About EGL Genetics
EGL Genetics specializes in genetic diagnostic testing, with nearly 50 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL Genetics offers a combined 1000 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients.

Equally important to improving patient care through quality genetic testing is the contribution EGL Genetics makes back to the scientific and medical communities. EGL Genetics is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>35,000 variants on >1700 genes) and is also the only laboratory with a free online database (EmVClass), featuring a variant classification search and report request interface, which facilitates rapid interactive curation and reporting of variants.

Instructions for Use
EGL Genetics is committed to ensuring clients and patients are kept up-to-date concerning the classification of sequence variants. EmVClass, the EGL Variant Classification Tool, provides free, online, no registration required, open-access to all sequence variants detected and analyzed by EGL Genetics. EmVClass is located at: www.geneticslab.emory.edu/EmVClass.

Once on the EmVClass webpage, enter the official gene symbol or transcript (e.g. NM_*) and click “Search” to see all of the variants reported and analyzed by EGL for that gene/transcript. Search functionality also accepts a cDNA position, an amino acid residue number, a class of variant (snp, del, ins, dup), or a complete nucleotide or protein change nomenclature. HGVS nucleotide ‘long’ and ‘short’ nomenclatures, as well as one-letter and three-letter amino acid protein change notations, are compatible with the search. The prefixes ‘c.’ and ‘p.’ are not required, but may be entered. Historic nomenclatures are respected whenever possible.

Submit a question regarding a variant or prompt a review of a variant of unknown significance (VOUS) by clicking the appropriate button on the returned data table.

Updated Reports
As more information from human exome and genome sequencing projects becomes publicly available and new research is conducted on previously reported DNA variants, knowledge of variant classification increases. This knowledge will allow variants previously classified as variants of unknown significance to be reclassified as pathogenic variants or benign polymorphisms.

When new data emerges to support a variant classification change, EGL Genetics will issue amended reports for each patient with that variant, upon request.