Global Data Resources for Human Genomics & Health

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Australian BioCommons Webinar

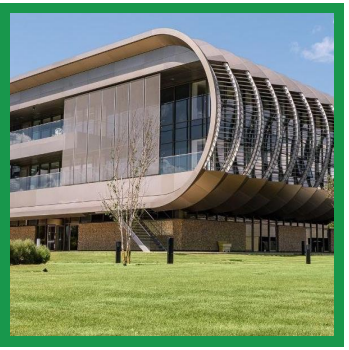
20 September 2024

EMBL-EBI

What is EMBL-EBI?

- World leading source of public biomolecular data
- Our vision is to benefit humankind by advancing scientific discovery and impact through bioinformatics.
- Part of the European Molecular Biology Laboratory (EMBL), Europe's flagship laboratory for the life sciences.

The European Molecular Biology Laboratory



EMBL-EBI

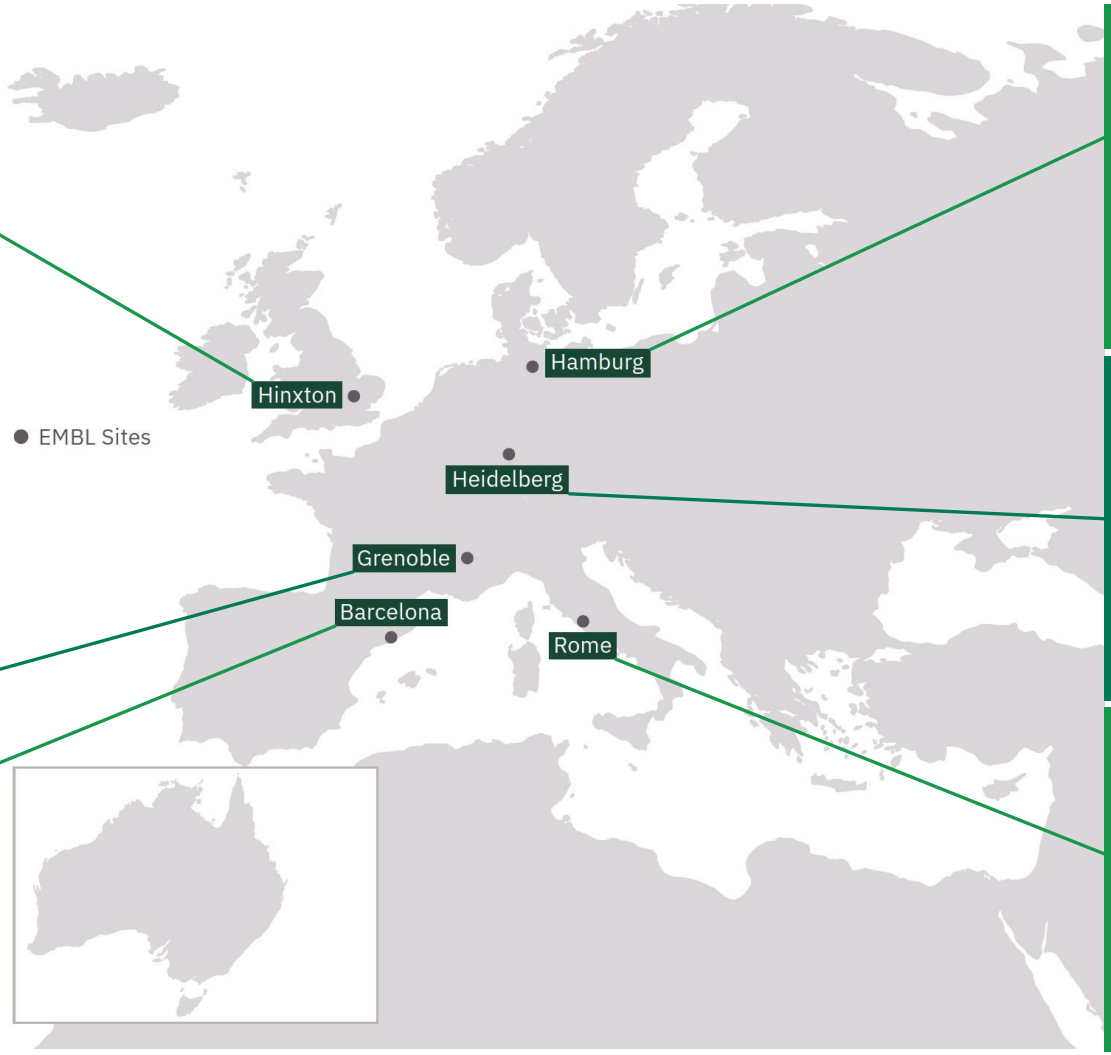
Bioinformatics

Grenoble

Structural biology

Barcelona

Tissue biology and disease modelling

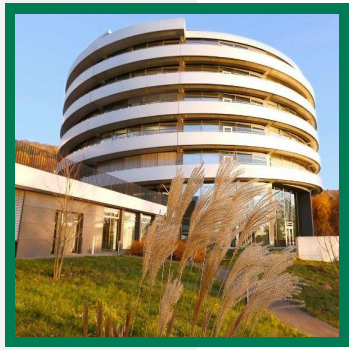


Hamburg

Structural biology

Heidelberg

Life sciences



Rome

Epigenetics and neurobiology

EMBL-EBI

EMBL member states

Member states (29)

Austria 1974
Denmark 1974
France 1974
Germany 1974
Israel 1974
Italy 1974
Netherlands 1974
Sweden 1974
Switzerland 1974
United Kingdom 1974
Finland 1984
Greece 1984
Norway 1985
Spain 1986
Belgium 1990
Portugal 1998
Ireland 2003
Iceland 2005
Croatia 2006
Luxembourg 2007
Czech Republic 2014
Malta 2016
Hungary 2017
Slovakia 2018
Montenegro 2018
Lithuania 2019
Poland 2019
Estonia 2023
Latvia 2024

Associate member state

Australia 2008

Prospect member states

Serbia

Bulgaria

EMBL-EBI's mission

Deliver data resources

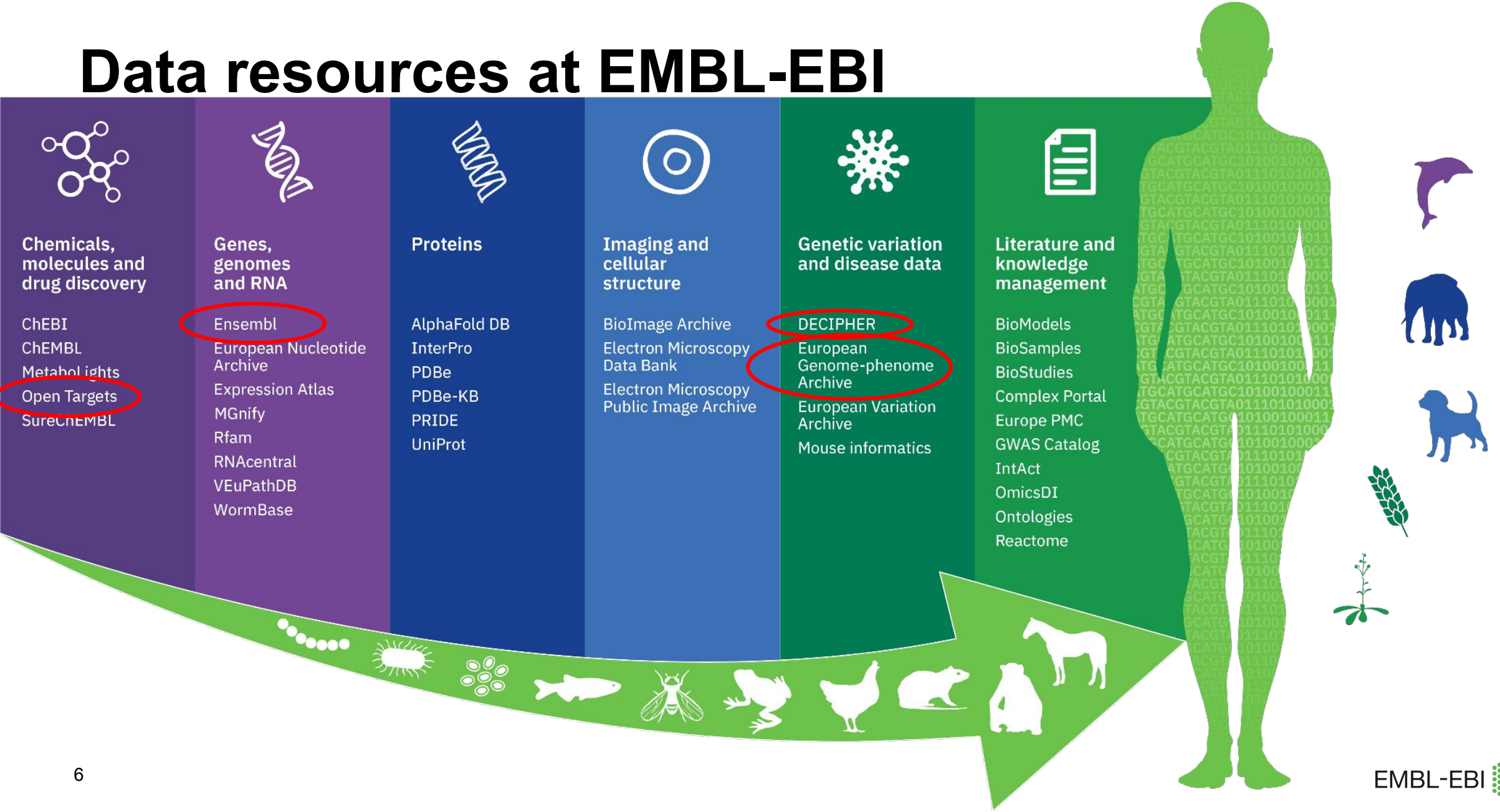
Perform excellent research

Train the next generation of scientists

Engage with industry

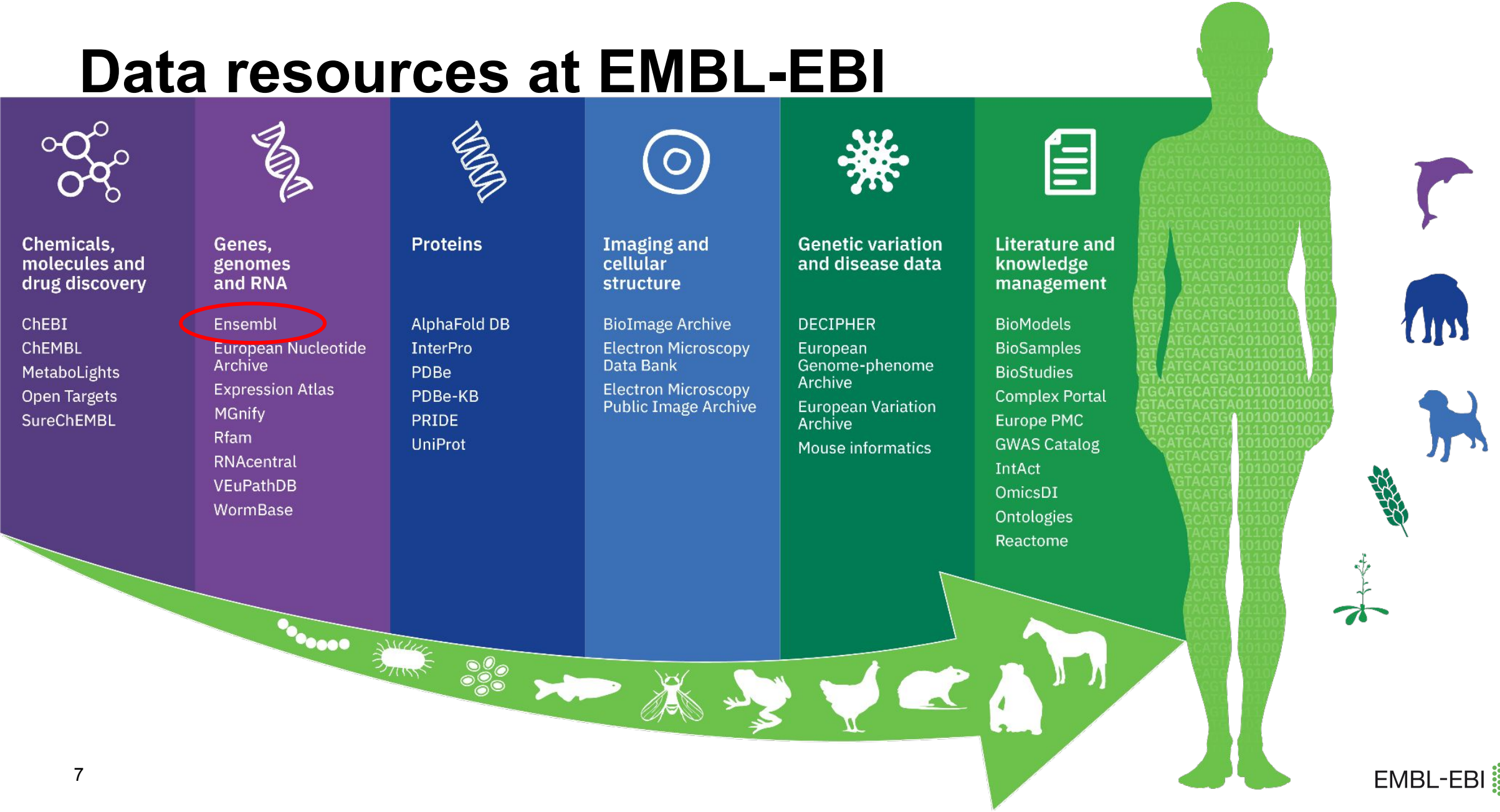
Coordinate bioinformatics in Europe

Data resources at EMBL-EBI



EMBL-EBI

Data resources at EMBL-EBI



EMBL-EBI

Tools

All tools

BioMart >

Export custom datasets from Ensembl with this data-mining tool

BLAST/BLAT >

Search our genomes for your DNA or protein sequence

Variant Effect Predictor >

Analyse your own variants and predict the functional consequences of known and unknown variants

Search

All species for

Go

e.g. **BRCA2** or **rat 5:62797383-63627669** or **rs699** or **coronary heart disease**

All genomes

-- Select a species --

Pig breeds

Pig reference genome and 12 additional breeds

View full list of all species

Favourite genomes

Human

GRCh38.p14

Still using GRCh37?

Mouse

GRCm39

Zebrafish

GRCz11

Ensembl is a genome browser for vertebrate genomes that supports research in comparative genomics, evolution, sequence variation and transcriptional regulation. Ensembl annotate genes, computes multiple alignments, predicts regulatory function and collects disease data. Ensembl tools include BLAST, BLAT, BioMart and the Variant Effect Predictor (VEP) for all supported species.

Ensembl Release 112 (May 2024)

- Many new fish genomes have been added to Ensembl
- Population frequency data is available for chicken, dog, goat and sheep through VEP
- Update to our current regulation annotation. The promoters now align with the 5' ends of known transcripts
- VEP will be updated to use the dbNSFP commercial data release

More release news on our blog

Ensembl Rapid Release

New assemblies with gene and protein annotation every two weeks.

Note: species that already exist on this site will continue to be updated with the full range of annotations.

Go

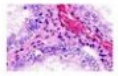
The Ensembl Rapid Release website provides annotation for recently produced, publicly available vertebrate and non-vertebrate genomes from biodiversity initiatives such as Darwin Tree of Life, the Vertebrate Genomes Project and the Earth BioGenome Project.

Rapid Release news on our blog

Compare genes across species

Find SNPs and other variants for my gene

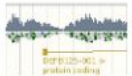
Gene expression in different tissues



Retrieve gene sequence

Find a Data Display

Use my own data in Ensembl



EMBL-EBI

Ensembl creates, integrates and distributes reference datasets and analysis tools that enable genomics. We are based at EMBL-EBI and our software and data are freely available.

Our acknowledgements page includes a list of current and previous funding bodies. How to cite Ensembl in your own publications.

