The RD Connect Platform

Steve Laurie

Variant Effect Predictor Training Course

Prague, 7th November 2017

cnag
centre nacional d'anàlisi genòmica
centro nacional de análisis genómicc

CRG
Centre for Genomic Regulation

The RD-Connect Platform

1) What is RD-Connect

2) What **you** can do with the RD-Connect Genome-Phenome Analysis Platform (GPAP)

3) Case study – the BBMRI-LPC WES call

RD-Connect: Infrastructure for Rare Disease Research

6-year project funded by EU 7th Framework Programme

An integrated platform connecting data, clinical information, registries, and biobanks for rare disease research

- Creating a central system for **reprocessing**, **storing**, **analysing** and **sharing** of ***-omics*** data, including the integration of **phenotypic** and **biosample** data

- Contributing to **International Rare Diseases Research Consortium (IRDiRC)** objectives of delivering 200 new therapies, and means to diagnose most rare diseases of genetic etiology by 2020

What are rare/orphan diseases?

RARE DISEASES

7% OF THE POPULATION ARE AFFECTED BY RARE DISEASES

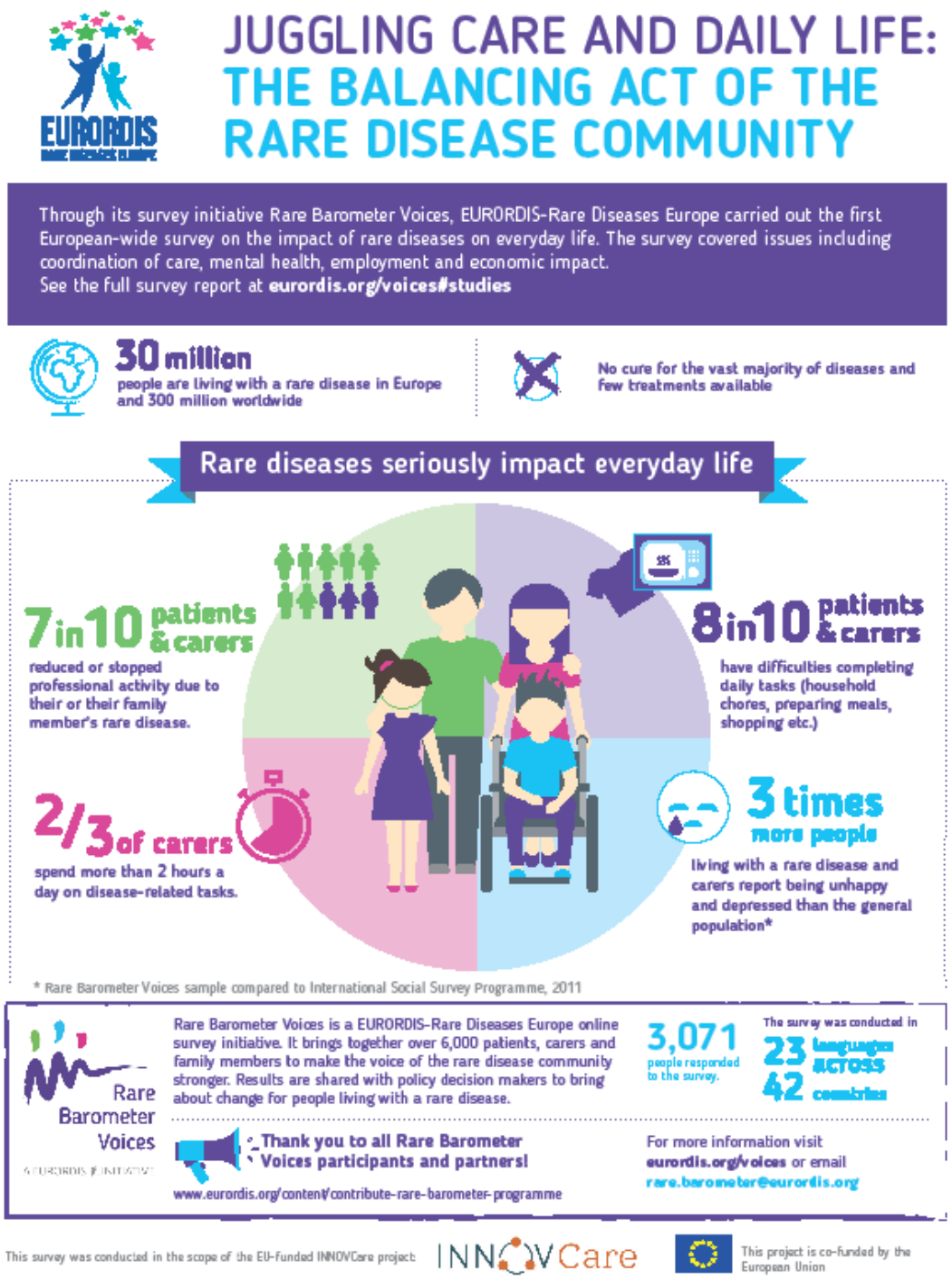
THE EU CLASSES A DISEASE AS 'RARE' WHEN LESS THAN 1 IN 2000 SUFFER

OVER 7000 DISEASES

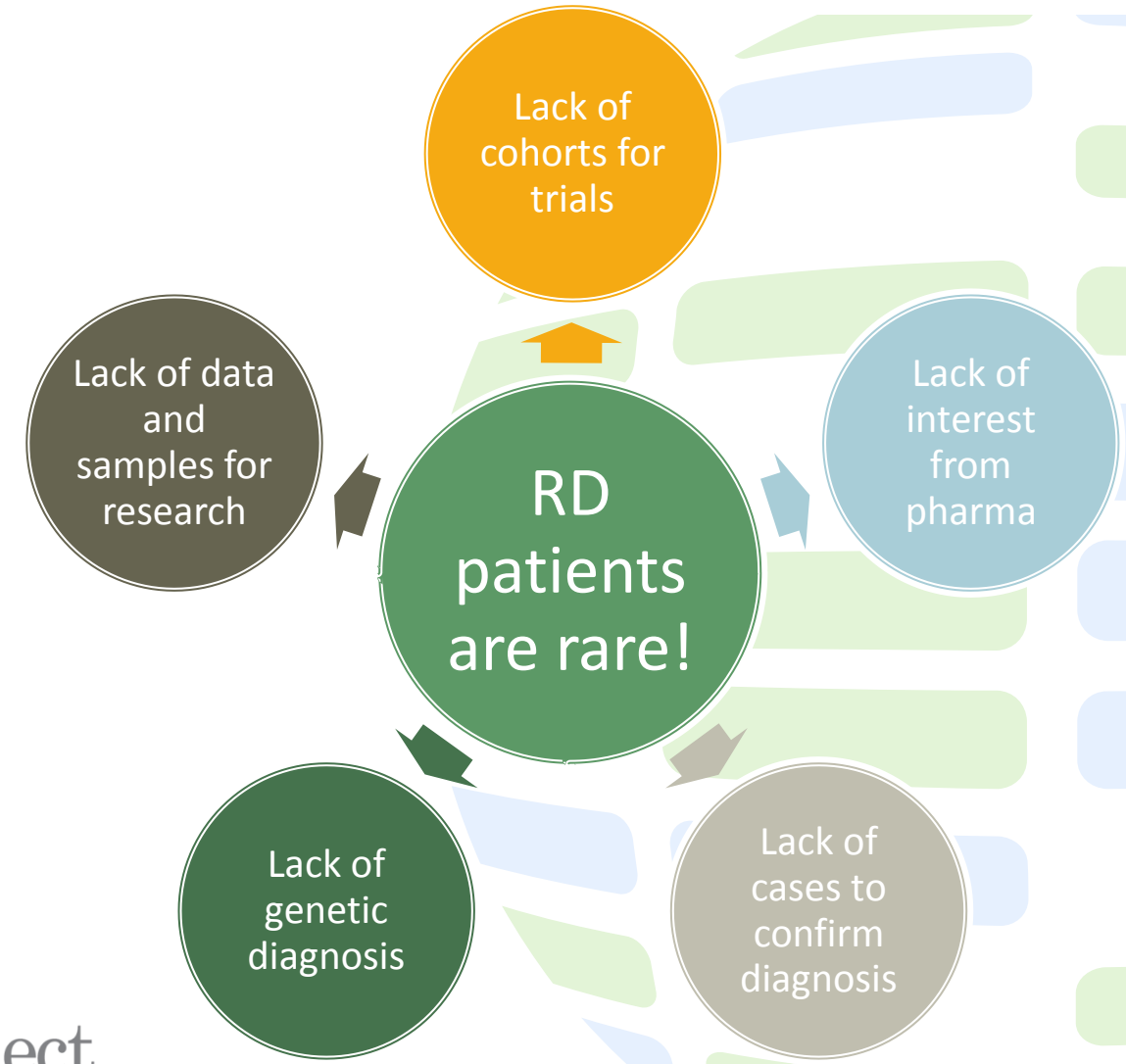
80% GENETIC IN ORIGIN.

OFTEN CHRONIC AND LIFE-THREATENING

Huge social impact on patients and their carers



Bottlenecks in RD research



Many bottlenecks are cross-cutting

… across diseases and across research domains

- A lot of them come down to **data**
- Not just scarcity of data, but lack of options to reuse the data that does exist
  - **Privacy protection issues**, particularly across borders
  - **Lack of infrastructure** for data sharing
  - **Lack of standards** and interoperability
  - **Reluctance to share** unpublished data
  - **Lack of capacity to analyse** large amounts of data
  - **Challenges of linking different datasets** in different places

RD Connect

Infrastructure for data sharing in rare disease research

Flagship IRDiRC project implementing IRDiRC policies and guidelines on data sharing

EU 7th Framework Programme, 12M EUR, 6 years

Genomic analysis and gene discovery

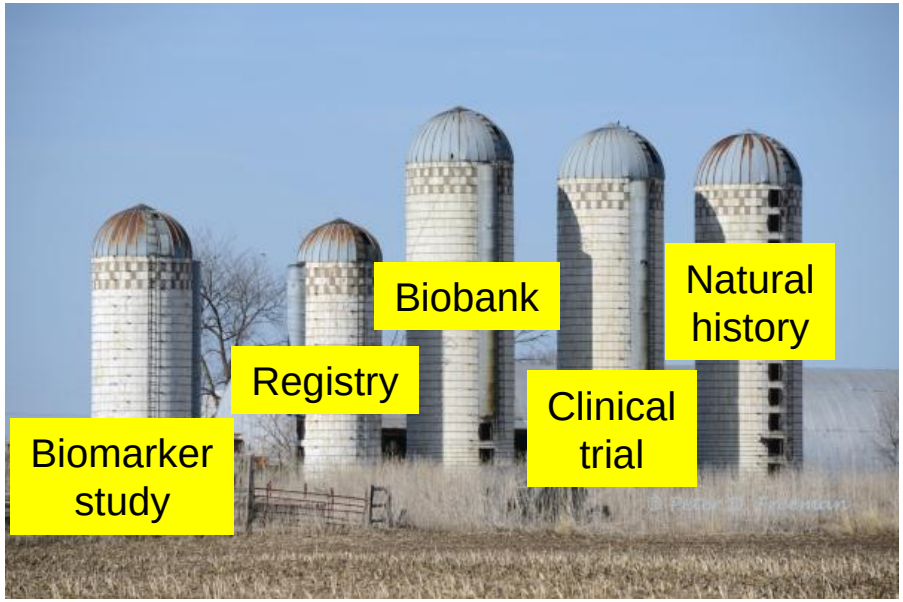
Standardized phenotypic data collection

Searchable catalogue of biosamples

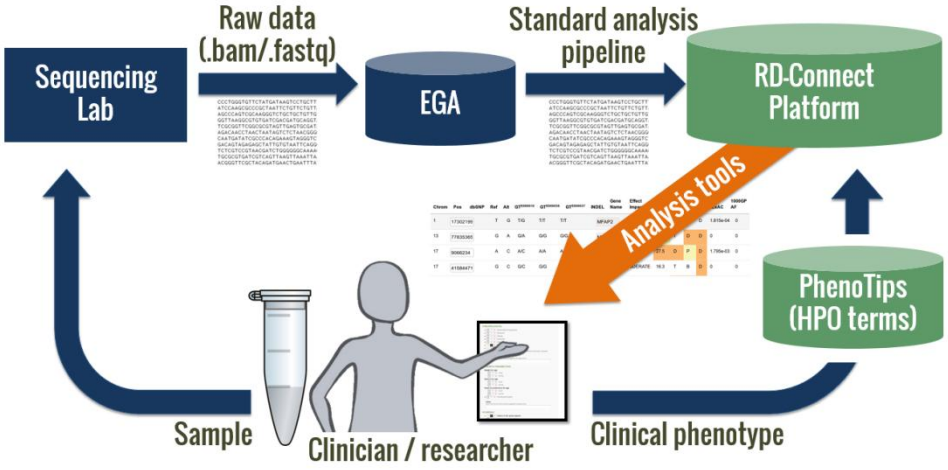
Data linkage across resources

Overcoming Silos

Data sharing for research and better data analysis



Omics data, clinical data and biosamples from an individual with RD



Disease-causing variants can be identified using the **Genome-Phenome Analysis Platform**

Samples findable in the **Biosample Catalogue**

Registry data findable in the **Registry and Biobank Finder**

RD-Connect Platform

RD Connect

Home	Genome-Phenome Analysis Platform	PhenoTips	Registry & Biobank Finder	Biosample

An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research

Welcome to the central platform for access to data submitted by RD-Connect's partner projects. The online Genome-Phenome Analysis Platform is now open for submissions from all users. Our automated registration system will come online shortly, but if you would like to access the interface now please email platform@rd-connect.eu and we will contact you to request the information we need to set you up on the system.

