

# UPDATE NEWLY SOLVED CASES IN RD-CONNECT GPAP

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

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

## 1. Update record manually

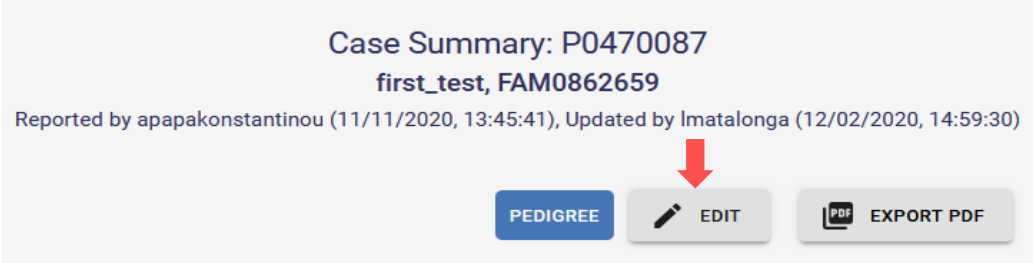
The manual update requires 2 steps:

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

### 1. Adding Diagnosis, Gene and case resolution in PhenoStore

1.1 Access PhenoStore at <https://platform.rd-connect.eu/phenostore/>

1.2 Go to “browse cases” and select the case you would like to edit.



Case Summary: P0470087  
first\_test, FAM0862659  
Reported by apapakonstantinou (11/11/2020, 13:45:41), Updated by Imatalonga (12/02/2020, 14:59:30)

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1.3. In the “Individual information” field, state the case has been solved by **ticking in the “Case solved” box.**

#### Solved status\*

Solved  Unsolved

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

· Relevant results:

Gene (HGNC)

DMD

Enter a gene symbol using HGNC nomenclature e.g. FMR1

Status

Causative

Genetic approach

WGS

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

## ➤ Diagnosis

### · Clinical diagnosis

#### Search ORDO

Orphanet:262 Duchenne and Becker muscular dystrophy

Diagnosis status

Confirmed

+ Add ORDO

### · Molecular diagnosis

#### Search OMIM

OMIM:310200 Muscular Dystrophy, Duchenne Type

Diagnosis status

Confirmed

+ Add OMIM

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

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 PhenoStore.

### 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.

The screenshot displays the RD-Connect GPAP search interface. At the top, there are filter controls: 'Filters' (dropdown), 'PRESET FILTERS' (toggle), 'RESET' (button), 'SHARE' (button), and 'RUN QUERY' (button). Below this, the search criteria are shown: 'Variant Type: high' and 'Population: gnomad\_af'. The main table has several tabs: 'Samples', 'Functional', 'Predictive', 'Population', 'Pathways', 'Protein interaction', and 'Diseases'. The 'Samples' tab is active, showing a table with columns 'RD-Connect ID', 'Participant ID', and 'GT'. The data row shows 'E000001', 'DNC0001', and '0/1'. Below this, there are more tabs: 'Phenotype', 'Analysis status', 'Variants (875)', 'Samples ()', and 'Exomiser'. The 'Variants (875)' tab is active, showing a pagination bar (1-5, Next, Last) and an 'EXPORT ALL' button. The variant table has columns: 'Chr', 'Position', 'dbSNP', 'Ref', 'Alt', 'Candidate', 'GT', 'INDEL', and 'Ger'. The first row shows '1', '135804', '.', 'G', 'A', a red arrow pointing to a 'TAG' button, 'G/A', and 'AL62'.

RD-Connect ID	Participant ID	GT
E000001	DNC0001	0/1

Chr	Position	dbSNP	Ref	Alt	Candidate	GT	INDEL	Ger
1	135804	.	G	A	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**

**Sample**

**Gene**

**Mode of Inheritance (required)**

**Origin**

**Clinical Significance (required)**

**CANCEL** **SUBMIT**

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

2.1 Access PhenoStore at <https://platform.rd-connect.eu/phenostore/>

2.2 Go to “browse cases” and select the case you would like to edit.

**Case Summary: P0470087**  
**first\_test, FAM0862659**

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2.3 In the “Genetic testing” field the gene previously introduced appears. Push the “Add variant” button.



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](mailto:platform@rd-connect.eu).