HGMD Pro Solution Overview for Clinical Labs

With over 314,707 expert-curated disease-causing mutations and more than 11,000 detailed summary reports of disease-associated/functional polymorphisms, HGMD® Pro is the most up-to-date and comprehensive collection of known and published pathogenic gene lesions responsible for human inherited disease. Cited in over 5000 publications in leading scientific journals, it is integral to any clinical assessment of germline variants. HGMD Pro provides valuable data for clinical interpretive and reporting use in exome screening studies, and optimizes mutational screening strategies. HGMD Pro is a widely used and trusted resource for medical and clinical geneticists, bioinformaticians, physicians and genetic counselors.

Value	Benefits	Features
Find pathogenic mutations of interest	Easily verify if an observed variant has been previously described as causative	Dataset is the only comprehensive collation of human germline variants that underly human inherited diseases, with expert curation of 127,000+ published articles describing the 307,000+ mutation entries
Evaluate mutation effect on disease quickly	Obtain an overview of the pathogenic mutational spectrum of a particular gene or disease	Industry's only variant database that continuously curates and reclassifies — providing high-quality data to the end-users and eliminating time wastage on benign variants/polymorphisms. This is achieved with a combination of human and AI search procedures for manual data curation (AI content is 4% of total variants)
Ensure thorough inspection of level of evidence for clinical impact	Quickly access detailed reports for disease associated human inherited mutations	Users can examine the phenotypes and links to references, mutational pages and curatorial comments

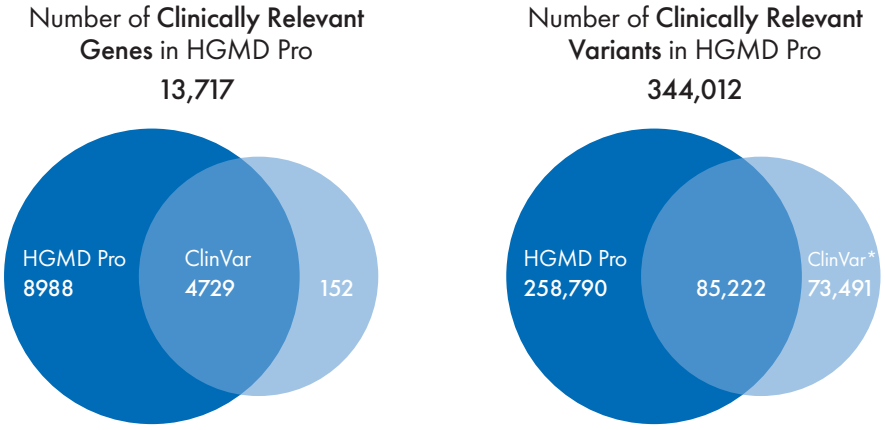

Figure 1. HGMD Pro has ~2.8x more genes with clinically relevant variants than ClinVar, and ~2.2x more clinically relevant variants. Data compiled as at October 2021.

"Clinically relevant variants" = pathogenic or likely pathogenic variants.

* Includes conflicting interpretation of pathogenicity.

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