Get started today

catalogue.rd-connect.eu

RD Connect | duchenne

Linking up rare disease resarch across the world

Catalogue | Search | Help Request

Search

duchenne

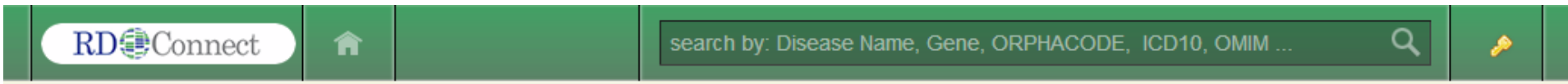
Search

Search for: duchenne

Search returned 113 hits (Registries:34 /Biobanks:8)

Australian National Duchenne Muscular Dystrophy Registry
Caroline Graham
https://nmdregistry.com.au/dmd/https://nmdregistry.com.au/index.html
Duchenne muscular dystrophy: Number of Patients, Donors 482
Diseases DiseaseName: Duchenne muscular dystrophy
DiseasesSynonym: Severe dystrophinopathy, Duchenne type, DMD
Organization OrganizationName: Australian National Duchenne Muscular Dystrophy Registry

Congenital Muscular Disease International Registry
Rachel Alvarez



ID # 77350 Date of Inclusion: 01/04/2015 Last Activities: 21/04/2017

NETWORK OF GENETIC BIOBANKS Telethon

Galliera Genetic Bank

The GGB is located at Genetics Laboratory – of Galliera Hospital in Genoa and collects samples from patients affected by chromosomal disorders several genetic diseases since 1983. To date the biobank

Overview [17] | Diseases [132] | Documents [0]

Website Link

Host institution

Laboratory of Human Genetics - E.O. Ospedali Galliera
Via Alessandro Volta, 6
16128 Genova
Italy

Tel.0039010563-4383

Fax.0039010563-4381

General Information

Acronym: **GGB**
Type of Host Institution: **Hospital**
Source of funding:
Target population:
Year of establishment: **1983**

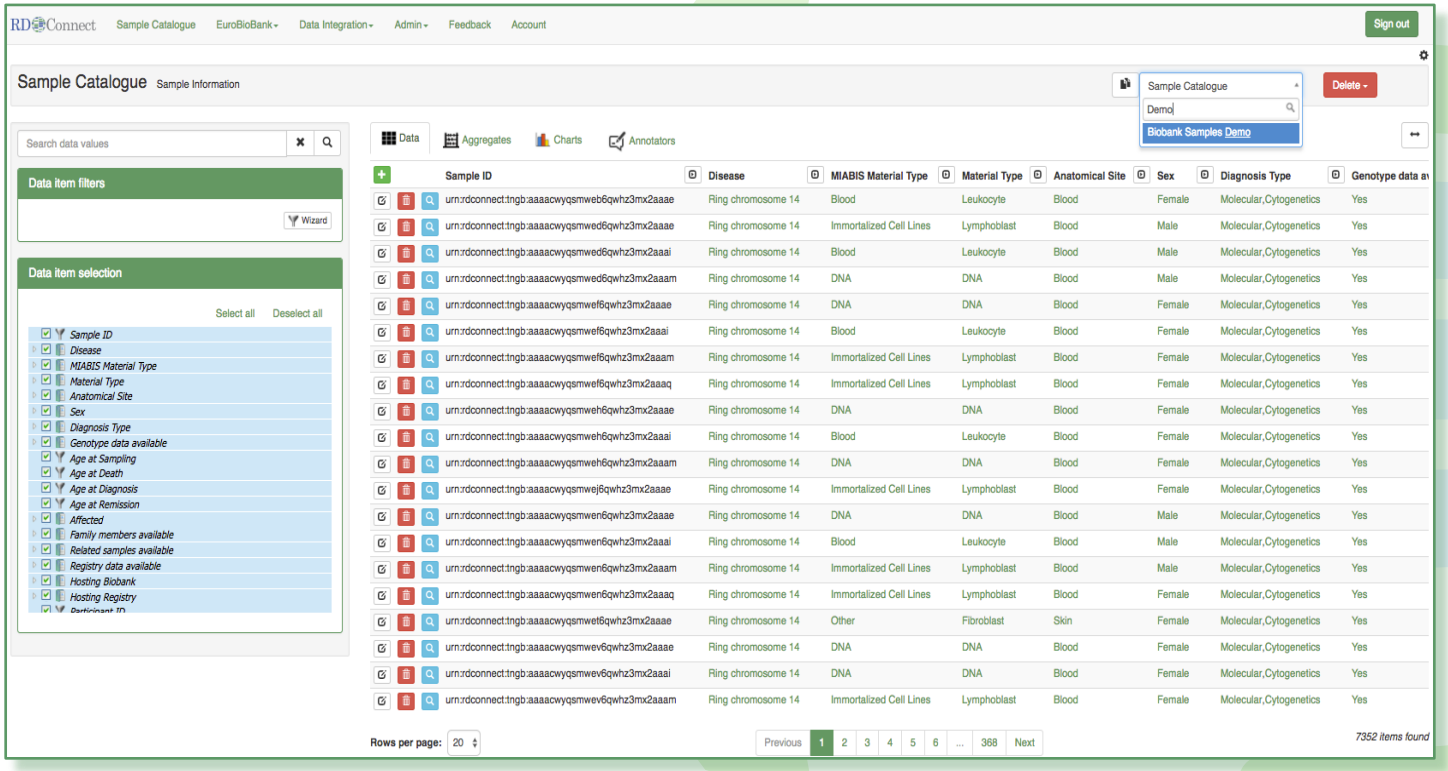
Personnel

Main contact

Chiara Baldo
Curator
chiara.baldo@galliera.it

Sample-level catalogue (under development)

The RD-Connect Sample Catalogue contains information on available samples across participating biobanks



RD-Connect

RD-Connect: Ethical and Legal Issues

- Database registered in the Agencia Española de Protección de Datos
- To submit and/or to access donor data, a **Code of Practice** and **Adherence Agreement** must be signed
- Documents were approved by Comité Ètic d'Investigació Clínica del Parc de Salut Mar in 2015
- **User-activity is logged**

RD-Connect enables:

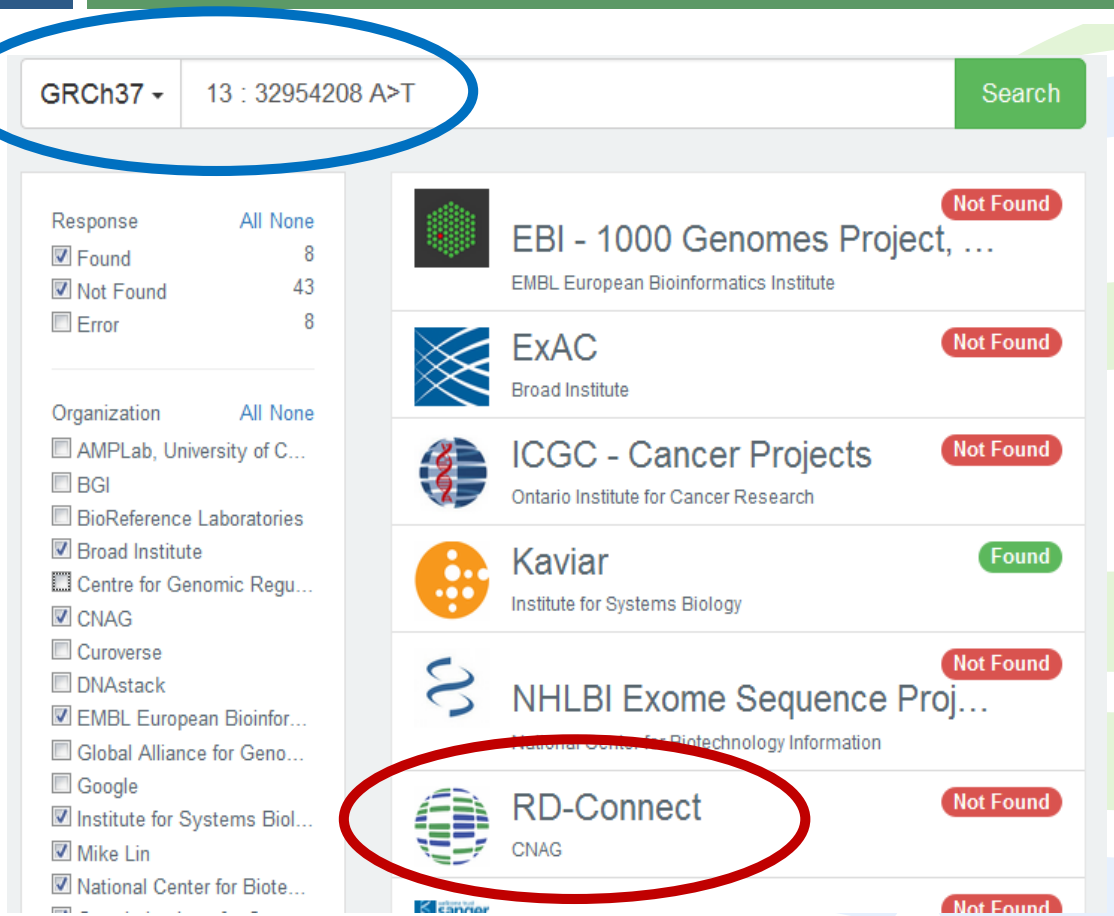
- Full data sharing and analysis within RD-Connect for **registered users**
- Partial data sharing outside RD-Connect (in accordance with ethical and legal limitations)
- A core requirement of having data incorporated into RD-Connect is that that **corresponding phenotypic descriptions for all donor samples must be entered** into the RD-Connect PhenoTips instance
- If you **share your data**, you are more likely to solve your cases e.g. through internal and external **matchmaking (MME)**

RD-Connect: Helping you share **your patients' data** (with consent)

RD-Connect enables:

- Full data sharing and analysis within RD-Connect for **registered users**
- Partial data sharing outside RD-Connect (in accordance with ethical and legal limitations)
- A core requirement of having data incorporated into RD-Connect is that that **corresponding phenotypic descriptions for all donor samples must be entered** into the RD-Connect PhenoTips instance
- If you **share your patients' data**, you are more likely to solve your cases e.g. through internal and external **matchmaking (MME)**

Data Sharing: GA4GH Beacon



Question:
Does any sample in your database have **variant V**?

Answer:
Yes / No

RD Connect

https://beacon-network.org/#/

Data Sharing: MatchMaker Exchange (IRDiRC, GA4GH)

Question:

Do you have a patient with a ***similar*** **phenotype** and ***similar*** **variants** to mine in your database?