GLOBAL CORE BIODATA RESOURCE

elixir Core Data Resource

https://beta.ensembl.org/

ENSEMBL

Genome data & annotation

About using Ensembl

Species selector

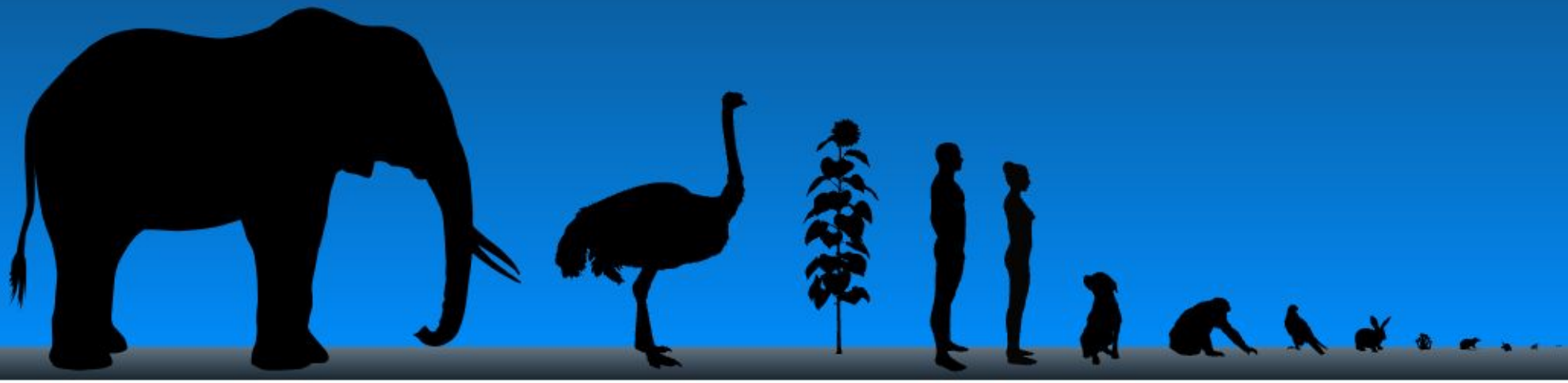
Create & manage your own species list

Genome browser

Look at genes & transcripts in their genomic context

Entity viewer

Get gene & transcript information



What is Ensembl?

```
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AGTAAGCTGTTACCGTTCCAGGAGATGGGACTGAATTAGAATTCAAACAAATTTTCCAGCGCTT
CTGAGTTTTACCTCAGTCACATAATAAGGAATGCATCCCTGTGTAAGTGCATTTTGGTCTTCTG
TTTTGCAGACTTATTTACCAAGCATTGGAGGAATATCGTAGGTAAAAATGCCTATTGGATCCAA
AGAGAGGCCAACATTTTTTGAAATTTTTAAGACACGCTGCAACAAAGCAGGTATTGACAAATTT
TATATAACTTTATAAATTACACCGAGAAAGTGTTTTCTAAAAAATGCTTGCTAAAAACCCAGTA
CGTCACAGTGTTGCTTAGAACCATAAACTGTTCCTTATGTGTGTATAAATCCAGTTAACAACAT
AATCATCGTTTGCAGGTTAACCACATGATAAATATAGAACGTCTAGTGGATAAAGAGGAAACTG
GCCCCTTGACTAGCAGTAGGAACAATTACTAACAAATCAGAAGCATTAATGT
```

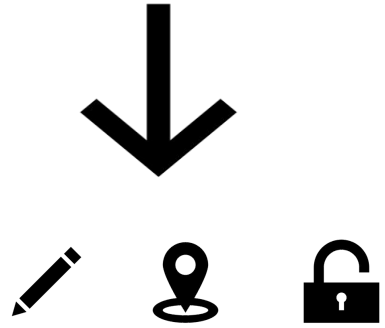

Ensembl annotates and maps genomic features from genome sequences

What is Ensembl?

```
AGTGCTTAATGTGGCTAGTGGCACCGGTTTGGACAGCACAGCTGTAAAATGTTCCCATCCTCAC
AGTAAGCTGTTACCGTTCCAGGAGATGGGACTGAATTAGAATTCAAACAAATTTTCCAGCGCTT
CTGAGTTTTACCTCAGTCACATAATAAGGAATGCATCCCTGTGTAAGTGCATTTTGGTCTTCTG
TTTTGCAGACTTATTTACCAAGCATTGGAGGAATATCGTAGGTAAAAATGCCTATTGGATCCAA
AGAGAGGCCAACATTTTTTGAAATTTTTAAGACACGCTGCAACAAAGCAGGTATTGACAAATTT
TATATAACTTTATAAATTACACCGAGAAAGTGTTTTCTAAAAAATGCTTGCTAAAAACCCAGTA
CGTCACAGTGTTGCTTAGAACCATAAACTGTTCCTTATGTGTGTATAAATCCAGTTAACAACAT
AATCATCGTTTGCAGGTTAACCACATGATAAATATAGAACGTCTAGTGGATAAAGAGGAAACTG
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```
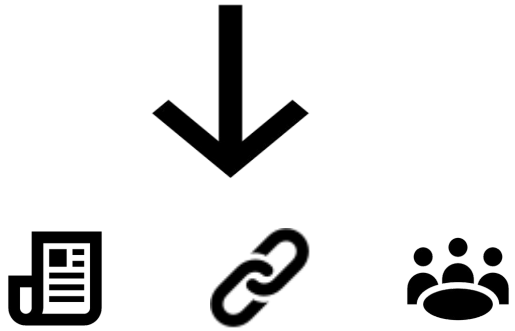
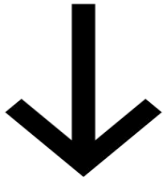



Ensembl is an ‘added value resource’ bringing together information from a wide range of other databases in a single site

What is Ensembl?

