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:

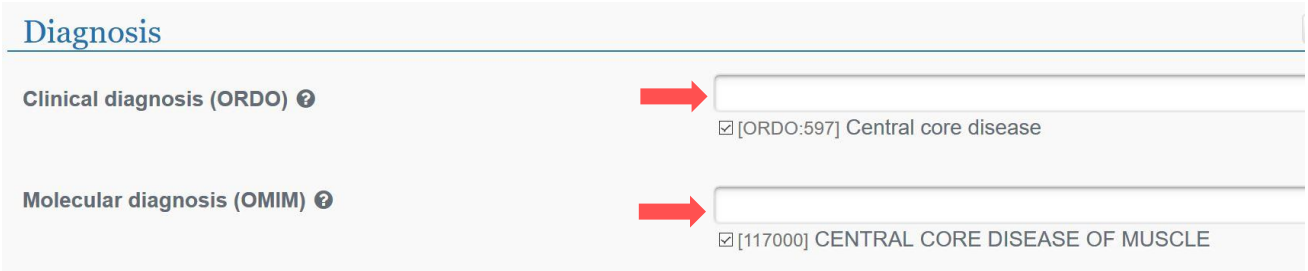
- Update the case resolution status and diagnostic in PhenoTips
- Enter the causative variant information in the GPAP

1. Adding Diagnosis, Gene and case resolution in PhenoTips

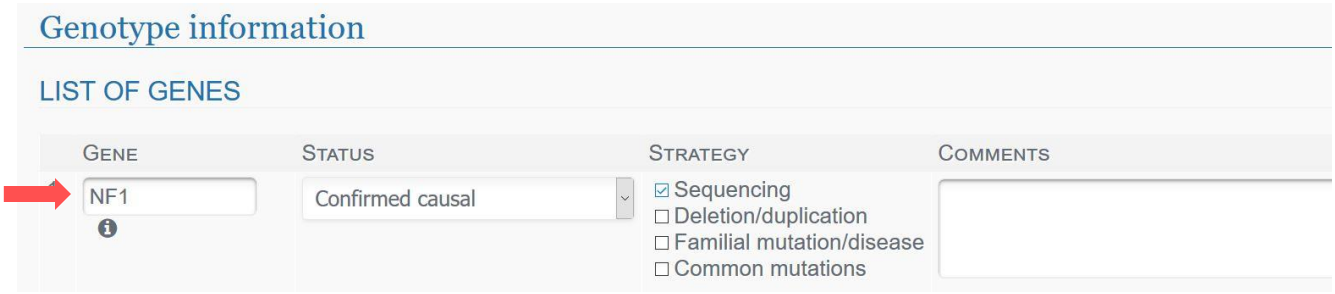
1.1 Access Phenotips at https://platform.rd-connect.eu/phenotips/

1.2 Select the Case you want to edit.

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



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



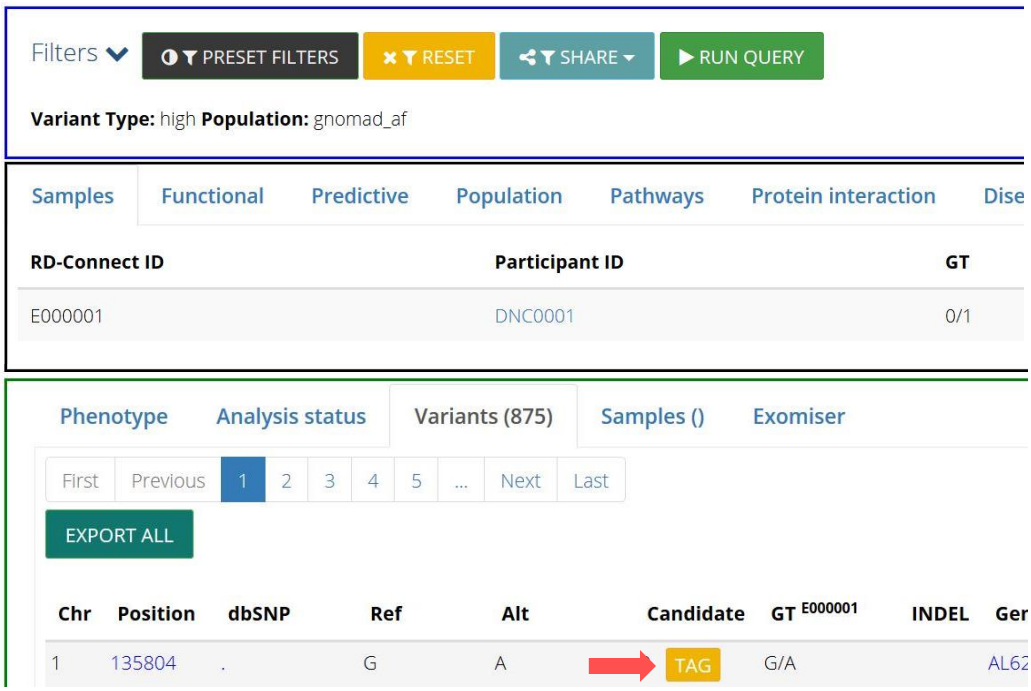
1.5. In the "Case resolution" field, state the case has been solved by **ticking in the "Case solved" box**.

