

The RD-Connect platform for data sharing

Solutions for data sharing in clinical research

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ESHG

Copenhagen 28. May 2017

centre nacional d'anàlisi genòmica
centro nacional de análisis genómico

cnag

```
3,123 0|0:123:123,123 0|0:123:123,123 0|1:123:123,123 0|1:49:52
0:123:123,123 0|0:123:123,123 0|0:123:123,123 0|0:123:123,123 0
3,123 0|0:123:123,123 0|0:123:123,123 0|0:123:123,123 0|0:52:12
0:123:123,123 0|1:123:123,123
HQ2 0|0:123:123,123 1|0:123:123,123:56:0.0852854;21:19 0
3,123 0|0:123:123,123 0|0:83:83,123 0|1:43:123,43 0|0:123:1
0:123:123,123 1|0:68:68,123 0|0:123:123,123 0|0:123:123,123 0
14,123 0|0:51:123,51 0|0:43:43,123 0|0:87:123,87 0|0:114:1
0:123:123,123 1|0:37:37,123 0|0:123:123,123 0|0:123:123,123 0
,37 0|0:123:123,123 1|0:123:123,123
HQ2 0|0:123:123,123 0|0:123:123,123:59:0.102882;5:3 0|0:113:1
0:123:123,123 0|0:123:123,123 0|0:123:123,123 0|0:76:105,76 0
3,123 0|1:123:123,123 0|0:76:76,123 0|0:123:123,123 0|0:123:1
0:123:123,123 0|0:123:123,123 0|0:123:123,123 1|0:123:123
,123 0|0:123:123,123 1|0:123:123,123 0|1:106:123,106
|1:123:123,123 0|0:113:123,113
T:GQ:HQ1,HQ2 0|0:123:1
```

```
ro@n8 indelcalling]$
ro@n8 indelcalling]$ cp /scratch/devel/fcastro/data/1000genomes/indelcalling/CEU* .
ro@n8 indelcalling]$ cp /scratch/devel/fcastro/data/1000genomes/indelcalling/README_* .
ro@n8 indelcalling]$ ls
ro@n8 indelcalling]$ cp /scratch/devel/fcastro/data/1000genomes/indelcalling/CEU.SRP000031.2010_03.indels.genotypes.vcf.gz.tbi CEU
ro@n8 indelcalling]$ pwd
/scratch/devel/fcastro/COPY_temp/indelcalling
ro@n8 indelcalling]$ cd /scratch/
```



Infrastructure for Rare Disease Research

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6-year project funded by EU 7th Framework Programme

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

- Contributing to **IRDiRC objectives** of delivering **200 new therapies** for rare diseases, and **means to diagnose most rare diseases** by 2020
- Creating a central system for **reprocessing, storage, analysis** and **sharing** of *-omics* data
 - Including integration of **phenotypic** and **biosample** data, and development of **new bioinformatic tools** to aid detailed analysis

