COHORT TOOL
IN RD-CONNECT GPAP

In RD-Connect GPAP you can create a specific cohort of participants based on phenotypic information, and you can use this cohort for downstream genomic analyses in the platform.

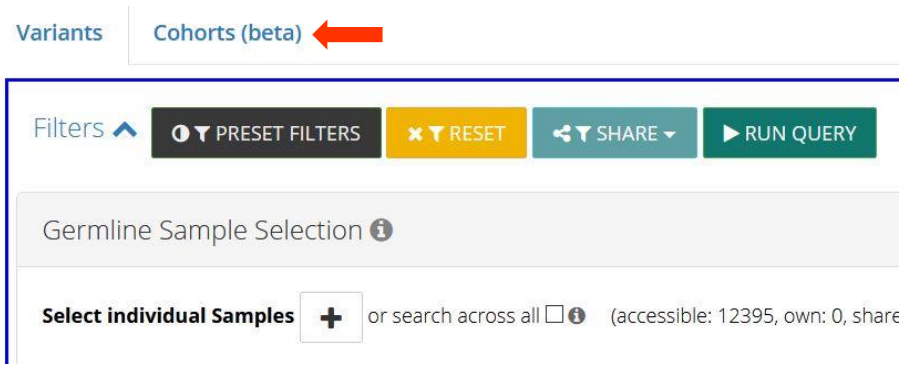
This approach will also enable you to contact submitters of patients of interest using the "Contact" button functionality (instructions at the end of this guide).

Using the "Cohort" tool

1. Creating a cohort

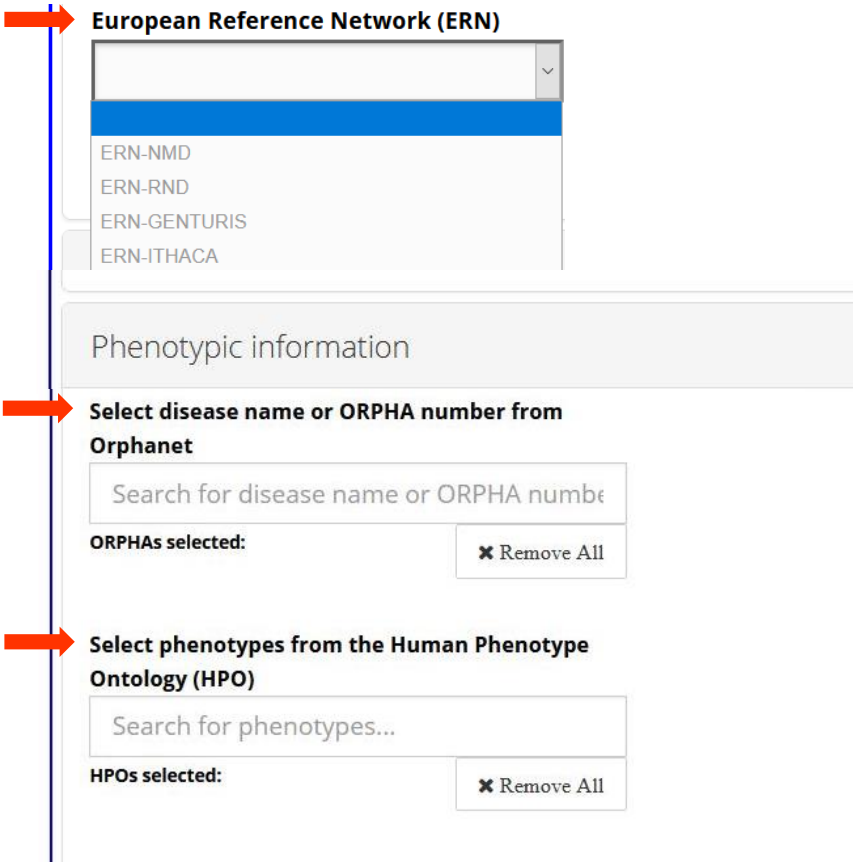
1.1 Access the RD-Connect GPAP (https://platform.rd-connect.eu/genomics/).