```
AGTGCTTAATGTGGCTAGTGGCACCGGTTTGGACAGCACAGCTGTAAAATGTTCCCATCCTCAC
AGTAAGCTGTTACCGTTCCAGGAGATGGGACTGAATTAGAATTCAAACAAATTTTCCAGCGCTT
CTGAGTTTTACCTCAGTCACATAATAAGGAATGCATCCCTGTGTAAGTGCATTTTGGTCTTCTG
TTTTGCAGACTTATTTACCAAGCATTGGAGGAATATCGTAGGTAAAAATGCCTATTGGATCCAA
AGAGAGGCCAACATTTTTTGAAATTTTTAAGACACGCTGCAACAAAGCAGGTATTGACAAATTT
TATATAACTTTATAAATTACACCGAGAAAGTGTTTTCTAAAAAATGCTTGCTAAAAACCCAGTA
CGTCACAGTGTTGCTTAGAACCATAAACTGTTCCTTATGTGTGTATAAATCCAGTTAACAACAT
AATCATCGTTTGCAGGTTAACCACATGATAAATATAGAACGTCTAGTGGATAAAGAGGAAACTG
GCCCCTTGACTAGCAGTAGGAACAATTACTAACAAATCAGAAGCATTAATGT
```



NIH U.S. National Library of Medicine | NCBI

Genome Data Viewer

www.ncbi.nlm.nih.gov/gdv

e!Ensembl

www.ensembl.org (vertebrates)

e!EnsemblGenomes

www.ensemblgenomes.org
(non-vertebrates)

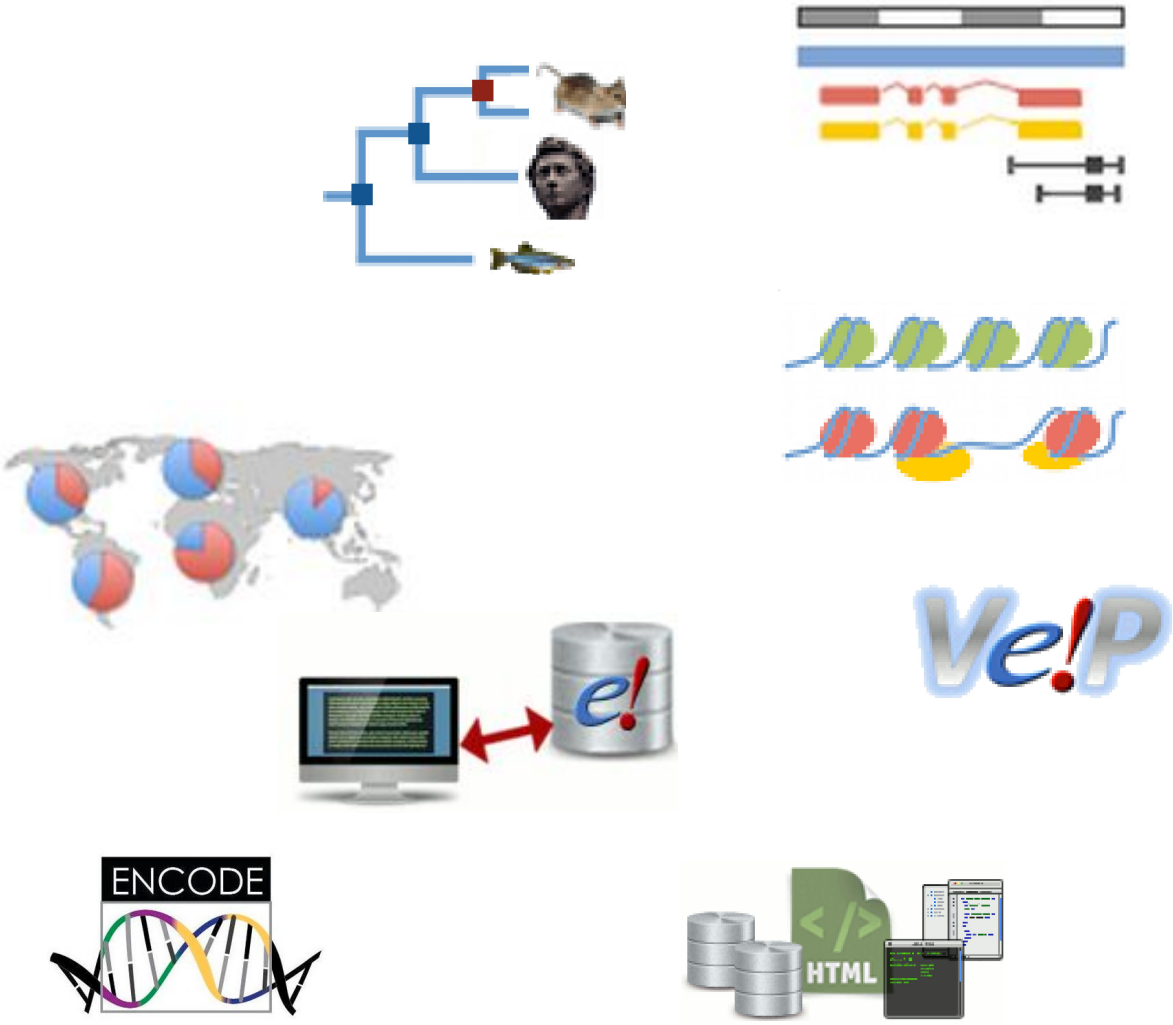
UCSC

www.genome.ucsc.edu

EMBL-EBI

Ensembl features

- Genomes and gene builds for >300 species
- Variation data
- Compara (alignments, gene trees, homologues)
- Regulatory builds
- BioMart (data export)
- Tools for data processing, e.g. Ensembl VEP
- Display your own data
- Programmatic access via APIs
- Completely Open Source (FTP, GitHub)



Ensembl Variant Effect Predictor (VEP)

dbSNP
Short Genetic Variations

1000 Genomes

gnomAD
genome aggregation database

HGMD®

NHGRI
GWAS
Catalog

ClinVar

orphanet

COSMIC

Europe PubMed Central

Reference Data

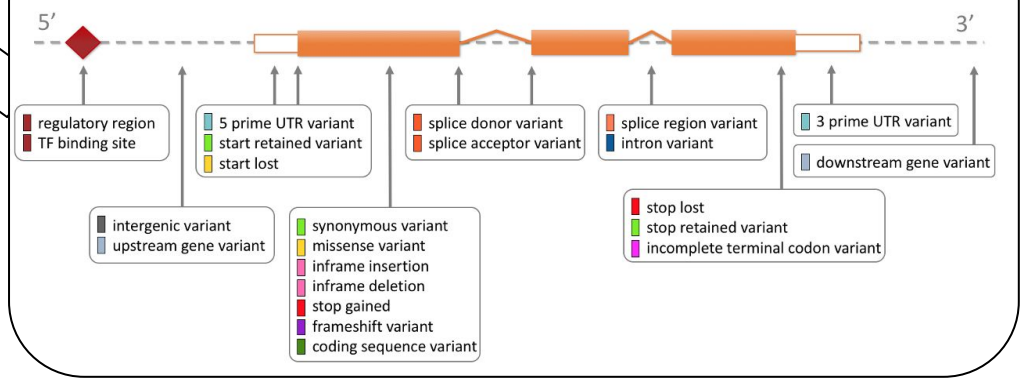
- Ensembl transcripts
- RefSeq transcripts
- Variants, citations frequencies
- Regulatory features + motifs
- Disease/ phenotype associations

Variants

Algorithms

- missense classifiers
- CADD
- splice classifiers
- conservation

Sequence ontology



Consequence	Symbol	Feature	HGVSp	Codons	SIFT	PolyPhen	gnomAD AFR AF	gnomAD FIN AF	Clinical significance
missense_variant	AGT	ENST00000366667	ENSP00000355627.4:p.Thr207Met	ACG/ATG	0.01	0.997	0.06221	0.1761	likely_benign

Acknowledgements

Funding

EMBL

Open Targets

National Human Genome Research Institute (NHGRI)

National Institute of Allergy and Infectious Diseases (NIAID)

Funded by the European Union

UK Research and Innovation

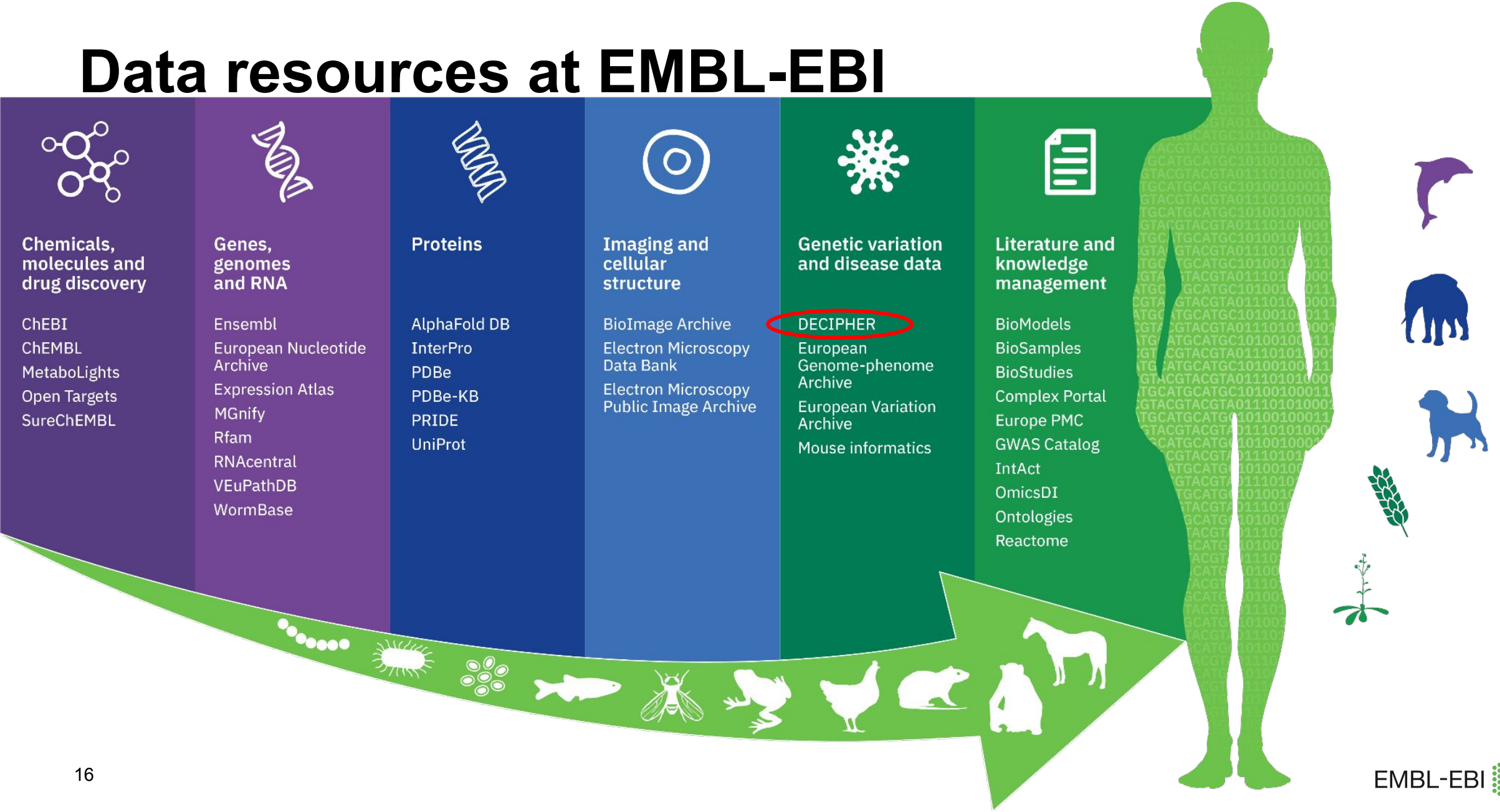
TRANSFORMING GENOMIC MEDICINE INITIATIVE

elixir

wellcome

EMBL-EBI

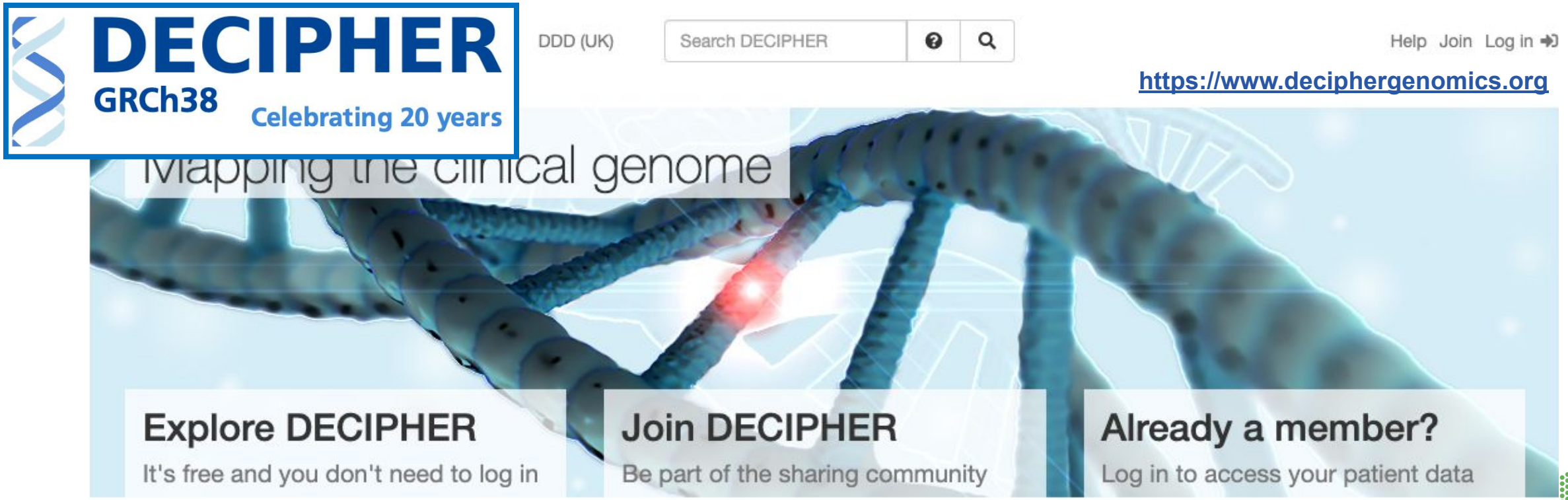
Data resources at EMBL-EBI



EMBL-EBI

DECIPHER is a web-based platform which helps **clinical** and **research teams** to **assess pathogenicity** and to **share** patient data

Help **integrate genotype & phenotype data** to determine the consequences of variants for health and disease



DECIPHER supports data sharing and linking

Matchmaker Exchange

Centers for Mendelian Genomics
JOHNS HOPKINS MEDICINE
Baylor College of Medicine
CARE forRARE
Undiagnosed Diseases Network INTERNATIONAL
Gene Matcher
Phenome Central
Patient Matcher
MyGene2
University of Washington Center for Mendelian Genomics
MyGene2
DECIPHER
EMBL-EBI
DECIPHER
DBCLS Database Center for Life Science
monarch INITIATIVE
Connected Knowledge Sources
RD-Connect GPAP
RD-Connect GENOME-PHENOME ANALYSIS PLATFORM
IRUD
IRUD
Japan Agency for Medical Research and Development
BCH seqr
Boston Children's Hospital Children's Rare Disease Cohorts Initiative
Broad seqr
BROAD INSTITUTE CENTER FOR MENDELIAN GENOMICS
ModelMatcher
Functional Study Node

GA4GH
DRIVER PROJECT

DECIPHER
GRCh38
Celebrating 20 years

G2P
EMBL-EBI
e!Ensembl
VeP
OMIM®
UniProt
Pfam
gnomAD
ExAC
HGNC HUGO Gene Nomenclature Committee
NCBI
ClinGen Clinical Genome Resource
ISCA
HGMD®
ClinVar