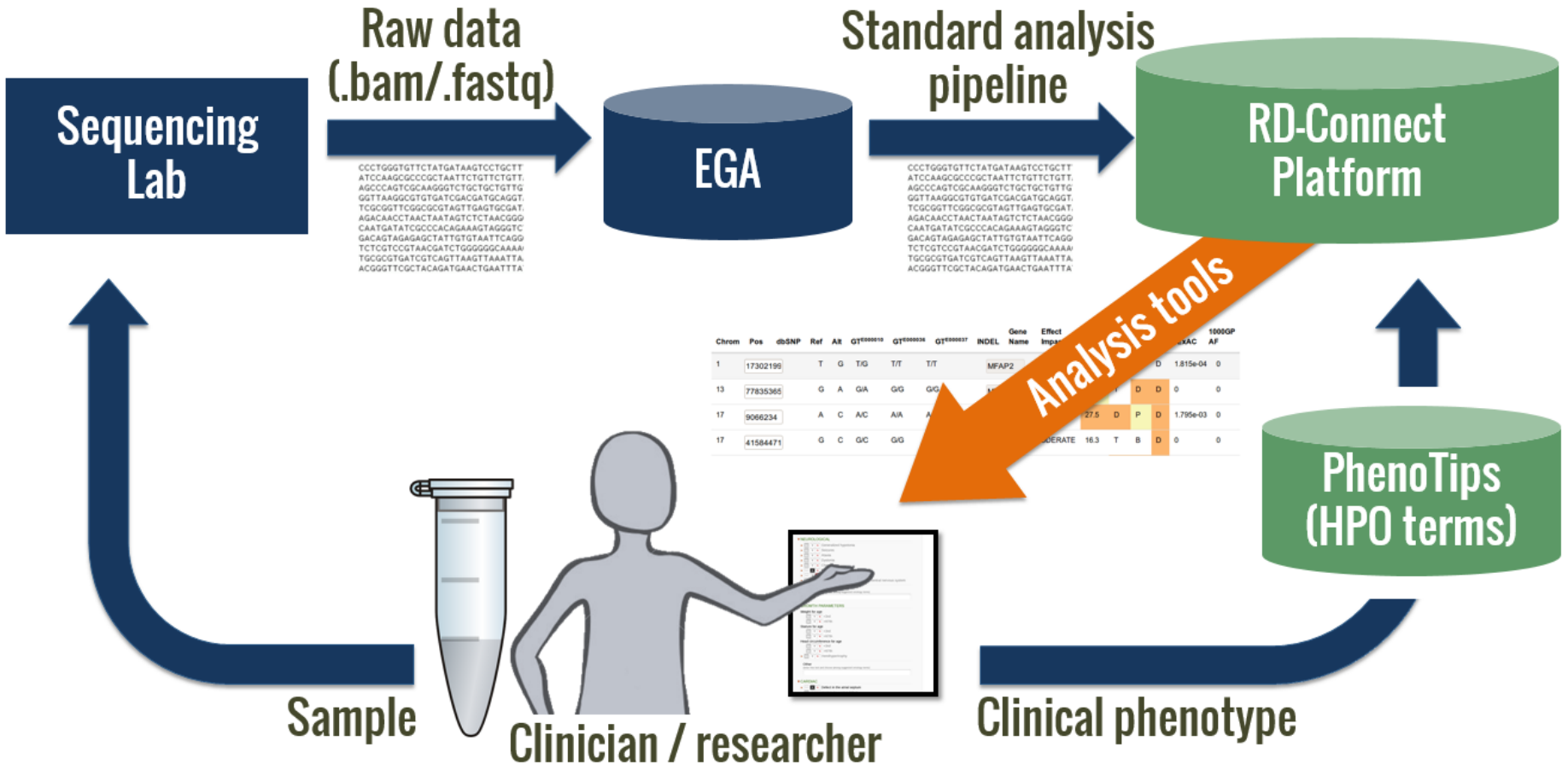
Answer:

No / Yes – here are some of the details of some ***similar*** **individuals**

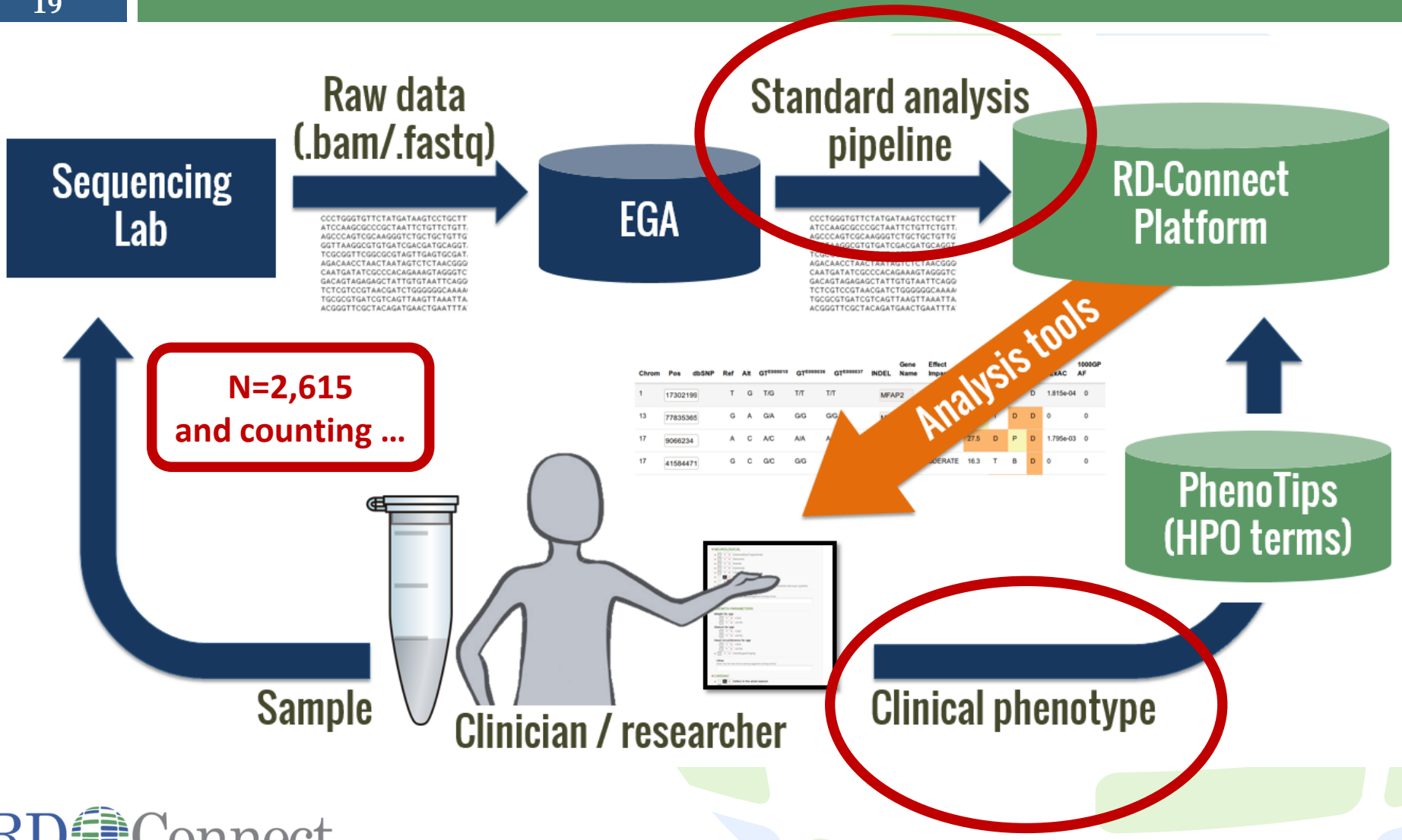
Philippakis, A.A. *et al*. (2015)

RD Connect

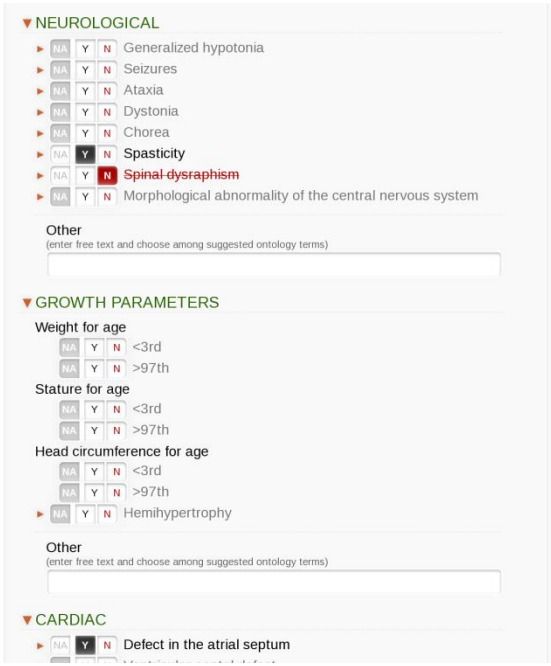
Genome-phenome data flow into RD-Connect

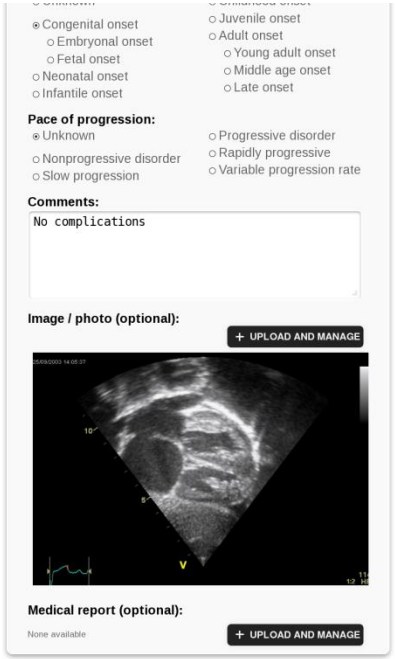


Genome-phenome data flow into RD-Connect



RD-Connect PhenoTips Instance





Deep phenotyping in **PhenoTips** (Brudno *et al.*) achieved using the **Human Phenotype Ontology** (HPO – Robinson, Köhler *et al.*)

Diseases classified using the **Orphanet Rare Disease Ontology** and **OMIM** identifiers

Information from PhenoTips can be sent directly to other tools within the platform (e.g. Exomiser, MME)

RD Connect

RD-Connect PhenoTips Instance

PHENOTIPS

» DATA » P0000700 NEW PATIENT REC

P0000700

Reported by **Irina Zaharieva** on 2014/06/16 10:13 · Last modified by **Rachel Thompson** on 2016/07/29 06:40

I confirm that patient consent has been obtained which allows next generation sequencing of the sample to which the data refers
I confirm that patient consent has been obtained which allows the sharing of this anonymised clinical information with international collaborators and researchers including those from commercial partners

Patient information

Identifier: DNC0010

Sex: Female

Clinical status

FAMILY STUDY

This patient is the Child **Of patient with identifier** DNC0040
This patient is the Child **Of patient with identifier** DNC0041

Global mode of inheritance:

Sporadic

NO **Consanguinity**

Global pace of progression

Progressive

Global age of onset:

