Introduction
2. RD-Connect GPAP
3. National projects and initiatives
    1. NAGEN
    2. IMPaCT Genómica
4. International projects and initiatives
    1. Solve-RD
    2. Screen4care
5. Others
6. Yakup’s journey to hope

GPAP instances

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IMPaCT Genómica

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Re-analysis of unsolved cases: Solve-RD

Solving the Unsolved Rare Diseases

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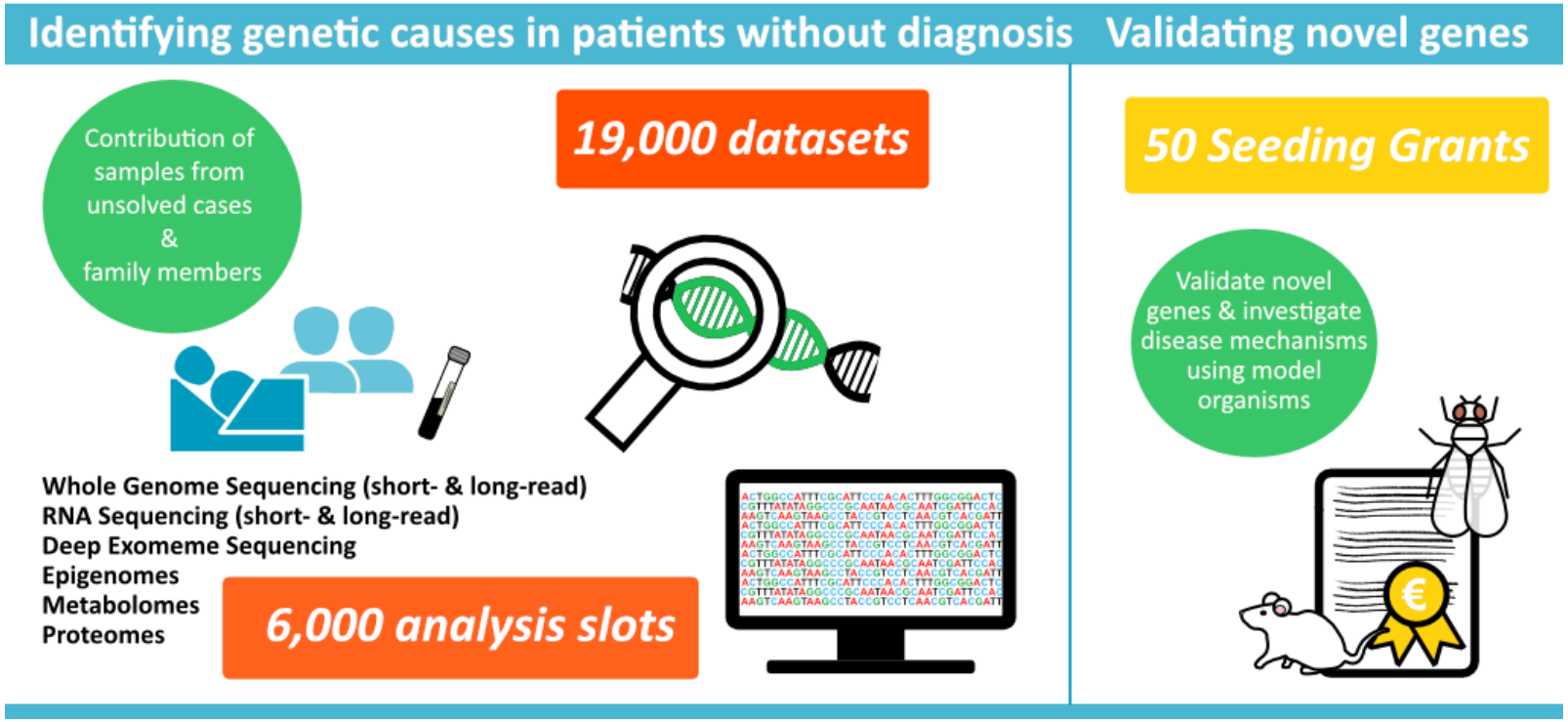
~50% of cases remain without diagnosis after routine hospital workflows



- Equity on diagnostics across EU
- CNAG participation:
  - Infrastructure (RD-Connect GPAP)
  - Analysis (WP lead: WES, WGS, RNAseq)
  - Sequencing (RNAseq)

Solve-RD

Solving the Unsolved Rare Diseases

- EU funded research project
- 1.1.2018 – 01.06.2024
- Coordinated by Olaf Riess & Holm Graessner (Tübingen)

- > 200 groups
  - 22 beneficiaries
  - 23 associated partners
  - 40 collaborators
- >300 (clinical) scientists

- **RD expertise and infrastructure**: RD-Connect, EGA, Orphanet, HPO, EuroGentest, Canadian Models and Mechanisms Network, EURORDIS

www.solve-rd.eu

@Solve_RD

Solve-RD

Solving the Unsolved Rare Diseases

European Reference Network | Neurological Diseases (ERN-RND)

EURO-NMD | Building bridges and breaking barriers in rare neuromuscular diseases

ITHACA

ERN-ITHACA focuses on rare congenital malformation syndromes and intellectual disability

GENTURIS

rare genetic tumour risk syndromes

EpiCARE

European Reference Network for rare and complex epilepsies

rita

The European Reference Network that aims at improving the care of patients with Rare Immunological Disorders

+ UDP-Spain and UDP-Italy

Solve-RD Analysis and Interpretation

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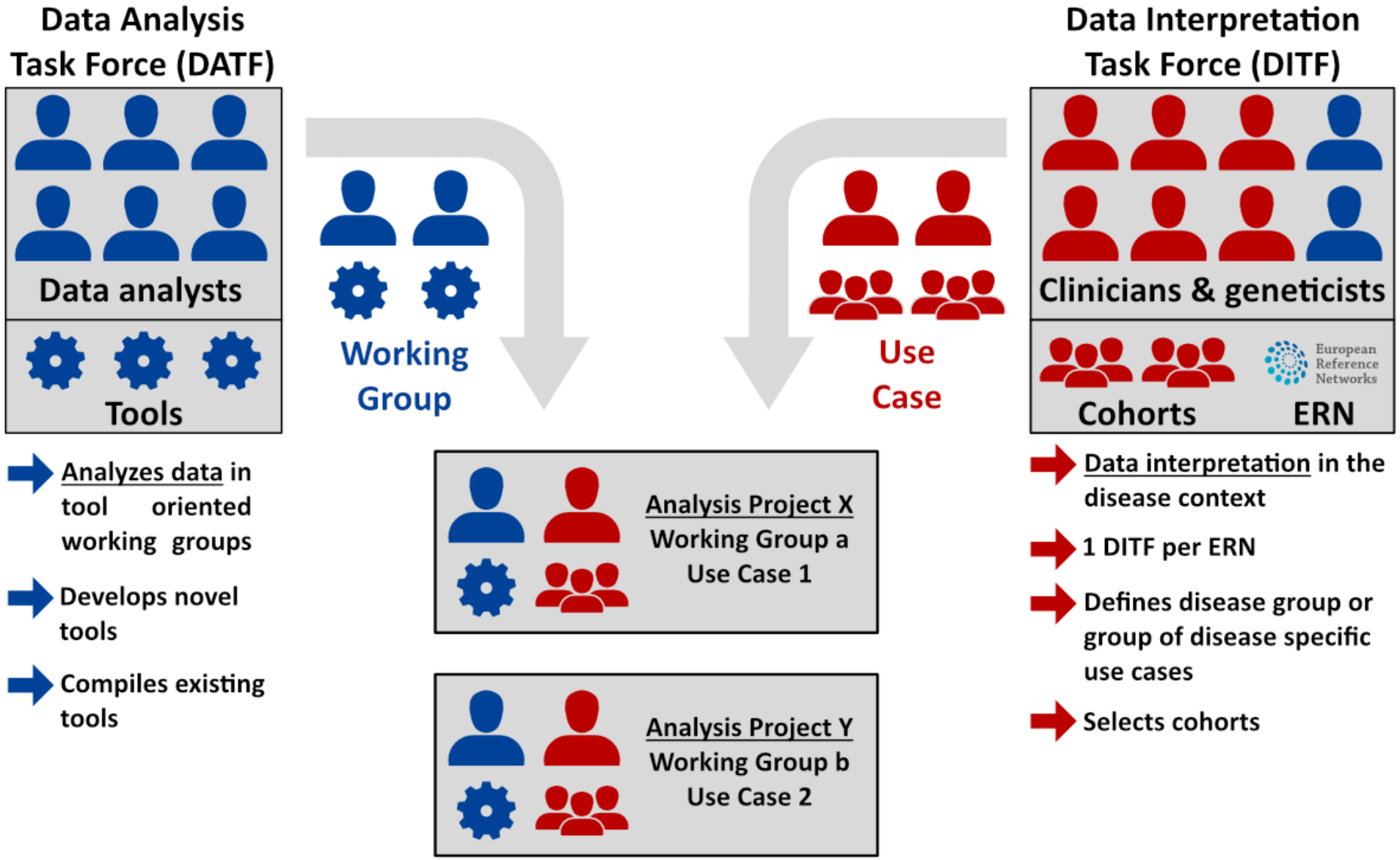