Introduction to Rare diseases: data fragmentation



**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
BIOSAMPLES,
DISEASE &
PATIENT
INFO, OMICS,
GENOTYPE-
PHENOTYPE**

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

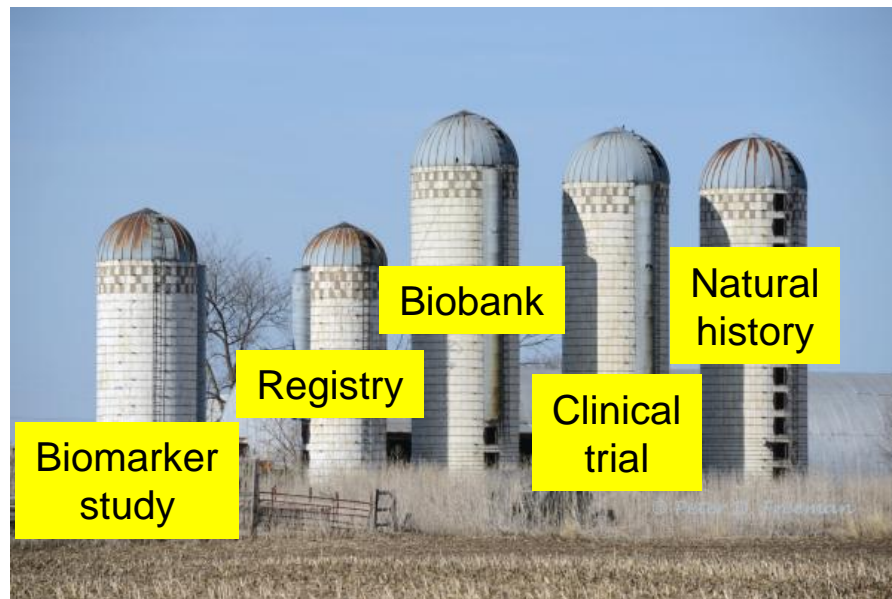
Standardized phenotypic data collection

Searchable catalogue of biosamples

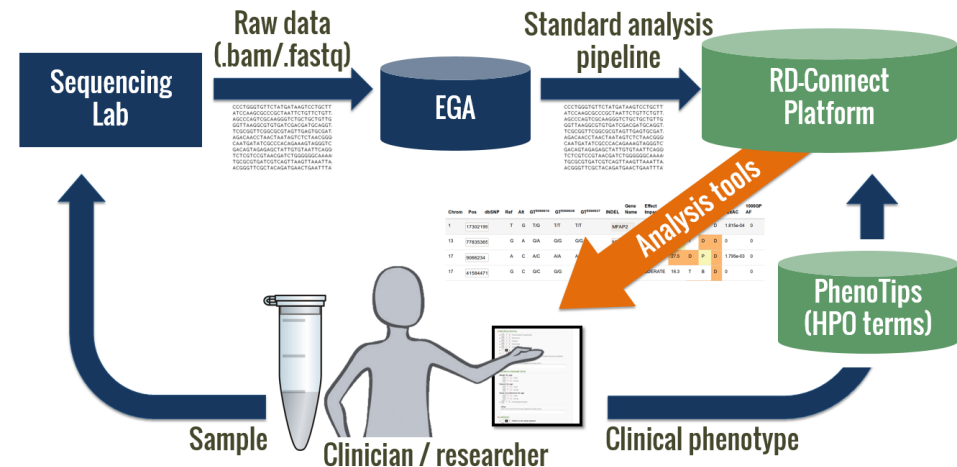
Data linkage across resources

Overcoming Silos

Data sharing for research and better data analysis



Omics data, clinical data and biosamples from individual with RD



Disease-causing variant can be identified using the genomics analysis platform

Sample is findable in the Sample Catalogue

Registry data in the ID-Cards directory of registries and biobanks



Data in the RD-Connect Platform

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Genomic data

WES, WGS,
gene panels



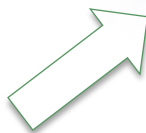
Clinical data

phenotypic data,
patient registries



Sample data

biobanks



Other omics data

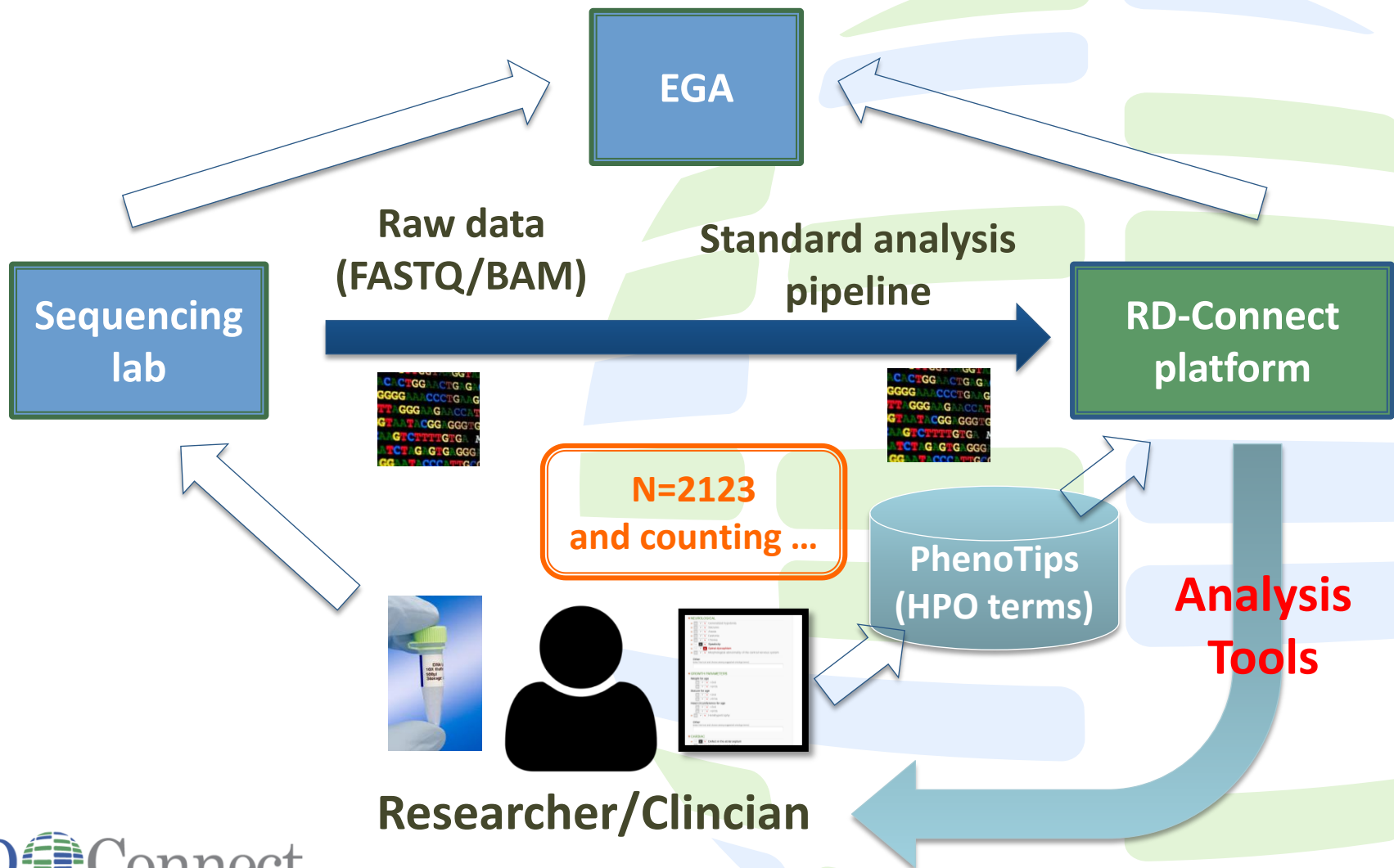
transcriptomics,
metabolomics,
proteomics ...