Locus Specific Database
dbSNP Short Genetic Variations

DECIPHER by the numbers

>49,000 anonymised, open records

>59,000 variants

>197,000 phenotypes (HPO)

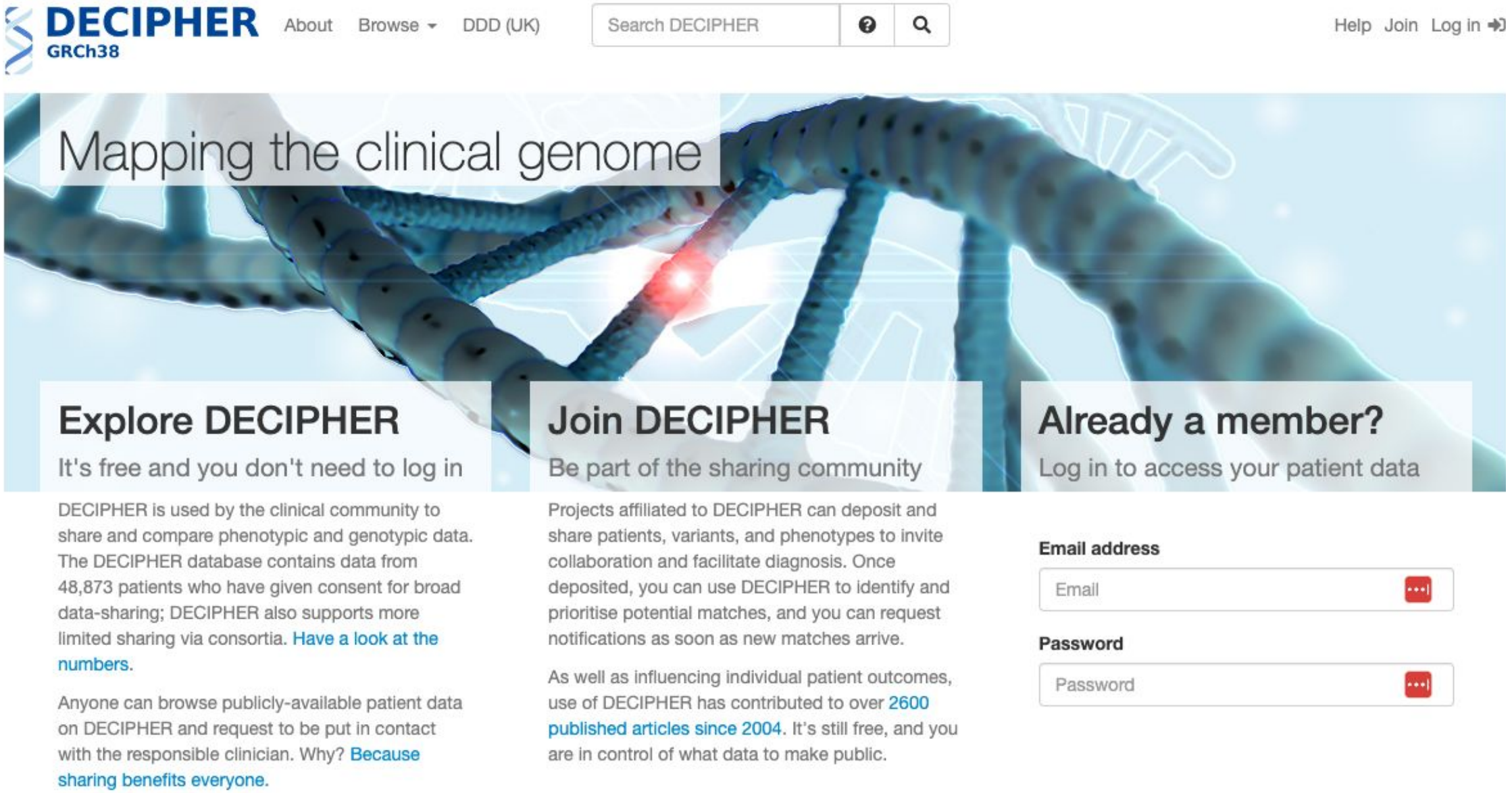
>3,500 publications citing DECIPHER

>320 depositing centres

45 countries with depositing centres

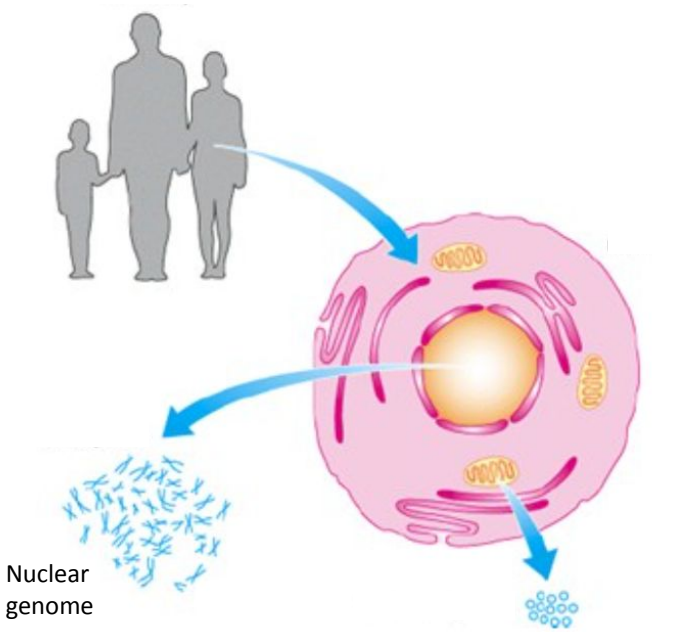
7 consortia, >73,000 patient records

What can I do/see in DECIPHER?



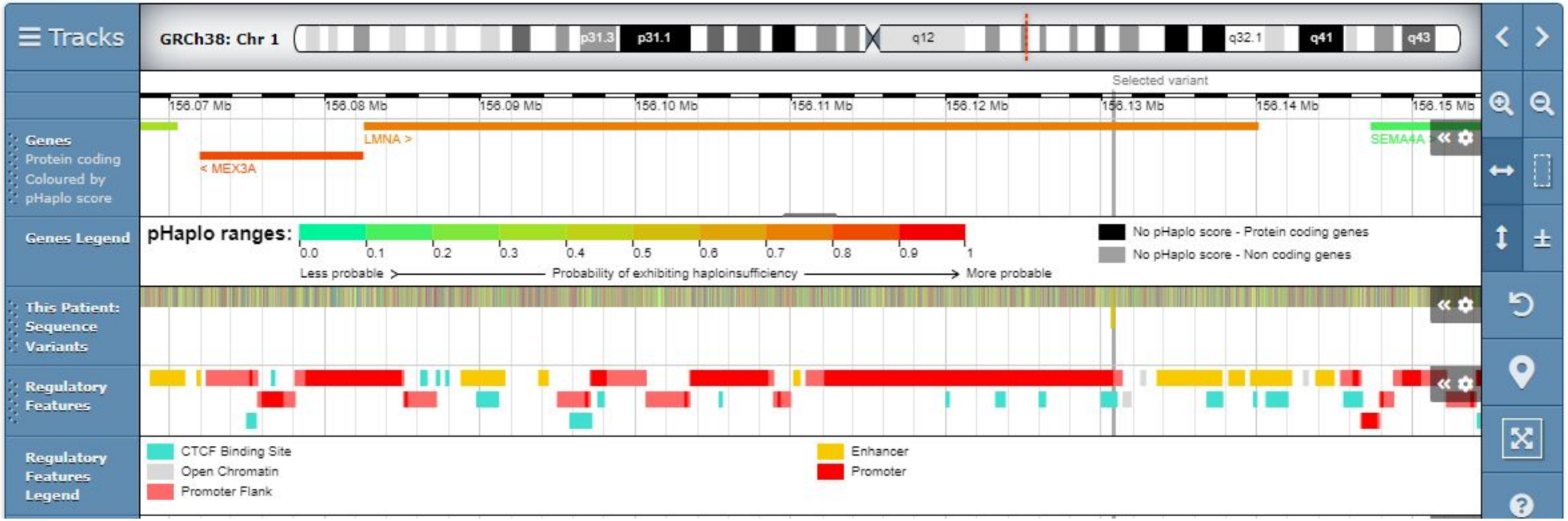
DECIPHER supports many types of genetic variation

Sequence Variant	Aneuploidy/Segmental aneuploidy	Insertion
Sequence Variant	Nullisomy	Mobile Element: Alu
Copy-Number Variant	Monosomy	Mobile Element: LINE1
Deletion	Disomy	Mobile Element: SVA
Duplication	Trisomy	Retrogene
Duplication/Triplication	Tetrasomy	Other Insertion
Triplication	**Uniparental Disomy**	**Short Tandem Repeat**
Amplification (> Triplication)	Isodisomy	Short Tandem Repeat
	Heterodisomy	**Inversion**
	Unknown	Inversion



Location	Type	Genes	Size	Annotations	Inheritance / Genotype	Pathogenicity / Contribution	Shared	Links
12 7045880 7045882 C A G Repeated > 50 times	Short Tandem Repeat Deposited 2020-06-17	ATN1	> 150 bp		Unknown Heterozygous	Click here		Show more
15 22749354 28438266	Uniparental Isodisomy Deposited 2020-06-17	119	5.69 Mb		Maternally inherited	Click here		Show more
16 56370773 56370773 G > A	Sequence Variant Deposited 2020-10-07	GNAO1	SNV	splice_donor_variant ENST00000262494 c.723+1G>A SpliceAI: ΔS donor loss 1.00 Not in gnomAD: see coverage	De novo Heterozygous	Likely pathogenic Full PM2 PP3 PS2 Last annotated: 2020-10-07		Show more
5 88100580 88100580	Insertion of LINE1 element Deposited 2020-06-17	MEF2C	6.02 kb insertion Forward strand		De novo Homozygous	Click here		Show more
11 118374637 118374637	Insertion Deposited 2020-06-17	KMT2A	450.00 kb insertion Sequence from 1:30000-480000		Unknown Homozygous	Click here		Show more
X 154156796 154227925	Inversion Deposited 2020-06-17	F8	71.13 kb		De novo, mosaic Hemizygous	Click here		Show more
6 156392359 158464899	Duplication/Triplication Deposited 2020-06-17	11	2.07 Mb	Dosage sensitivity score: 1.00 Sampling probability: > 5%	De novo, mosaic Homozygous	Click here		Show more

DECIPHER supports visualisation on GRCh38



Transcript	Protein change	VEP Consequence	Other annotations	
ENST00000368300.9 - NM_170707.4 MANE Select · Selected transcript ENST00000368300.9:c.490G>A	D/N at position 164 of 664 ENSP00000357283.4:p.Asp164Asn	missense_variant	Sift PolyPhen CADD REVEL SpliceAI	Deleterious (0.02) Benign (0.063) 25.8 0.55 ≤ 0.2
ENST00000677389.1 - NM_005572.4 MANE Plus Clinical ENST00000677389.1:c.490G>A	D/N at position 164 of 572 ENSP00000503633.1:p.Asp164Asn	missense_variant	Sift PolyPhen CADD REVEL SpliceAI	Deleterious (0.01) Benign (0.037) 25.8 0.55 ≤ 0.2

DECIPHER supports granular phenotype display

Patient 6 | Mother 0 | Father 0

Patient phenotypes

Abnormality of head or neck

Cleft palate | Dimple chin

Abnormality of limbs

Broad thumb

Abnormality of the nervous system

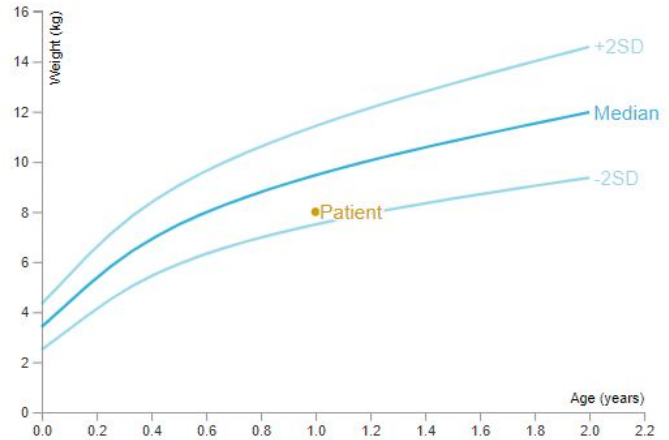
Delayed speech and language development | Specific learning disability

Abnormality of the skeletal system

Clinodactyly of the 5th finger

Milestones | Measurements

Milestones	Measurements
Social smile	Growth
Sat Independently	Height
Walked Independently	✔ Weight
First words	✔ Head circumference
	Visual Function
	Visual acuity
	Area of preserved field
	Area of scotoma
	Flicker ERG timing
	Fundus imaging
	Area of atrophy (AF)