Congenital onset

Clinical symptoms and physical findings

RD-Connect PhenoTips Instance

Clinical symptoms and physical findings

COGNITIVE DYSFUNCTION

NO Intellectual disability
⚠ Normal cognitive development

MUSCLE BULK

Distal arthrogryposis
Arthrogryposis multiplex congenita

SKELETAL DEFORMITIES

Congenital hip dislocation

WEAKNESS

Face
Neck

MOTOR ABILITY

Inability to walk
⚠ can crawl and stand with KAFOs at 4 yrs

OTHER SIGNS

Recurrent lower respiratory tract infections
Muscular hypotonia

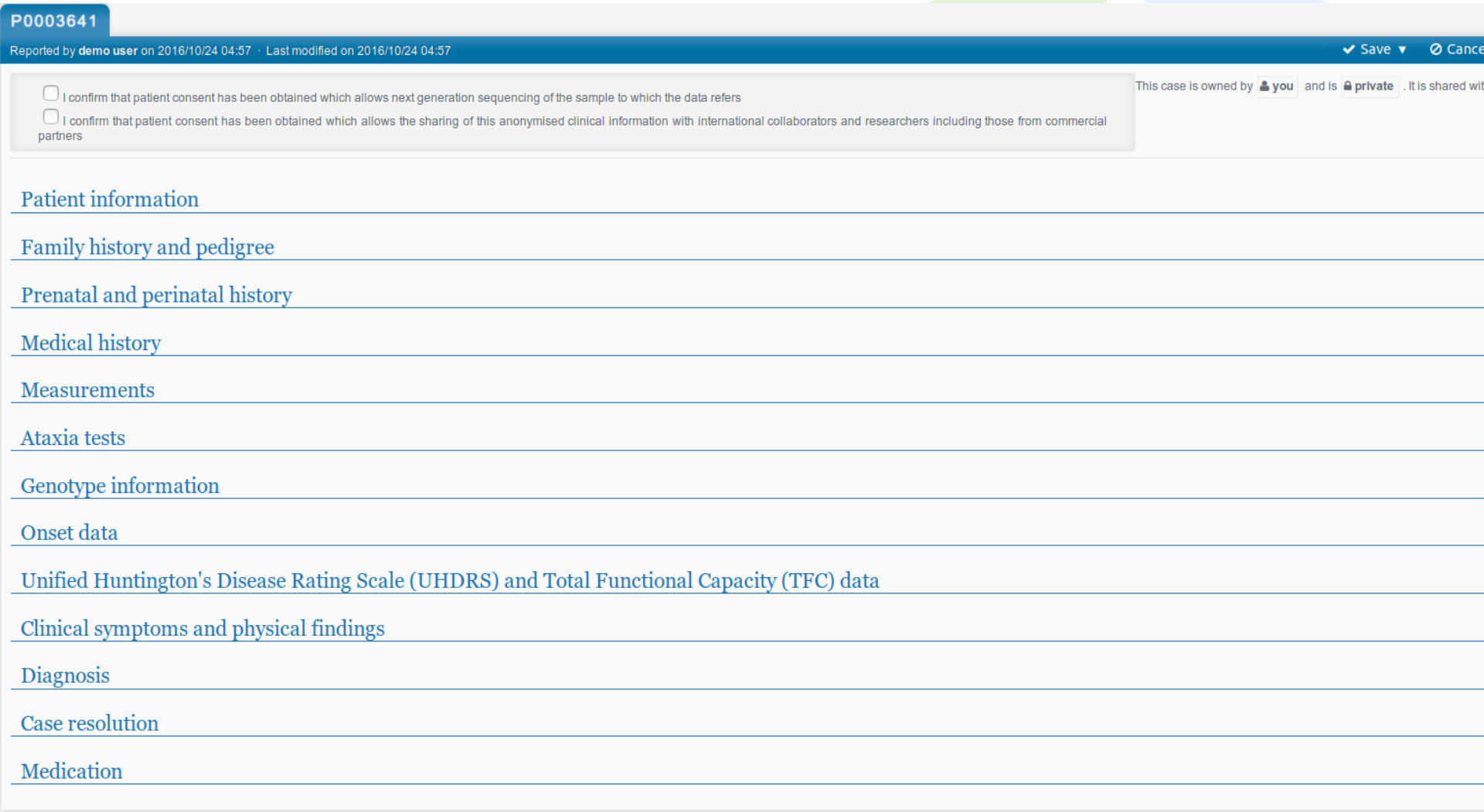
INVESTIGATIONS

Muscle biopsy
⚠ Suggestive of CMY

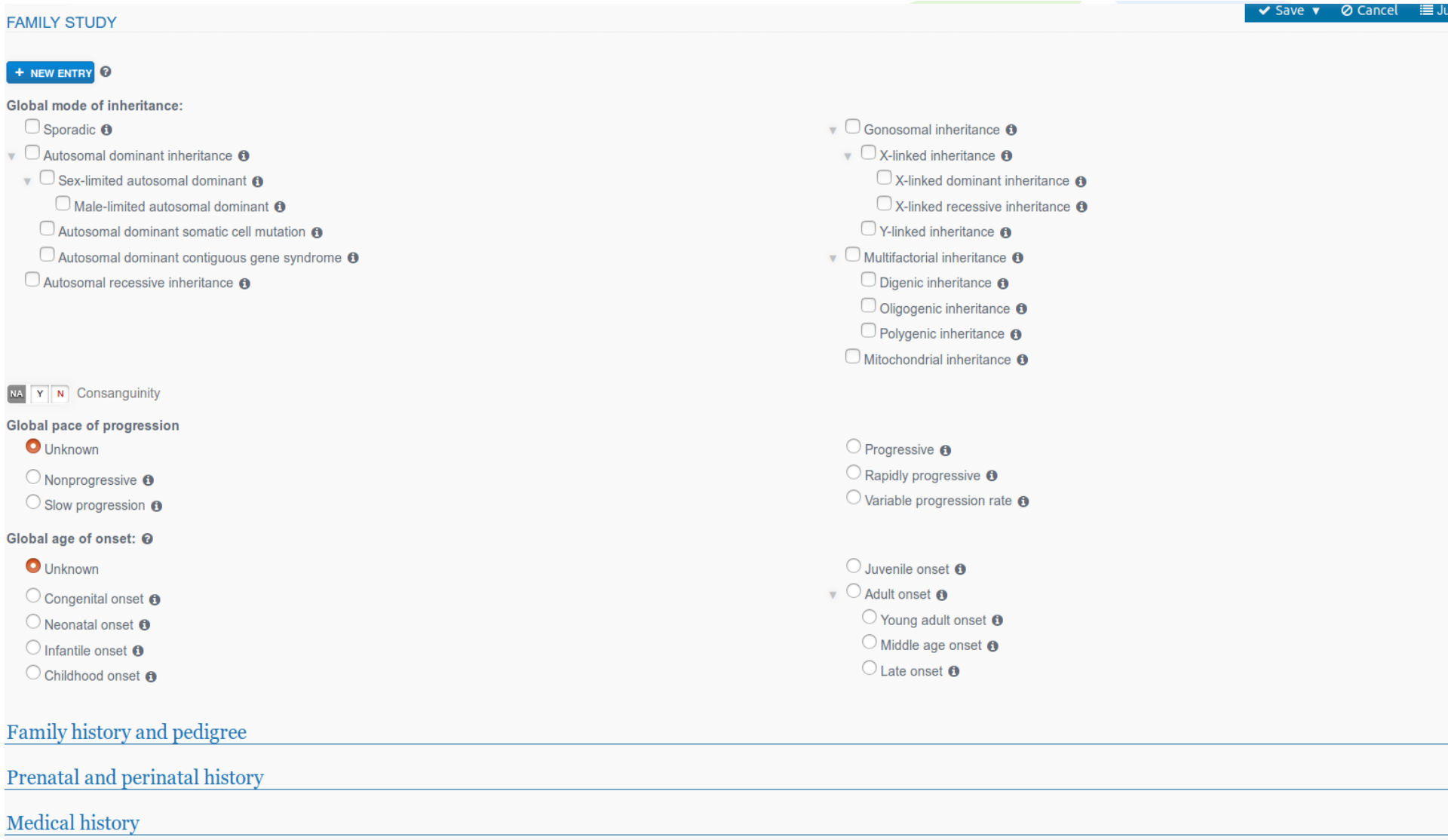
Diagnosis

OMIM disorder: [117000] #117000

RD-Connect PhenoTips Instance



RD-Connect PhenoTips Instance



RD-Connect PhenoTips Instance

Clinical symptoms and physical findings

Save

This patient is clinically normal

Or select observed phenotypes:

Quick phenotype search:

Enter keywords and choose from the suggested ontology terms

BROWSE CATEGORIES

EXPAND ALL | COLLAPSE ALL

- ▶ OCULAR SIGNS
- ▶ BULBAR SIGNS
- ▶ COGNITIVE DYSFUNCTION
- ▼ MUSCLE BULK

Muscle atrophy

- NA Y N Proximal upper limb atrophy
- ▶ NA Y N Neck muscle atrophy
- NA Y N Proximal lower limb muscle atrophy
- NA Y N Paraspinal muscle atrophy
- ▼ NA **Y** N Distal upper limb muscle atrophy
  - ▶ NA **Y** N **Hand muscle atrophy**
- NA Y N Scapular muscle atrophy
- ▼ NA Y N Distal lower limb muscle atrophy
  - NA Y N Tibialis atrophy
- NA Y N Facial muscle atrophy

Muscle Hypertrophy

- ▶ NA Y N Proximal upper limb muscle hypertrophy
- NA Y N Neck muscle hypertrophy
- ▶ NA Y N Proximal lower limb muscle hypertrophy
- NA Y N Paraspinal muscle hypertrophy
- ▶ NA Y N Distal upper limb muscle hypertrophy
- NA Y N Scapular muscle hypertrophy
- ▶ NA Y N Distal lower muscle hypertrophy
- NA Y N Facial muscle hypertrophy

Other

enter free text and choose among suggested ontology terms

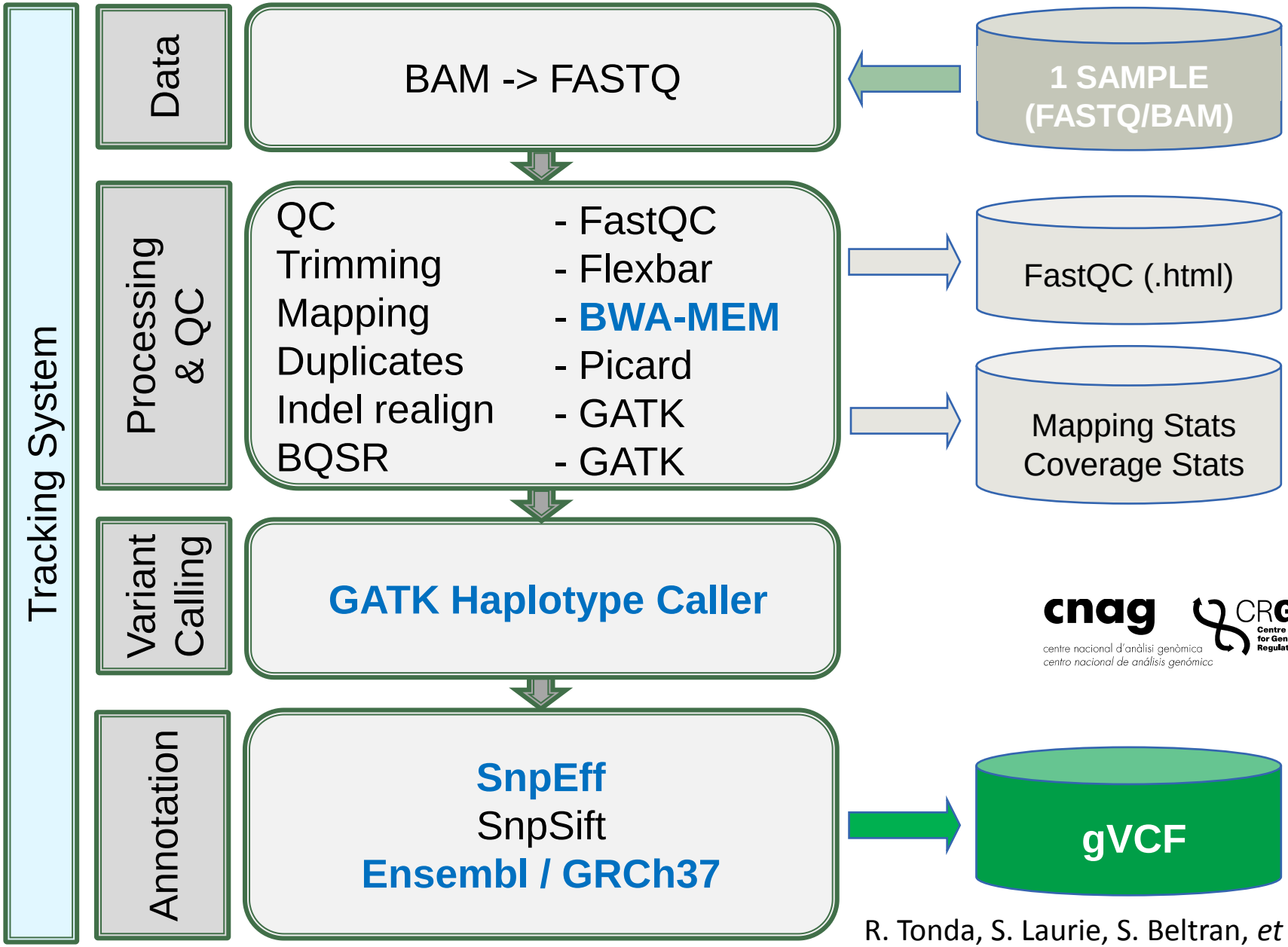
CURRENT SELECTION

How informative is your phenotypic description:

