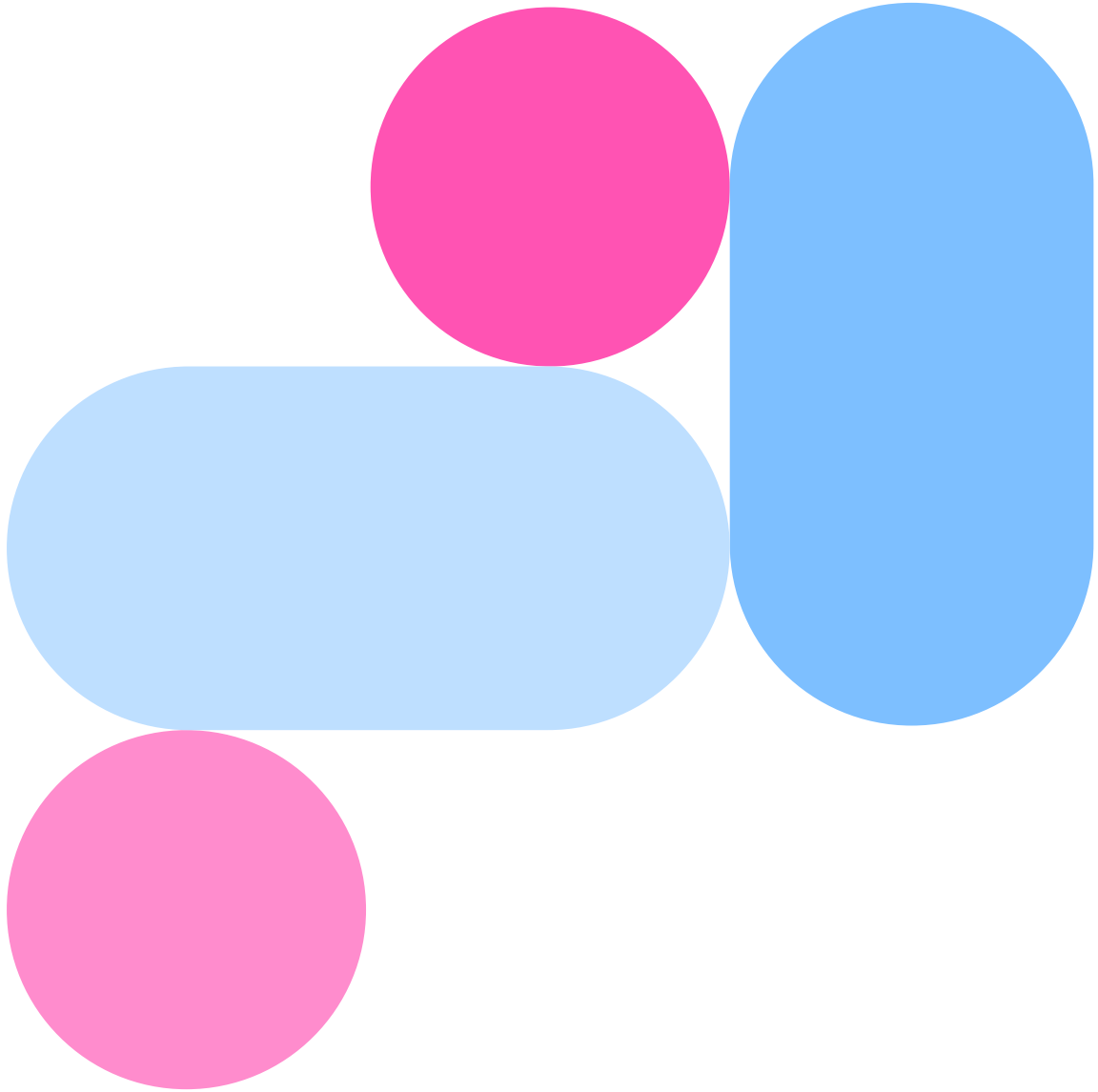
Genomics England

Auto-populating ROQs

Mapping the questions between Congenica and the Interpretation Portal

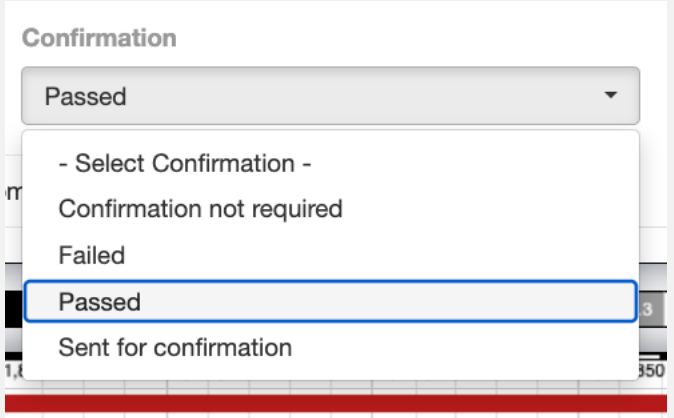


Question 1. Did the test confirm that the variant is present?

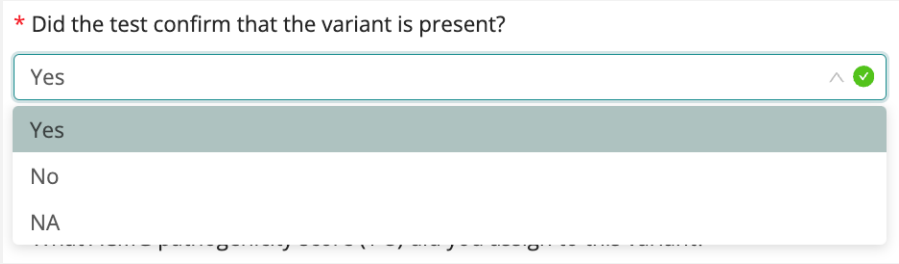
Here the mapping:

Response in Congenica	Response in the ROQs
Confirmation not required	NA
Failed	No
Passed	Yes
Sent for confirmation	NA

Question in Congenica



Question in the ROQs