	Area within hyperAF ring
	Central subfield thickness
	Macular thickness
	Ellipsoid zone length (H)
	Ellipsoid zone length (V)



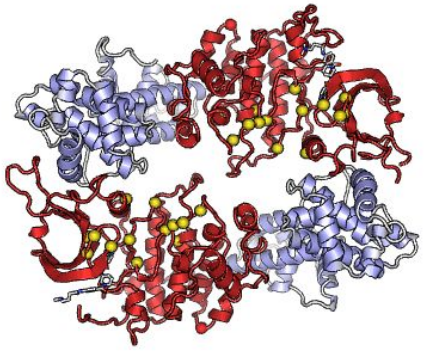
Weight

Age	Value	Percentile	SD
1 year	8 kg	5	-1.63 SD

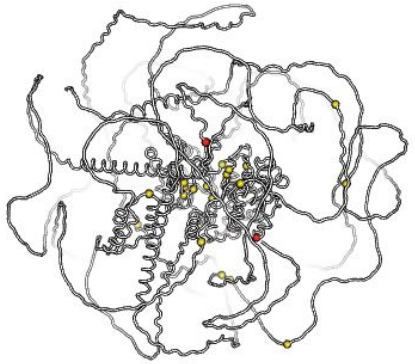
DECIPHER supports protein structure visualisations

CDK13

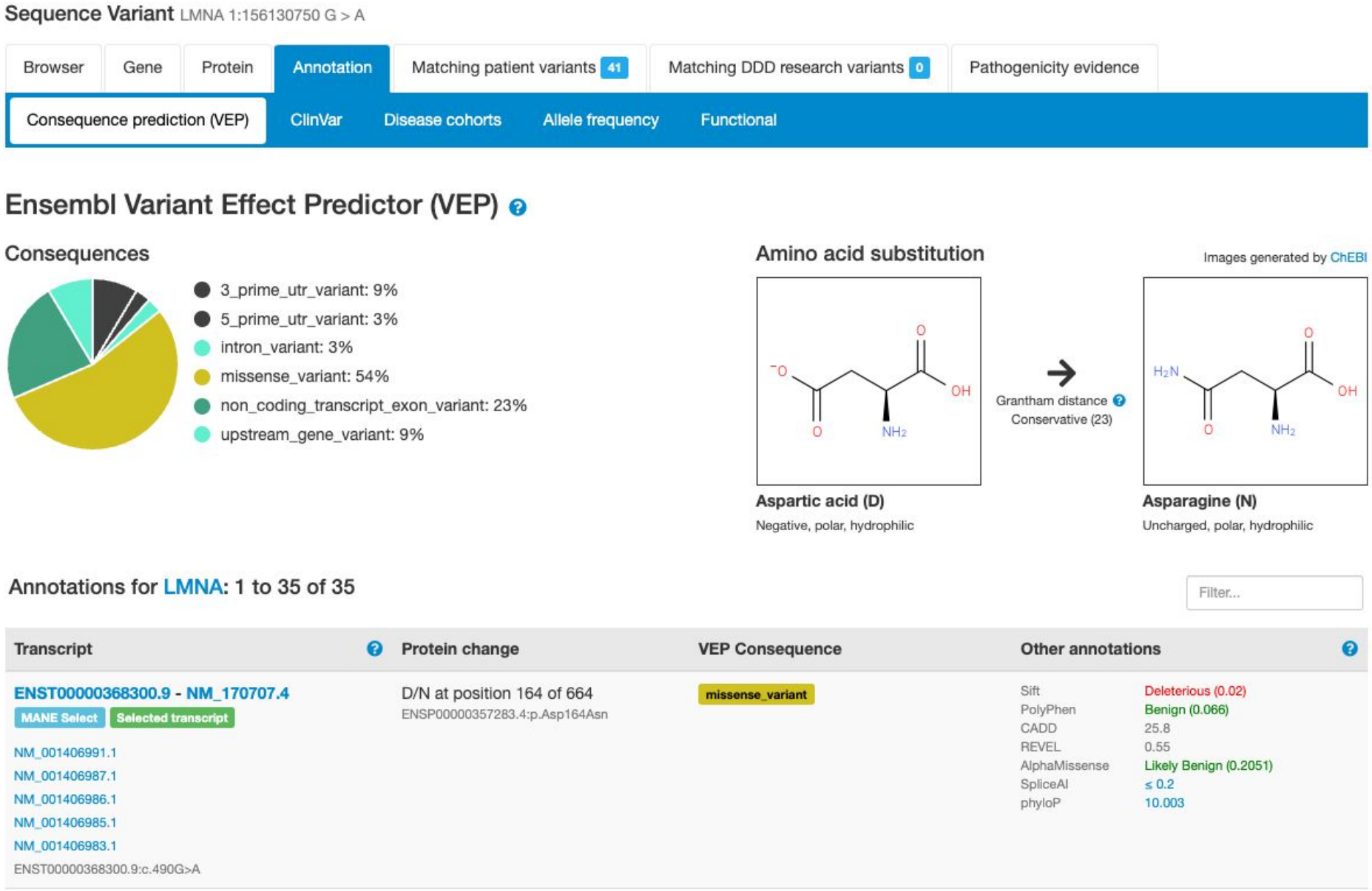
PDBe



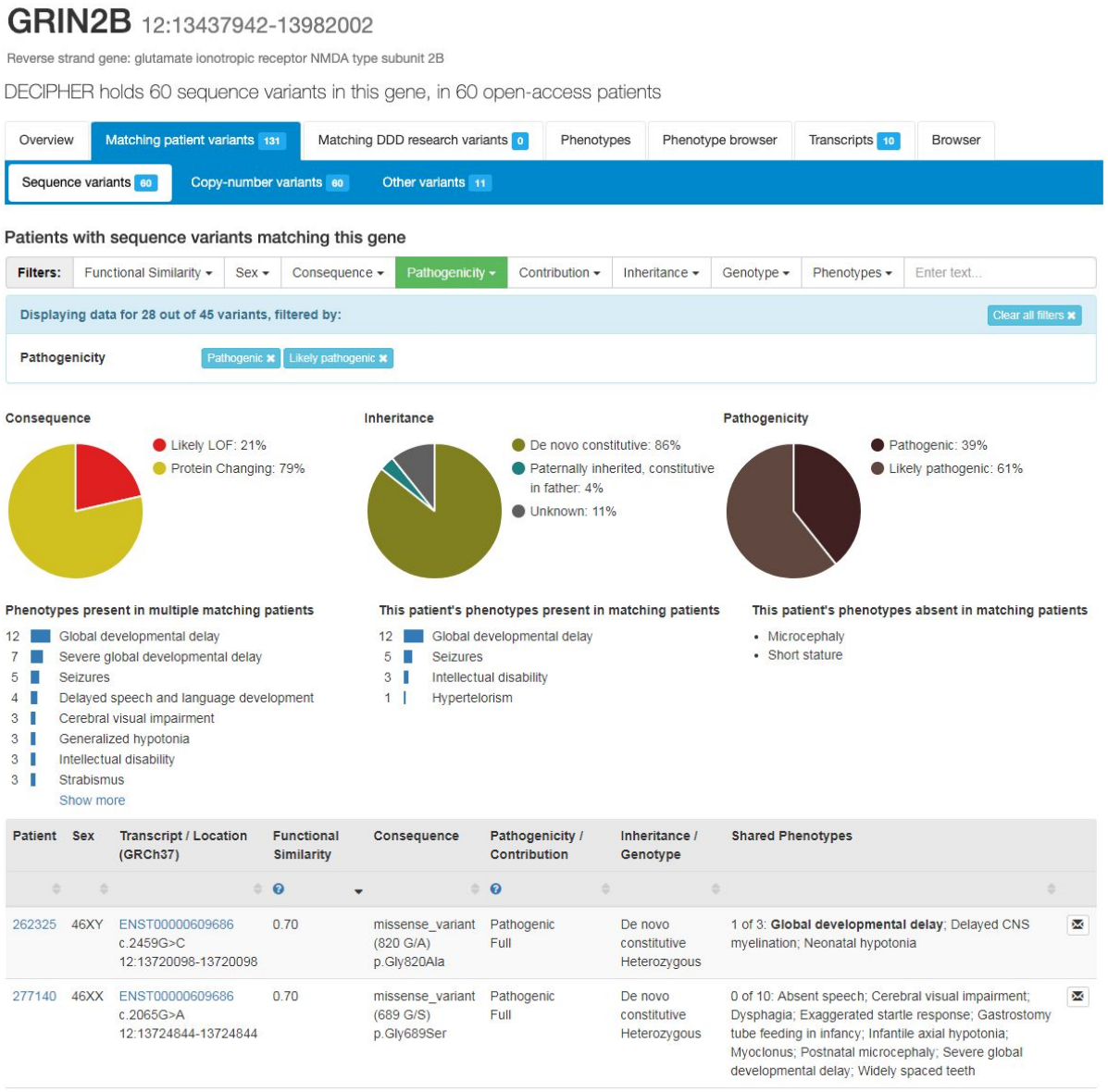
AlphaFold



DECIPHER supports Ensembl Variant Effect Predictor



DECIPHER supports matching patients



- Inform diagnosis
- New phenotypes associated with disease
- New variant-disease pairings

DECIPHER links to disease management resources

ATP7B 13:51930436-52012125

Reverse strand gene: ATPase copper transporting beta

Formerly known as: **WND**

Also known as: **ENSG00000123191**

Function: "Copper ion transmembrane transporter involved in the export of co
from hepatocytes into the bile in response to copper overload." *Source: Uni*

DECIPHER holds 4 sequence variants in this gene, in 4

Overview | Matching patient variants 70 | Matching DDD research v

Clinical | Protein / Genomic

Gene/disease association

Gene2Phenotype
-

OMIM
606882

Morbid
Wilson disease (Autosomal recessive)

GeneReviews
- Hereditary Dystonia
- Wilson Disease

ClinGen gene/disease
- Wilson disease **Definitive; AR**

ClinGen Dosage Sensitivity
- Haploinsufficiency: Gene associated with autosomal recessive phenotype (30)
- Triplosensitivity: No evidence for dosage pathogenicity (0)

GenCC
Definitive: 1
Strong: 2
Supportive: 1
(Assessed by ClinGen, Genomics England PanelApp, Orphanet, Invitae, with respect to Autosomal recessive inheritance)

ClinGen Clinical Actionability

ATP7B has been curated by ClinGen, with respect to clinical actionability.

Filter...

Disease	MONDO ID	Status / Consensus Assertion	Context	Report	Outcome / Intervention
Wilson Disease	MONDO:0010200	Complete / Strong Actionability	Adult	2022-02-14 AC028	Morbidity and mortality from copper deposition / Referral to specialist for evaluation and to guide appropriate therapy including diet, copper chelation and zinc therapy
Wilson Disease	MONDO:0010200	Complete / Definitive Actionability	Pediatric	2022-02-14 AC028	Morbidity and mortality from copper deposition / Copper chelation and zinc therapy

LOEUF 1.19
sHet 0.004
pHaplo 0.16
pTriplo 0.68

- Gene Tests
- Genomics England PanelApp
- LSDB
- Entries in DECIPHER for this gene
- Open Targets Platform

Interpretation and management resources

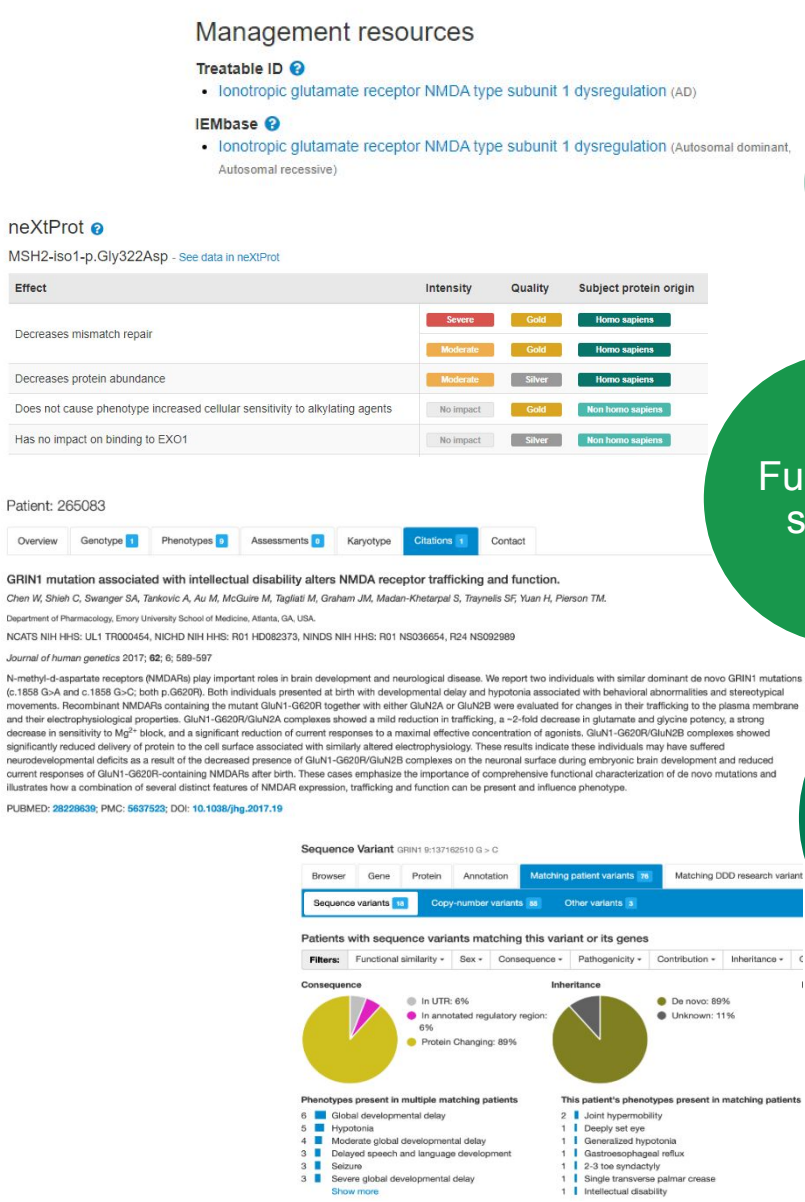
Treatable ID
- Copper-transporting ATPase subunit beta deficiency (synonym: Wilson disease) (AR)

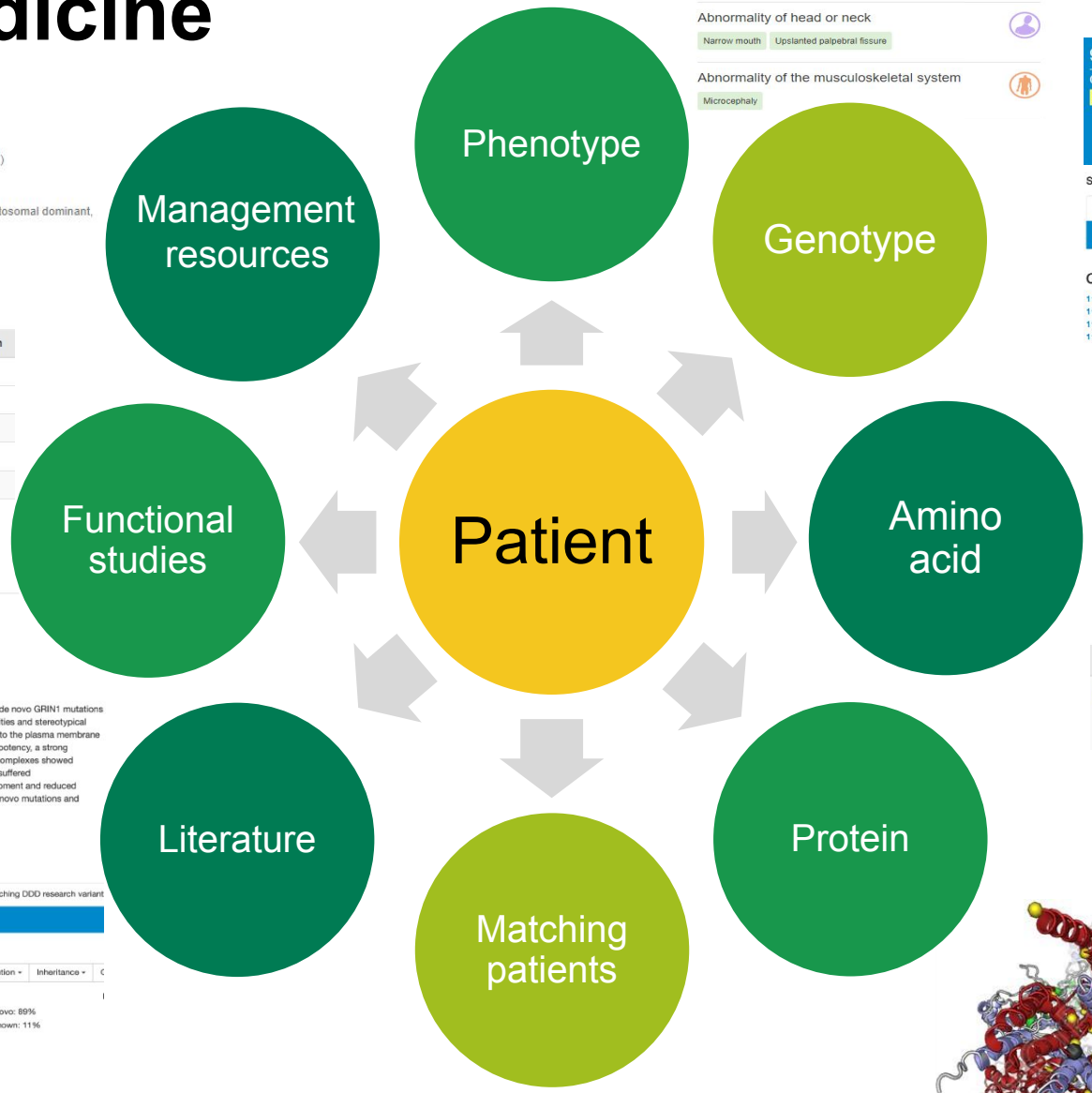
IEMbase
- Copper-transporting ATPase subunit beta deficiency (Autosomal recessive)

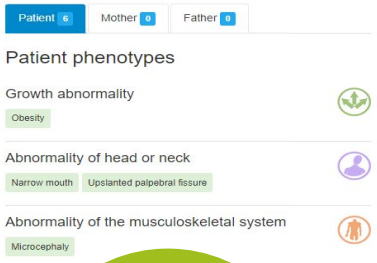
ACMG Secondary Findings
- Wilson disease (AR)

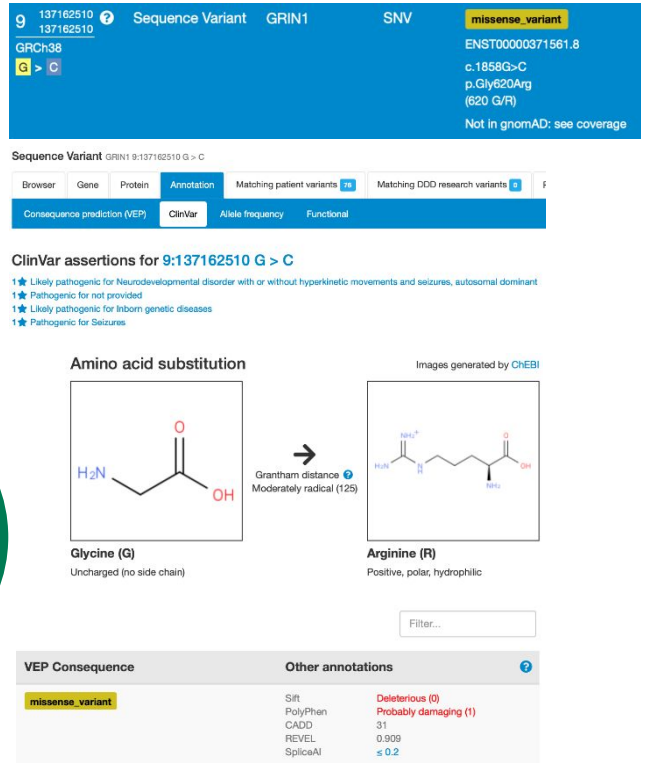
ClinGen Clinical Actionability Report
- Wilson Disease

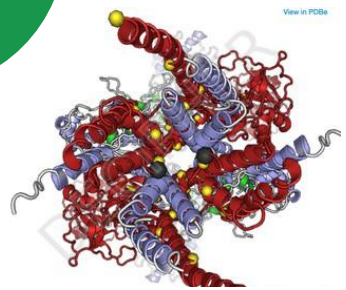
DECIPHER: Data integration for Genomic Medicine











Acknowledgements: DECIPHER

★ Patients and families for permission to include their data in DECIPHER

★ Members of the global DECIPHER community

★ Resources which DECIPHER uses

DECIPHER Leadership & Core Team

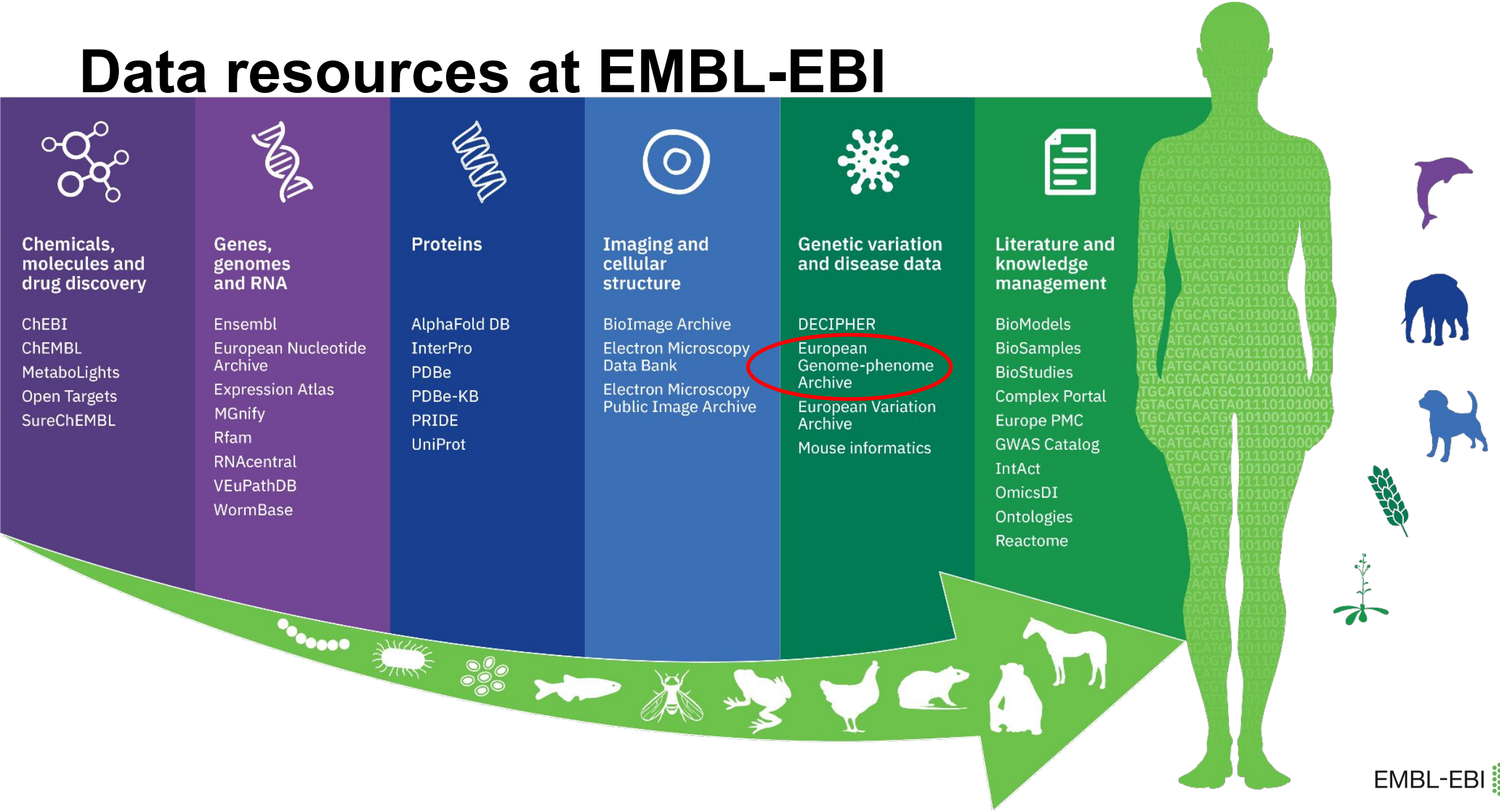
Recent alumni: Daniel Perrett, Simon Brent, Ben Hutton

Website: https://www.deciphergenomics.org

X/Twitter: @deciphergenomic

Email: contact@deciphergenomics.org

Data resources at EMBL-EBI



