

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

A screenshot of a dropdown menu titled "Confirmation". The menu is open, showing several options: "- Select Confirmation -", "Confirmation not required", "Failed", "Passed", and "Sent for confirmation". The "Passed" option is highlighted with a blue border. The dropdown is set against a background of a grid.

Question in the ROQs

A screenshot of a question in the ROQs interface. The question is: "* Did the test confirm that the variant is present?". Below the question is a list of possible answers: "Yes", "No", and "NA". The "Yes" option is selected and highlighted in a dark grey bar. A green checkmark icon is visible in the top right corner of the answer selection area.

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

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

Yes



Yes

No

NA

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_unknown_clinical_significance
Likely benign	likely_benign_variant
Benign	benign_variant
Excluded	not_assessed

Question in Congenica

The screenshot shows a dropdown menu titled "Pathogenicity". The selected option is "Likely pathogenic" (orange square). The menu is open, showing a list of options: "Pathogenic" (red square), "Likely pathogenic" (orange square, highlighted with a blue border), "Uncertain significance" (light orange square), "Likely benign" (blue square), "Benign" (dark blue square), and "Excluded" (grey square). Below the list is a "Passed" status indicator.

Question in the ROQs

The screenshot shows a question in the ROQs: "* What ACMG pathogenicity score (1-5) did you assign to this variant?". The dropdown menu is open, showing a list of options: "likely_pathogenic_variant" (highlighted with a green bar), "pathogenic_variant", "variant_of_unknown_clinical_significance", "likely_benign_variant", "benign_variant", and "not_assessed". A green checkmark is visible in the top right corner of the dropdown menu.

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

Article suggestions **Pubmed search** Google search

8956044

Pubmed

 **Three new mutations (P183T, V150L, 528insG) and eleven sequence polymorphisms in Italian patients with galactose-1-phosphate uridytransferase (GALT) deficiency.**

Authors: Maceratesi P, Sangiuolo F, Novelli G, Ninfali P, Magnani M, Reichardt JK, Dallapiccola B
Journal: Human mutation (1996)

Question in the ROQs

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

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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

Contribution to phenotype

Full

- Select Contribution to phenotype -

Full

Partial

Uncertain

None

Question in the ROQs

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

Yes

Yes

No

Partially

Unknown