Curation Working Group

The goal of the Curation Working Group is to develop gene-disease associations for GLA and GBA as well as standardize variant and case curation for individuals tested for Gaucher disease or Fabry disease. Members include medical experts, scientists, and curators with the expertise to create and sustain a consistent, accurate record of test results and clinical information.

The group meets monthly to review relevant user feedback and make decisions about any suggested changes to the curation process. Meetings are scheduled and minutes are recorded by the coordinator and maintained internally. Any decisions impacting GLA or GBA variants will be made available in the variant information of https://www.centogene.com/centolsd.html.

By defining the types and amount of evidence needed to support a gene-disease association, the group ensures the clinical validity for GLA and GBA causing Fabry and Gaucher disease, respectively. A strong link between gene and disease is necessary for an accurate diagnosis. Incorporating new evidence as it becomes available creates the most accurate gene-disease associations.

In addition, the group oversees the curation of variants detected in GLA and GBA as well as the curation of individuals screened for Fabry disease or Gaucher disease. This process includes the development of a custom, curation interface facilitating the systematic recording of variants, evidence used in classification, and clinical information. The curation of internal data with external information, including allele frequency, in silico predictions, and publications, ensures data consistency, accuracy, and quality.