Solve-RD DATF Working Groups

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WG1: SNV/indel re-(analysis) (Leslie Matalonga, CNAG)
WG2: CNV re-(analysis) (Steven Laurie, CNAG)
WG3: Relatedness and Regions of Homozygosity analysis (Stephan Ossowski, EKUT, Tübingen)
WG4: *De novo* trio analysis (Christian Gilissen, Radboud UMC, Nijmegen)
WG5: Meta-analysis for novel genes & gene burden analysis (Christian Gilissen)
WG6: Transcriptomics (Anna Esteve, CNAG)
WG7: Epigenomics
WG8: Somatic Mutations - Novel DeepWES
WG9: Structural Variants - Novel WGS
WG10: Novel long read WGS analyses

SolveRD
Solving the Unsolved Rare Diseases

European Reference Networks

RND | ITHACA
EURO-NMD | GENTURIS

>300 collaborators
37 institutes

12 European countries

RD Data

Comprehensive analysis suite

2-level expert review

Individuals affected by rare diseases (*n* = 6,449) from 6,004 families

Genomic datasets
9,351 ES
523 GS

human phenotype ontology

Phenotypes
(Av. 6 HPO)

Pedigree information

Five data analysis working groups

DATF-WGs

Four ERN-expert interpretation groups

ERN-DITFs

4,253 expert review hours

ERNs

2,512 disease genes

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Solve-RD reanalysis results – 6004 families

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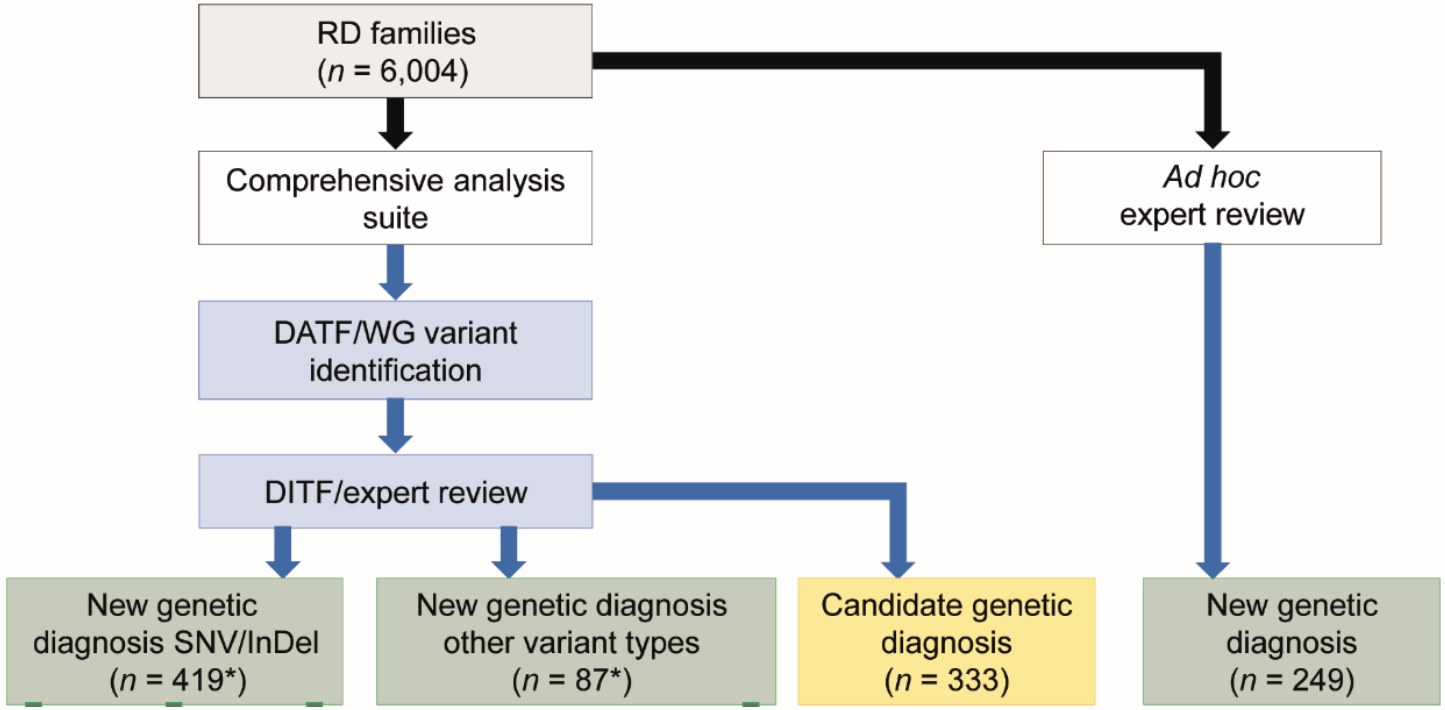

755 new solved cases!

These cases had previously undergone a WES or WGS in the routinary diagnostic setting of their country

IMPORTANCE OF EXPERTS COLLABORATION!

Data interpretation - collaborative

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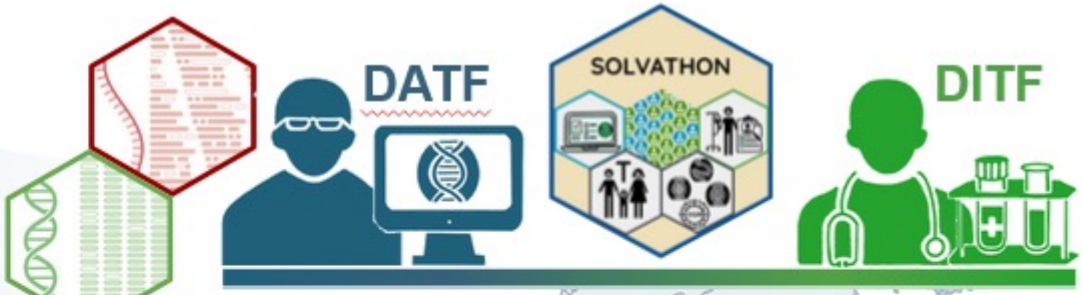
On-site or online Solvathon workshop bring together analysts and data clinical researchers

Solvathon 1 - **RNA sequencing**

Participants: 57
Families: 216
Solved: 5



Solvathon 2 - **SR Genomes**

Participants: 72
Families: 764
Solved: 9

Solvathon 3 - **LR Genomes & OGM**

Participants: 55
Families: 197
Solved: 9

Solvathon 4 - **Multiomics**

Participants: 59
Families: 474
Solved: 5

European Reference Networks
for rare or low prevalence complex diseases

NMD: Neuromuscular diseases, **RND:** Neurological diseases, **RITA:** Rare immunological disorders, **ITHACA:** Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders, **GENTURIS:** Genetic Tumour Risk Syndromes, **EpiCARE:** Rare and complex epilepsies