Geno:pheno data flow in RD-Connect

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Data Submission Workflow

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SUBMISSION PLATFORM

[ABOUT](#) [WELCOME](#) [FAQ](#) [LOGOUT](#)

Submission Data to RDconnect [See instructions](#)

1- Submit Participant Set

2- Submit Experiment Set

3- Upload data

Manage your RDconnect data

Participant

Experiment

Uploaded files



Submit participants (linked to ID-Cards patient registries)

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SUBMISSION PLATFORM

Submit Participant/s to RDconnect [See instructions](#)

+ +10 +100 VALIDATE AND CONTINUE EDITING

SAVE

Group assigned to the entries cnag

PhenoTips ID * ?	Patient Registry ?	Matchmaker exchange ?	Sex * ?	Study ?	Mode of inheritance ?	Consanguinity ?
<input type="text" value="P0000700"/> <i>Phenotips_ID already exists in the database</i>	<input type="text" value="× NBS Connect(ID=11509) ×"/>	<input type="checkbox"/>	<i>This field is required.</i>			
<input type="text" value="P0000701"/>	<input type="text" value="× European patient registry on autoimmune ×"/> <input type="text" value="× RDCRN Contact registry(ID=14682)"/>	<input checked="" type="checkbox"/>	male	CMD/CM	unknown	unknown



RD-Connect:PhenoTips Instance

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▼ NEUROLOGICAL

▶ ☐ NA ☒ Y ☐ N Generalized hypotonia

▶ ☐ NA ☒ Y ☐ N Seizures

▶ ☐ NA ☒ Y ☐ N Ataxia

▶ ☐ NA ☒ Y ☐ N Dystonia

▶ ☐ NA ☒ Y ☐ N Chorea

▶ ☐ NA ☒ Y ☐ N Spasticity

▶ ☐ NA ☒ Y ☒ N **Spinal dysraphism**

▶ ☐ NA ☒ Y ☐ N Morphological abnormality of the central nervous system

Other
(enter free text and choose among suggested ontology terms)

▼ GROWTH PARAMETERS

Weight for age

▶ ☐ NA ☒ Y ☐ N <3rd

▶ ☐ NA ☒ Y ☐ N >97th

Stature for age

▶ ☐ NA ☒ Y ☐ N <3rd

▶ ☐ NA ☒ Y ☐ N >97th

Head circumference for age

▶ ☐ NA ☒ Y ☐ N <3rd

▶ ☐ NA ☒ Y ☐ N >97th

▶ ☐ NA ☒ Y ☐ N Hemihypertrophy

Other
(enter free text and choose among suggested ontology terms)

▼ CARDIAC

▶ ☐ NA ☒ Y ☐ N Defect in the atrial septum

Onset

◉ Congenital onset

○ Embryonal onset

○ Fetal onset

○ Neonatal onset

○ Infantile onset

○ Juvenile onset

○ Adult onset

○ Young adult onset

○ Middle age onset

○ Late onset

Pace of progression:

◉ Unknown

○ Nonprogressive disorder

○ Slow progression

○ Progressive disorder

○ Rapidly progressive

○ Variable progression rate

Comments:

No complications

Image / photo (optional):

+ UPLOAD AND MANAGE

Medical report (optional):

None available

+ UPLOAD AND MANAGE

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. variant prioritization, MME)

