UNIVERSITÄT LEIPZIG

Medizinische Fakultät

Universitätsklinikum Leipzig

Medizin ist unsere Berufung.

MorbidGenes Panel:
A monthly updated list of diagnostically relevant genes derived from diverse sources

Robin-Tobias Jauss, Bernt Popp, Konrad Platzer, Rami Abou Jamra
Institute of Human Genetics, University of Leipzig Hospitals and Clinics, Leipzig, Germany

Upshot

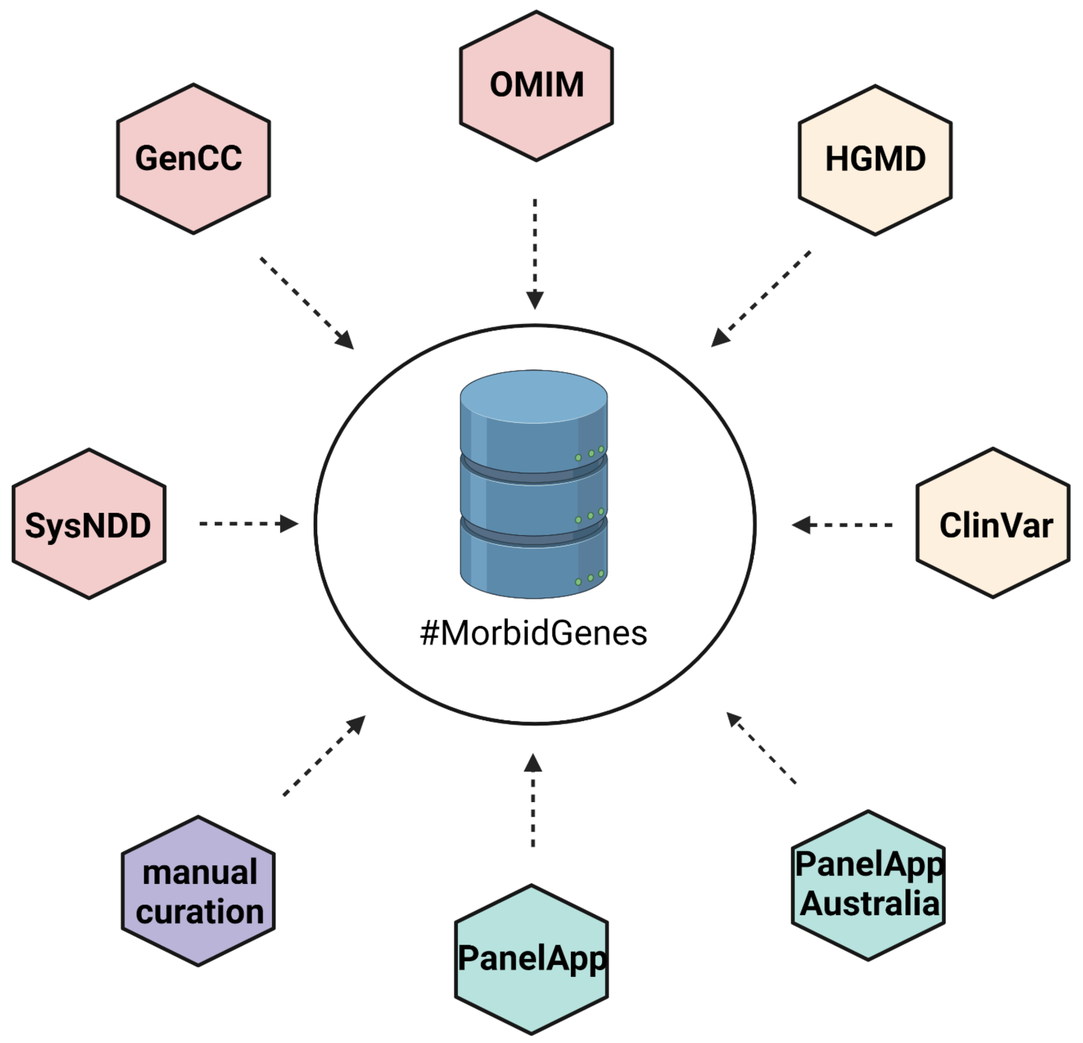
The MorbidGenes panel is a comprehensive and open overview of clinically relevant genes based on a growing list of sources, aiding researchers and clinicians in routine diagnostics. The panel is freely available at **www.morbidgenes.org**

Background

Identifying clinically relevant genetic variants is crucial for a fast and reliable genetic diagnosis. With exome sequencing now standard, diagnostic labs are in need of a, in principle, to-the-day-accurate list of genes associated with rare diseases. Manual curation efforts are slow and often disease specific, while efforts relying on single sources are too inaccurate and may result in false-positive genes.

