

## **Description of Additional Supplementary Files**

File Name: Supplementary Data 1

Description: List of tools and versions applied in the bioinformatics pipeline.

File Name: Supplementary Data 2

Description: List of 4,250 OMIM disease genes used in clinical analyses section containing Approved\_Symbol, Gene\_Name, MIM\_number and Phenotypes.

File Name: Supplementary Data 3

Description: Pathogenic variants in OMIM Disease genes with frequencies above 10% in SABE dataset.

File Name: Supplementary Data 4

Description: Reclassified variants after manual curation in genes with dominant mode of inheritance.

File Name: Supplementary Data 5

Description: Pathogenic variants and categories after manual curation in genes with dominant mode of inheritance.

File Name: Supplementary Data 6

Description: Pathogenic findings on ACMG-59 genes.

File Name: Supplementary Data 7

Description: Variant-based incidence of selected genes associated with recessively inherited disorders.

File Name: Supplementary Data 8

Description: OMIM genes with mobile element insertion events in exonic regions.