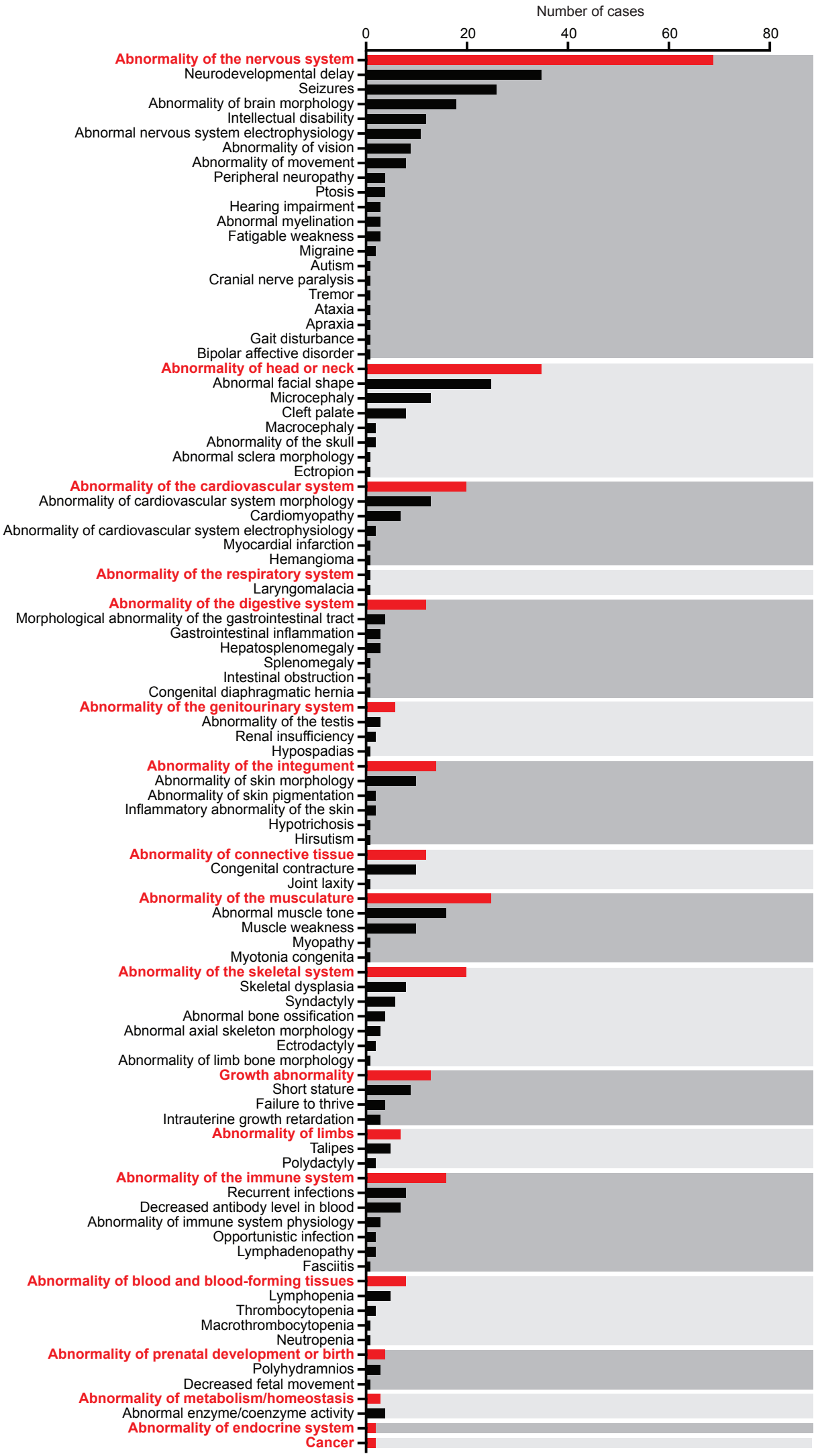
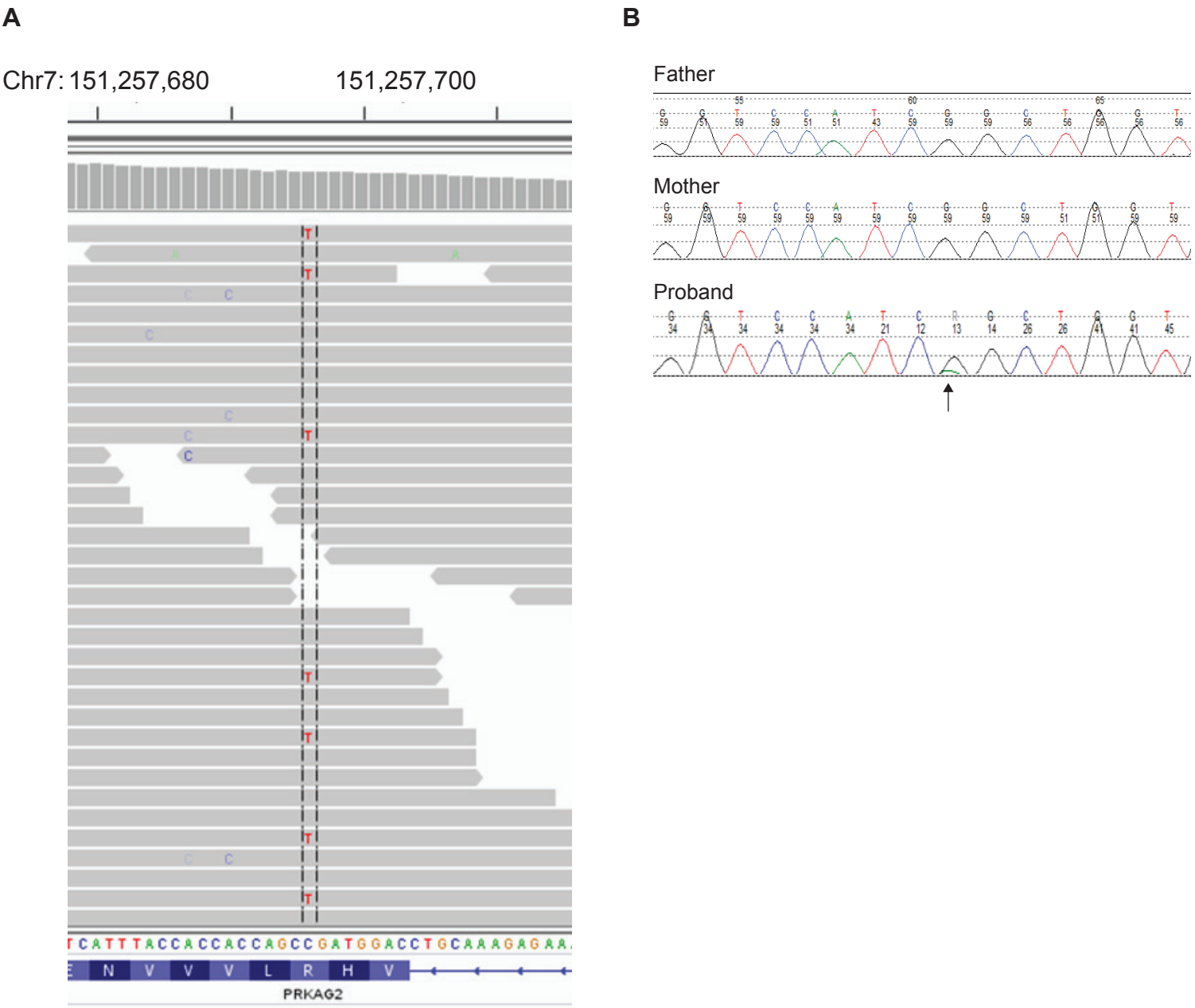



**Supplementary Figure 1. Clinical phenotypes for all cases referred to GM-MDT (n=132).** Human phenotype ontology terms used.




**Supplementary Figure 2. Mosaic *PRKAG2* variant c.1592G>A, p.(Arg531Gln).** (**A**) Aligned ES reads for the proband visualised in IGV show the variant base change was present in 22/122 (18%) of Illumina reads mapping to the *PRKAG2* gene (5' to 3' runs right to left). (**B**) Sanger sequencing confirmation (the reverse complement of the sequence in panel A, 5' to 3' from left to right) illustrates the presence of the variant adenosine base (indicated by the arrow).