Phenotype

Basics >

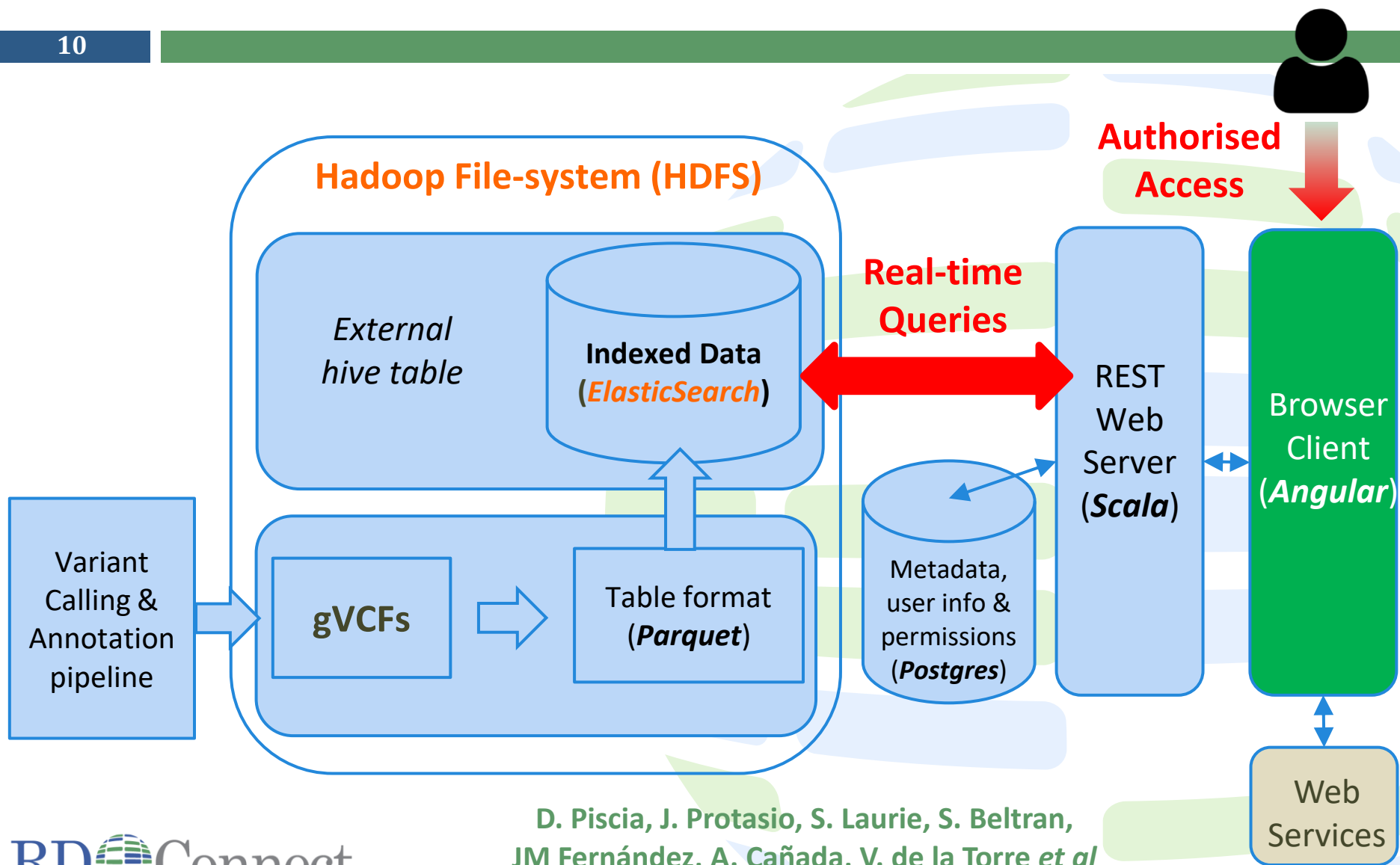
Diagnosis >

Clinical symptoms >



Genomics platform architecture

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D. Piscia, J. Protasio, S. Laurie, S. Beltran,
JM Fernández, A. Cañada, V. de la Torre *et al*



Select samples, set filters and share queries

11

Filters ^

PRESET FILTERS **RESET** **SHARE** **▶ RUN QUERY**

☐ **High stringency**

☐ **Medium stringency**

☐ **Low stringency**

Select individual ☐ **Add ClinVar(score 4-5)**

all ☐ ? (accessible: 1451, own: 0, shared: 174, visible to all: 1277)

compound het.

Affected	ID	0/0	0/1	1/1	Min DP	Min GQ
<input checked="" type="checkbox"/>	E000010 DNC001C	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	20	50
<input type="checkbox"/>	E000036 DNC004C	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	20	50
<input type="checkbox"/>	E000037 DNC0041	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	20	50



Filters: variant type

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- ClinVar can be used for filtering, and ClinVar categories are shown
- Started conversations to explore integration of HGMD

Variant Type ?