Omics: **RNA** **SR-GS** **LR-GS** **OGM** **Methylation**

Data interpretation – collaborative - RDMM

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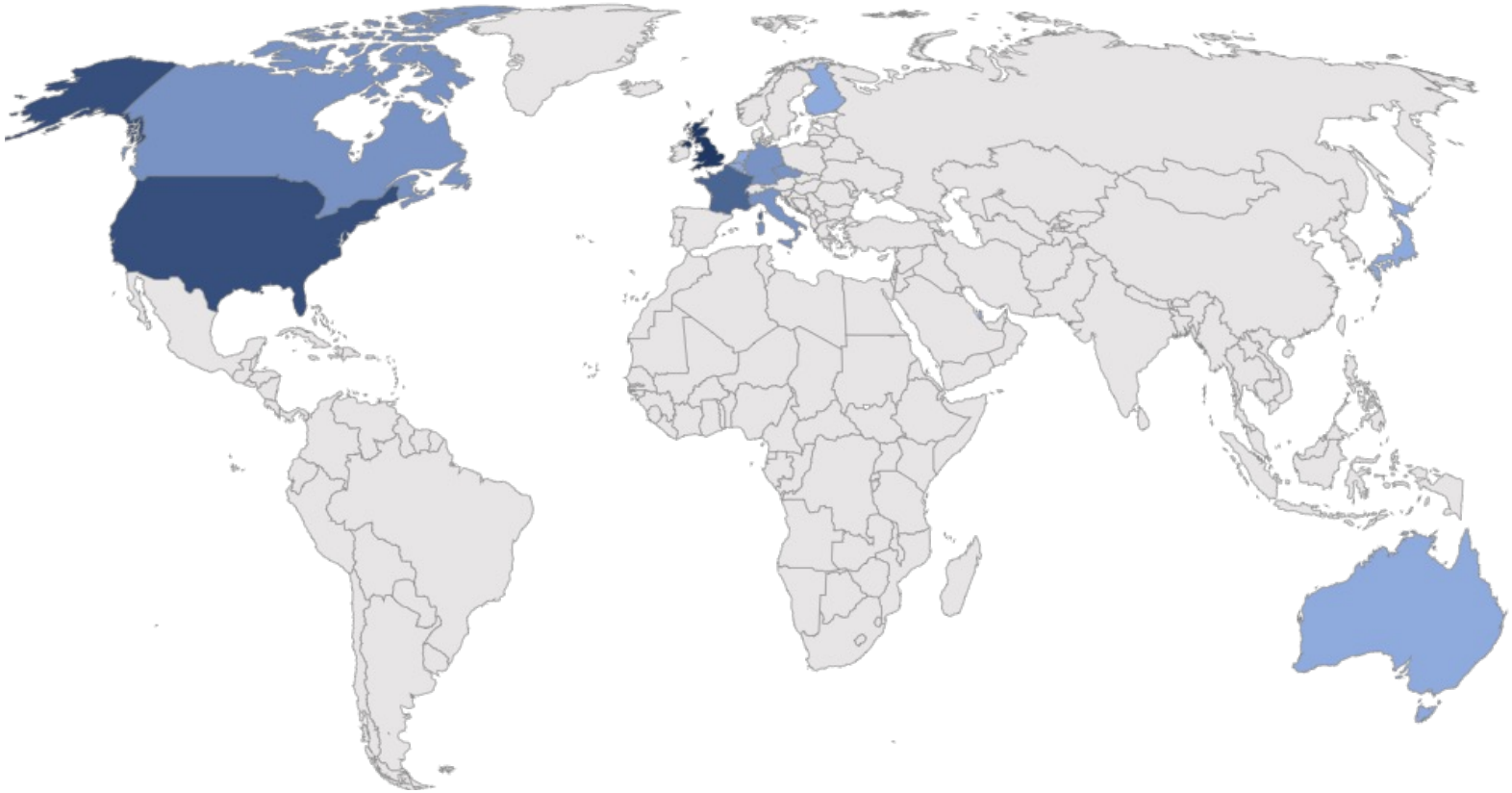
RDMM-Europe brokerage service

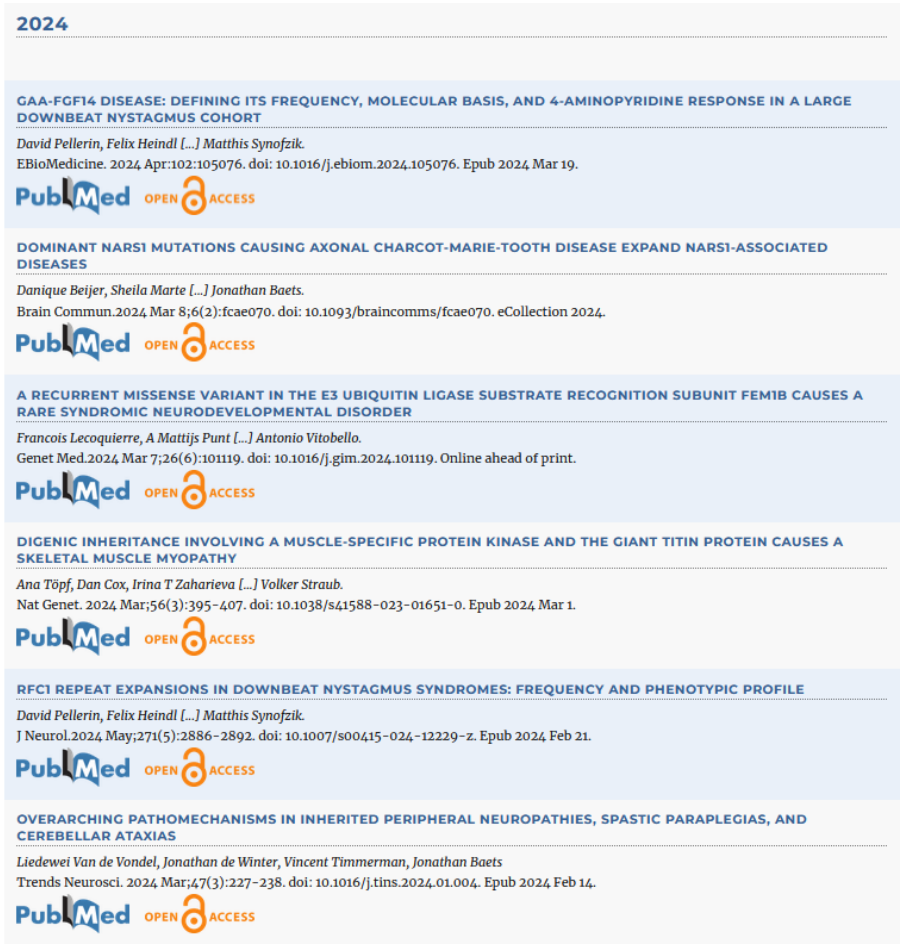
The European Rare Disease Models & Mechanisms Network (RDMM-Europe) has been established by Solve-RD to boost research in rare diseases, discover new disease-causing genes and obtain evidence for pathogenicity through functional validation.



Solve-RD Publications

Generating **knowledge** for the RD community

https://solve-rd.eu/results/scientific-publications/



2024

GAA-FGF14 DISEASE: DEFINING ITS FREQUENCY, MOLECULAR BASIS, AND 4-AMINOPYRIDINE RESPONSE IN A LARGE DOWNBEAT NYSTAGMUS COHORT

David Pellerin, Felix Heindl [...] Matthis Synofzik.
EBioMedicine. 2024 Apr:102:105076. doi: 10.1016/j.ebiom.2024.105076. Epub 2024 Mar 19.
PubMed OPEN ACCESS

DOMINANT NARS1 MUTATIONS CAUSING AXONAL CHARCOT-MARIE-TOOTH DISEASE EXPAND NARS1-ASSOCIATED DISEASES

Danique Beijer, Sheila Marte [...] Jonathan Baets.
Brain Commun.2024 Mar 8;6(2):fcae070. doi: 10.1093/braincomms/fcae070. eCollection 2024.
PubMed OPEN ACCESS

A RECURRENT MISSENSE VARIANT IN THE E3 UBIQUITIN LIGASE SUBSTRATE RECOGNITION SUBUNIT FEM1B CAUSES A RARE SYNDROMIC NEURODEVELOPMENTAL DISORDER