MUSCLE BULK

- Distal upper limb amyotrophy DELETE ADD DETAILS
- Hand muscle atrophy DELETE ADD DETAILS

RD-Connect Variant Calling Pipeline



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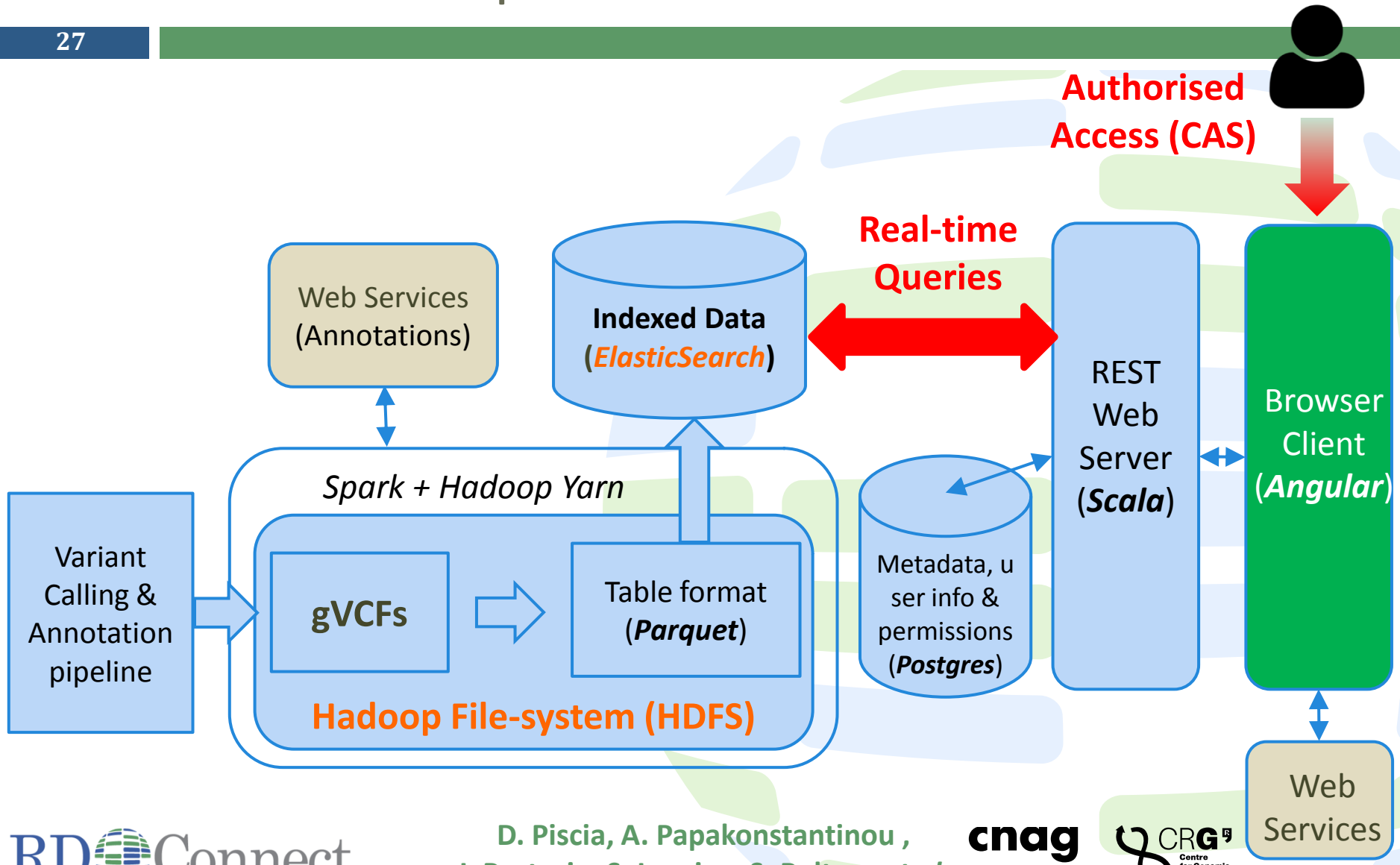
CRG Centre for Genomic Regulation

R. Tonda, S. Laurie, S. Beltran, *et al.*

RD-Connect: Genomics platform architecture



RD-Connect GPAP Interface

RD-Connect GENOMICS ABOUT WELCOME S.BELTRAN {PLATFORM V0.13.0, DATASET 20170426 } FAQ LOGOUT

Filters PRESET FILTERS RESET SHARE ▶RUN QUERY

Variant Type: coding high moderate **Population:** exac **SNV->MT:** A D **SNV->SIFT:** D **SNV->PP2:** D P

Samples | Functional | Predictive | Population | Diseasecard | Candidate | Links | ALFA

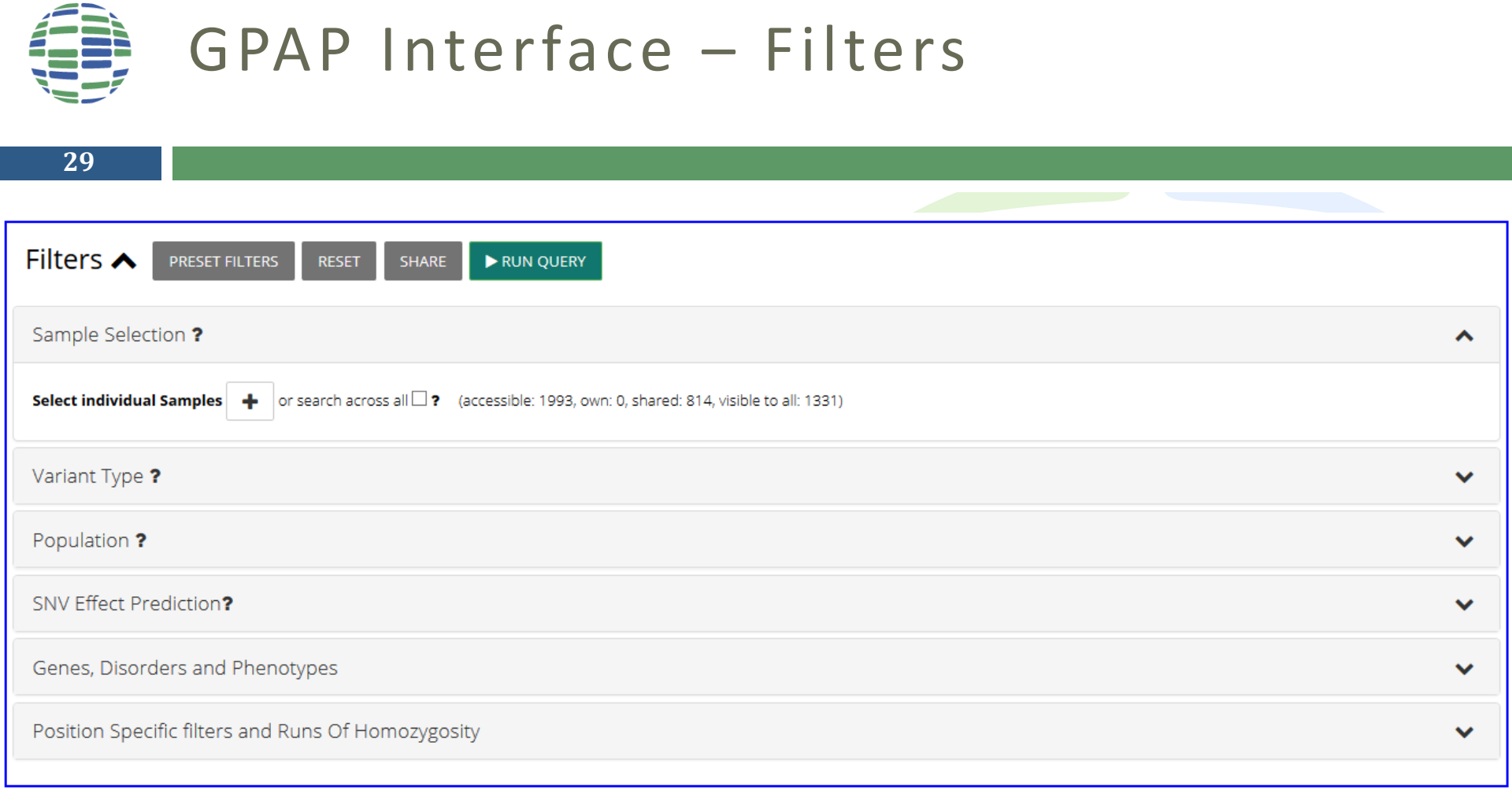
Gene Name	Transcript ID	Effect Impact	Consequence	Feature Type	HGVS coding	Amino Acid change	Amino Acid length	Genotype Number	Exon Rank	CDS Position	Transcript BioType
MFAP2	ENST0000037553	MODERATE	missense_variant	transcript	c.313A>C	p.Thr105Pro	183	1	7/9	313/552	protein_coding
MFAP2	ENST0000037553	MODERATE	missense_variant	transcript	c.310A>C	p.Thr104Pro	182	1	6/8	310/549	protein_coding
MFAP2	ENST0000043854	MODERATE	missense_variant	transcript	c.310A>C	p.Thr104Pro	182	1	7/9	310/549	protein_coding

Variants (11) | Exomiser

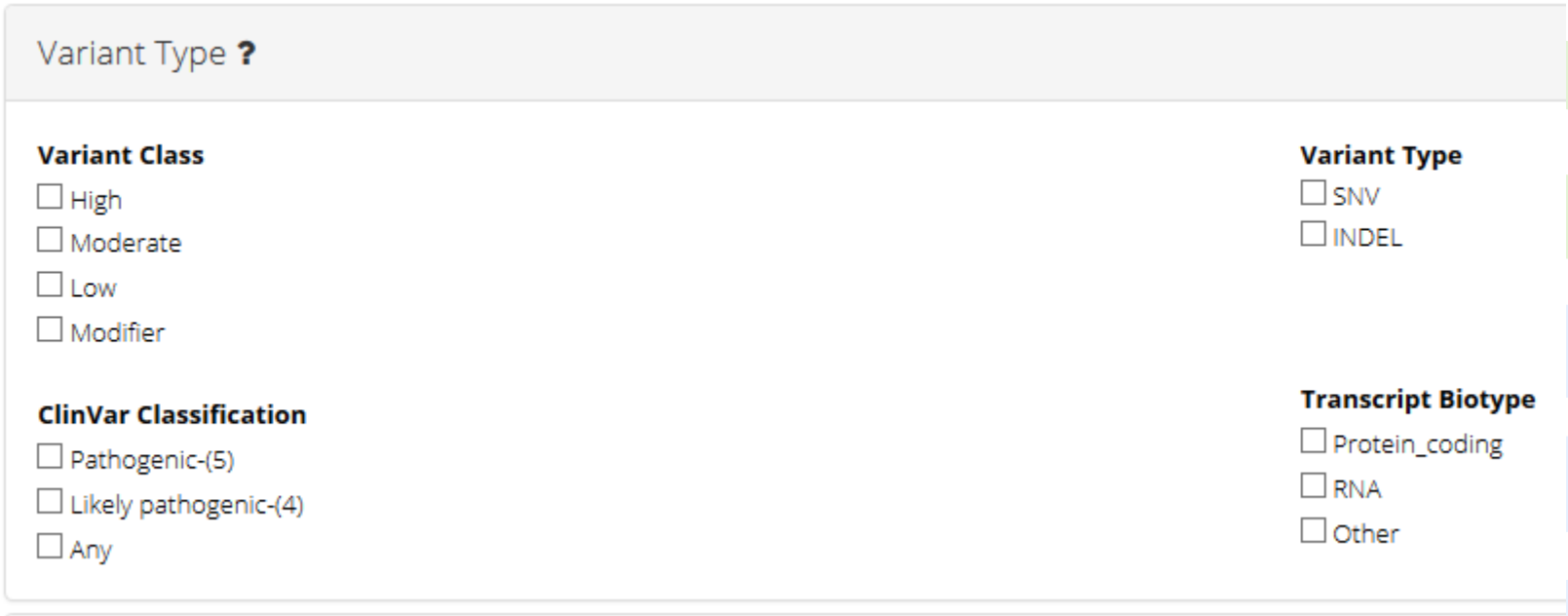
First Previous 1 Next Last EXPORT ALL

Chr	Pos	dbSNP	Ref	Alt	Candidate	$GT^{E000010}$	$GT^{E000036}$	$GT^{E000037}$	INDEL	Gene Name	Effect Impact	ClinVar	CADD	SIFT	PP2	MT	ExAC	1000GP AF
1	17302199	.	T	G	0 ADD	T/G	T/T	T/T		MFAP2	MODERATE		26.9	D	D	D	NA	0
11	93535027	.	C	A	0 ADD	C/A	C/C	C/C		MED17	MODERATE		34	D	D	D	NA	0
13	77835365	.	G	A	0 ADD	G/A	G/G	G/G		MYCBP2	MODERATE		25.6	T	D	D	NA	0
13	110437802	.	A	C	0 ADD	A/C	A/A	A/A		IRS2	MODERATE		24.6	D	D	D	NA	0
13	113210444	.	G	T	0 ADD	G/T	G/G	G/G		TUBGCP3	MODERATE		< 20	T	B	D	NA	0
17	9066234	.	A	C	0 ADD	A/C	A/A	A/A		NTN1	MODERATE		26.7	D	P	D	NA	0

