

UPDATE NEWLY SOLVED CASES IN RD-CONNECT GPAP

There are two possibilities according to the number of cases to update:

1. [Manually through PhenoTips](#)
2. [Bulk-upload submitting an excel file to coordination](#)

1. Update record manually

The manual update requires 2 steps:

- [Update the case resolution status and diagnostic in PhenoTips](#)
- [Enter the causative variant information in the GPAP](#)

1. Adding Diagnosis, Gene and case resolution in PhenoTips

1.1 Access PhenoTips at <https://platform.rd-connect.eu/phenotips/>

1.2 Select the Case you want to edit.



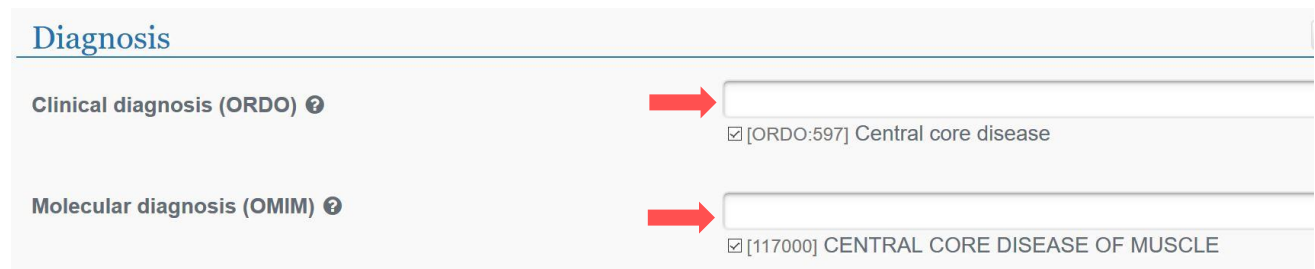
P0018701

Reported by Carles Garcia on 2020/02/26 13:27 · Last modified on 2020/03/20 14:57


Study: None


This case is owned by you and is private

1.3. In the “Diagnosis” field, enter the corresponding **ORDO** and **OMIM**.



Diagnosis

Clinical diagnosis (ORDO) 

Molecular diagnosis (OMIM) 

[ORDO:597] Central core disease

[117000] CENTRAL CORE DISEASE OF MUSCLE

1.4. In the “Genotype information” field push the “Add Gene” button and **add all the necessary information**.

Genotype information

LIST OF GENES

GENE	STATUS	STRATEGY	COMMENTS
<input type="text" value="NF1"/>	Confirmed causal	<input checked="" type="checkbox"/> Sequencing <input type="checkbox"/> Deletion/duplication <input type="checkbox"/> Familial mutation/disease <input type="checkbox"/> Common mutations	

1.5. In the “Case resolution” field, state the case has been solved by **ticking in the “Case solved” box**.

Case resolution

Case solved: ?

2. Adding the causal variant information in RD-Connect GPAP or Phenotips

If the variant is a SNV or Indel it should be added in RD-Connect GPAP (TAG). Otherwise, the variant information should be added in Phenotips.

A) Adding the variant in RD-Connect GPAP (only if mutation is SNV or Indel)

2.1 Access RD-Connect GPAP and search for your variant by applying the necessary filters.

2.2 Once obtained, click in the “TAG” button.

Filters PRESET FILTERS RESET SHARE RUN QUERY

Variant Type: high Population: gnomad_af

Samples	Functional	Predictive	Population	Pathways	Protein interaction	Dise
RD-Connect ID	Participant ID		GT			
E000001	DNC0001		0/1			

Phenotype Analysis status Variants (875) Samples () Exomiser

First Previous **1** 2 3 4 5 ... Next Last

EXPORT ALL

Chr	Position	dbSNP	Ref	Alt	Candidate	GT ^{E000001}	INDEL	Ger
1	135804	.	G	A	<input checked="" type="checkbox"/> TAG	G/A		AL62

2.3 Then, fill in the form with all the relevant information for the variant.

Identify Variant as Causal/of Interest

User Name

Date: 23 / 03 / 2020

Sample: E000001

Gene: AL627309.1

Mode of Inheritance (required): autosomal dominant

Origin: de novo

Clinical Significance (required): pathogenic

CANCEL **SUBMIT**

B) Adding the variant in Phenotips (if mutation is not SNV or Indel)

2.1 Access Phenotips at <https://platform.rd-connect.eu/phenotips/>

2.2 Select the Case you want to edit.


P0018701
Reported by **Carles Garcia** on 2020/02/26 13:27 · Last modified on 2020/03/20 14:57  Edit  Del



Study: None  This case is owned by **you** and is **private** . It

2.3 In the “Genotype information” field it will appear the gene previously introduced. Push the “Add variant” button.

Genotype information

LIST OF GENES

GENE	STATUS	STRATEGY	COMMENTS
1 NF1 	Confirmed causal	<input checked="" type="checkbox"/> Sequencing <input type="checkbox"/> Deletion/duplication <input type="checkbox"/> Familial mutation/disease <input type="checkbox"/> Common mutations	

 **+ ADD VARIANT** 

2.4 In the form appearing you can **introduce all the relevant information** for the variant.

Variants in (1) HIDE VARIANTS

	cDNA	INTERPRETATION	ZYGOSITY
1.1	c. 4237C>T	Pathogenic	heterozygous
Reference genome:	GRCh38 (hg38)		
Chromosome:			
Start position:			
End position:			
Protein:	p.Gly438Pro		
Transcript:	NM_000000.0		
dbSNP:	rs2837824		
Effect:			

2. Bulk-upload submitting an excel file to coordination

2.1 Fill in the excel file with the requested information. The document is accessible through the RD-Connect Website at:

https://rd-connect.eu/wp-content/uploads/2020/07/template_solved_cases_to_submit_v1.xlsx

Causative variant information: enter requested variant annotation OR TAG variant in the GPAP and paste the corresponding link														
ERN	RD-Connect experiment ID*	PhenoTips ID	gene*	chromosome*	START_genomic coordinate*	END_genomic coordinate	genome version	REF	ALT	zygosity*	disease inheritance*	clinical significance* (according to ACMG guidelines)	OR TAG the variant in GPAP and paste the corresponding link	OMIM associated* (if existing)

There are 9 mandatory fields: Experiment ID, Gene(s), Chromosome, START position, Zygosity, Clinical significance of the variant and OMIM.

Fields in blue are not mandatory if the variant has been TAG in the RD-Connect GPAP.

IMPORTANT: Please use VCF annotation format, for example start/stop/ref/alt :

notation used by VCF

1 100 TA T

2.2 Please send the file to platform@rd-connect.eu.