Variant Class
☒ High
☒ Moderate
☐ Low
☐ Modifier

Variant Type
☐ SNV
☐ INDEL

ClinVar Classification
☒ Pathogenic-(5)
☐ Likely pathogenic-(4)
☐ Any

Transcript Biotype
☒ Protein_coding
☐ RNA
☐ Other

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
X	6451869	rs35874450	C	T	0	ADD	C/T	C/C	C/C	VCX3A	MODERATE	2	< 20	T	P		NA	0



Filters: lists of genes

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Genes, Disorders and Phenotypes

Gene Name(s)

e.g.:OTUD3

Select a predefined gene list

ACMG Medically Actionable Genes (n=59)
BabySeq Class A and B Genes (n=889)
Digenic gene list, Feb2017 (n=136)
Imprinted-confirmed list, Feb2017 (n=80)
Imprinted-all, Feb2017 (n=253)
Medically Interpretable Genome (n=5,419)
Muscle Gene Table, July2016 (n=416)
Muscle Gene Table, Sep2015 (n=403)
Mitocarta 2.0 (n=1158)
Tubingen HSP Version 6 (n=140)
Tubingen SCA Version 8 (n=183)

Added more lists of genes

OMIM and HPO related genes accessed
through OMIM and PhenoTips APIs

Search OMIM

#254300 MYASTHENIC SYNDROME,

Genes linked to 254300 : DOK7, C4orf25, CMS10

Search HPO

HP:0009053 Distal lower limb musculature wasting

Genes linked to HP:0009053 : SLC46A1, IFT172, DCAF8, ANTXR2, GLE1, RPS6KA3, FAM134B, ZFYVE26, TBK1, TFG, AP1S2, SLC12A6, PRKACA, SIL1, MCCC2, CPT1A, ENTPD1, MUSK, PHKA1, CPT1C, SOX10, HSPG2, SLC5A7, AFG3L2, AR, TRNL1, WDR81, BIN1, PRKAR1A, SUCLG1, WDPCP, TMEM126B, DNA2, PFN1, GNE, MTMR2, DHH, PIK3R2, C9ORF72, C12ORF65, TTPA, NEU1, COX3, COX2, COX1, PIP5K1C, C5ORF42, ABCA1, BBS2, BBS1, EGR2, DMPK, INSR, SLC33A1, PTCR2, BBIP1, ERLIN2, ACADSB, SETX, FAM111B, GJB1, WNK1, NDUFAF4, TDP1, NDUFAF5, MKS1, NDUFAF2, OGDH, CD28, SPRTN, SFXN4, NDUFAE3, ATM, ALDH18A1,



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	ENST0000037553	MODERATE	missense_variant	transcript	c.310A>C	p.Thr104Pro	182	1	6/8	310/549	protein_coding
MFAP2	ENST0000037553	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	Ensembl		C	A	0 ADD	C/A	C/C	C/C		OMIM	MODERATE		34	D	D	D	NA	0
13	ExAC		G	A	0 ADD	G/A	G/G	G/G		Ensembl	MODERATE		25.6	T		D	NA	0
13	gnomAD		A	C	0 ADD	A/C	A/A	A/A		PubMed	MODERATE		24.6	D		D	NA	0
13	UCSC		G	T	0 ADD	G/T	G/G	G/G		HGMD	MODERATE		< 20	T		B	NA	0
17	11351444		A	C	0 ADD	A/C	A/A	A/A		Entrez	MODERATE		26.7	D		P	NA	0
17	9066234		A	C	0 ADD	A/C	A/A	A/A		NTN1	MODERATE		21.4	D		D	NA	0
17	3295088		A	C	0 ADD	A/C	A/A	A/A		GeneCards	MODERATE		20.6	T		B	NA	0
17	GWAS Central		G	C	0 ADD	G/C	G/G	G/G		TMEM132B	MODERATE		24.4	D		D	NA	0
17	GA4GH Beacon		G	C	0 ADD	G/C	G/G	G/G		COSMIC	MODERATE		27	T		D	NA	0
17	VarSome		G	C	0 ADD	G/C	G/G	G/G		ClinVar	MODERATE		< 20	T		P	NA	0
22	30768124	.	T	G	0 ADD	T/G	T/T	T/T		ExAC	MODERATE			D		D	NA	0
X	6451869	rs35874450	C	T	0 ADD	C/T	C/C	C/C		GTEx	MODERATE			T		D	NA	0
										gnomAD	MODERATE	2		T		P	NA	0
										GWAS Central								
										ATLAS								
										WikiPathways								
										Open PHACTS								

RD

Connect



Exomiser for prioritising variant lists

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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