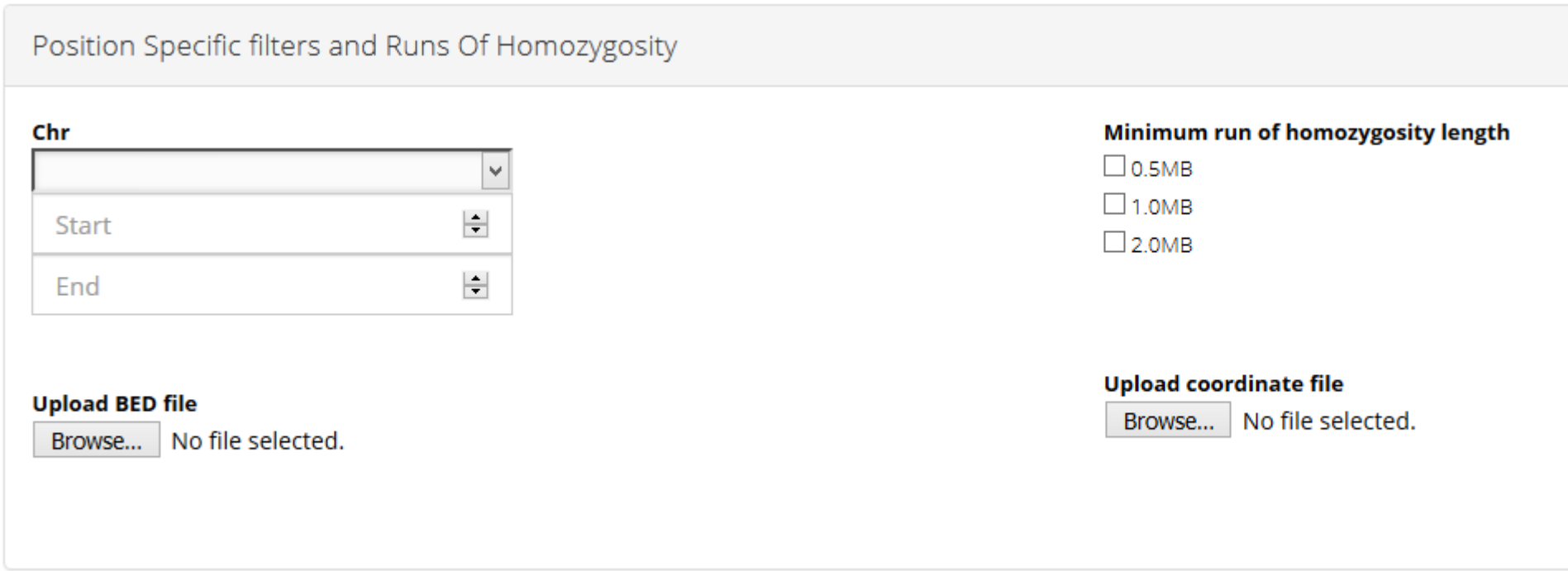
GPAP Interface – Filters



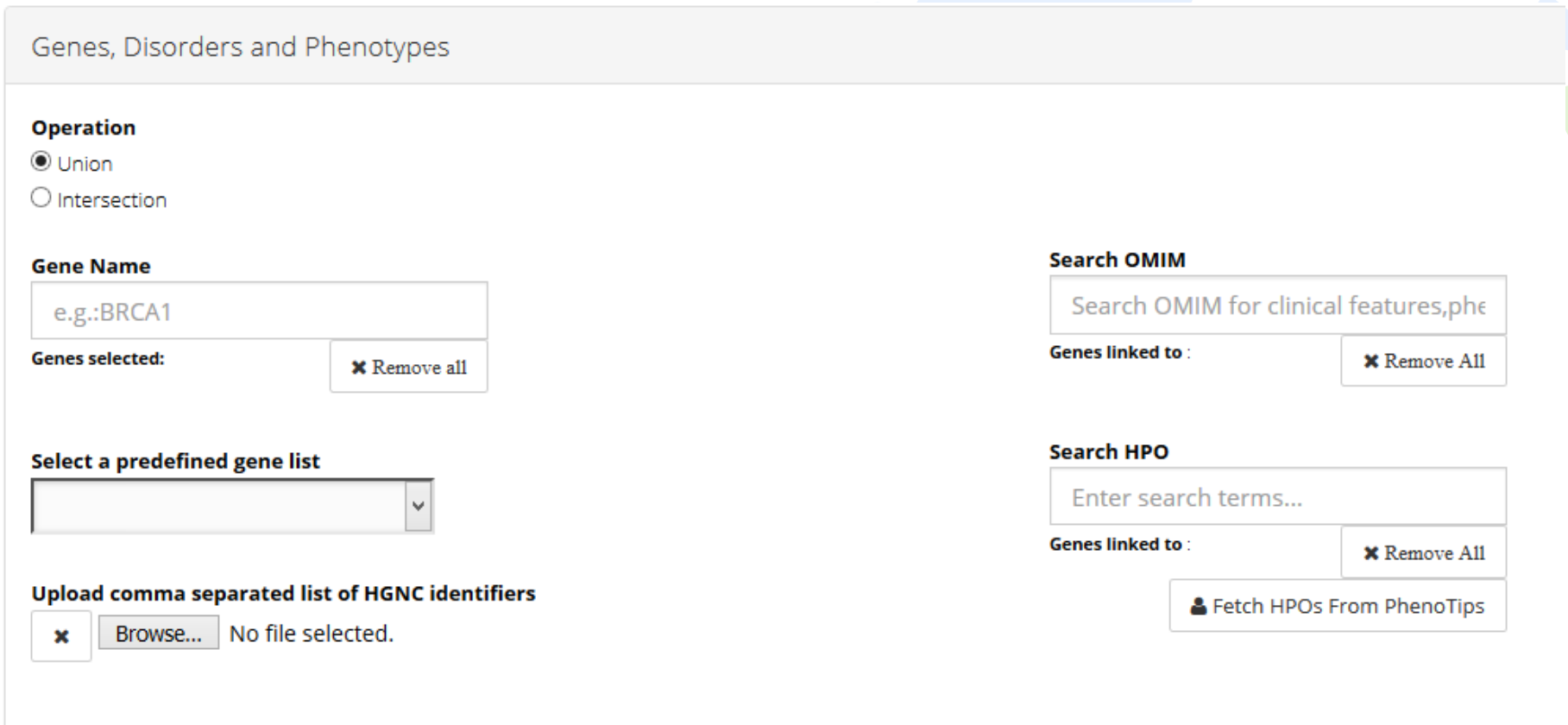
GPAP Filters – Variant Type



GPAP Filters – Runs Of Homozygosity



GPAP Filters – Genes & Phenotypes



Results (integrated data and links)

Samples | Functional | Predictive | Population | Diseasecard | Candidate | Links | ALFA

Gene Name	Transcript ID	Effect Impact	Consequence	Feature Type	HGVS coding	Amino Acid change	Amino Acid length	Genotype Number	Exon Rank	CDS Position	Transcript BioType
MFAP2	ENST0000037553	MODERATE	missense_variant	transcript	c.313A>C	p.Thr105Pro	183	1	7/9	313/552	protein_coding
MFAP2	ENS 5534	MODERATE	missense_variant	transcript	c.310A>C	p.Thr104Pro	182	1	6/8	310/549	protein_coding
MFAP2	ENS 542	MODERATE	missense_variant	transcript	c.310A>C	p.Thr104Pro	182	1	7/9	310/549	protein_coding

ExAC
Ensembl
HSF

Variants (11) | Exomiser

First | Previous | 1 | Next | Last

EXPORT ALL

Chr	Pos	dbSNP	Ref	Alt	Candidate		$GT^{E000010}$	$GT^{E000036}$	$GT^{E000037}$	INDEL	Gene Name	Effect Impact	ClinVar	CADD	SIFT	PP2	MT	ExAC	1000GP AF
1	17302199	.	T	G	0	ADD	T/G	T/T	T/T		MFAP2	MODERATE		26.9	D	D	D	NA	0
11			C	A	0	ADD	C/A	C/C	C/C			MODERATE		34	D	D	D	NA	0
13			G	A	0	ADD	G/A	G/G	G/G			MODERATE		25.6	T	D	D	NA	0
13			A	C	0	ADD	A/C	A/A	A/A			MODERATE		24.6	D	D	D	NA	0
13			G	T	0	ADD	G/T	G/G	G/G			MODERATE		< 20	T	B	D	NA	0
17			A	C	0	ADD	A/C	A/A	A/A			MODERATE		26.7	D	P	D	NA	0
17			A	C	0	ADD	A/C	A/A	A/A			MODERATE		21.4	D	D	D	NA	0
1			G	C	0	ADD	G/C	G/G	G/G			MODERATE		20.6	T	B	D	NA	0
1			G	C	0	ADD	G/C	G/G	G/G			MODERATE		24.4	D	D	D	NA	0
22	30768124	.	T	G	0	ADD	T/G	T/T	T/T			MODERATE		27	T	D	D	NA	0
X	6451869	rs35874450	C	T	0	ADD	C/T	C/C	C/C			MODERATE	2	< 20	T	P		NA	0

Ensembl
ExAC
gnomAD
UCSC
NCBI
DGVa
GWAS Central
GA4GH Beacon
VarSome

OMIM
Ensembl
PubMed
HGMD
Entrez
GeneCards
COSMIC
ClinVar
ExAC
GTEx
gnomAD
GWAS Central
ATLAS
WikiPathways
Open PHACTS

RD Connect

Exomiser for Variant Prioritisation

Variants (9) | Exomiser

Run Exomiser on filtered results

HPO terms are extracted from the first affected sample that is selected. If you want to run the analysis on another sample, please select it as first.

For performance reasons, Exomiser can only run with a number of variants up to 200.