Additional Question. Did you carry out technical confirmation of this variant via an alternative test?

If the response to Question 1 is YES or NO, then the following question:

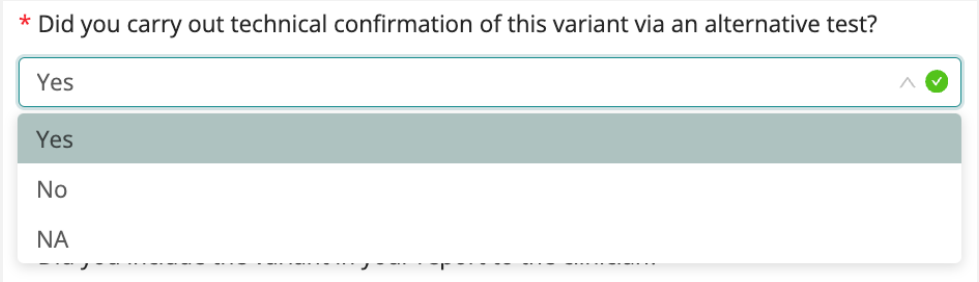
Did you carry out technical confirmation of this variant via an alternative test?

will be auto-populated to Yes

If the response to Question 1 is NA, then the following question:

will be auto-populated to Na

Question in the ROQs

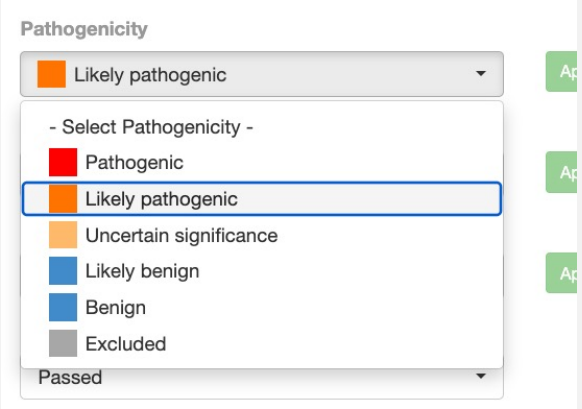


Question 2. What ACMG pathogenicity score (1-5) did you assign to this variant?