Francois Lecoquierre, A Mattijs Punt [...] Antonio Vitobello.
Genet Med.2024 Mar 7;26(6):101119. doi: 10.1016/j.gim.2024.101119. Online ahead of print.
PubMed OPEN ACCESS

DIGENIC INHERITANCE INVOLVING A MUSCLE-SPECIFIC PROTEIN KINASE AND THE GIANT TITIN PROTEIN CAUSES A SKELETAL MUSCLE MYOPATHY

Ana Töpf, Dan Cox, Irina T Zaharieva [...] Volker Straub.
Nat Genet. 2024 Mar;56(3):395-407. doi: 10.1038/s41588-023-01651-0. Epub 2024 Mar 1.
PubMed OPEN ACCESS

RFC1 REPEAT EXPANSIONS IN DOWNBEAT NYSTAGMUS SYNDROMES: FREQUENCY AND PHENOTYPIC PROFILE

David Pellerin, Felix Heindl [...] Matthis Synofzik.
J Neurol.2024 May;271(5):2886-2892. doi: 10.1007/s00415-024-12229-z. Epub 2024 Feb 21.
PubMed OPEN ACCESS

OVERARCHING PATHOMECHANISMS IN INHERITED PERIPHERAL NEUROPATHIES, SPASTIC PARAPLEGIAS, AND CEREBELLAR ATAXIAS

Liedewei Van de Vondel, Jonathan de Winter, Vincent Timmerman, Jonathan Baets
Trends Neurosci. 2024 Mar;47(3):227-238. doi: 10.1016/j.tins.2024.01.004. Epub 2024 Feb 14.
PubMed OPEN ACCESS

>200 scientific publications!
CNAG directly involved in >30

Knowledge accessible for clinical practice:
New disease-causing genes, ClinVar database etc.

Beyond diagnosis: TREATABOLOME

treatabolome
linking genotypes to treatments

SolveRD
Solving the Unsolved Rare Diseases

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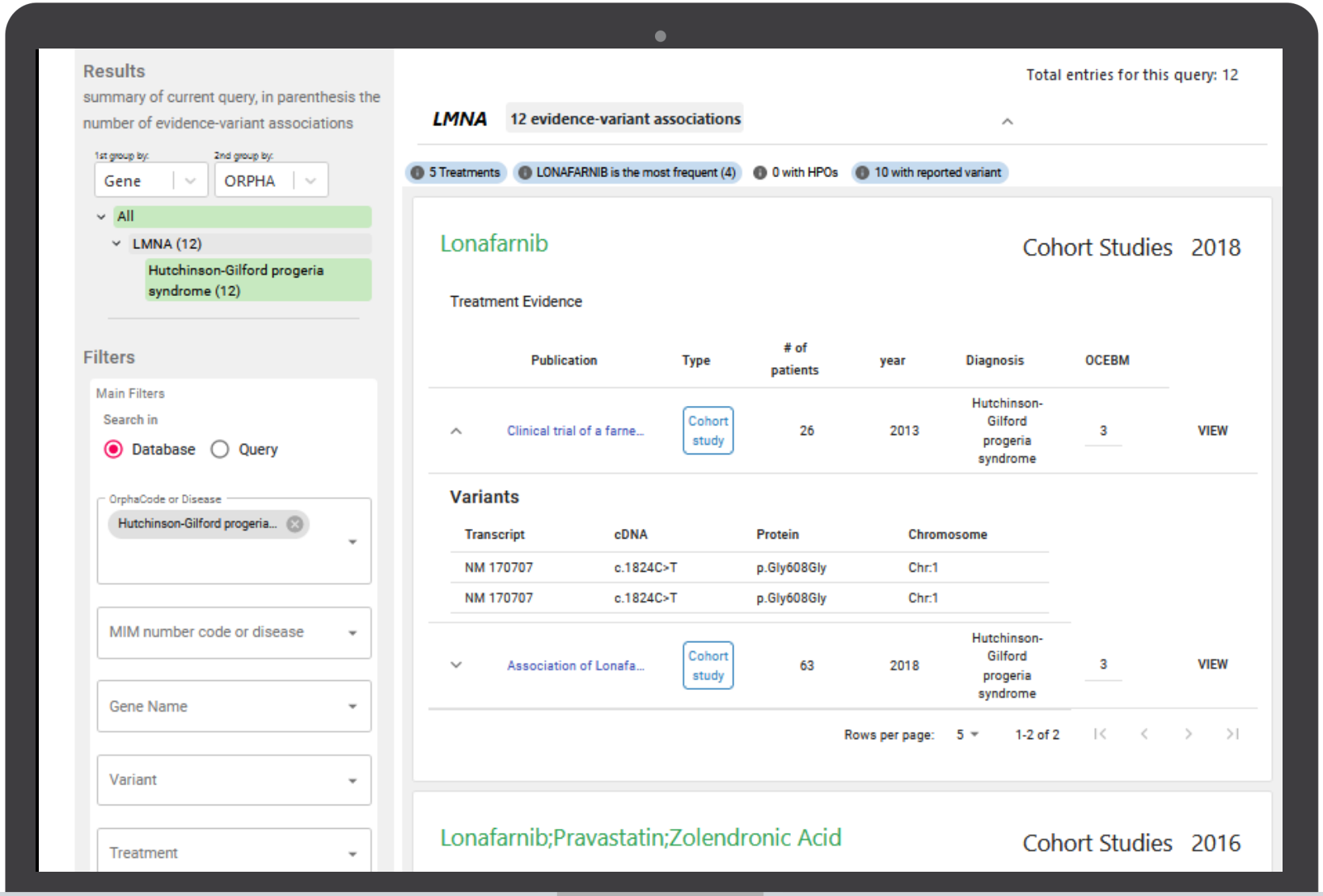
ERDERA
European Rare Diseases
Research Alliance

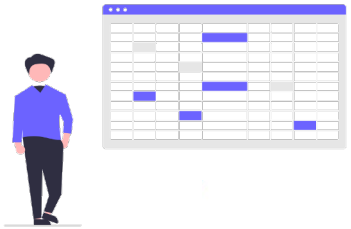
Inserm
Institut national de la santé et de la recherche médicale