Set Parameters

Inheritance model:

Autosomal dominant

Prioritise genes:

PhenIX (compare phenotypes against human only)

Exomiser will run with the following HPO terms: *HP:0000297 HP:0000467 HP:0001252 HP:0001374 HP:0002540 HP:0002783 HP:0002804 HP:0005684*

SUBMIT | RESULTS

HPO terms and inheritance model extracted from PhenoTips through API

Data in the RD-Connect GPAP

NeuroOmics
1000 exomes

EURenOmics
1000 exomes

MyoSeq, Newcastle
1000 exomes

SeqNMD, Broad Institute
500 exomes

RD Connect

NCNP, Japan
500 exomes

CIMG, Slovenia
300 exomes

CNAG and BBMRI-LPC
1200 exomes

WE WANT YOU

2016 BBMRI-LPC Whole Exome Sequencing Call

Sequencing the exome of 900 rare disease samples in collaboration with EuroBioBank and RD-Connect

Objectives

- ✓ to **promote the usage of biobanks for rare diseases**
  - samples deposited in the EuroBiobank network
- ✓ to promote the utilization of NGS for the identification of novel causative variants and genes through **free-of-charge sequencing of 900 exomes**
- ✓ to molecularly diagnose RD patients analysis through the RD-Connect GPAP
- ✓ **to promote data sharing for rare disease research** through the EGA and RD-Connect and phenotyping with HPO

2016 BBMRI-LPC Whole Exome Sequencing Call

Sequencing the exome of 900 rare disease samples in collaboration with EuroBioBank and RD-Connect

Eligibility

- ✓ Coordinated projects, with **2-3 PIs from different European countries**
- ✓ Each **project will focus on a particular disorder** and will include at least **3 genetically undiagnosed index cases**
- ✓ Samples must have been obtained with "**informed consent" of donors, including sharing of the data in controlled access repositories and databases**
- ✓ DNA **samples must be available in the biobanks from the EuroBioBank network,** or researchers must commit to deposit them , if project is awarded
- ✓ **Phenotypic description of the donors will have to be provided** using the Human Phenotype Ontology (HPO)

2016 BBMRI-LPC Whole Exome Sequencing Call

Sequencing the exome of 900 rare disease samples in collaboration with EuroBioBank and RD-Connect

Timeline

Call launch	June 23rd 2016
Project submission	by July 25th 2016
Review Process	by September 2nd 2016
Sample Submission	by September 30th 2016
Sequencing	by December 30th 2016
Data release	by February 28th 2017

2016 BBMRI-LPC Whole Exome Sequencing Call

Sequencing the exome of 900 rare disease samples in collaboration with EuroBioBank and RD-Connect

Outcome

- ✓ 17 transnational projects selected
- ✓ 899 samples sequenced in total
- ✓ 10 projects sequenced at CNAG (545 samples)
- ✓ 7 projects sequenced at the Wellcome Trust Sanger Institute (345 samples)

2016 BBMRI-LPC Whole Exome Sequencing Call

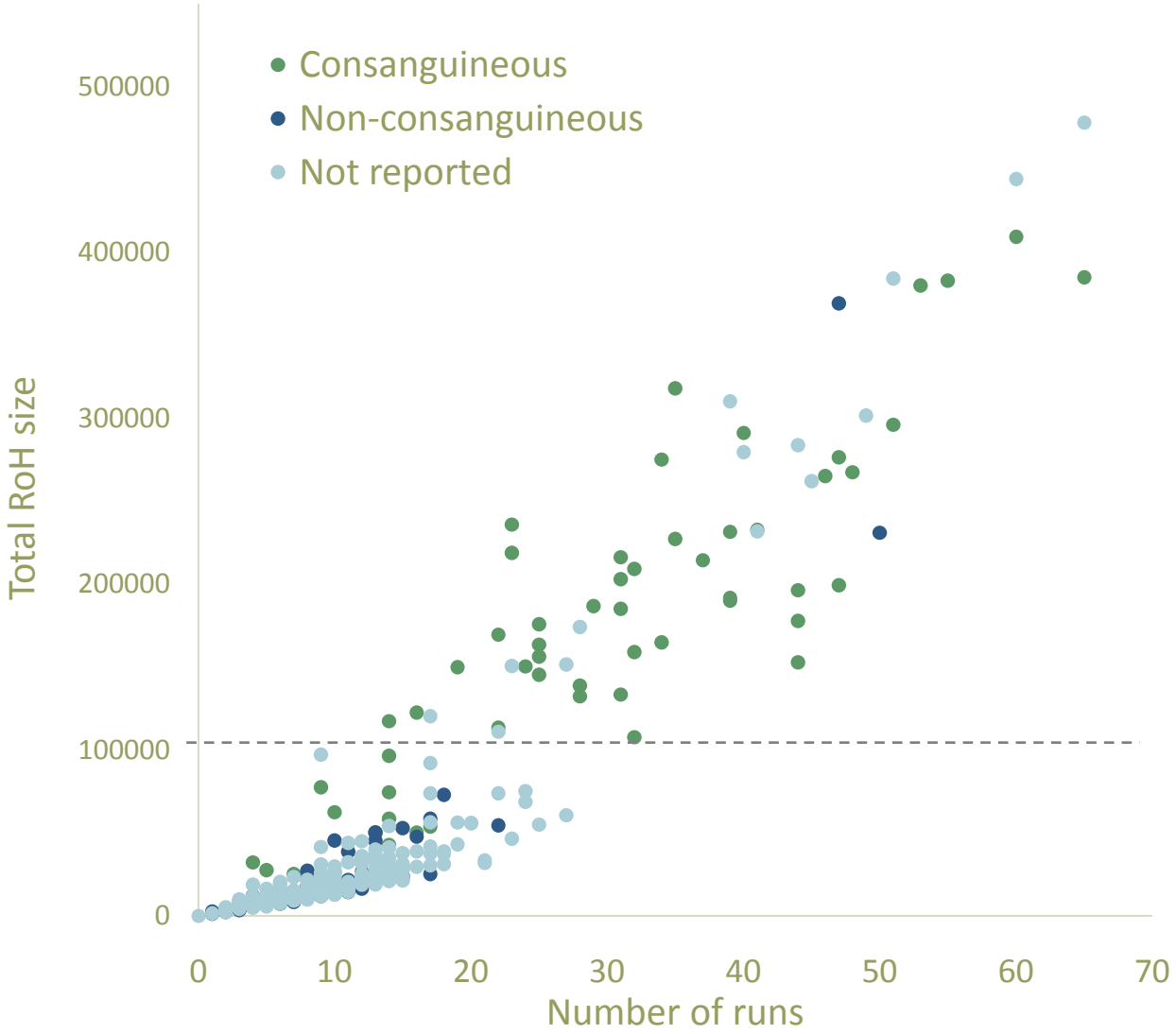
TITLE OF AWARDED PROJECT	PRINCIPAL INVESTIGATORS
Exome sequencing of a cohort of Rett syndrome-like patients	Armstrong Morón JS, Tejada M, Renieri A
Genetic Heterogeneity of the familial gastric neuroendocrine tumors	Benitez J, Valdés Socin H, Calvete O
Molecular diagnosis of albinism	Montoliu José L, Carracedo Alvarez Á, Arveiler B
Deciphering the molecular causes of Ophthalmogenetic Diseases: Exome Sequencing analysis for gene discovery	Ayuso C, Rivolta C
Exome trios in 50 patients with gastroschisis	Lapunzina P, Scarano G
Identification of novel genes in patients with Congenital Myasthenic Syndrome (CMS)	Lochmüller H, Senderek J
Identification of Molecular Pathology of Undiagnosed Patients with Mitochondrial Disorders by Whole Exome Sequencing	Dursun A, Soler D
Undiagnosed cases with complex phenotypes including intellectual disability	Posada De La Paz M, Forzano F, Mari F
Identification of Genetic Causes of Undiagnosed Epileptic Encephalopathies	Yalnizoglu D, Antonietta Mencarelli M, Koksal Ozgul R
Unravelling the genetic cause of a neuropathic pain phenotype segregating in an extended multigenerational family through WES	Cormand Rifà B, Serra J
Identification of genes involved in congenital disorders of glycosylation and 3-methylglutaconic aciduria	Tort F, Morava Kozicz E, Vilarinho L
Undiagnosed cases of congenital and dystrophic neuromuscular diseases	Mora M, Muntoni F, Scerri C
Gene Characterization in Carbohydrate metabolic alterations (neonatal diabetes & congenital hyperinsulinemic) in early childhood	Castaño L, Barbetti F, Polak M
Identification and characterization of the underlying genetic and molecular defect in undiagnosed inherited ataxias by WES	Matilla-Dueñas A, Houlden HJ
Whole exome sequencing for clarification of rare causes of axonal Charcot-Marie-Tooth disease	Espinós C, Seeman P
Identification of additional genes involved in Pyruvate Kinase Deficiency phenotypic variability	Segovia JC, Bianchi P, van Wijk R
Identification of New Genes involved in Infant and Adult Sudden Cardiac Death	Gimeno Blanes JR, Corral De La Calle J, Elliott PM

BBMRI-LPC exome call: Preliminary Results

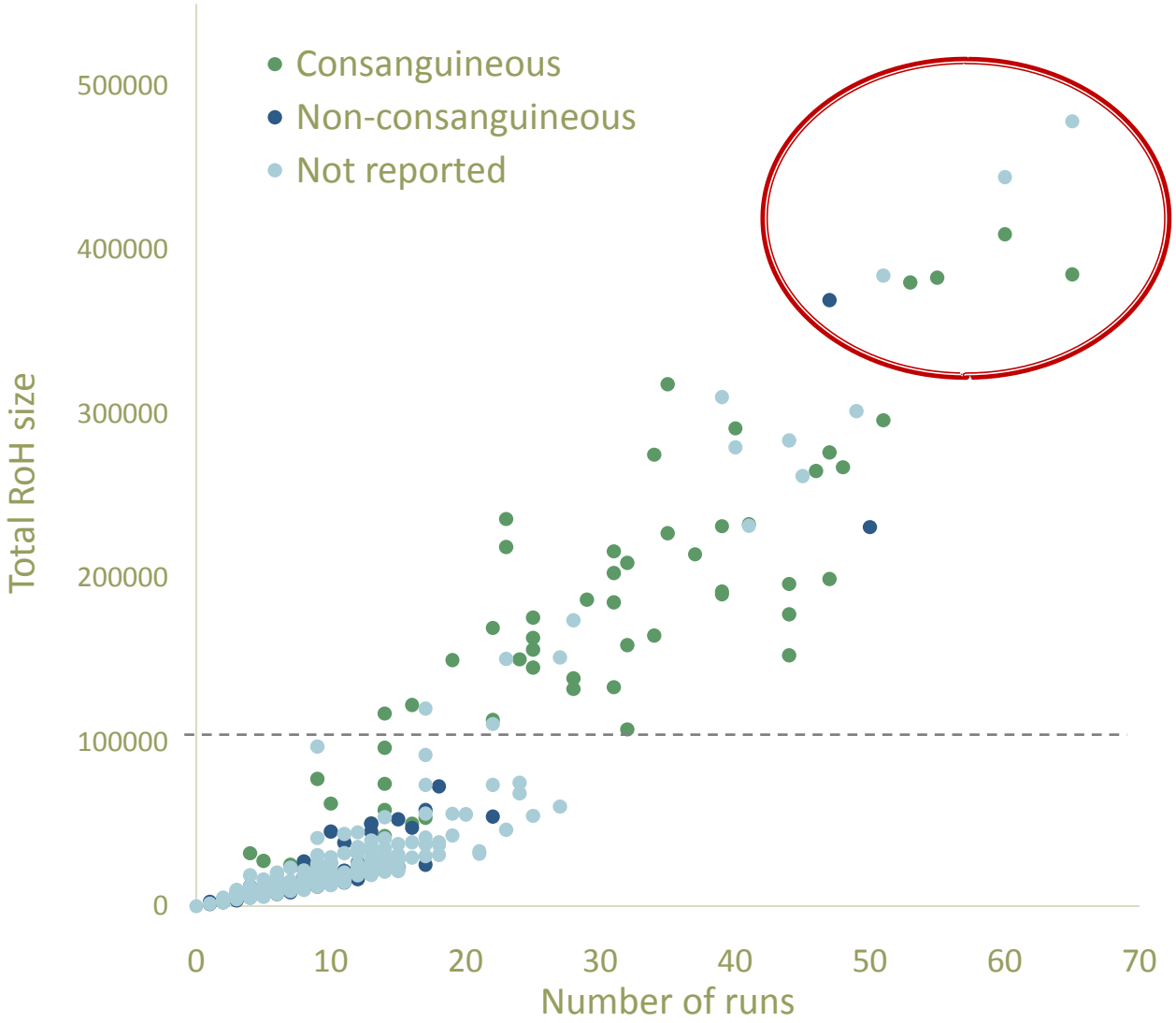
PROJECT	PI.1	CLINICAL REFERRAL	TOTAL SAMPLES	TOTAL FAMILIES	PEDIGREES ANALYSED	TOTAL CASES WITH CANDIDATE VARIANTS (ALL)	TOTAL CASES WITH CANDIDATE VARIANTS (%)	TOTAL VERY STRONG CANDIDATES	TOTAL VERY STRONG CANDIDATES (%)
BBMRI_01	Carmen Ayuso	Ophthalmogenetic Diseases	41	10	10	4	40	4	40
BBMRI_02	Hanns Lochmuller	Congenital Myasthenic Syndrome	87	47	47	16	34	16	34
BBMRI_03	Ali Dursun	Mitochondrial Disorders	50	41	41	25	61	19	46
BBMRI_04	Manuel Posada	Intelectual disability	60	26	8	5	62,5	4	50
BBMRI_05	Dilek Yalnizoglu	Epileptic Encephalopathies	50	32	32	19	59	12	37
BBMRI_06	Bru Cormand	Neuropathic pain	15	1	1	0	0	0	0
BBMRI_07	Marina Mora	Neuromuscular diseases	81	52	52	24	46	13	25
BBMRI_08	Antonio Matilla	Ataxia	50	28	28	16	57	11	39
BBMRI_09	José C. Segovia	Pyruvate Kinase Deficiency	31	Not released					
BBMRI_10	JR Gimeno Blanes	Sudden Cardiac Death	83	Pending					
		Total	548	237	219	**109**	50	**79**	36