1.2 Enter the "Cohorts" tab.



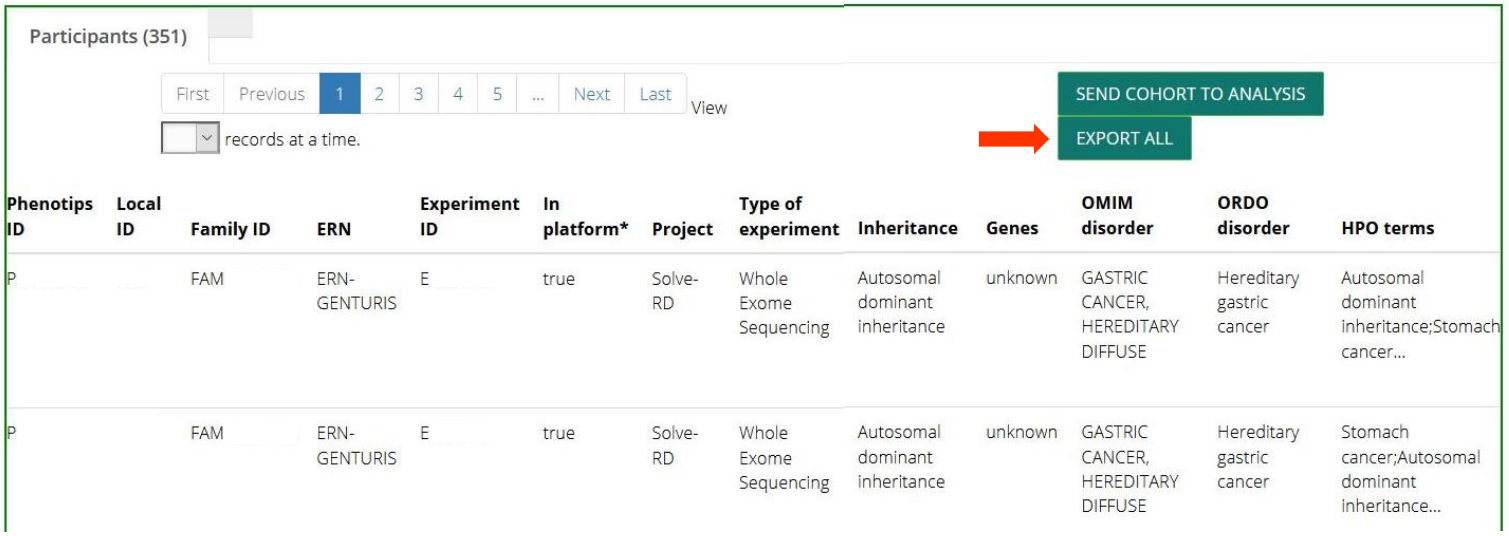
1.3 Select the filters for your cohort.

- You can select All GPAP data or only your experiments.
- You can filter by European Reference Network (ERN).
- You can filter by clinical disorders (ORDO ontology from Orphanet) or symptoms (Human Phenotype Ontology – HPO).



1.4 "Run query" to obtain the cohort and scroll to the bottom of the page.