CHEO RESEARCH INSTITUTE
INSTITUT DE RECHERCHE

MÉDECINE SORBONNE UNIVERSITÉ



Beyond diagnoses



treatabolome
linking genotypes to treatments

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Entries
3623

1192
Variants

745
publications

48 Orpha terms
and
88 OMIM terms

77
Genes

180
Treatments

Beyond diagnoses

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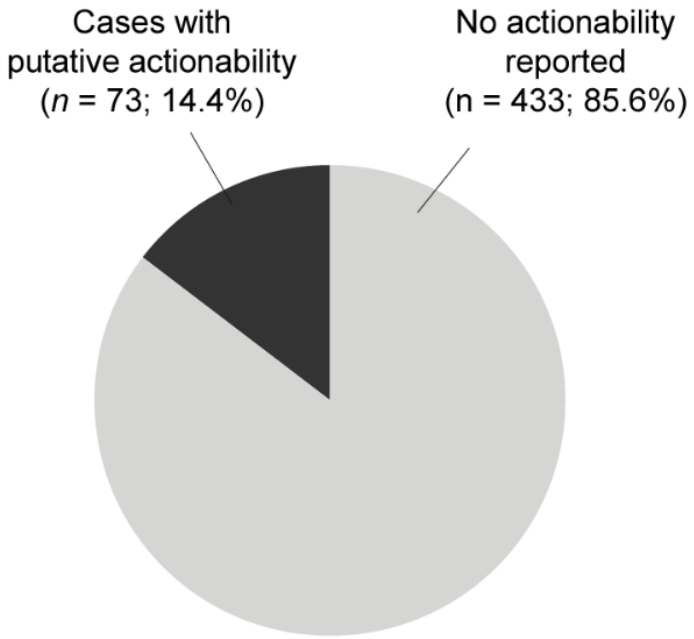
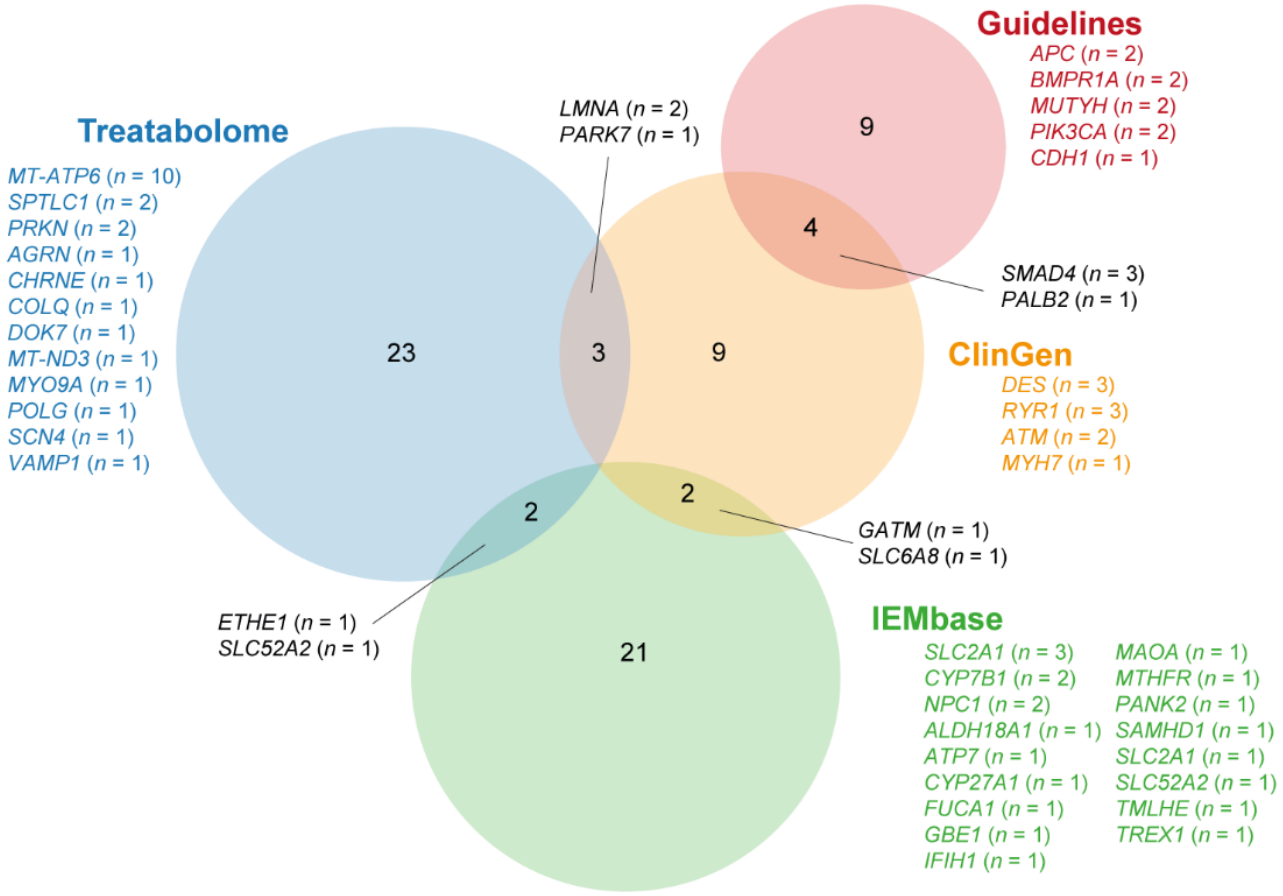
506 newly diagnosed families





The Solve-RD project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 779257.

ERDERA
European Rare Diseases Research Alliance

178 Organisations

40 funders

81 research performing organisations

9 patients' organisations

3 research infrastructures

22 private for-profit partners (industry & SME)

23 other (univ, hospital, non-profit, public administration)

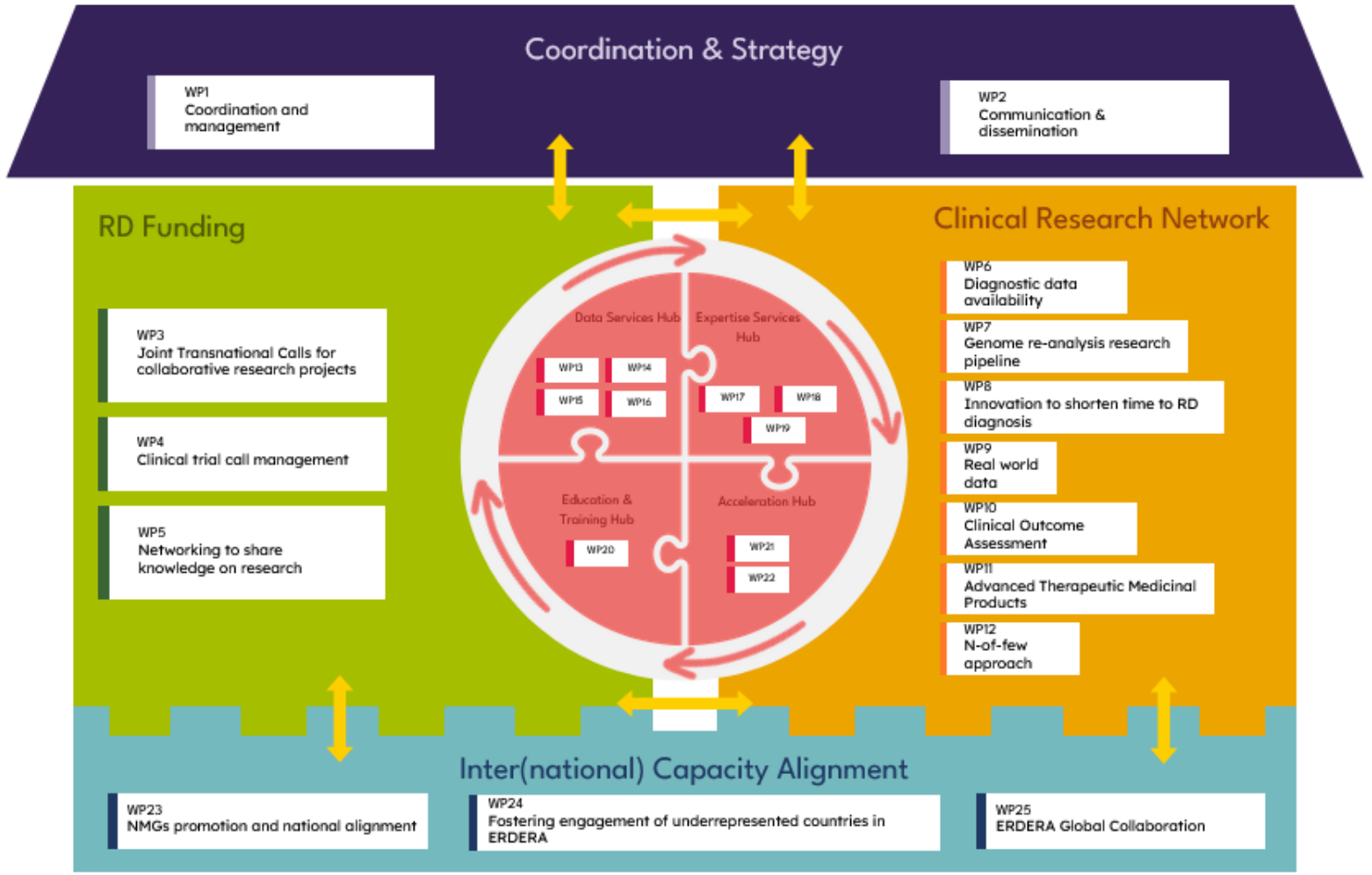
37 Countries

26 EU member states

8 associated countries

3 non-EU*

*at the time of proposal submission



WP13 Rare Diseases-Virtual Platform (RD-VP): Finding and accessing the data ecosystem