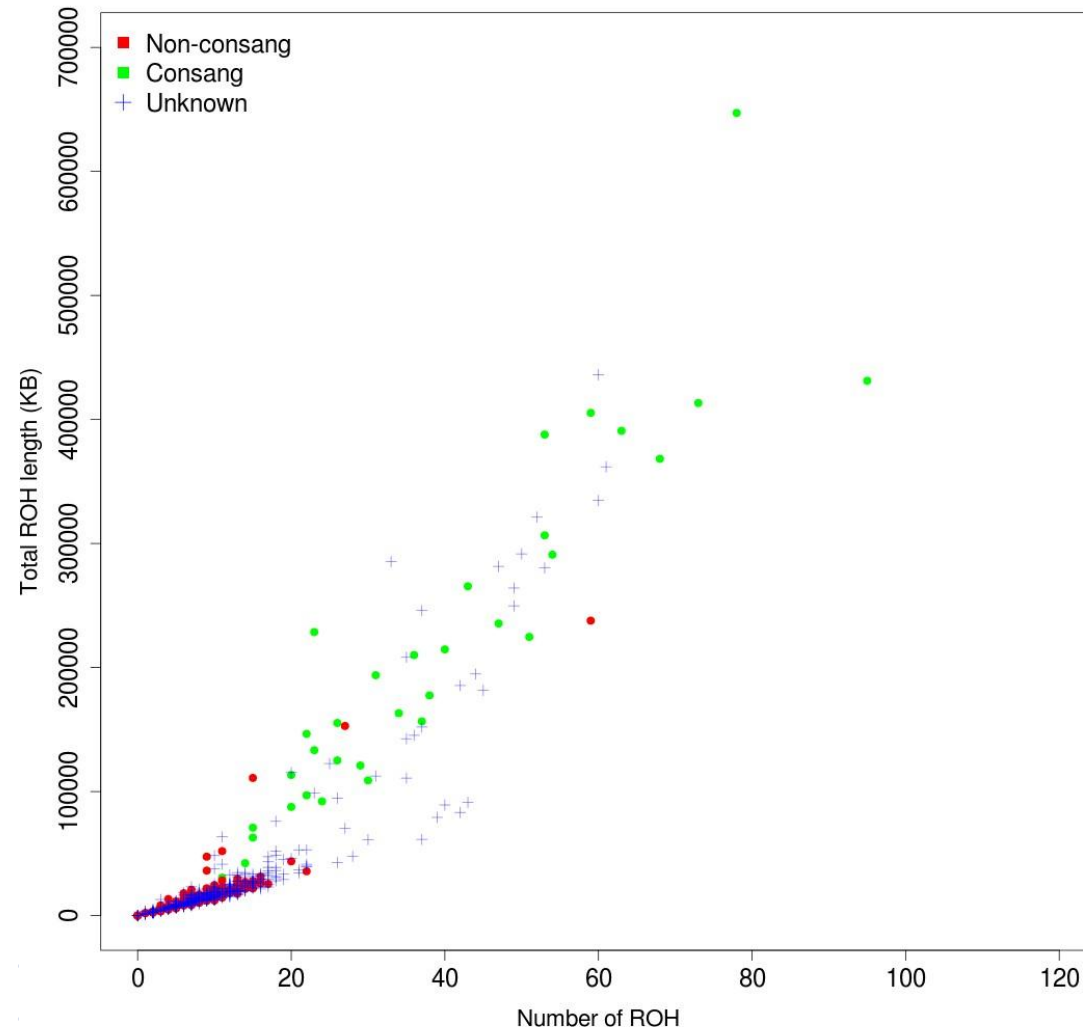
HPO terms and inheritance model extracted from PhenoTips through API



Runs of homozygosity

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Total ROH length versus number of ROH (1000KB)



- Allows identification of consanguineous cases even when not identified as such
- Narrows down regions to focus gene discovery search
- NeurOmics PIs with interesting candidates in these regions have been contacted – feedback welcome



Standard filters – 101 variants

Homozygous blocks – 9 variants

17	4805260	G	A	A/A	A/A	CHRE	MODERATE
----	---------	---	---	-----	-----	------	----------

32	D	D	D	NA	0
----	---	---	---	----	---

BBMRI-LPC Whole Exome Sequencing Call for RD (2016)



Goal:

to promote the utilization of cutting-edge next-generation sequencing technology for the identification of novel causative variants and genes and to molecularly diagnose rare disease patients. BBMRI-LPC also wants to promote biobanking for rare diseases, the use of rare diseases biobanks and responsible data sharing.

To sequence and analyse:

900 exomes in 17 coordinated projects.

Sequencing and analysis carried out at the CNAG-CRG and the Wellcome Trust Sanger Institute (WTSI).

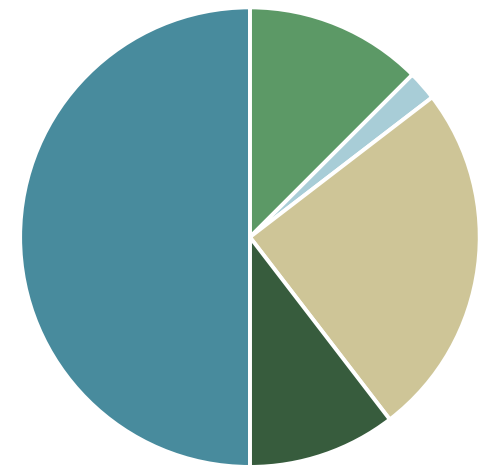
Project results are being released through RD-Connect, where researchers can analyse their data