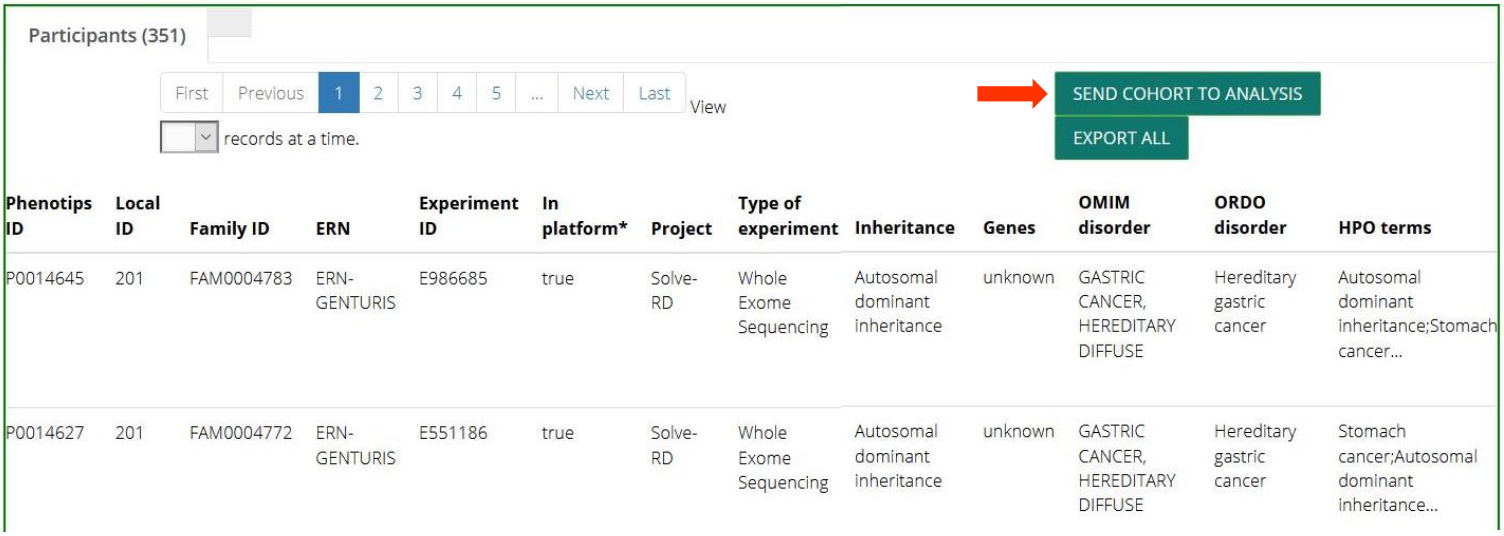
- You will see the results in a table.
- You can export all data by using the "Export all" button.



Phenotips ID	Local ID	Family ID	ERN	Experiment ID	In platform*	Project	Type of experiment	Inheritance	Genes	OMIM disorder	ORDO disorder	HPO terms
P		FAM	ERN-GENTURIS	E	true	Solve-RD	Whole Exome Sequencing	Autosomal dominant inheritance	unknown	GASTRIC CANCER, HEREDITARY DIFFUSE	Hereditary gastric cancer	Autosomal dominant inheritance;Stomach cancer...
P		FAM	ERN-GENTURIS	E	true	Solve-RD	Whole Exome Sequencing	Autosomal dominant inheritance	unknown	GASTRIC CANCER, HEREDITARY DIFFUSE	Hereditary gastric cancer	Stomach cancer;Autosomal dominant inheritance...