WP14 Data readiness services

WP15 Data sharing and analysis services

WP16 Knowledge bases and ontologies for RD research

WP17 Mentoring and consultancy

WP18 Regulatory support service

WP19 Methodological Support

WP20 Education and training in rare diseases research

WP21 Technology accelerator

WP22 Public-Private Collaboration Accelerator

Co-funded by the European Union

Screen4care – the project

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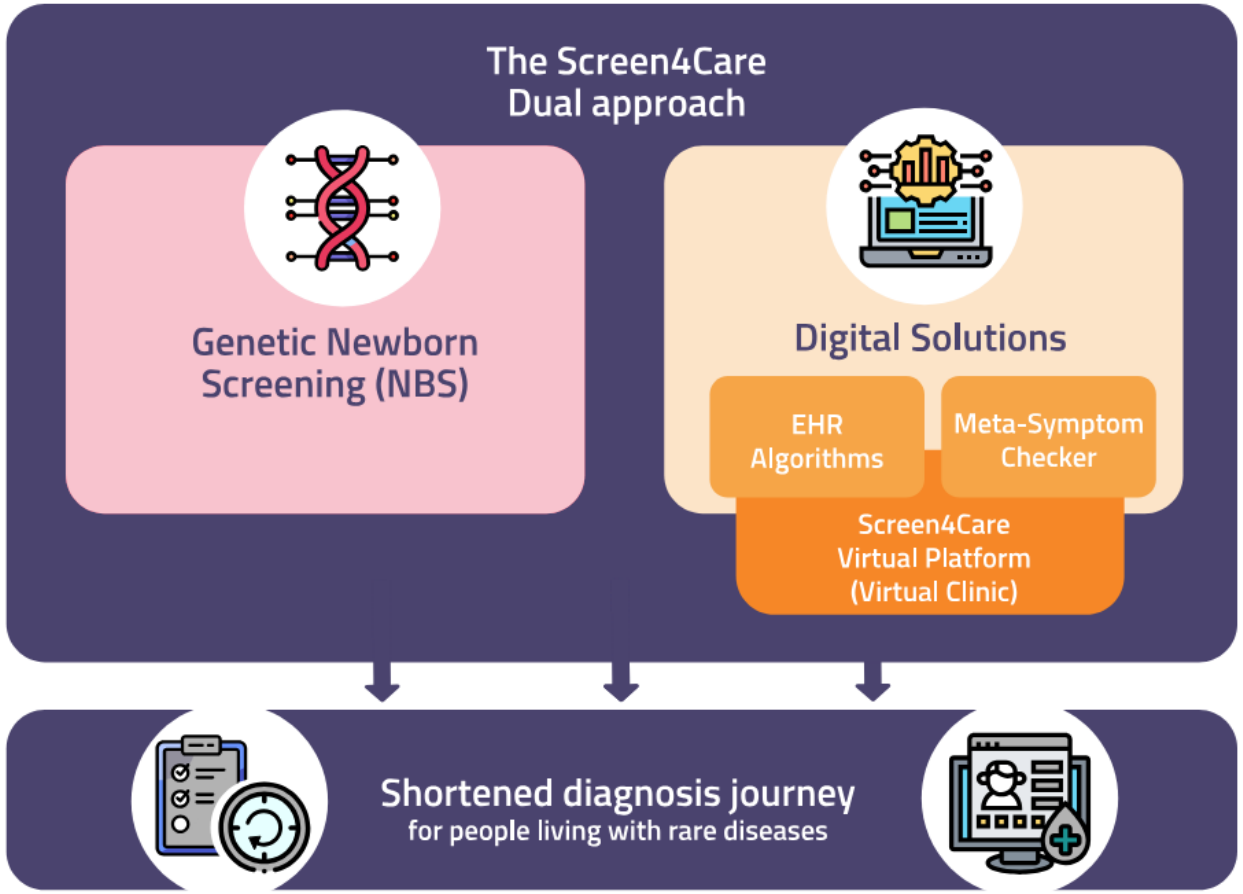
SCREEN 4CARE

The Innovative Medicines Initiative (IMI) is Europe's largest public-private initiative aiming to improve health by speeding up the development of, and patient access to, innovative medicines, particularly in areas where there is an unmet medical or social need.



Screen4care – gNBS: TREAT panel

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SCREEN 4CARE

245 genes

Saier, C., Sansen, S., Berghout, J. et al. Orphanet J Rare Dis 20, 231 (2025).

Disease category	Number of genes
Blood and coagulation disorders	33
Cardiological disorders	4
Endocrinological disorders	29
Immunological disorders	26
Kidney disorders	9
Metabolic disorders	106
Neurologic, neurodegenerative and neuromuscular disorders	25
Syndromic disorders	6
Others	7

Screen4care –Recruitment

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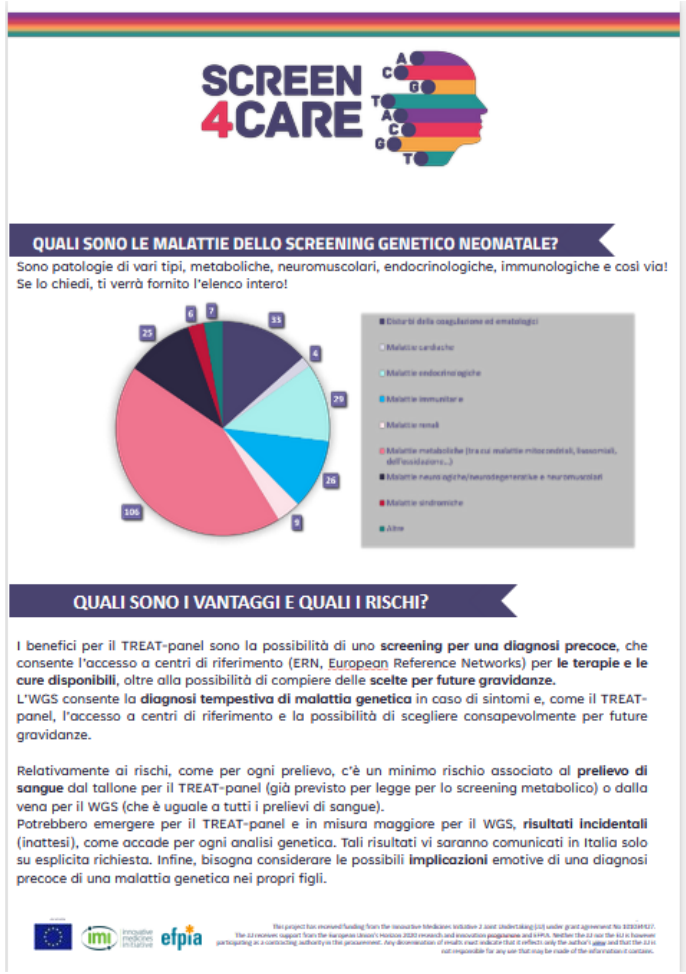
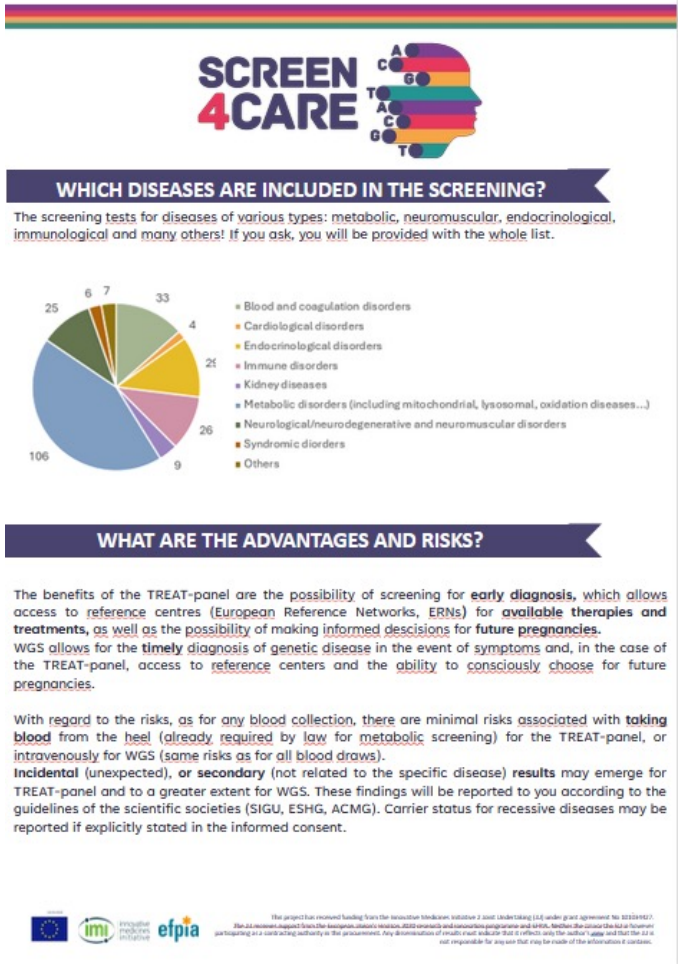