CANDIDATE VARIANTS	Very good candidates + VUS in described genes or P and LP in genes associated with diseases that partially fit the clinical symptoms-> need validation from clinician
VERY STRONG CANDIDATES	P or LP variants in described genes or VUS in described genes that fit the phenotype of the patient

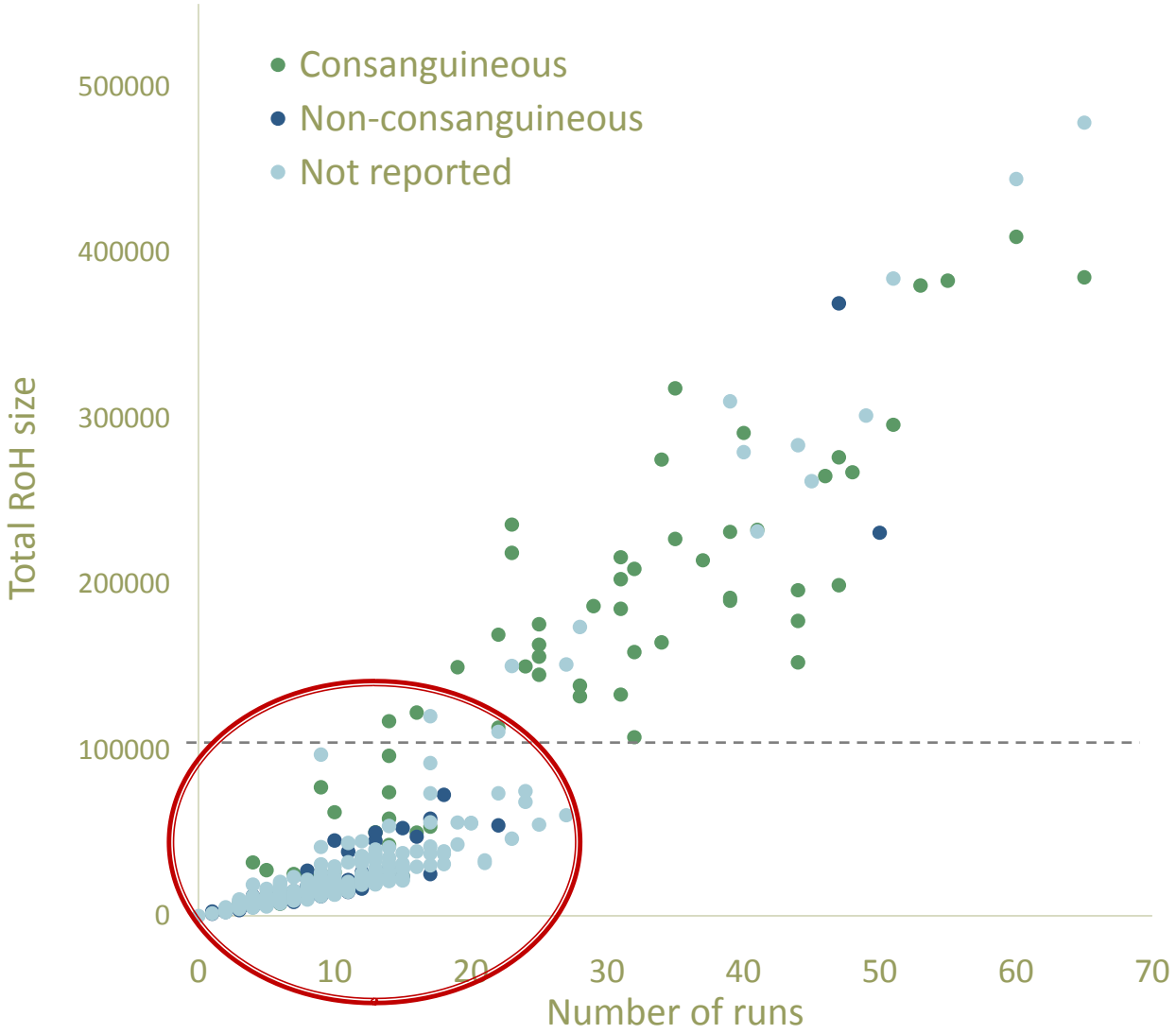
Concordance of length of RoH with consanguinity reported in PhenoTips



Concordance of length of RoH with consanguinity reported in PhenoTips



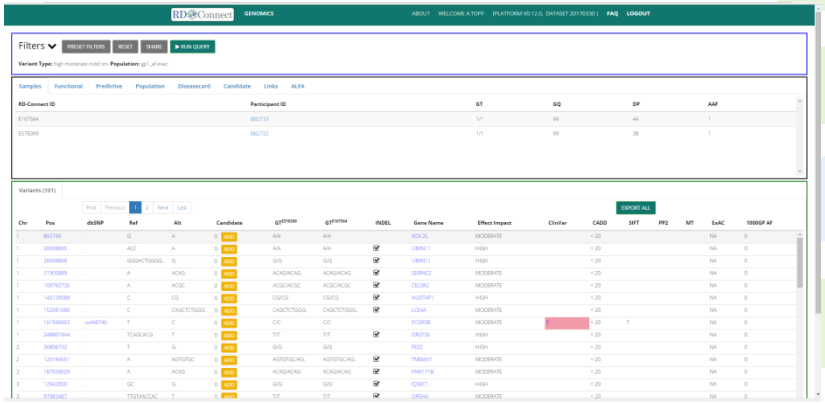
Concordance of length of RoH with consanguinity reported in PhenoTips

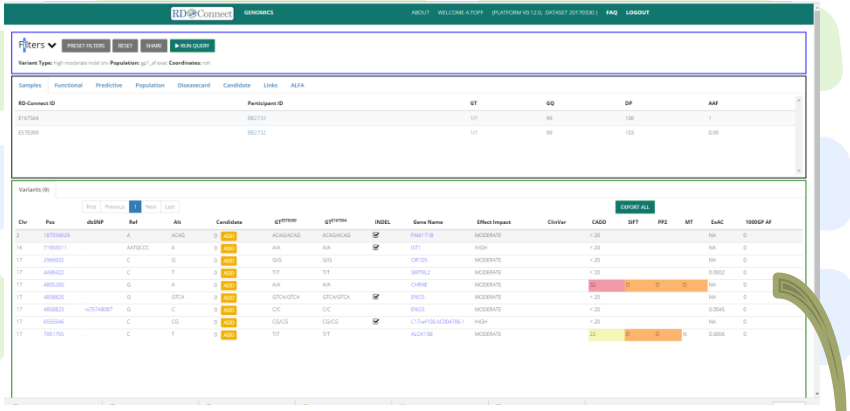


Application of RoH filter

Standard filters – 101 variants

Homozygous blocks – 9 variants

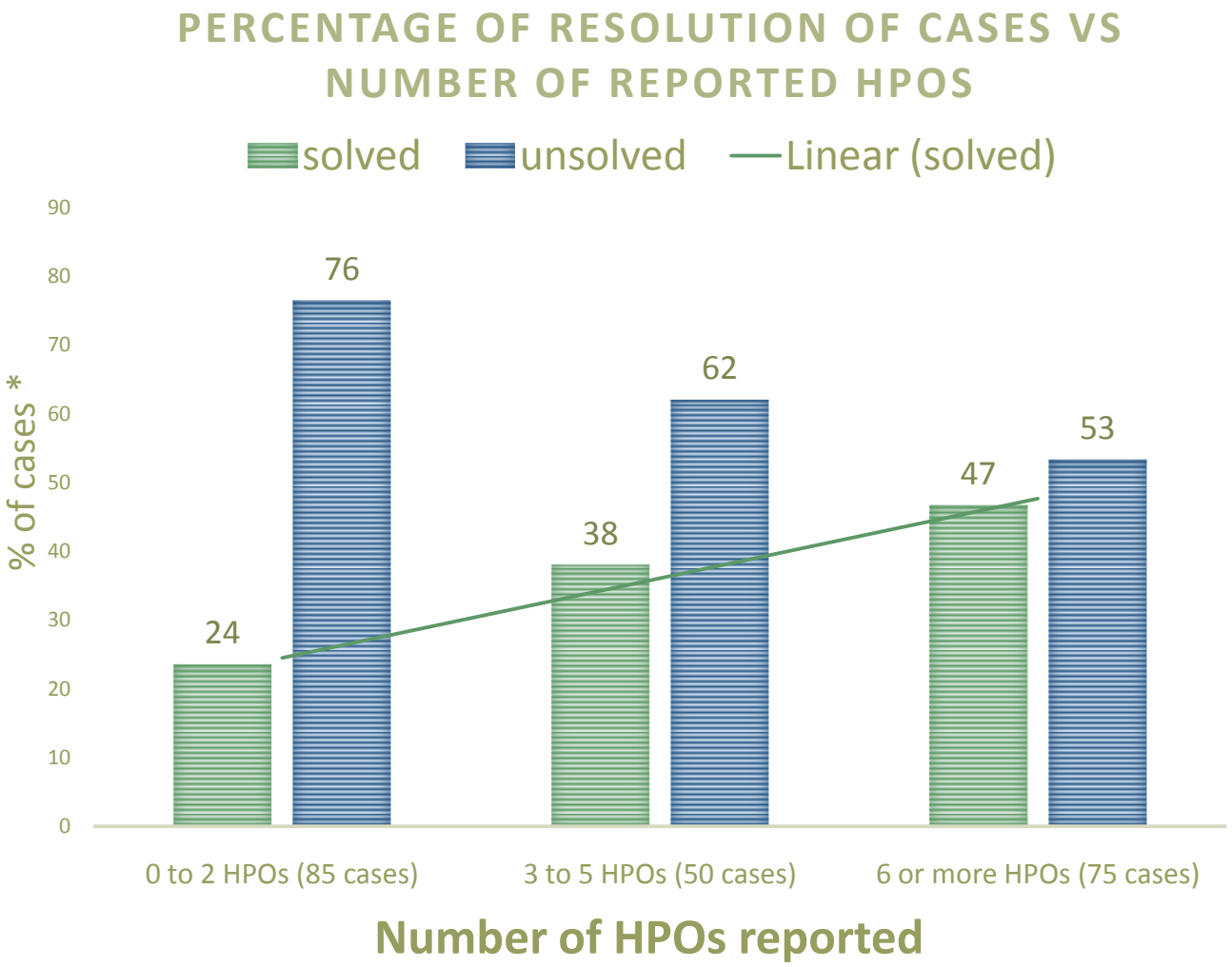




17	4805260	G	A	A/A	A/A	CHRNE	MODERATE	32	D	D	D	NA	0

RD Connect

Importance of Detailed Phenotypic Description



***Only cases with "strong candidates" have been taken into account**

RD-Connect: Become a partner

- Registration details are available on-line – first point of contact is **platform@rd-connect.eu**
  - Check your consent forms - do they cover data sharing for research purposes?
  - Check your phenotypic data - do you have a detailed phenotype for each participant?
  - Check your data - do you have access to the BAM / CRAM / FastQ files from your sequencing experiments?
  - Contact us - platform@rd-connect.eu for further details

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WP5: Unified platform
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WP6 Ethical/legal/social
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WP7: Impact/Innovation
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