2. Using the Cohort as a query in GPAP

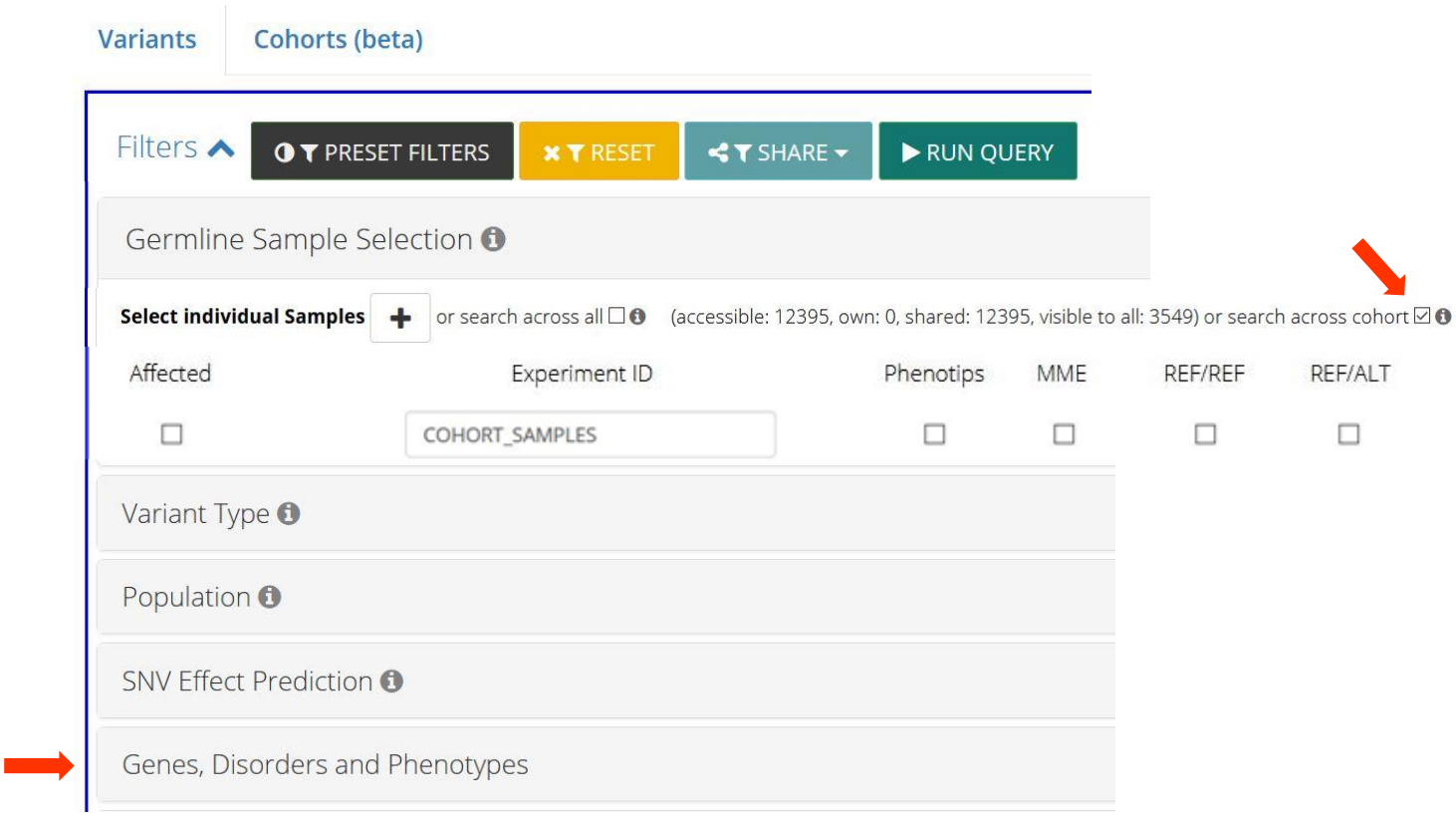
2.1 Push the "Send cohort to analysis" button to send the query to the "Variant" tab.



Phenotips ID	Local ID	Family ID	ERN	Experiment ID	In platform*	Project	Type of experiment	Inheritance	Genes	OMIM disorder	ORDO disorder	HPO terms
P0014645	201	FAM0004783	ERN-GENTURIS	E986685	true	Solve-RD	Whole Exome Sequencing	Autosomal dominant inheritance	unknown	GASTRIC CANCER, HEREDITARY DIFFUSE	Hereditary gastric cancer	Autosomal dominant inheritance;Stomach cancer...
P0014627	201	FAM0004772	ERN-GENTURIS	E551186	true	Solve-RD	Whole Exome Sequencing	Autosomal dominant inheritance	unknown	GASTRIC CANCER, HEREDITARY DIFFUSE	Hereditary gastric cancer	Stomach cancer;Autosomal dominant inheritance...