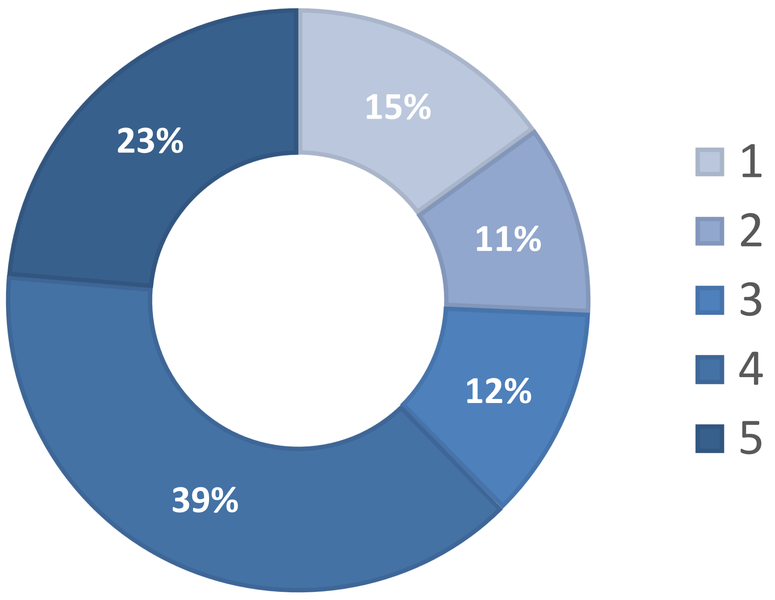
We established the MorbidGenes panel based on a list of publicly available databases: OMIM, PanelApp, SysNDD, ClinVar, HGMD and GenCC. A simple logic allows inclusion of genes with sufficient evidence based on a voting algorithm. By providing an API endpoint, users can directly access the list and metadata for all relevant information on their genes of interest. The genes are scored based on the number of sources supporting the pathogenicity (MorbidScore).

Data Sources

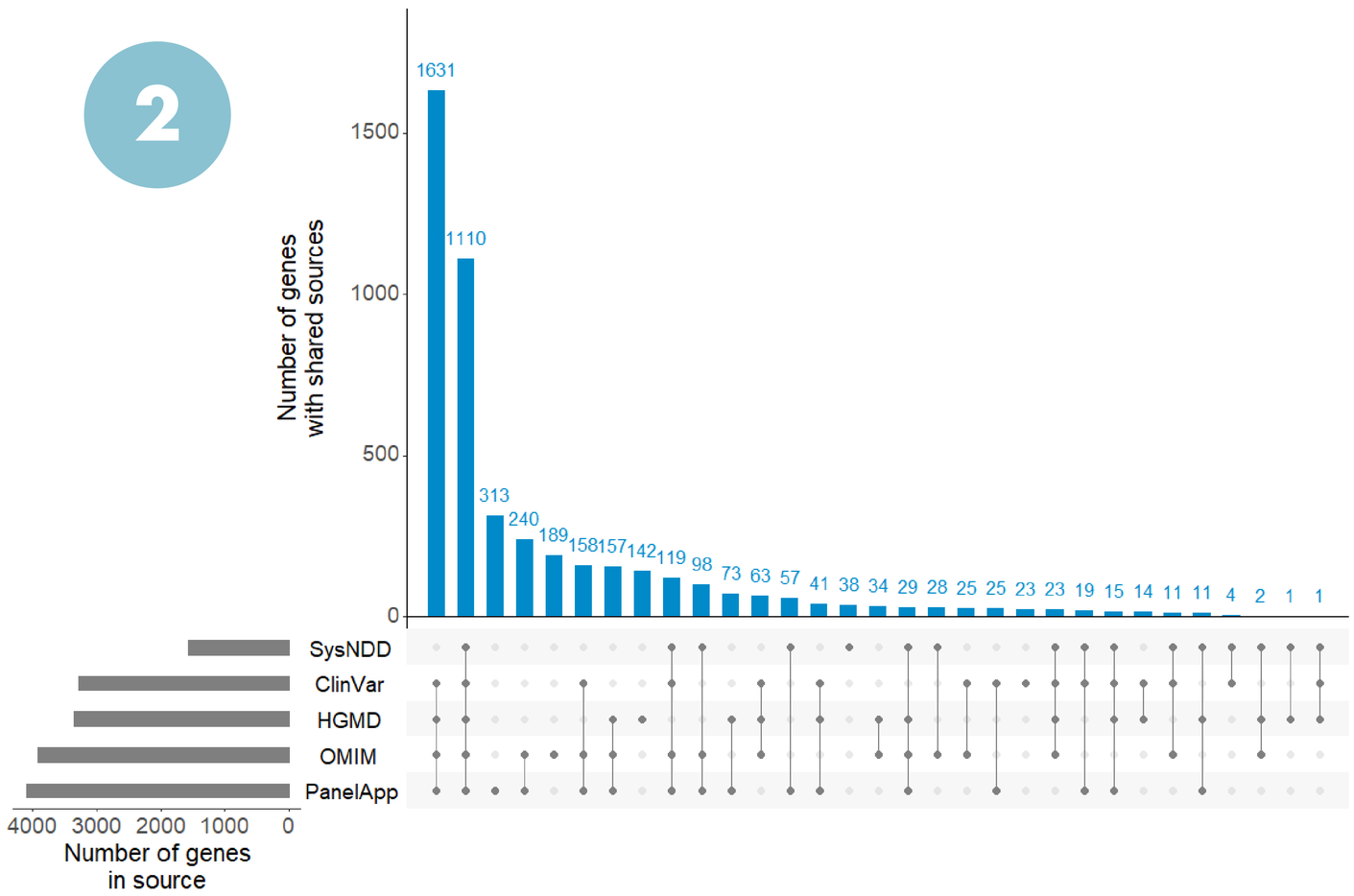


Results & Discussion

1



2



The MorbidGenes Panel currently contains 4712 genes (version 2022-05, as of May 2022) with minimal sufficient evidence on disease causality to classify them as diagnostically relevant. The MorbidScore (Fig. 1) provides a robust count on the top 5 data sources (ClinVar, HGMD, OMIM, PanelApp and SysNDD) supplementing evidence on the pathogenicity per gene, as Fig. 2 demonstrates the need to include as many and diverse data sources as possible for such virtual panels.

Dr. rer. nat. Robin-Tobias Jauss | Universitätsklinikum Leipzig | Institut für Humangenetik | Philipp-Rosenthal-Straße 55 04103 Leipzig | Robin-Tobias.Jauss@medizin.uni-leipzig.de