HGMD®:

Human Gene Mutation Data

The human gene mutation database (HGMD®) represents an up-to-date and comprehensive collection of known and published pathogenic gene lesions responsible for human inherited disease.

HGMD® provides information of practical diagnostic importance to medical and clinical geneticists, diagnosticians, bioinformaticians, researchers in human and molecular genetics and physicians and genetic counselors interested in a particular inherited condition in a given patient or family. HGMD® is a widely used, trusted resource that has been cited in over 5000 publications in leading scientific journals.

HGMD® is available as a free public version with restricted content and limited search options for academic use only and as a fully functional professional version that requires annual subscription through BIOBASE.

Key Capabilities

- Easily verify whether an observed mutation has been previously described to be responsible for causing human inherited disease
- Obtain an overview of the pathogenic mutational spectrum of a particular gene or disease
- Quickly access detailed reports for disease-associated human inherited mutations

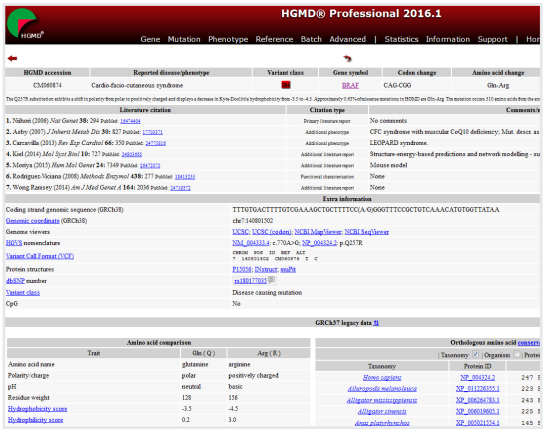

Figure1. HGMD® Professional sample mutation report

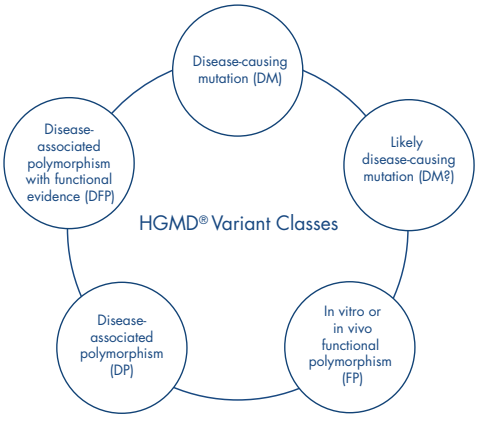

Figure2. Types of mutation within HGMD®

From Our Customers

"HGMD® professional provides the most comprehensive database of human disease associations and is an invaluable resource in both clinical and research-grade genetics and genomics activties."

– Dr. Ali Torkamani,
CSO at Cypher Genomics

"We rely on HGMD® professional heavily for reporting our clinical tests. We are currently working on next generation sequencing projects, identifying genes for disease-causing mutations and disease-associated / functional polymorphisms."

- We Yaping Yang, PhD,
Baylor College of Medicine