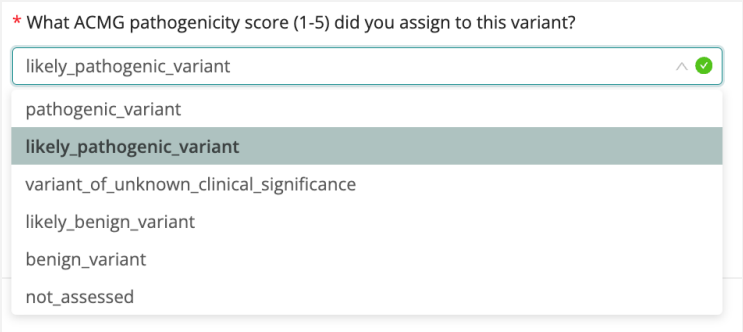
Here the mapping:

Response in Congenica	Response in the ROQs
Pathogenic	pathogenic_variant
Likely Pathogenic	likely_pathogenic_variant
Uncertain significance	variant_of_uknown_clinical_significance
Likely benign	likely_benign_variant
Benign	benign_variant
Excluded	not_assessed

Question in Congenica



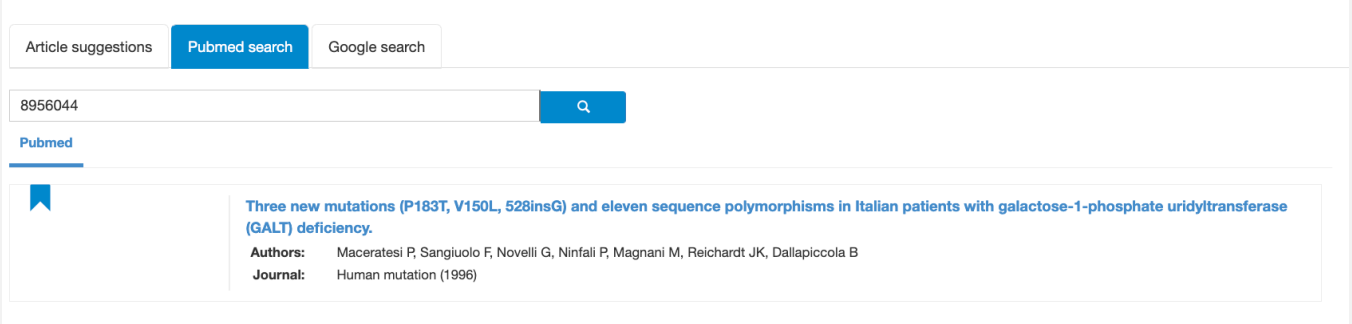
Question in the ROQs



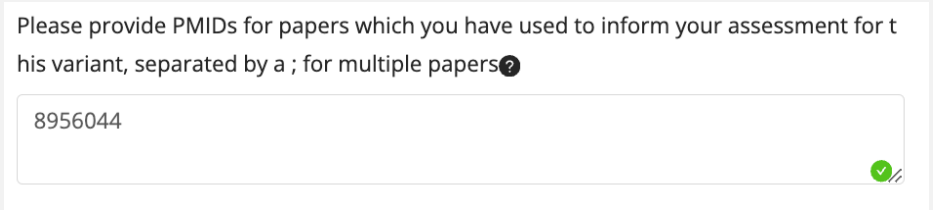
Question 3. Please provide PMIDs for papers which you have used to inform your assessment for this variant, separated by a ; for multiple papers

The PMID for all selected papers in Congenica will now automatically appear in the ROQs.

Question in Congenica



Question in the ROQs



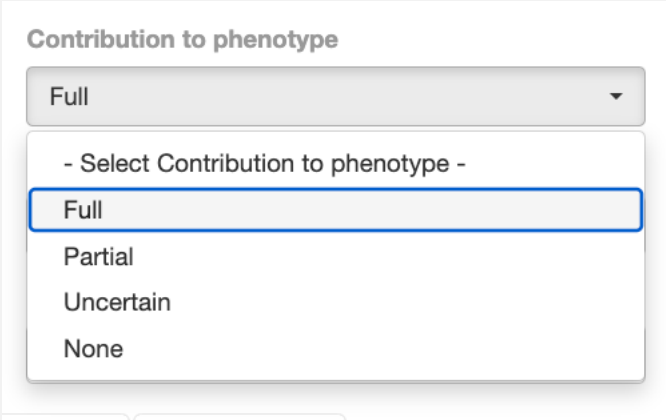
The picture can't

Question 4. Did you report the variant(s) as being partially or completely causative of the family's presenting phenotype(s)?

Here the mapping:

Response in Congenica	Response in the ROQs
Full	yes
Partial	partially
Uncertain	unknown
None	no

Question in Congenica



Question in the ROQs