Sharing data about humans requires care

Exposure of personal or sensitive data is **potentially harmful**

Individuals have the **right to protection** of their personal data

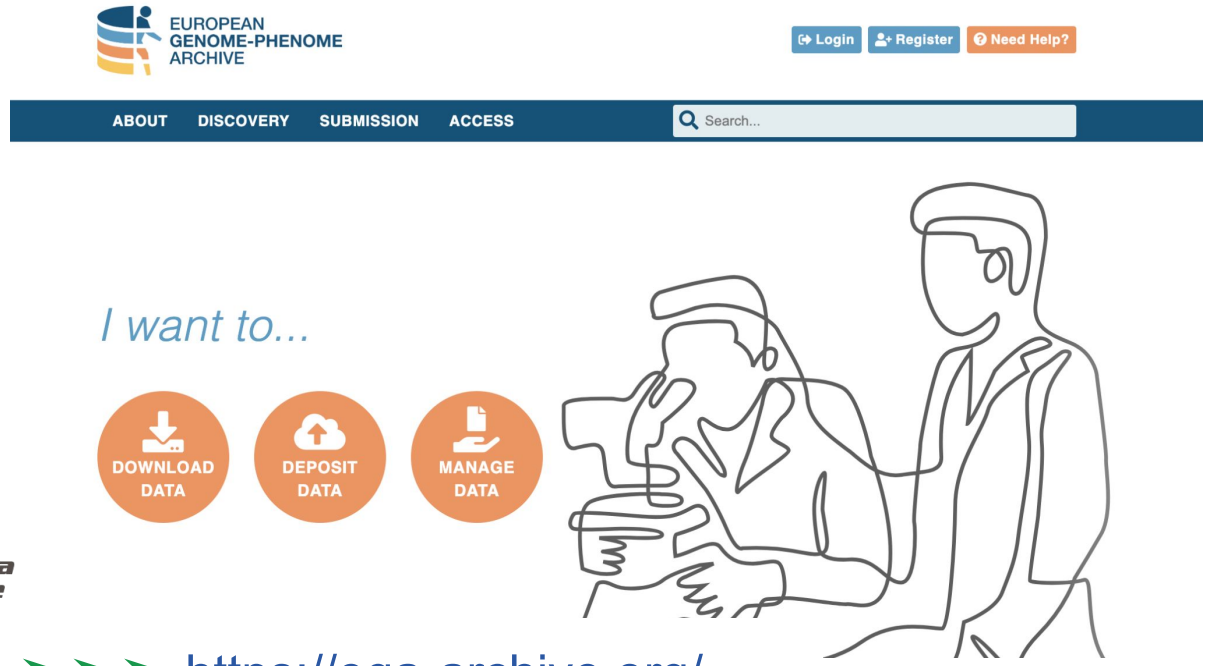
Governments pass legislation that **codifies data protection rights**

Support aims of research & health care initiatives

EGA manages sensitive human data

EUROPEAN GENOME-PHENOME ARCHIVE

Mission: Permanent archiving and sharing of personally identifiable genetic, phenotypic, and clinical data generated for the purposes of biomedical research projects or in the context of research-focused healthcare systems



GA4GH DRIVER PROJECT

elixir Core Data Resource

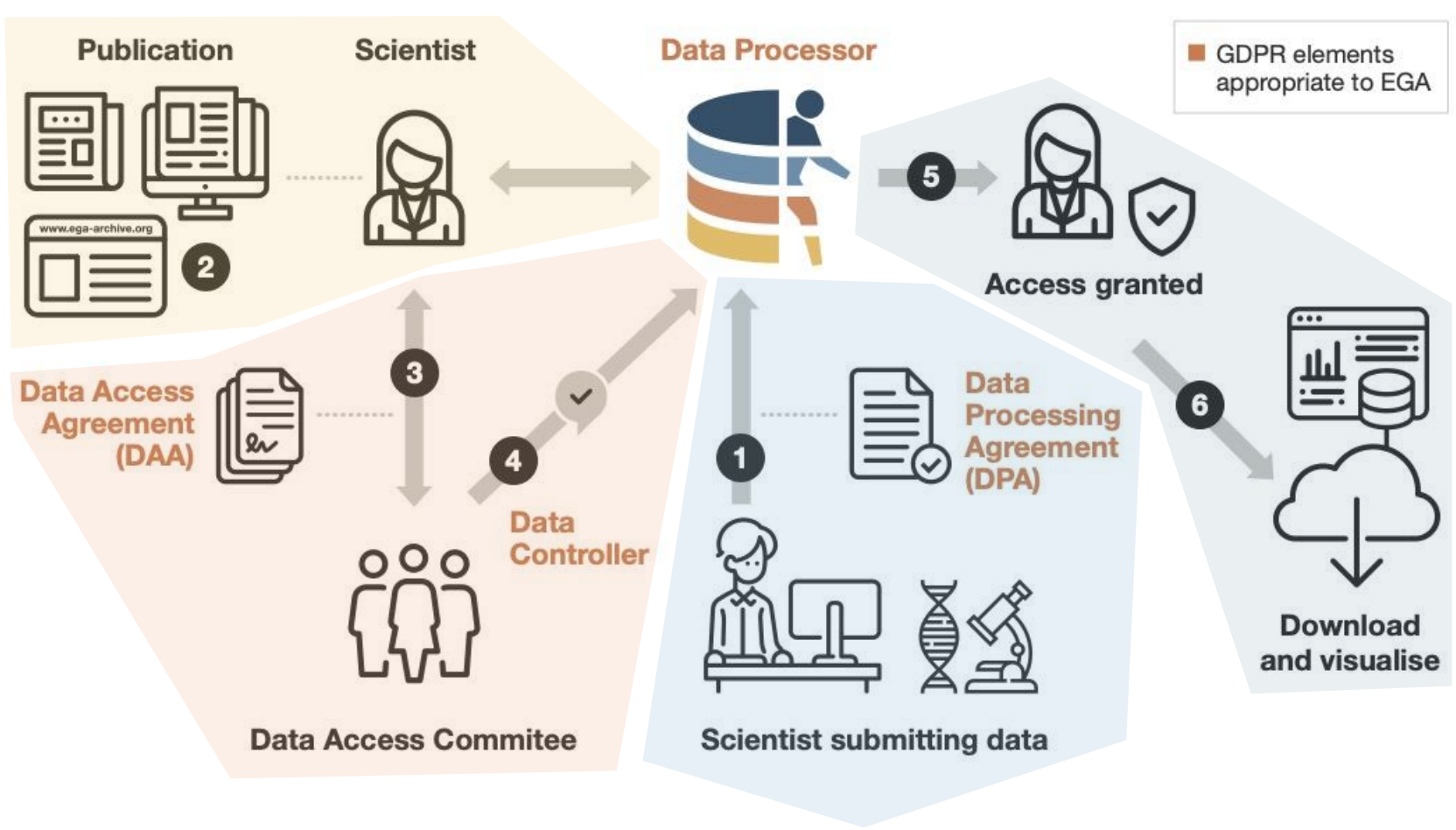
2008: EGA launched at EMBL-EBI

2013: CRG & EMBL-EBI enter formal collaboration to co-manage EGA

➤➤➤ https://ega-archive.org/

EMBL-EBI

EGA data access model



Data submission

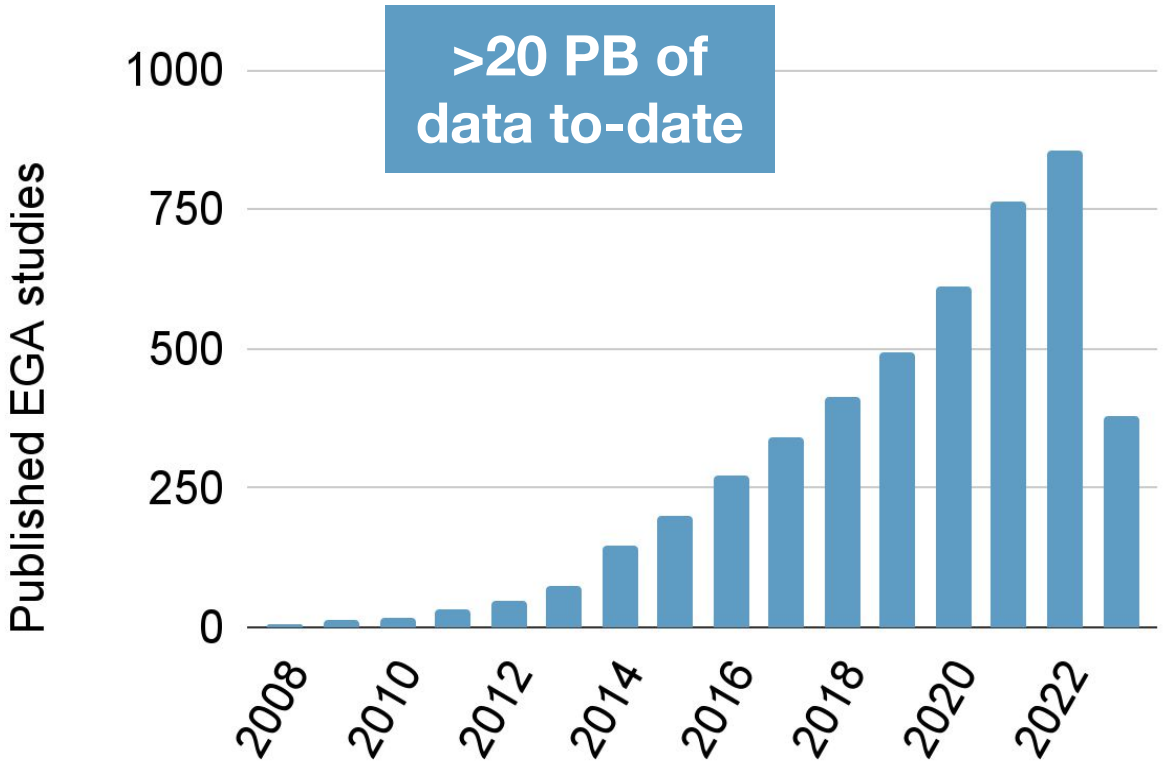
Data discovery

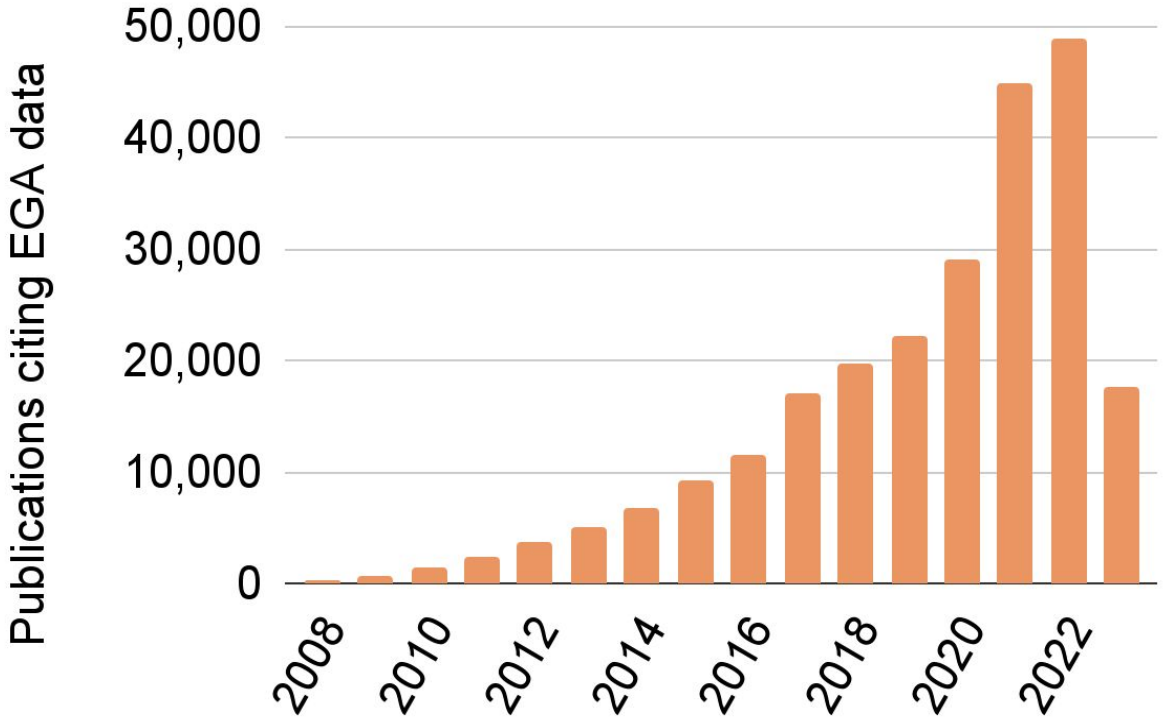
Data access

Data distribution

EMBL-EBI

EGA grows and remains key to life science research





Last Updated : June 20 2023

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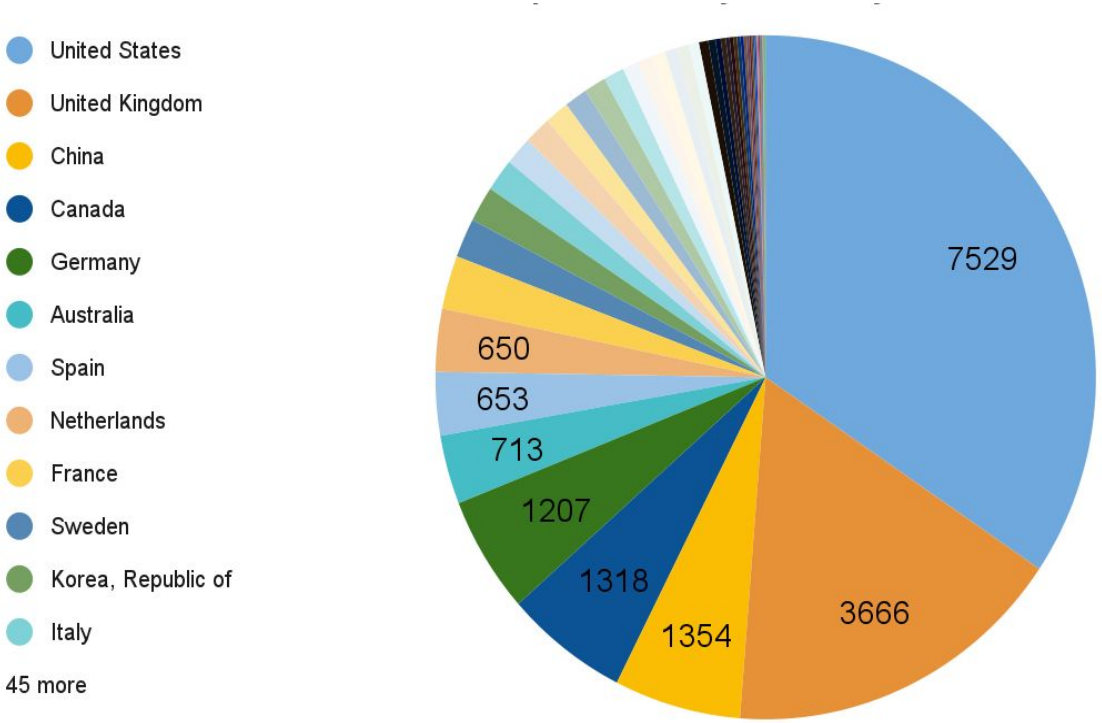
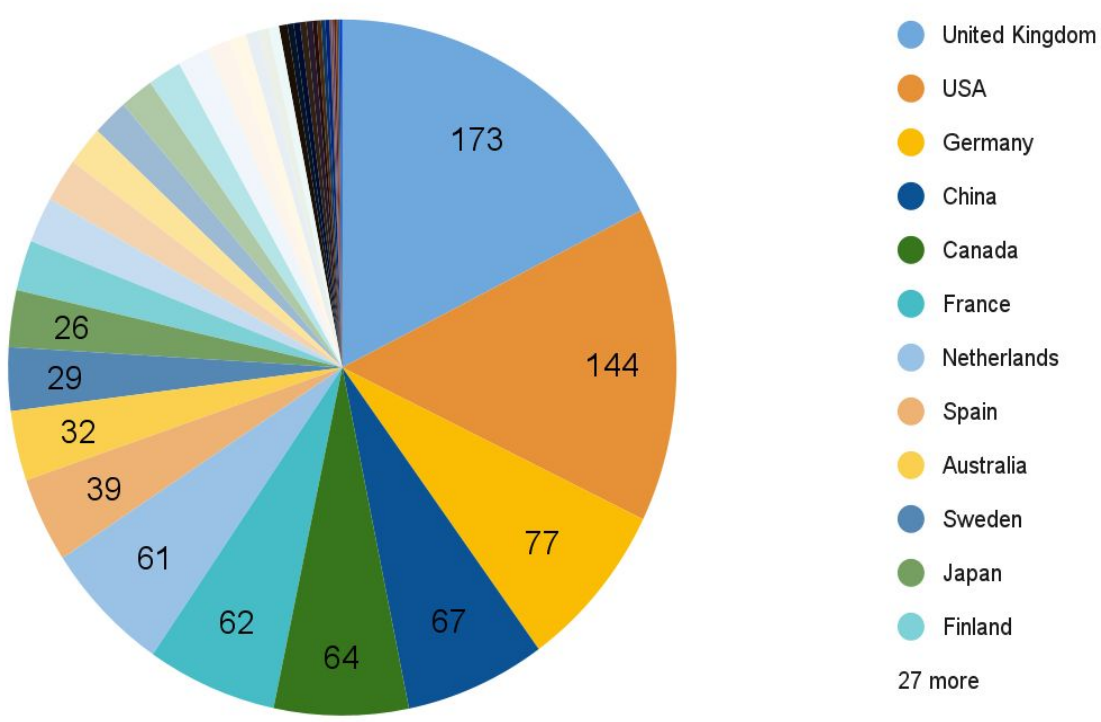
Data deposited and requested by global community

Data requesters by country



Data submitters by country

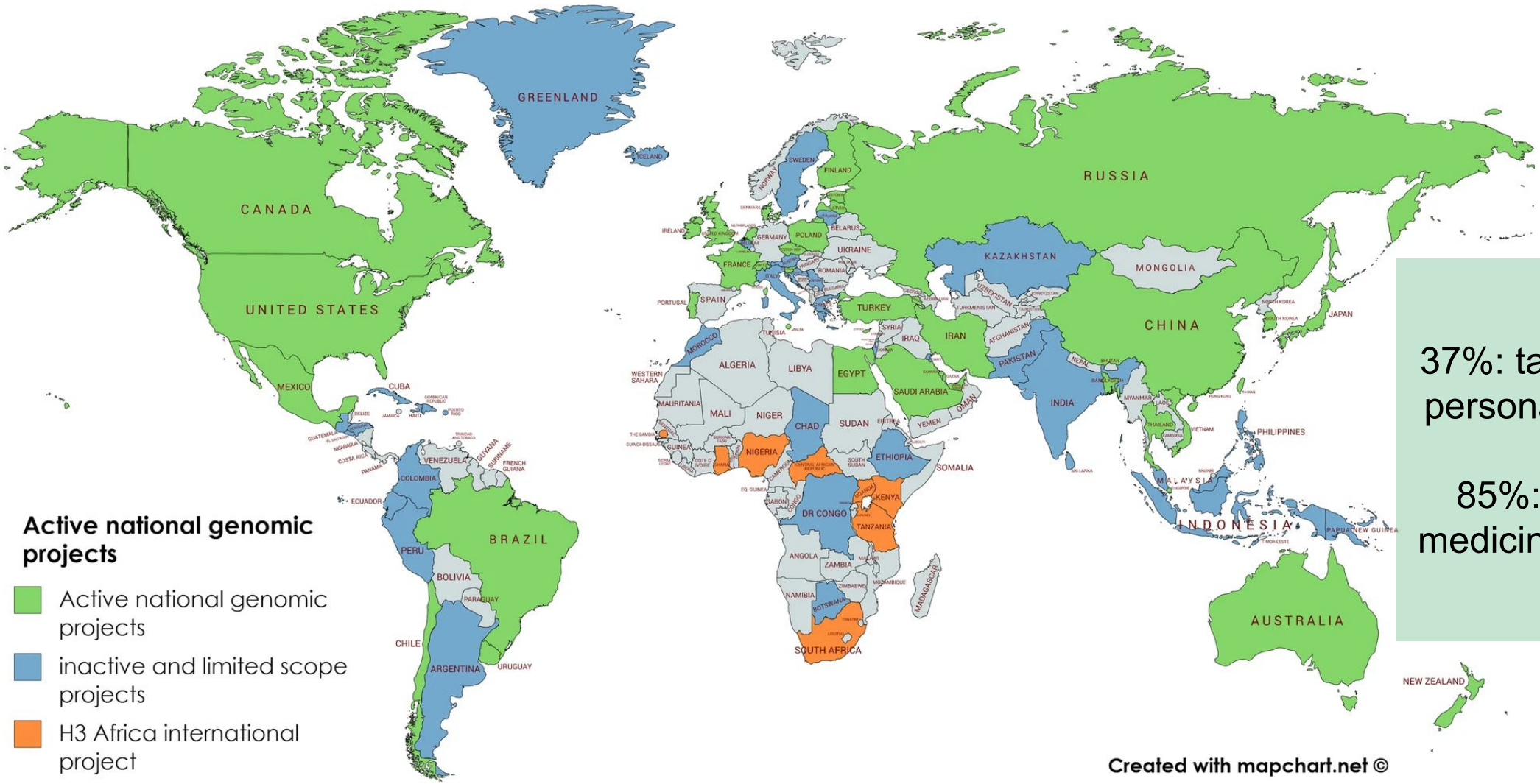


Last Updated : November 2023

Image credit :Aina Jene Cortada

EMBL-EBI

More genomics data generated in healthcare context



37%: tangible plans for personalized medicine

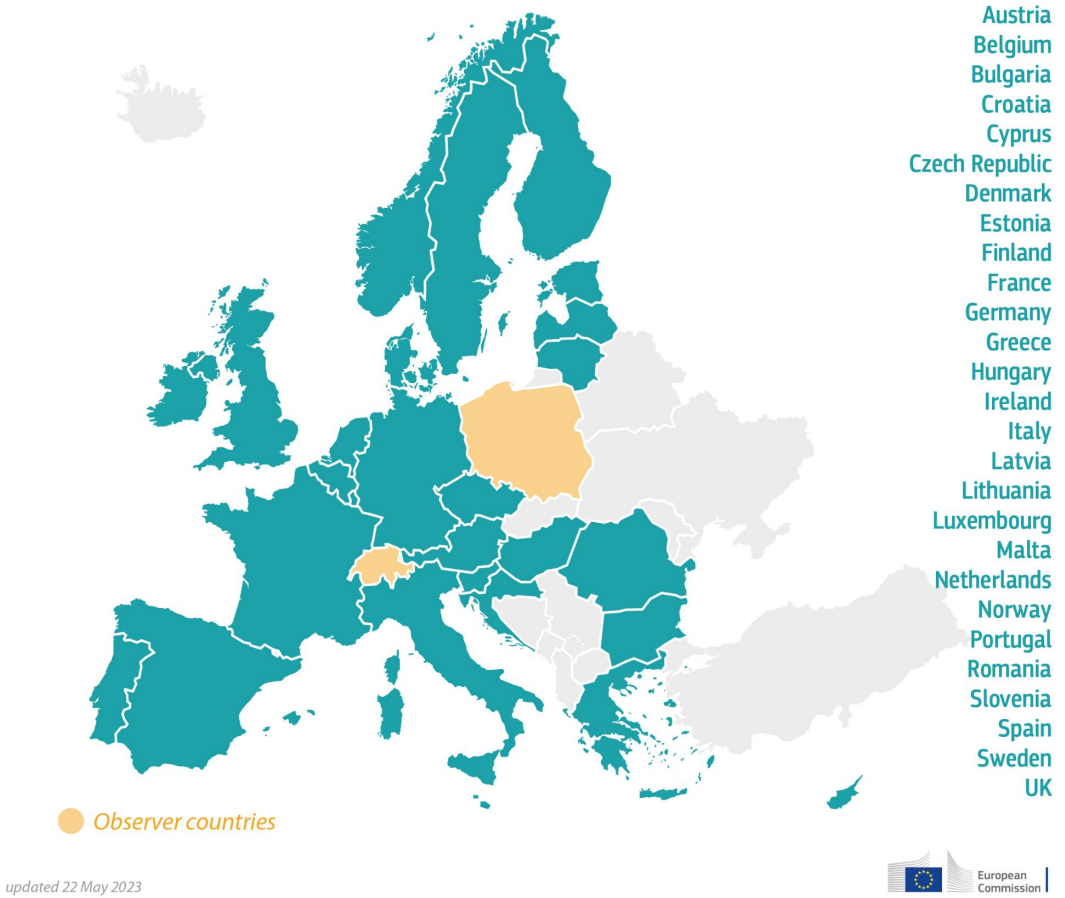
85%: personalized medicine as a rationale

Kovanda et al. How to design a national genomic project—a systematic review of active projects. Hum Genomics (2021). https://doi.org/10.1186/s40246-021-00315-6

EMBL-EBI

European 1+MG initiative sets vision for data sharing

Cross-border access to genomic data, implementation of genomics-based health. Supported by 27 countries to date



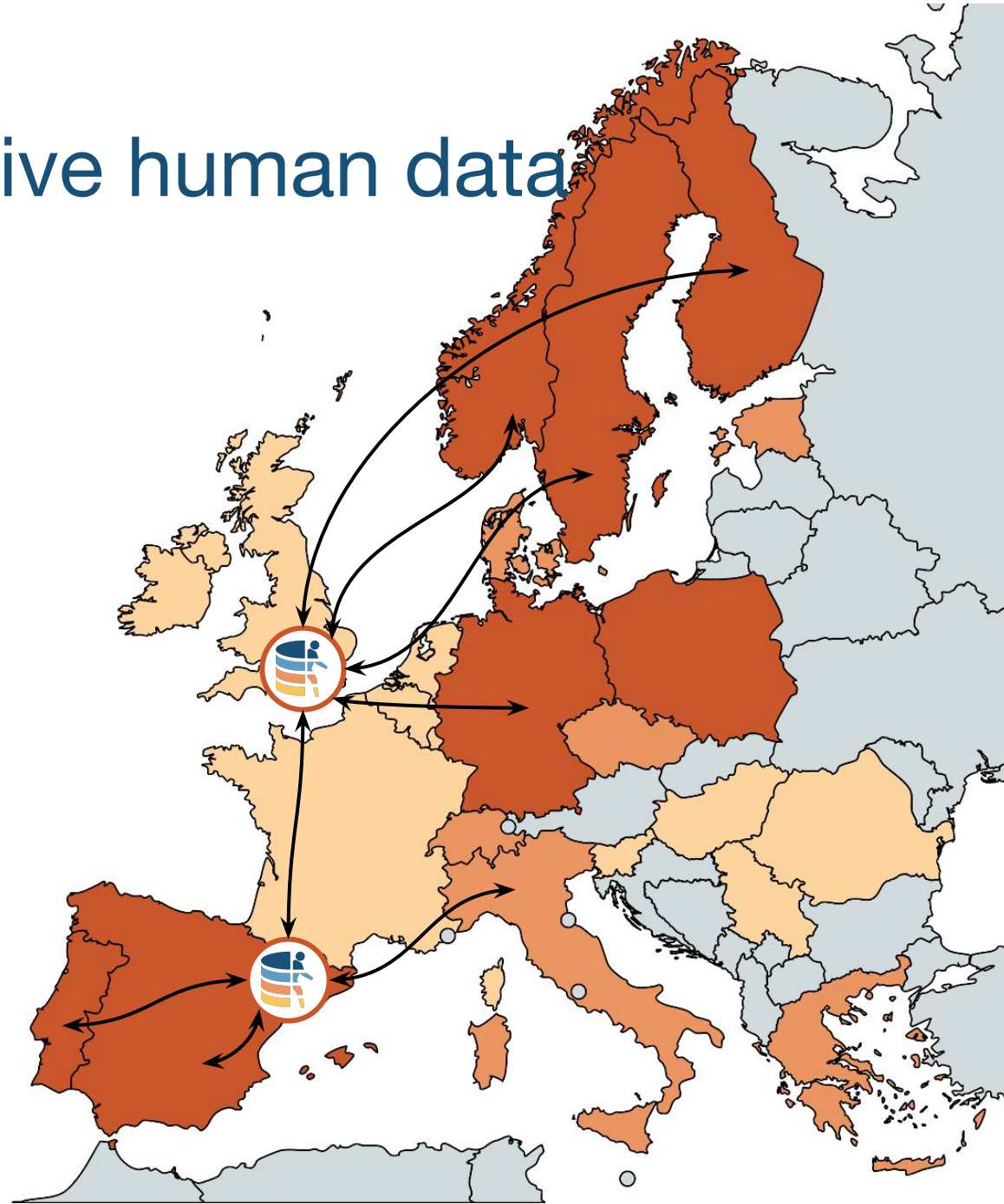
Ambition

Enable secure access to high-quality genomics and the corresponding clinical data across Europe for better research, personalised healthcare and health policy making

EMBL-EBI

Federated EGA:
discovery & access for sensitive human data

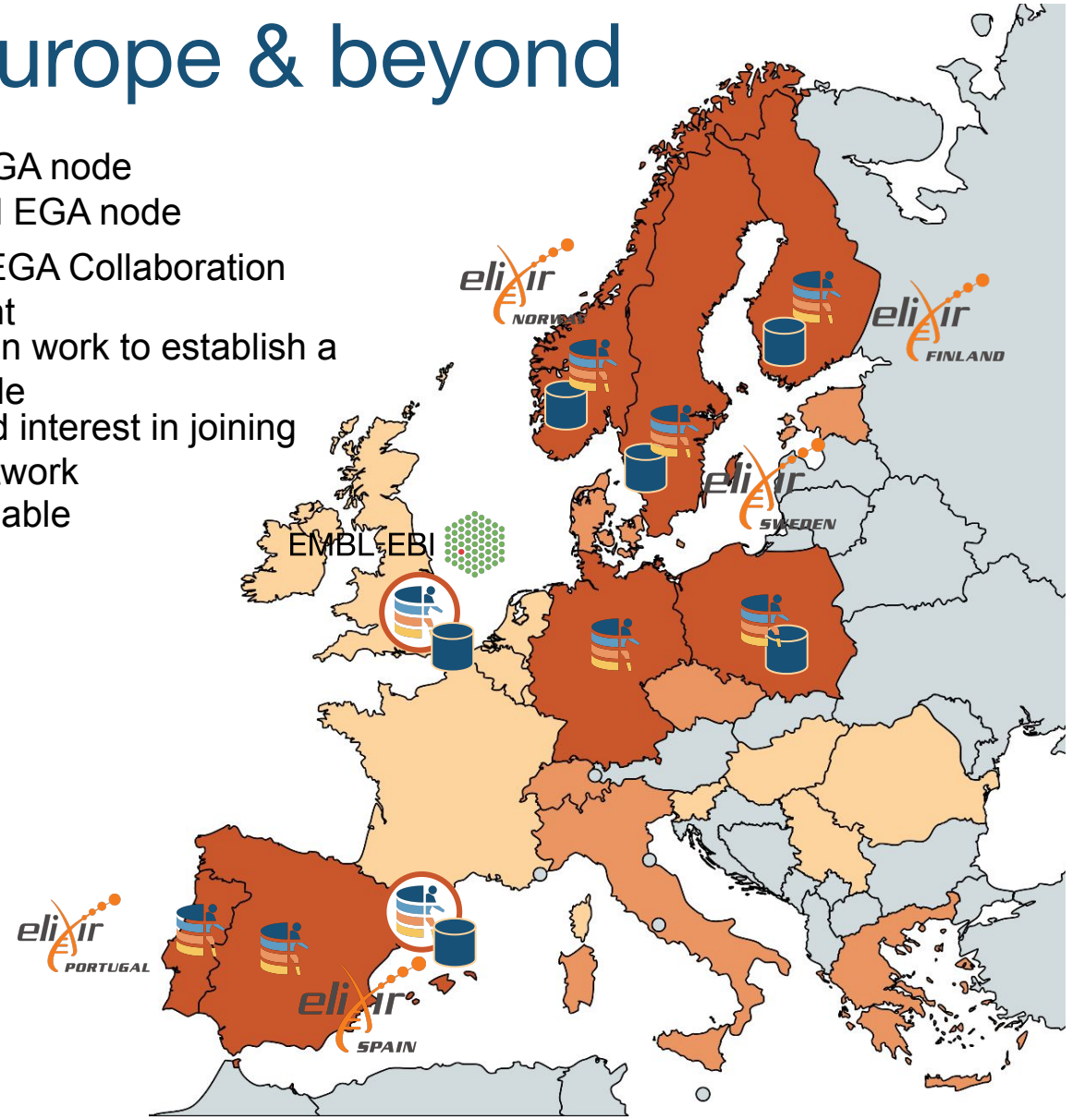
Federated EGA strives to support the discovery of and secure access to human data globally, while respecting national data protection regulations, with the goal of accelerating disease research and understanding and improving human health.



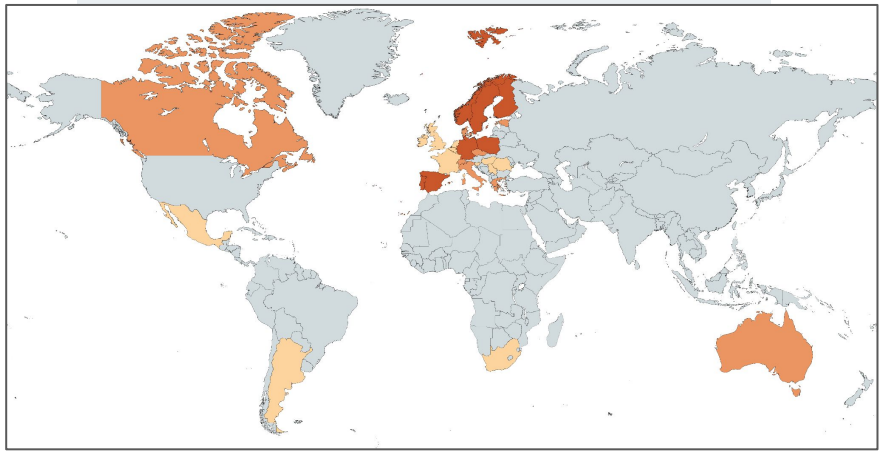
Federated EGA collaboration officially launched in September 2022

FEGA engagement across Europe & beyond

- Central EGA node
- Federated EGA node
- Signed FEGA Collaboration Agreement
- Engaged in work to establish a FEGA node
- Expressed interest in joining FEGA Network
- Data available



>24 nodes engaged and counting!



Last Updated: July 2024

Standards are key to interoperability

Globa
for Ge
Collabora

Standards

The EGA is a long-standing supporter of the **Global Alliance fo**
genetic data through the development of interoperable global st
Driver Projects and has contributed to the development and imp

Below is a list of the GA4GH standards and APIs that are curren

Technical Standards	Purpose
Large Scale Genomics	
htsget	A protocol for secure, efficient, and reli access to sequencing read and variati
Read File Formats (SAM/BAM/CRAM)	Specifications for storing next-generati sequencing read data.