BBMRI-LPC 1st subcohort (Newcastle/Munich)

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- Congenital myasthenic syndrome (majority), myopathy and neuropathy cases
- Investigators (Senderek, Lochmüller) signed adherence forms with RD-Connect; data on RD-Connect platform
- Samples deposited in Biobank (EuroBioBank/RDC catalogue)
- Sequencing (WES) with CNAG
- Clinical data uploaded on PhenoTips
- 87 samples: 47 kindred; 55 affected



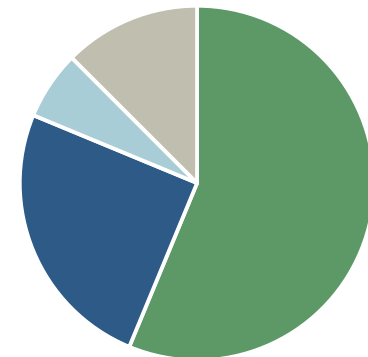
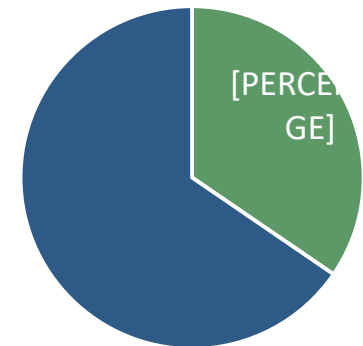
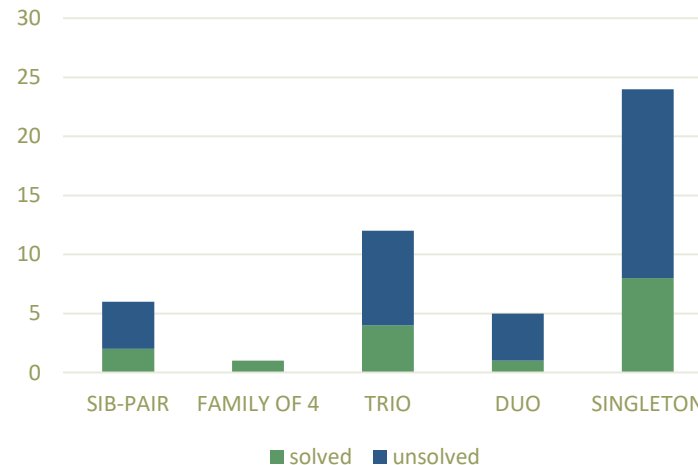
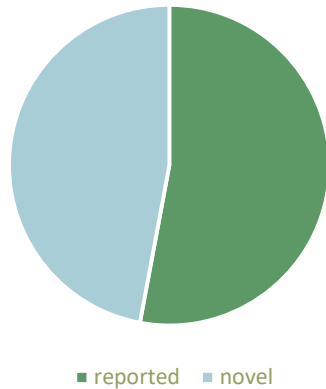
■ SIB-PAIR ■ FAMILY OF 4 ■ TRIO ■ DUO ■ SINGLETON



Detection rate

20

- Data analysed on the RD-Connect genomic platform
- 16 families solved so far
- 9 novel variants identified



■ homozygous ■ heterozygous ■ compound heterozygous ■ de novo

Project	Phenotype	PI	Total N	Status
NeurOmics	NMD/NDD	Multiple	?1000	680 WES available; 65 WGS available pending HW upgrade; further upload by deCODE required
EURenOmics	Kidney	Multiple	?1000	453 panel/26 WES received; processing pending
Neptune	Kidney	Sampson	450	Pending receipt
CNAG RD	Multiple RD	Multiple	300	Pending consent
Neurogenetics	Neurogenetic/ mitochondrial	Horvath: Newcastle	336 WES	Received; processing pending
SeqNMD	NMD	Newcastle	169 WES	122 available; 47 failed – reprocessing pending
MYO-SEQ	LGMD	Straub: Newcastle	1000 WES (all index cases)	27 solved cases available; additional solved pending; unsolved pending PI consent
Titinopathies	NMD/ titinopathy	Udd/Hackman/ Savarese	(I) 76 panel / 15 WES / 12 WGS (II) 2000 WES/WGS	(I) Received; processing underway (II) To be submitted
Rare Immuno	Rare immunodeficiencies	Hambleton: Newcastle	160 WES	Received 38; processing pending
BBMRI-LPC	17 projects / multiple RD	Multiple	900 WES	About half are now complete
Consequitur	Consanguineous neurogenetic	Lochmüller: Newcastle	500 WES	New project; sequencing pending
SERBORDISinn	Multiple RD	Pavlovic: Belgrade	9 WES	Available
Sayer Group	Kidney	Sayer: Newcastle	30 WES	Patient consent pending
ISCIII	Multiple RD	Posada: Madrid	23 WES	Available
Italian NMD	NMD	Nigro	tbc	Negotiation underway
Telethon UDP	Syndromic undiagnosed	Multiple	tbc	Negotiation underway



RD-Connect : Data sharing

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RD-Connect enables:

- Full data sharing and analysis within RD-Connect for authorized users
- Partial data sharing outside RD-Connect, in accordance with ethical and legal limitations



Search across samples (per gene/s) with all filters

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- Find all cases with a candidate mutation in a gene of interest

Filters

PRESET FILTERS

RESET

SHARE

RUN QUERY

Sample selection: special-gene-Z-query-all-samples

Sample Selection ?

