

Variant Interpretation Working Group

The goal of the Variant Interpretation Working Group is to support the definition and refinement of the ACMG Variant Classification Guidelines at CENTOGENE as applied to variants in GLA and GBA genes. The group is composed of medical experts and scientists with the combined expertise to develop and maintain a consistent, robust classification pipeline.

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

Based on CENTOGENE's extensive experience in diagnosing lysosomal storage diseases, the group works to establish transparent classification processes for variants in GLA and GBA genes. As such, the group is also responsible for gene and disease specific modifications of the ACMG Guidelines, improving accuracy in classification.

The group oversees the reclassification of variants in GLA and GBA genes. Reclassification is essential to a diagnostic workflow; continuously growing data must be incorporated into existing classifications. All reclassification of variants in GLA or GBA genes will be reviewed monthly by the working group, with high risk reclassifications being reviewed immediately.