MATCHMAKER EXCHANGE (MME)
IN RD-CONNECT GPAP

> Matchmaker exchange (MME) enables gene discovery through the anonymized exchange of genomic and phenotypic information (https://www.matchmakerexchange.org/).
>
> This discovery network is accessible through the RD-Connect GPAP.

Authorizing a participant to be searchable by MME

This authorization is provided at the time of submission.

1. Access the RD-Connect GPAP Data Management (https://platform.rd-connect.eu/datamanagement/).

2. Enter Step-1 "Submit Participant Set".

3. For each participant, click in the "MatchMaker Exchange" box to allow the participant to be searchable by MME.

Using MME in RD-Connect GPAP

1. Access the MME menu

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

1.2. In the Variant tab select your experiment by pushing the "+" button.

1.3. If the corresponding participant has been authorized for MME, an MME icon will appear. Press to access the MME query menu.
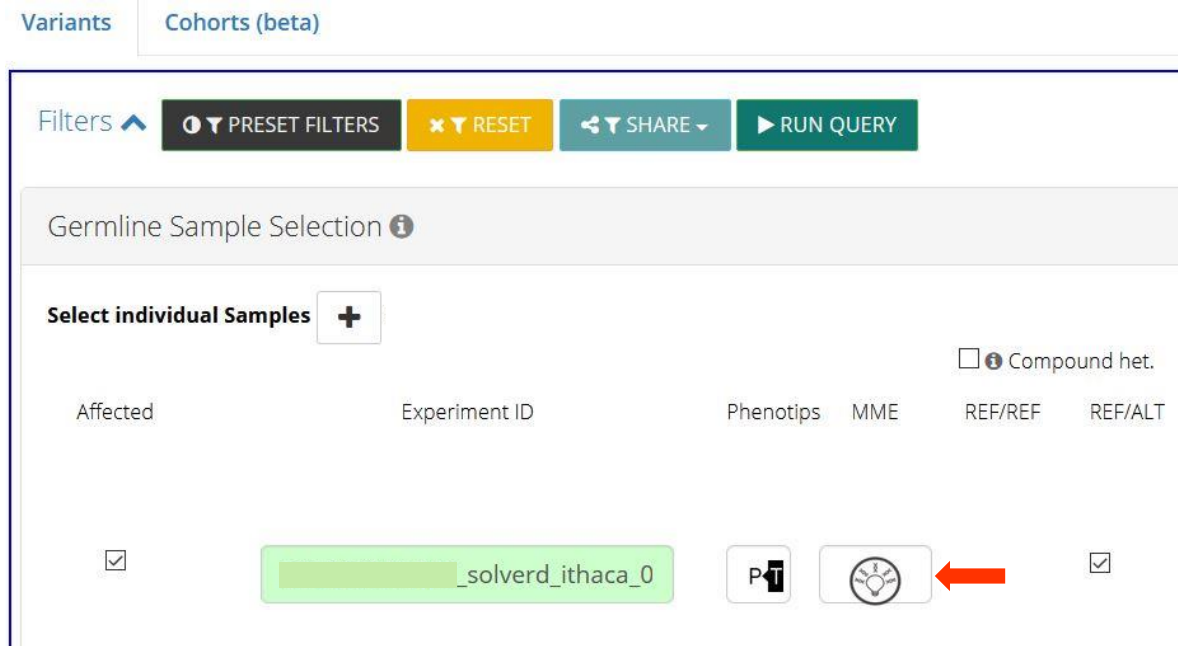
2. Submit a MME request from the query menu

2.1. A MME request form will appear. Several fields are automatically populated based on the information entered in the system; these fields are also editable. In the MME query menu, you can also enter additional information and select the database you would like to query:

- Target Endpoint: You can do internal queries (RD-Connect GPAP) or query other available databases (currently PhenomeCentral and Decipher).
- Add gene(s): You can add specific genes to query.
- Add HPOs: You can add specific HPOs to query.

2.2 Then push "Submit" to obtain the results.

3. MME query results

3.1. The results will show a list of participants with a Score value:

- Score value can have a maximum value of 1, and it is based on:
  - Gene-match: if the gene matches, the score adds 0,5 value.
  - Phenotypic similarity: if there are similar HPOs, score value increases until a maximum of 0,5 value.

Contact	Patient	Score	Submitter	Phenotype	Genes
CONTACT		0.72	RD-Connect Matchmaker Exchange	Muscle fiber atrophy ,Abnormality of muscle morphology ,Chewing difficulties ,Abnormality of muscle fibers ,Episodic flaccid weakness ,Proximal muscle weakness ,Type 2 muscle fiber atrophy ,EMG: decre...	CHRND
CONTACT		0.70	RD-Connect Matchmaker Exchange	Fatigable weakness ,EMG: decremental response of compound muscle action potential to repetitive nerve stimulation ,Ptosis ...	CHRND
CONTACT		0.66	RD-Connect Matchmaker	Congenital bilateral ptosis ,EMG: decremental response of	CHRND

4. Using the "Contact" button

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

- 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.

Contact	Patient	Score	Submitter	Phenotype	Genes
CONTACT		0.72	RD-Connect Matchmaker Exchange	Muscle fiber atrophy ,Abnormality of muscle morphology ,Chewing difficulties	CHRND