SCREEN 4CARE

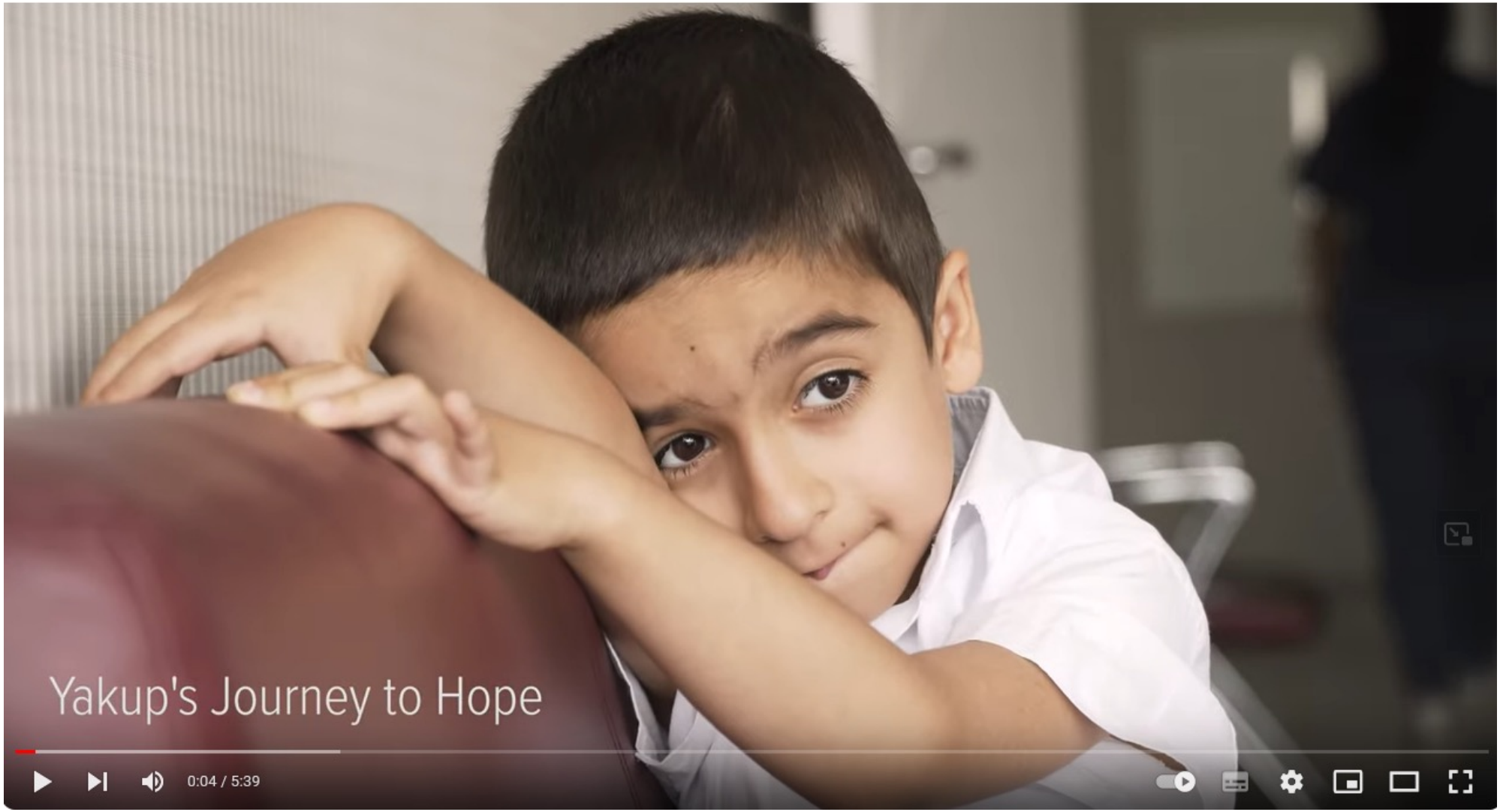
20,000 newborns

At birth centers in Germany, Italy and France during pregnancy and after birth:

- Flyers in different languages to explain complicated concepts in an "easy" way
- Videos
- Follow-up

Yakup's Journey to Hope

https://www.youtube.com/watch?v=GLgB6mvVx5E



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Científic de Barcelona
Clúster I
#SequencingForABetterLife

Other international initiatives where CNAG participates

cnaa

Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.

Search...

ABOUT US HOW WE WORK GA4GH TOOLKIT NEWS & EVENTS COMMUNITY CONTACT US

Enabling responsible genomic data sharing for the benefit of human health

The Global Alliance for Genomics and Health (GA4GH) is a policy-framing and technical standards-setting organization, seeking to enable responsible genomic data sharing within a human rights framework.

Genomic Data Toolkit →

Regulatory & Ethics Toolkit →

Data Security Toolkit →

Patient matchmaking to confirm diagnosis

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Matchmaker Exchange
Genomic discovery through the exchange of phenotypic & genotypic profiles

Global Alliance for Genomics & Health
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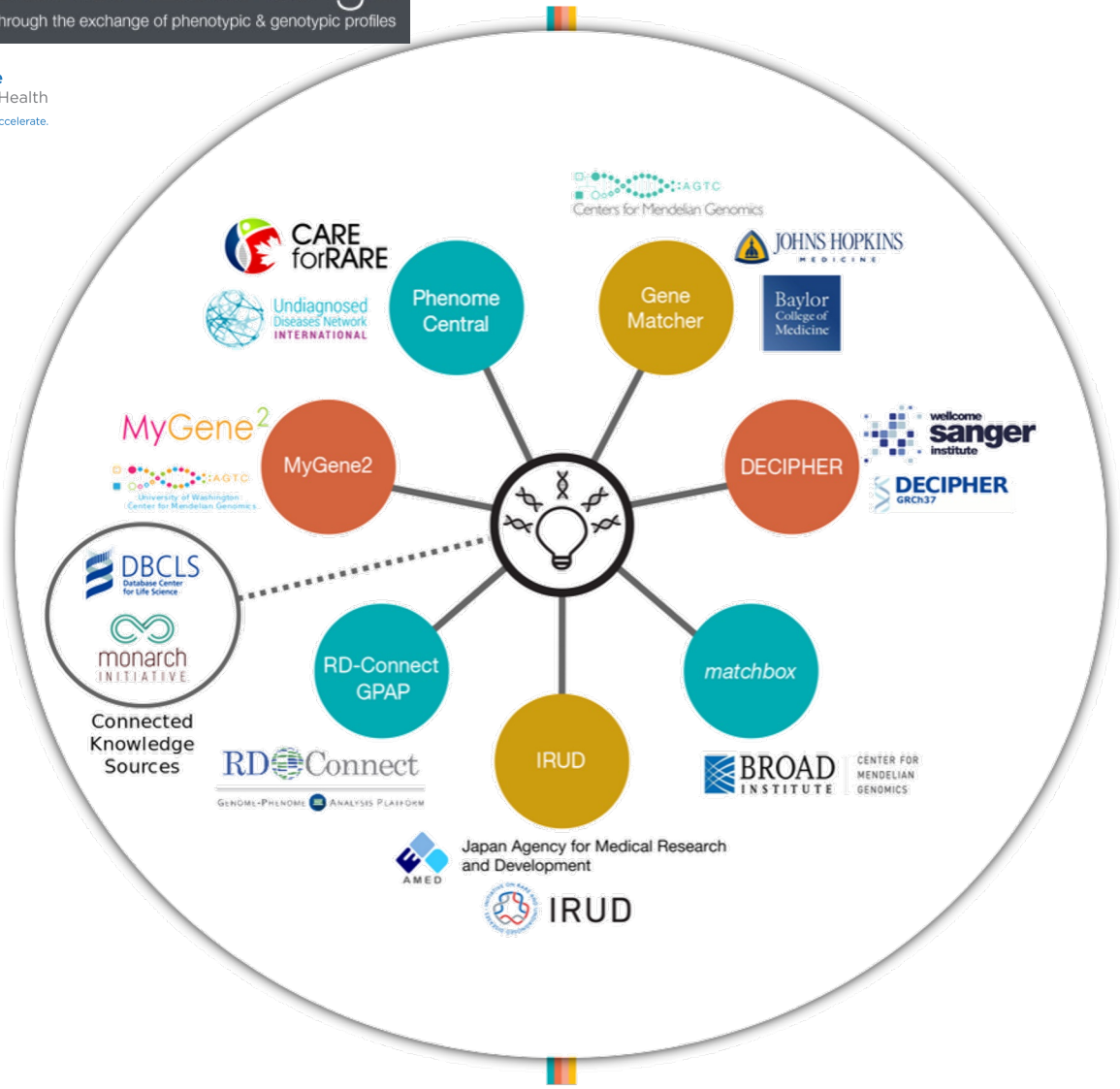
IRDiRC



