REFERENCE LABORATORY
GENETICS

RefLab Database®

Detailed information on millions of gene variants validated by our experts

Disease related variants

Performing the correct clinical interpretation of a genetic variant can be a long and complex procedure. Misinterpretation or delayed diagnosis can have negative consequences on the quality of life of the patient and their relatives.

In order to facilitate the correct interpretation of genetic variants and to bring our extensive experience in clinical diagnosis closer to the scientific-medical community, we have developed RefLab Database®, our database of rare disease genotypes and phenotypes.

RefLab Database® correlates the clinical information of our patients, described using HPO terminology, with the detailed annotation of millions of genetic variants.

How does RefLab Database® work?

RefLab Database® provides a comprehensive overview of the clinical validity and causality of detected genetic variants with associated symptoms. In addition, it is a valuable tool for identifying new genes involved in pathogenic processes by correlating new genetic variants with specific and well-defined phenotypes.

The RefLab Database® software correlates the clinical information of our patients, described using HPO terminology, with the detailed annotation of millions of genetic variants, validated by our experts.

RefLab Database® is in continuous growth with:

- The clinical data and genetic results of samples analysed by Reference Laboratory.
- The main international genomic databases.
- The most recent medical and scientific bibliography.

Our database allows:

- Sharing of our >40 years of experience in clinical diagnosis.
- Reduction in the number of VOUS in genetic analysis.
- Facilitation of the annotation and interpretation of genetic variants.
- Acquisition of information from an international patient cohort.

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