Variation File Formats (VCF/BCF)	The specifications for Variant Call Forn (VCF) and its binary counterpart BCF.
Crypt4GH v1.0	Enables direct byte-level compatible ra access to encrypted genetic data store community standards (e.g. CRAM, VC

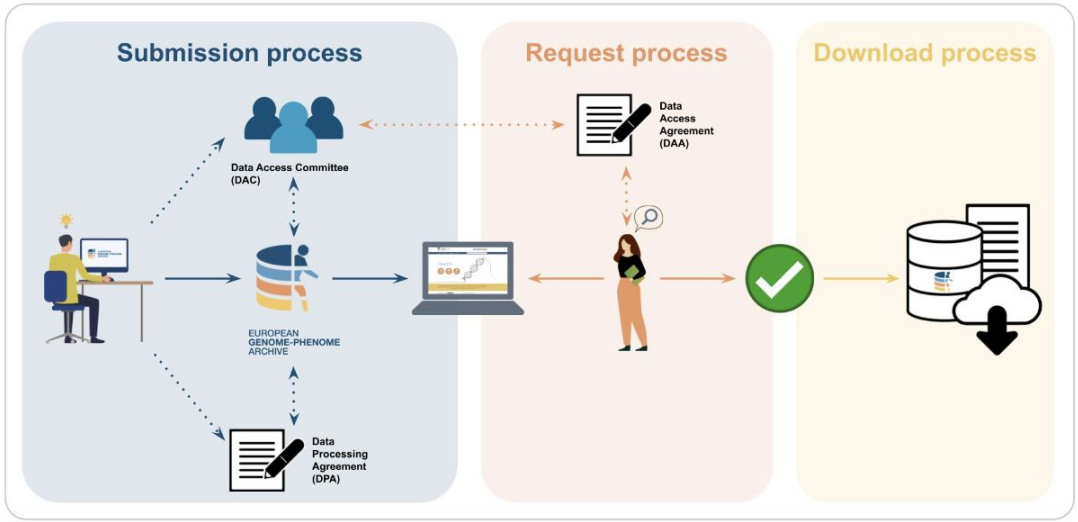
Our community | Driver Projects

European Genome-phenome Archive (EGA)

Provides a secure service for storing and sharing human genetic, phenotypic, clinical, and other "omics" data for research projects

The EGA is a global network for the permanent archiving and sharing of personally identifiable genetic, phenotypic, and clinical data generated for biomedical research projects or in the context of research-focused healthcare systems.. Since its launch in 2008, the EGA has collected data from over 6,000 different research projects at nearly 1,000 institutions worldwide. The EGA makes this data discoverable, accessible, and reusable for researchers who need it, adhering to the findable, accessible, interoperable, and reusable (FAIR) principles and maximising the value of the collected data.

EGA is funded by ongoing **institutional support** from EMBL-EBI and the Centre for Genomic Regulation (CRG) and support through collaborative **projects and partnerships** .



European Genome-phenome Archive (EGA) information

DRIVER PROJECT WEBSITE

EUROPEAN GENOME-PHENOME ARCHIVE

LOCATION	Europe
THEMATIC AREA	Rare Disease Cancer Complex Traits Basic Biology
CHAMPIONS	Thomas Keane Jordi Rambla Aina Jené

Acknowledgements: Federated EGA

EMBL-EBI

CRG Centre for Genomic Regulation

BSC Barcelona Supercomputing Center
Centro Nacional de Supercomputac

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

CSC

elixir SPAIN

Federated European Genome-phenome Archive Sweden

Fundació "la Caixa"

GHGA THE GERMAN HUMAN GENOME-PHENOME ARCHIVE

INB Spanish National Bioinformatics Institute

isc

Federated European Genome-phenome Archive Portugal

Federated European Genome-phenome Archive Norway

elixir CONVERGE

Acknowledgements: EGA

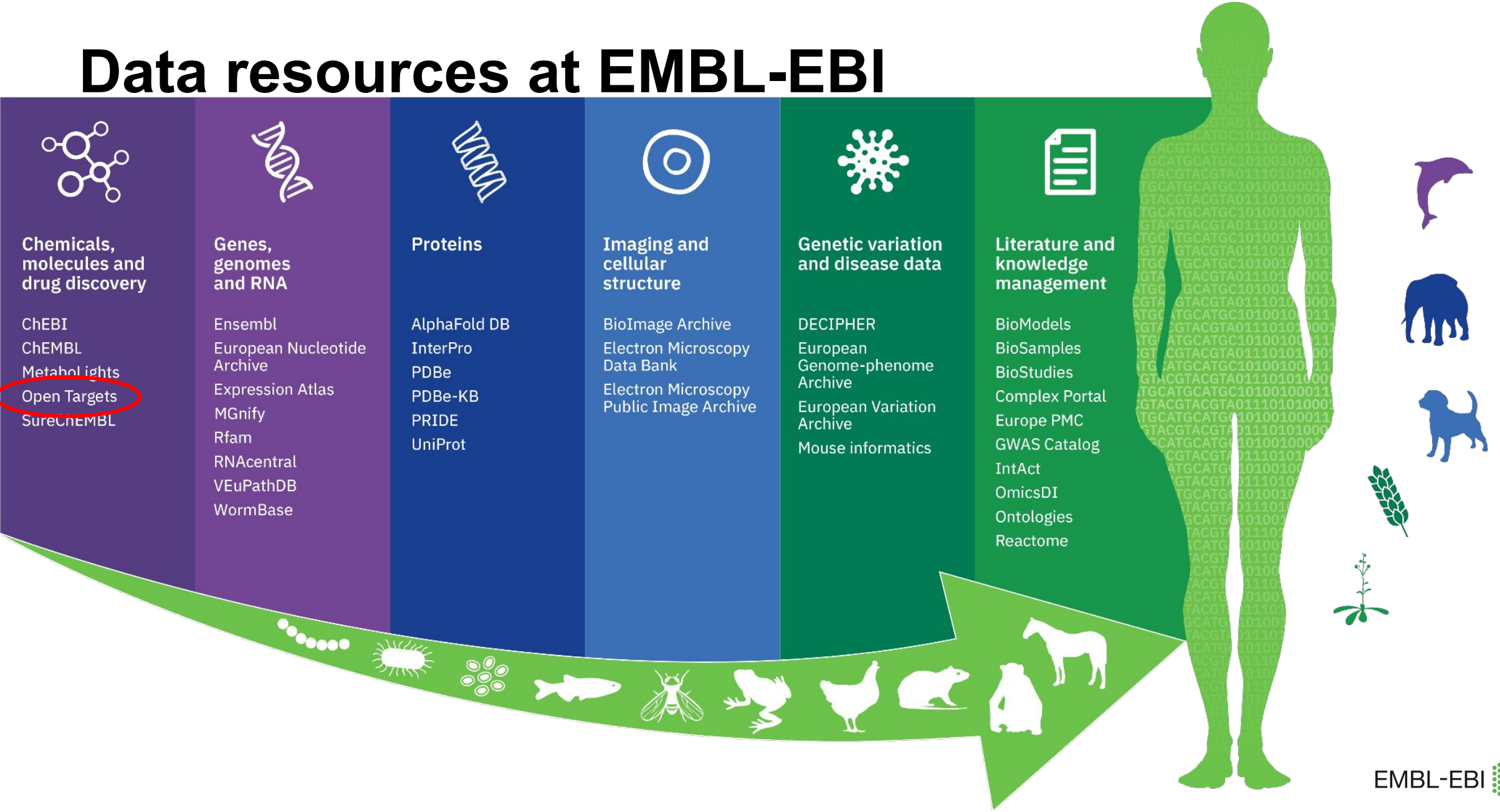
Core organizations:

Additional sources:

And critical support from the following sources:

LinkedIn: https://www.linkedin.com/company/european-genome-phenome-archive-ega/
Web: https://ega-archive.org/
Tw: @EGAarchive

Data resources at EMBL-EBI



What is Open Targets?

Developing safe and effective drugs is difficult and expensive

We are dedicated to changing this with innovative experimental and informatics approaches

Open Targets is an innovative, large-scale, multi-year, public-private partnership that uses human genetics and genomics data for systematic drug target identification and prioritisation.

Visit the Open Targets Platform which integrates public domain data to enable target identification and prioritisation, or Open Targets Genetics which identifies targets based on GWAS and functional genomics. We complement data integration with large scale systematic experimental approaches to support target identification, prioritisation and validation. Check out our latest papers describing our experimental target identification approaches in oncology, neurodegeneration, and immunity and inflammation.

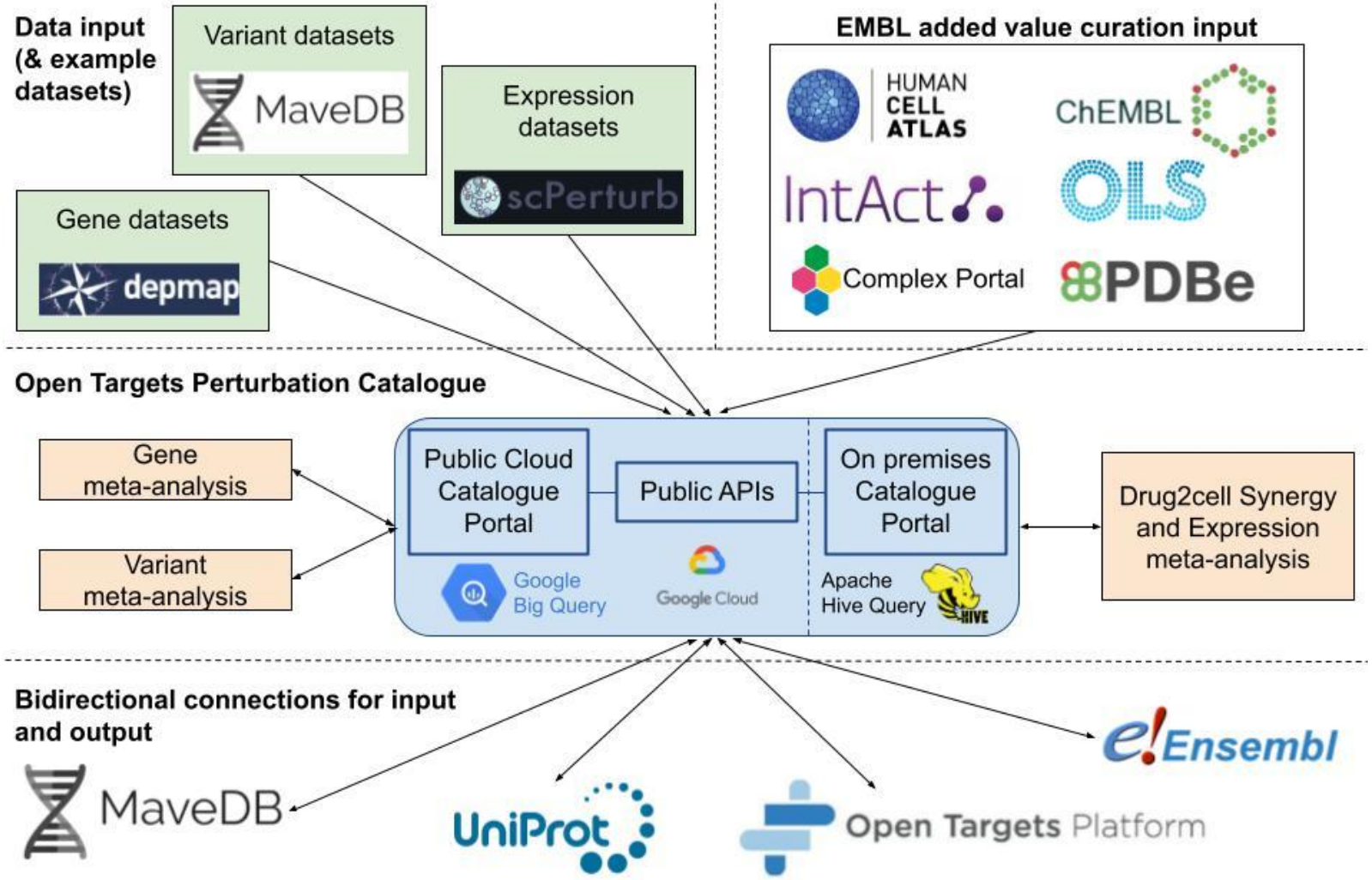
Find targets for a given disease
Visit the Open Targets Platform

Discover genetic evidence for targets
Visit Open Targets Genetics

Explore our research projects
Read our publications

Open Targets Perturbation Catalogue