Select individual Samples

or search across all ? (accessible: 1451, own: 0, shared: 174, visible to all: 1277)

? Compound het.

Affected	ID	0/0	0/1	1/1	Min DP	Min GQ	Min AAF	Max AAF
----------	----	-----	-----	-----	--------	--------	---------	---------

ALL_SAMPLES

20

50

0.2

0.8



RD-Connect : Data sharing

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RD-Connect enables:

- Full data sharing and analysis within RD-Connect for authorized users
- Partial data sharing outside RD-Connect, in accordance with ethical and legal limitations



Data Sharing: GA4GH Beacon

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Posted: May 29, 2015

Beacon Project

Being implemented on the website of the world's top genomic organizations to test the willingness of international sites to share genetic data.



About this Project

The **Beacon project** is a project to test the willingness of international sites to share genetic data in the simplest of all technical contexts. It is defined as a simple public web service that any institution can implement as a service. The service is designed merely to accept a query of the form "Do you have any genomes with an 'A' at position 100,735 on chromosome 3" (or similar data) and responds with one of "Yes" or "No." A site offering this service is called a "beacon". This open web service is designed to be technically simple, easy to implement, and to not return privacy violating information.

For current Beacons, and a short guide about how to light a Beacon, please visit:

[Beacon Network »](#)

Question:

Have you seen this **variant** in any sample in your database?

Answer:

Yes / No

<https://genomicsandhealth.org/work-products-demonstration-projects>





MatchMaker Exchange in RD-Connect (in pre-production)

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RD-Connect

GENOMICS

ABOUTWELCOME TEST{PLATFORM V0.8.0, DATASET 20170315_ELASTIC2}

Filters ^

PRESET FILTERSRESETSHARE▶ RUN QUERY

Sample Selection ?

Select individual Samples + or search across all ? (accessible: 211, own: 1, shared: 0, v ? Compound het.

AffectedID0/00/1

☐ E000001 NA12123

☒ E000040 MUN0788

Variant Type ?

Population ?

SNV Effect Prediction?

Genes, Disorders and Phenotypes

Chromosome Coordinates

SamplesFunctionalPredictivePopulationDiseasecardCandidate

RD-Connect IDParticipa

E000001P000034

E000040P000012

MME patient matching response (v1)

Patientrdconnect-annual

target endpointRD-Connect -> PhenomeCentral

Gene(s).(recovered from RD-Connect), Complete if necessarySDHA, CYC1, UQCR10, UQCRH

HPO term(s).(recovered from RD-Connect), Complete if necessaryHP0001508 Failure to thrive; HP0002151 Increased serum lactate; HP0002878 Respiratory failure

Matches in partner MME databases

Case ID	Contact	Relevance
P0001609	Phenome Central Initiative	41%
P0002596	Phenome Central Initiative	38%
P0001769	Phenome Central Initiative	34%
P0001866	Contact owner for further information	30%
P0003585	Phenome Central Initiative	30%
P0001505	Phenome Central Initiative	28%
P0000220	Phenome Central Initiative	28%
P0002516	Phenome Central Initiative	27%

CANCELACCEPT



Ethical and Legal Issues

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- 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.
- Activity of the users is logged.



platform.rd-connect.eu

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RD Connect

Home

Genomics

PhenoTips

ID-Cards

Biosample

Contact

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 genomics analysis interface is now open to 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

Contributors

cnag

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I. Gut



WP1: Coordination

Hanns Lochmüller

(Newcastle and TREAT-NMD)

WP2: Patient registries

Domenica Taruscio (ISS and EPIRARE)

WP3: Biobanks

Lucia Monaco

(Fondaz. Telethon & EuroBioBank)

WP4: Bioinformatics

Christophe Bérout

(INSERM Marseille)

WP5: Unified platform

Ivo Gut (CNAG Barcelona)

WP6 Ethical/legal/social

Mats Hansson (Uppsala)

WP7: Impact/Innovation

Kate Bushby

(Newcastle and EUCERD/ EJARD)



WP2: Benchmarking

Soren Brunak & Alfonso Valencia

WP5: Elixir Interoperability Backbone

Barend Mons, Carole Goble, Helen Parkinson

WP8: Rare Diseases Use Case

Ivo Gut & Marco Roos

WP9: Human Data Use Case

Jordi Rambla & Helen Parkinson

WP11: Training Platform

Chris Ponting & Patricia Palagi