2.2 The cohort is now available to query by adding filters like in any other search.

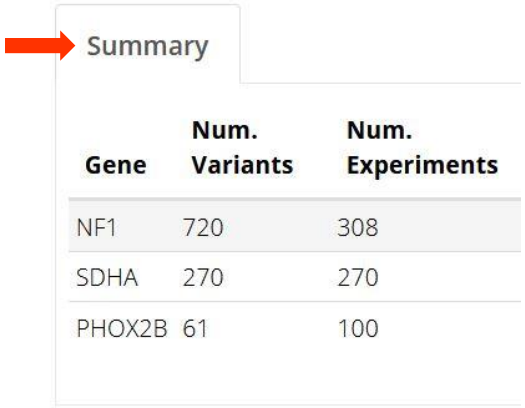
- It is **mandatory to add at least one gene** as a filter in the "Gene, disorders and Phenotypes section".
- You can reload the cohort by clicking on the "search across cohort" box.
- "Run query" to obtain the results.



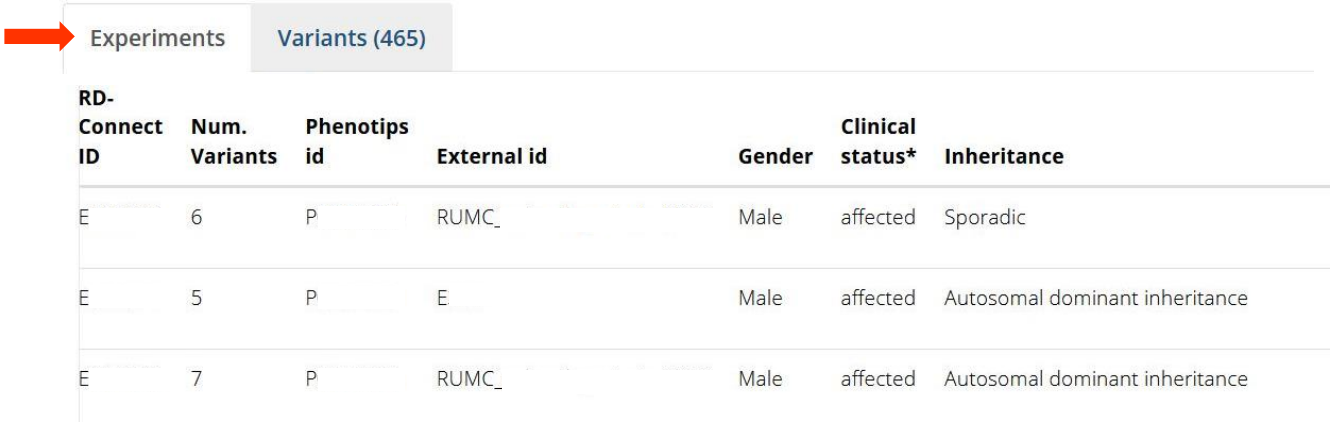
3. Results

3.1 The results area will show different sections:

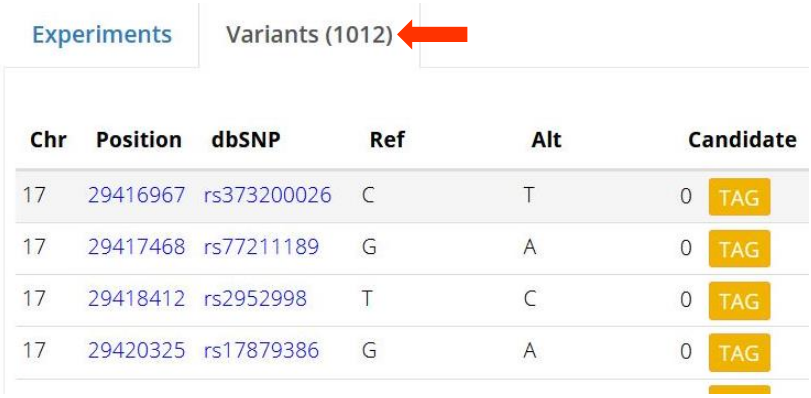
- "Summary" tab: Genes containing the variants.



- "Experiments" tab: Information of participants bearing the variants.



- "Variants" tab: Individual variants found according to the filters used.