"Integrate data from functional genetics screens and post-perturbational experiments, including data from CRISPR, MAVE, and Perturb-seq, to deliver gold standard machine learning-ready datasets, and a highly customisable public cloud platform with distributed data warehousing technology for cross metadata queries"



Acknowledgements: Open Targets Perturbation Catalogue

EMBL-EBI

Alexey Sokolov
James Stephenson
Prabhat Totoo
Maria-Jesus Martin
Mallory Freeberg

Genentech

David Richmond
Hector Corrada Bravo
Tim Sterne-Weiler

GSK

Aleix Lafita
Daniel Seaton
Philippe Sanseau

Human Technopole

Francesco Iorio

Open Targets

Daniel Suveges

Pfizer

Charul Jani
Isac Lee
Jason Arroyo
Paul Yenerall

Sanofi

Charlie Fulco
TJ Iyyanki

MaveDB

Alan Rubin (WEHI Australia)
Doug Fowler (University of Washington)

Wellcome Sanger Institute

Ally Dunham
Leopold Parts
Mo Lotfollahi

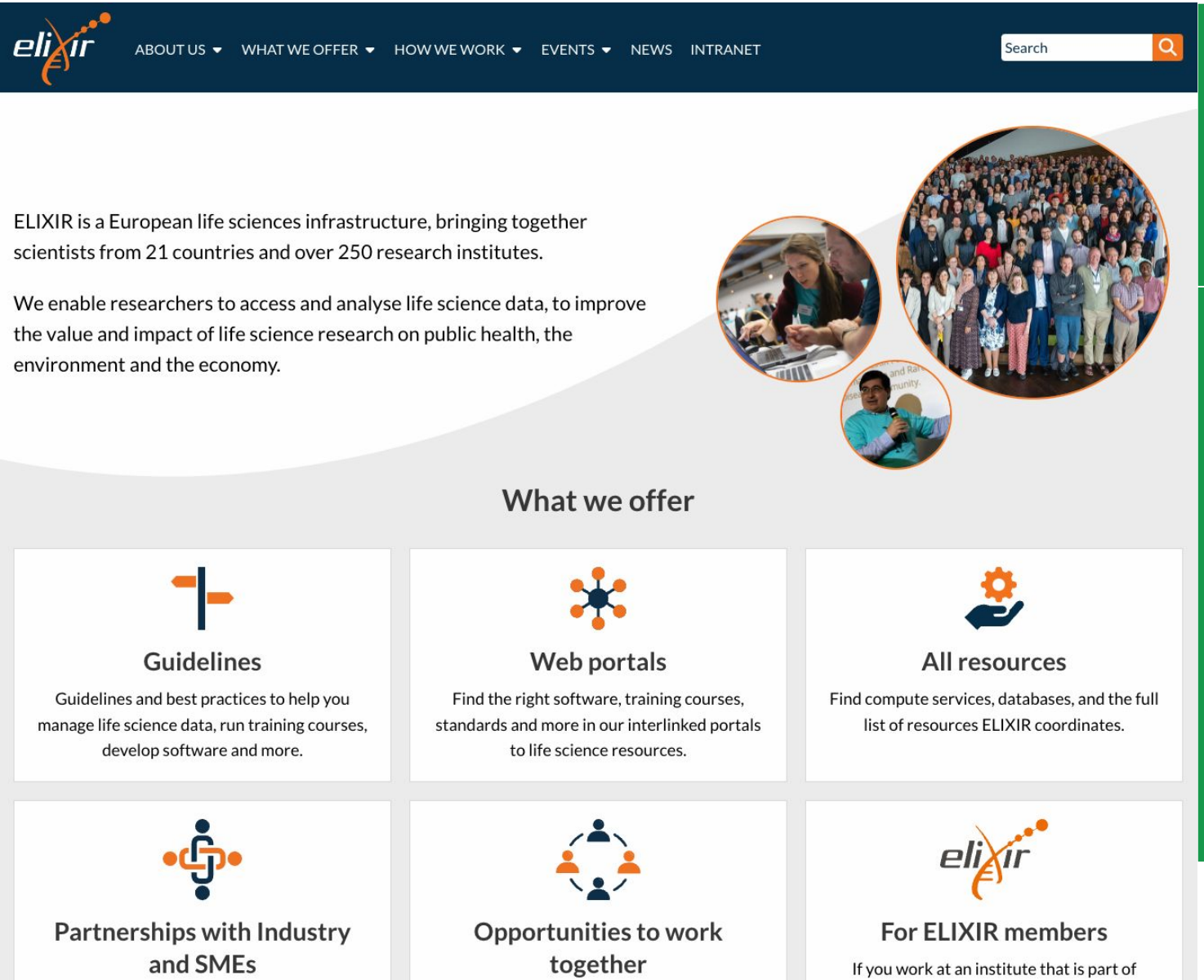
Website: Coming soon!

What makes these human genomics and health data resources global?

EMBL-EBI

Active participants in global initiatives

ELIXIR infrastructure for life sciences



ELIXIR Core Data Resources

Ensembl, EGA

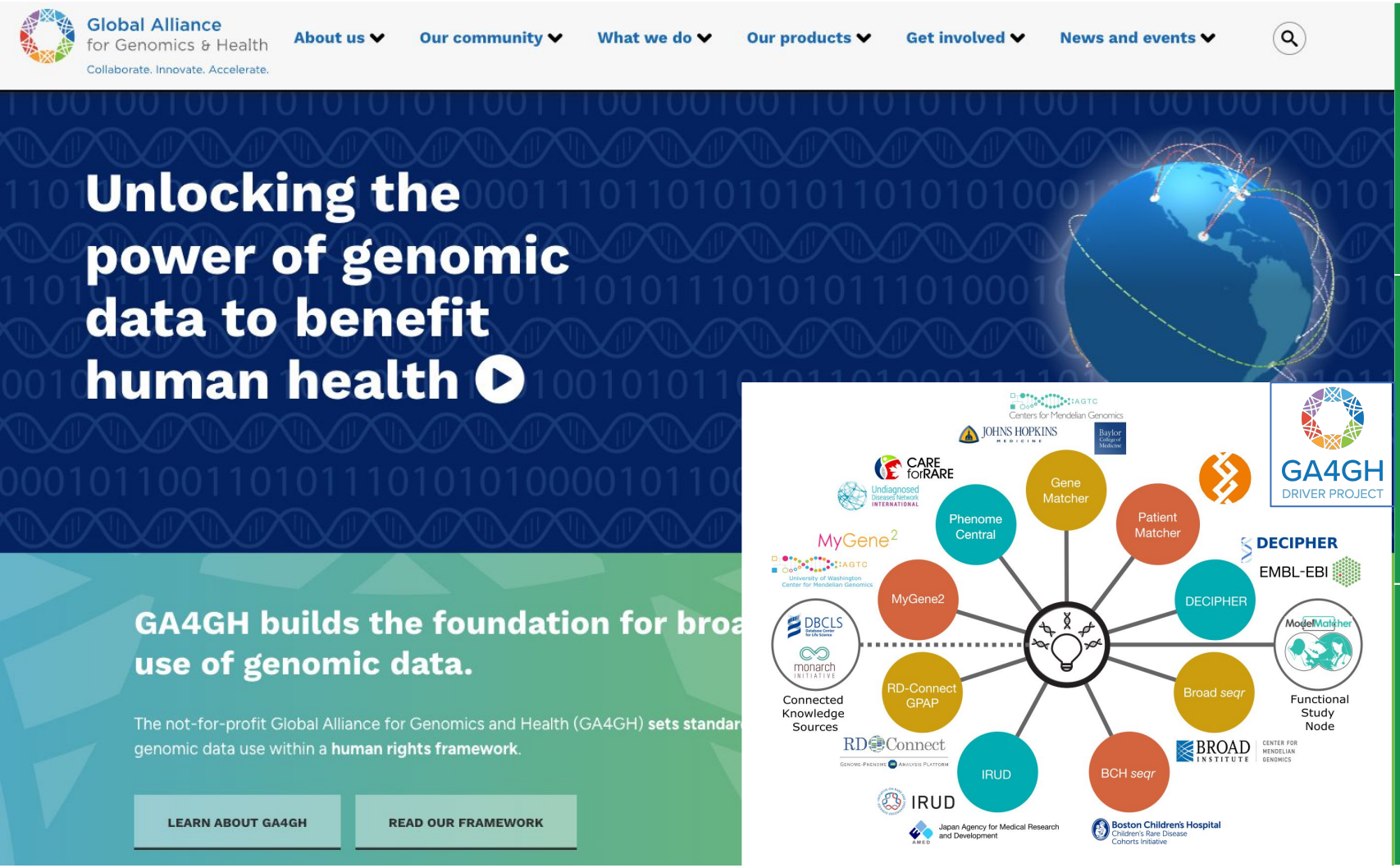
International collaborations

- Australian BioCommons
- Global Organisation for Bioinformatics Learning, Education and Training (GOBLET)
- Research Data Alliance (RDA)
- GA4GH Strategic Partnership

https://elixir-europe.org/

EMBL-EBI

Global Alliance for Genomics and Health



Driver Projects

EGA, Matchmaker Exchange (DECIPHER)

Communities of Interest

Rare Disease,Cancer, Clinical Genomics Laboratory

Strategic Partners

ELIXIR, RDA, NIH, Health Data Research UK

https://www.ga4gh.org/

★ Patients and families for permission to include their data in DECIPHER

★ Members of the global DECIPHER community

★ Resources which DECIPHER uses

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X/Twitter: @deciphergenomic

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Funding

Open Targets

National Human Genome Research Institute (NHGRI)

National Institute of Allergy and Infectious Diseases (NIAID)

Funded by the European Union

EMBL

UK Research and Innovation

TRANSFORMING GENOMIC MEDICINE INITIATIVE

elixir

wellcome

Acknowledgements: Federated EGA

EMBL-EBI

CRG Centre for Genomic Regulation

Barcelona Supercomputing Center Centro Nacional de Supercomputac

elixir Global Alliance for Genomics & Health

CSC

elixir SPAIN

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ISC

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Federated European Genome-phenome Archive Norway

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EMBL-EBI

Acknowledgements: EGA

LinkedIn: https://www.linkedin.com/company/european-genome-phenome-archive-ega/

Web: https://ega-archive.org/

Tw: @EGAarchive

EMBL-EBI

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Wellcome Sanger Institute

Ally Dunham
Leopold Parts
Mo Lotfollahi

Website: Coming soon!

Additional sources:

UNIÓN EUROPEA

Generalitat de Catalunya

"la Caixa"

elixir

HORIZON 2020

SEVERO OCHOA

FNIH

RD Connect

And critical support from the following sources:

Barcelona Supercomputing Center

upf. Universitat Pompeu Fabra Barcelona

INB

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Genentech A Member of the Roche Group

MSD

GSK

Pfizer

sanofi

EMBL-EBI 55

EMBL-EBI 52

Australian BioCommons, especially
Bernie Pope ~ Melissa Burke ~ Christina Hall ~ Jess Holliday

Happy to take Questions