HGMD Professional 2020.3 Release

The Fall 2020 Release of the Human Gene Mutation Database (HGMD) Professional is available, expanding the world's largest collection of human inherited disease mutations to 298,409 entries–*that's 9,063 more than the previous release.*

For over 30 years, HGMD Professional has been used worldwide by researchers, clinicians, diagnostic laboratories and genetic counselors as an essential tool for the annotation of next-generation sequencing (NGS) data in routine clinical and translational research. Founded and maintained by the Institute of Medical Genetics at Cardiff University, HGMD Professional provides users with a unique resource containing expert-curated mutations all backed by peer-reviewed publications where there is evidence of clinical impact.

Whether searching for an overview of known mutations associated with a particular disease, interpreting clinical test results, looking for the likely causal mutation in a list of variants, or seeking to integrate mutation content into your custom NGS pipeline or data repository–HGMD is the defacto-standard repository for heritable mutations that can be adapted to a broad range of applications.

Solve more cases faster, with data you can trust

- **298,409**

  detailed mutation reports

- **31,644**

  new mutation entries in 2019 alone

- **10,500+**

  summary reports listing all known inherited disease mutations

Expert-curated content updated quarterly

HGMD is powered by a team of expert curators at Cardiff University. Data are collected weekly by a combination of manual and computerized search procedures. In excess of 250 journals are scanned for articles describing germline mutations causing human genetic disease. The required data are extracted from the original articles and augmented with the necessary supporting data.

The number of disease-associated germline mutations published per year has more than doubled in the past decade (Figure 1). As rare and novel genetic mutations continue to be uncovered, having access to the latest scientific evidence is critical for timely interpretations of next-generation sequencing (NGS) data.

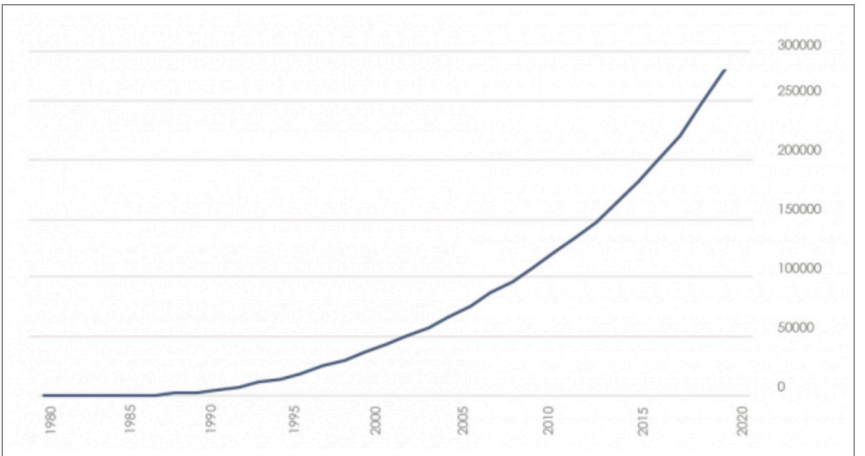

Figure 1. Mutation entries in HGMD Professional. The number of inherited disease-associated germline mutations published per year has more than doubled since 2010 (within 10 years).

View the complete HGMD Professional statistics here.