



If you wish to participate please contact us!

antonio.marques-atalaia@inserm.fr

gisele.bonne@inserm.fr

Read more:
solve-rd.eu

Treatabolome Task Group:

Antonio Atalaia (INSERM, France)

Gisèle Bonne (INSERM, France)

Hanns Lochmuller (CHEO, Ottawa, Canada)

Leigh Carmody (Jackson Laboratory, USA)

Peter Robinson (Jackson Laboratory, USA)

Rachel Thompson (Newcastle University, UK)

Sergi Beltran (CRG-CNAG, Spain)

Steven Laurie (CRG-CNAG, Spain)

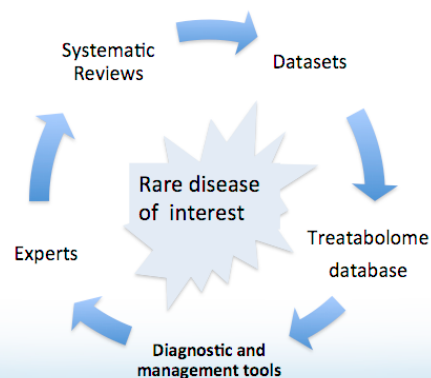
Solve  RD

Solving the unsolved Rare Diseases

TREATABOLOME

a rare diseases treatment awareness project

Solve-RD is an European project using H2020 funding working directly with four **European Reference Networks (ERNs)** - European networks of care providers - that were set up to share and enhance the knowledge and resources used for treating rare diseases.



Solve-RD WP3 aims at changing patient lives via the establishment of a "treatabolome" database flagging treatable genes and variants, which will contribute to the translation of genomics data into the clinical setting.

Experts lead on writing **systematic reviews** of existing gene and variant-specific treatments for rare diseases adopting the **FAIR Guiding Principles for scientific data management and stewardship** for **datasets** production. A **Treatabolome** database will be assembled from these for future inclusion in existing **diagnostic and management tools**, so that when a gene or variant diagnosis is reached the existing variant and gene specific treatments are flagged alongside the diagnosis. This will reinforce visibility of the growing number of therapies in place for rare diseases, therefore contributing for treatment awareness in the field.