accurately characterized for clinical reporting and genetic research.

## 2.3 Annotation and interpretation

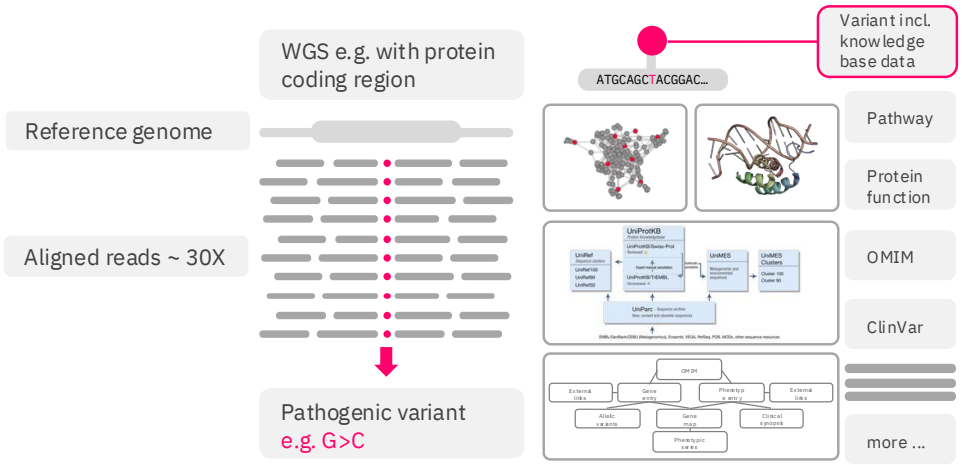


Figure 2: Variant annotation and interpretation.

In our approach to DNA annotation and interpretation, we employ tools such as Variant Effect Predictor (VEP) to systematically incorporate data from roughly 160 genetic databases, aiming to improve the clarity and utility of variant interpretations. Our protocols conform to ACMG/AMP standards, facilitating accurate clinical and research classifications. This methodical application is handled by our `Guru` software for genomic data to ensure reliable identification of pathogenic variants, grounded in detailed annotation and rigorous analysis (figure 2.3).

## 2.4 Qualifying variants

This report relied on qualifying variant filter protocol `Design QV SNV INDEL v1` (figure 2.4). This is our comprehensive framework for the identification and qualification of SNV and INDEL variants in genomic studies. It uses a flexible approach, incorporating quality control and multiple filtering stages to refine datasets for downstream analysis. The protocol is tailored to accommodate various research needs, balancing stringent filtering for rare disease

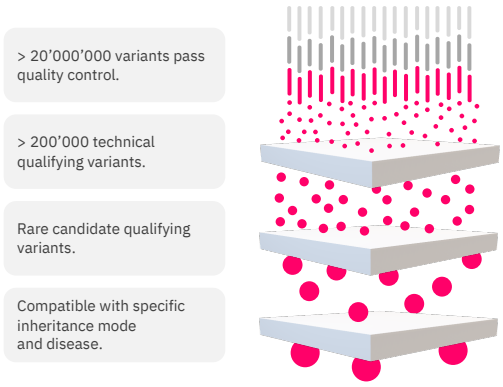


Figure 3: Qualifying variants (QV).