Question: Do you have a patient with similar phenotype and genotype to "mine"?

Answer: a list of scored matches and the possibility of contacting the submitter of the dataset for follow-up.

Needs:

- Tagged genetic variants
- Machine-readable phenotypic information

Human Mutation Volume 36, Issue 10, pages 915-921, 17 SEP 2015 DOI: 10.1002/humu.22858
http://onlinelibrary.wiley.com/doi/10.1002/humu.22858/full#humu22858-fig-0001

1+ Million Genomes

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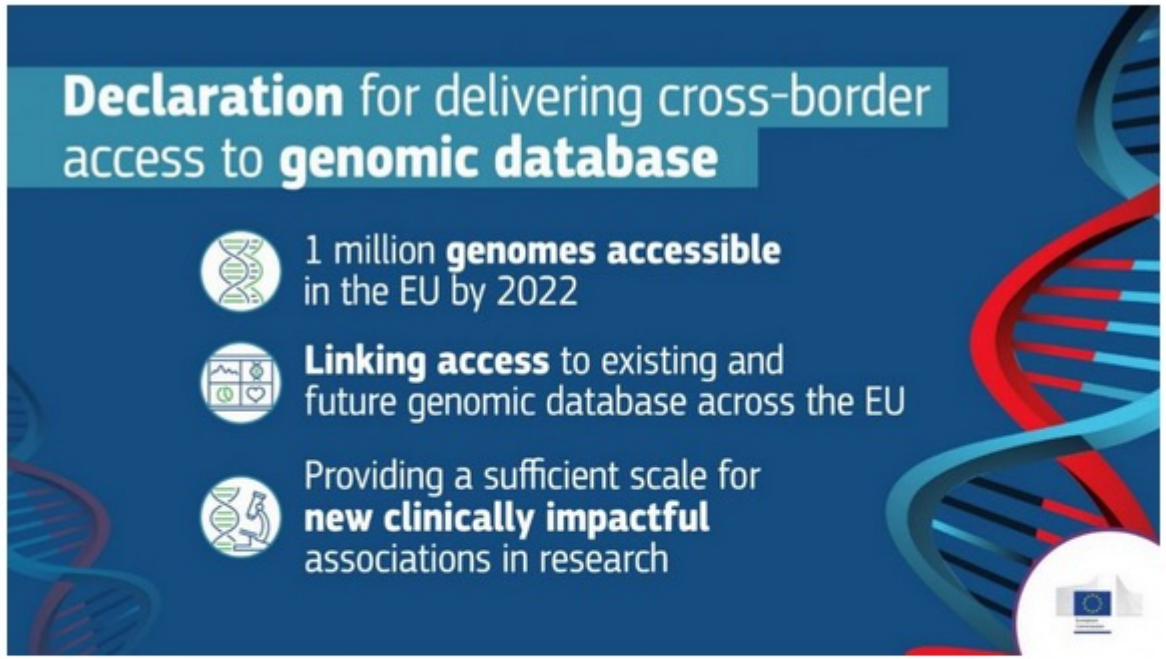


Since its launch on Digital Day 2018, the "1+ Million Genomes" initiative has grown into a real cooperation mechanism involving all 20 signatory Member States and Norway. These countries meet on a regular basis in order to make sure that the aim of the declaration – having at least 1 million sequenced genomes available in the EU by 2022- is achieved.

https://ec.europa.eu/digital-single-market/en/european-1-million-genomes-initiative

Leveraging European infrastructures to access 1 million human genomes by 2022

*Gary Saunders[1], Michael Baudis[2], Regina Becker[3], Sergi Beltran[4,5], Christophe Béroud[6,7], Ewan Birney[8], Cath Brooksbank[8], Søren Brunak[9,10], Marc Van den Bulcke[11], Rachel Drysdale[1], Salvador Capella-Gutierrez[12], Paul Flicek[8], Francesco Florindi[13], Peter Goodhand[14,15], Ivo Gut[4,5], Jaap Heringa[16], Petr Holub[13], Jef Hooyberghs[17], Nick Juty[18], Thomas M. Keane[8], Jan O. Korbel[19], Ilkka Lappalainen[20], Brane Leskosek[21], Gert Matthijs[22], Michaela Th. Mayrhofer[13], Andres Metspalu[23], Arcadi Navarro[24,25,26], Steven Newhouse[8], Tommi Nyrönen[20], Angela Page[15,27], Bengt Persson[28], Aarno Palotie[29], Helen Parkinson[8], Jordi Rambla[26], David Salgado[6], Erik Steinfelder[13], Morris A. Swertz[30], Alfonso Valencia[12,31], Susheel Varma[8], Niklas Blomberg[1] and Serena Scollen[1]**

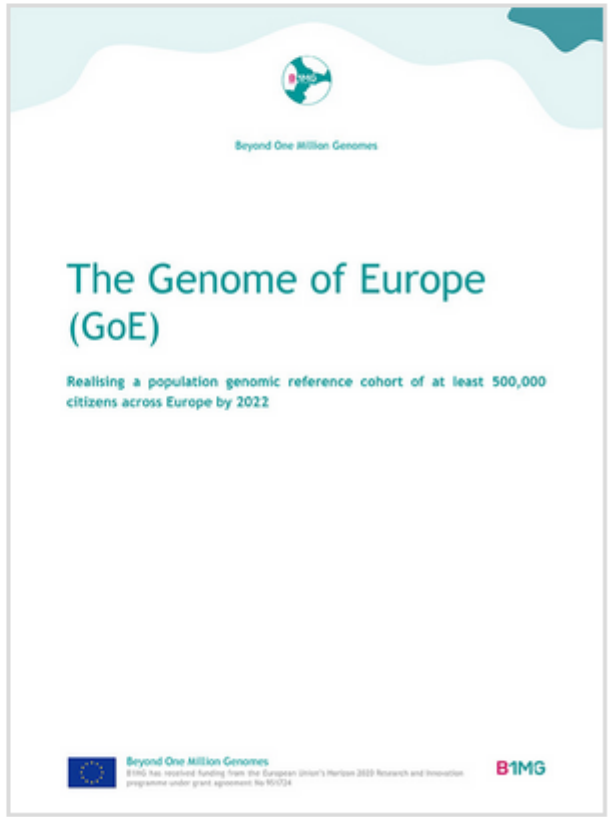
Nature Reviews Genetics volume 20, pp 693–701 (2019)

The Genome of Europe

The Genome of Europe initiative aims to build a European network of national genomic reference cohorts of at least 500,000 citizens. These reference cohorts will be selected to be representative of the European population.

To achieve this, each country involved will establish a population cohort that reflects the genetic composition of its population, including both healthy and diseased individuals. The country will then connect this data into a European cohort, thus establishing a collective European reference dataset.

The 500,000 sequenced genomes collected will contribute to the million genomes aimed for in the 1+MG initiative.



Find out more

- Support to 1+MG
- 1+MG Roadmap
- 1+MG Partner projects