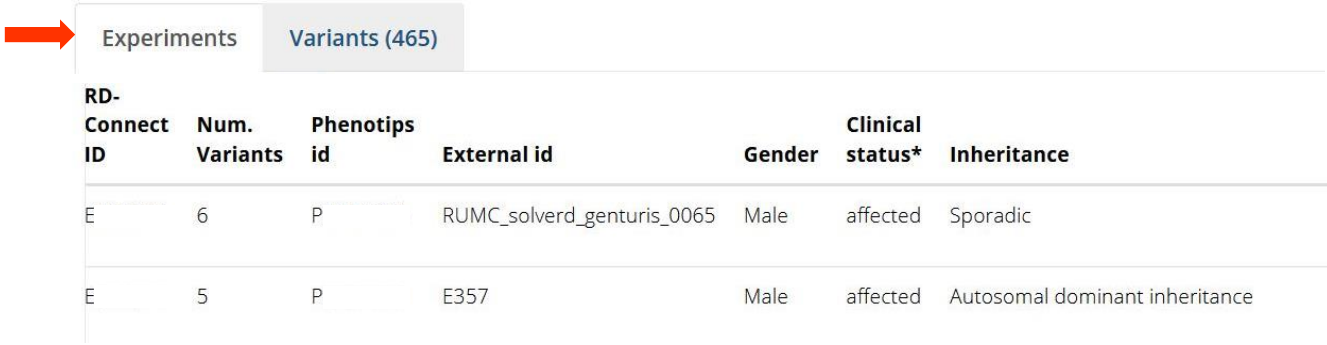
- Data related to the variants: In the middle-page area you will find a section composed by different tabs, each one containing specific information related to the selected variant.

Gene Name	Transcript ID	Effect Impact	Consequence	Feature Type	HGVS coding	Human Splicing Finder	Amino Acid change
SDHA	ENST00000264932	MODIFIER	upstream_gene_variant	transcript			664

Using the "Contact" button

If you find an interesting participant, you can use the "Contact" button to interact with its submitter.

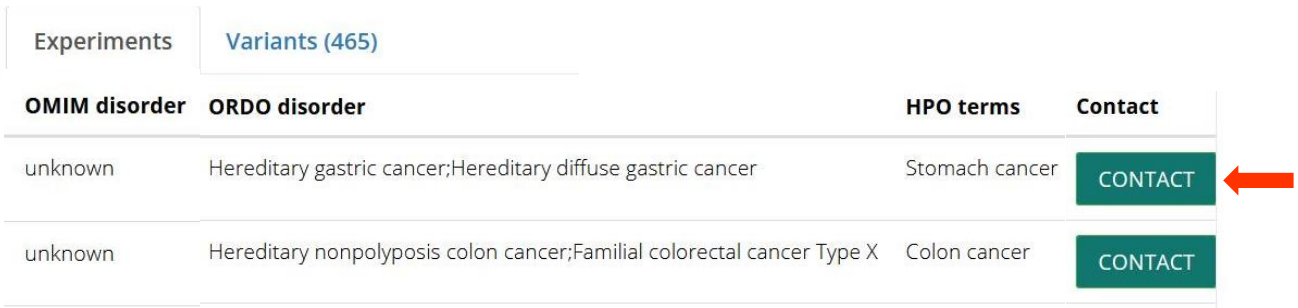
1. Access the "Experiments" tab after performing the "Search across cohort" query.



RD-Connect ID	Num. Variants	Phenotips id	External id	Gender	Clinical status*	Inheritance
E	6	P	RUMC_solverd_genturis_0065	Male	affected	Sporadic
E	5	P	E357	Male	affected	Autosomal dominant inheritance

2. In this tab move to the right side of the table, where you will see the "Contact" button.

- After pushing the "Contact" button an automatic email will be sent to both you and the participant's submitter.
- The participant's submitter will also be able to contact you regarding this matching-query.



OMIM disorder	ORDO disorder	HPO terms	Contact
unknown	Hereditary gastric cancer;Hereditary diffuse gastric cancer	Stomach cancer	CONTACT
unknown	Hereditary nonpolyposis colon cancer;Familial colorectal cancer Type X	Colon cancer	CONTACT