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

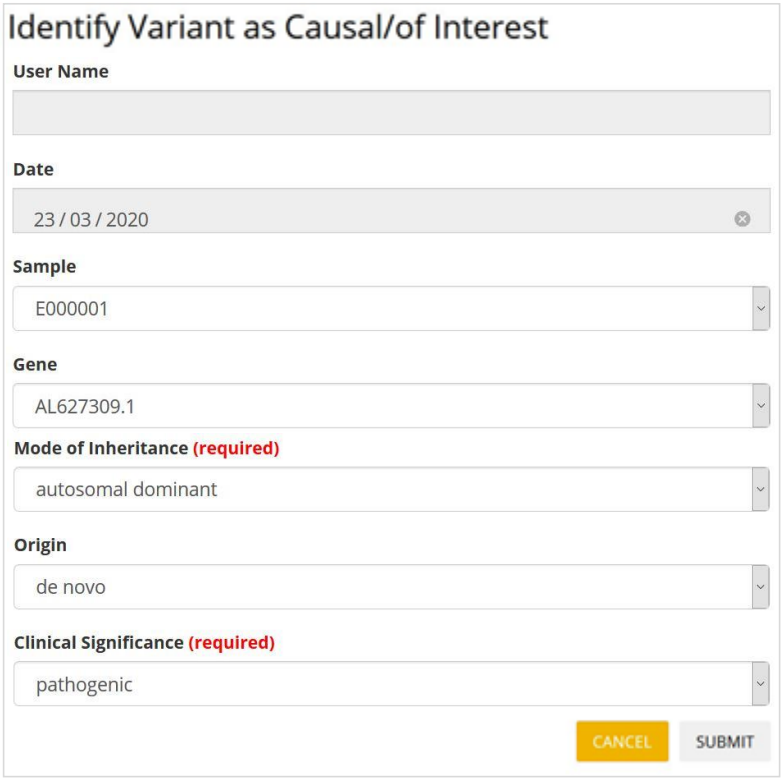
If the variant is a SNV or Indel it should be added in RD-Connect GPAP (TAG). Otherwise, the variant information should be added in Phenotips.

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.



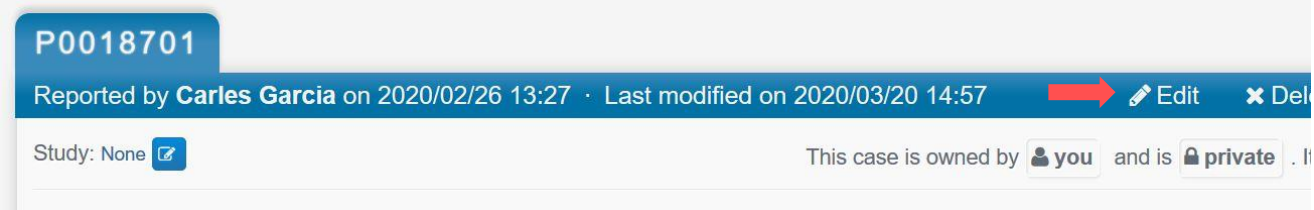
2.3 Then, **fill in the form with all the relevant information** for the variant.



