

Description of Additional Supplementary Files

File name: Supplementary Data 1

Description: list of genes with counts of variants >90% likelihood of being missed as calculated by StratoMod

File name: Supplementary Data 2

Description: list of pathogenic variants with >90% likelihood of being missed by Hifi or Illumina as calculated by Stratomod (used to generate Supplemental File 1)

File name: Supplementary Data 3

Description: Raw INDEL data corresponding to Figure 4c (which only shows SNVs)

File name: Supplementary Data 4

Description: Raw SNV data for Figure 4c

File name: Supplementary Data 5

Description: Raw INDEL data for Figure 5 containing all gnomAD variants that are also in ClinVar

File name: Supplementary Data 6

Description: Raw INDEL data for Figure 5 containing all ClinVar variants that are also in gnomAD

File name: Supplementary Data 7

Description: Raw SNV data for Figure 5 containing all gnomAD variants that are also in ClinVar

File name: Supplementary Data 8

Description: Raw SNV data for Figure 5 containing all ClinVar variants that are also in gnomAD