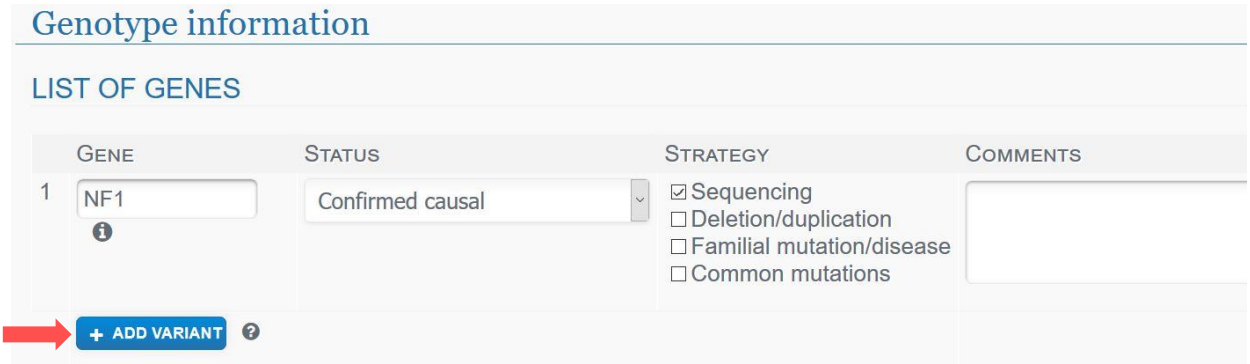
B) Adding the variant in Phenotips (if mutation is not SNV or Indel)

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

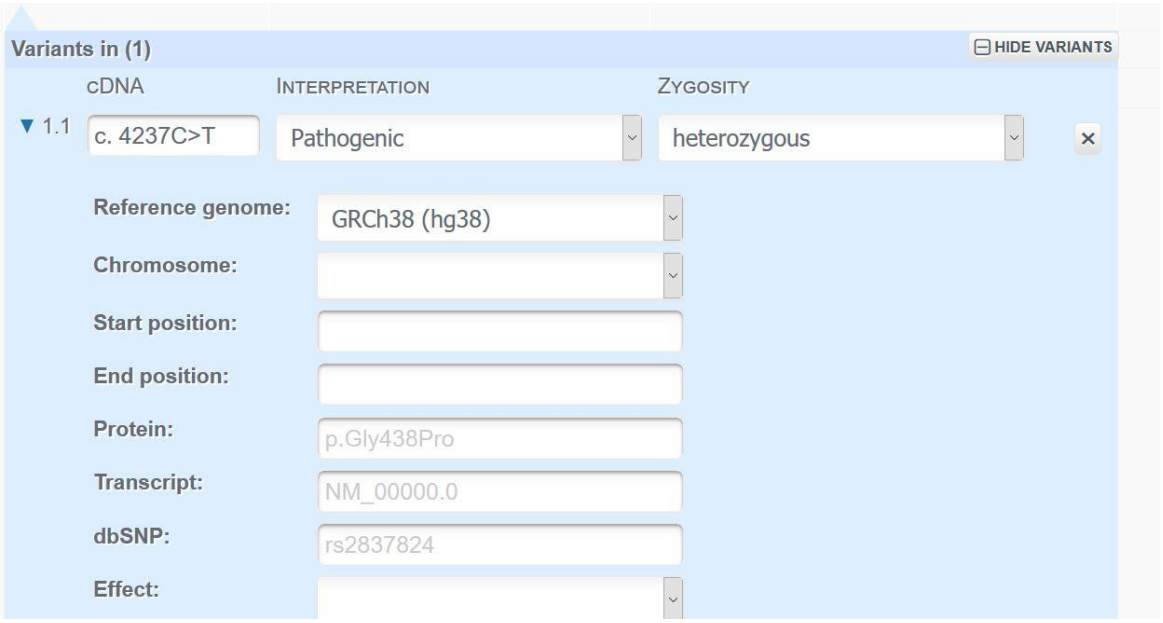
2.2 Select the Case you want to edit.



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



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



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.