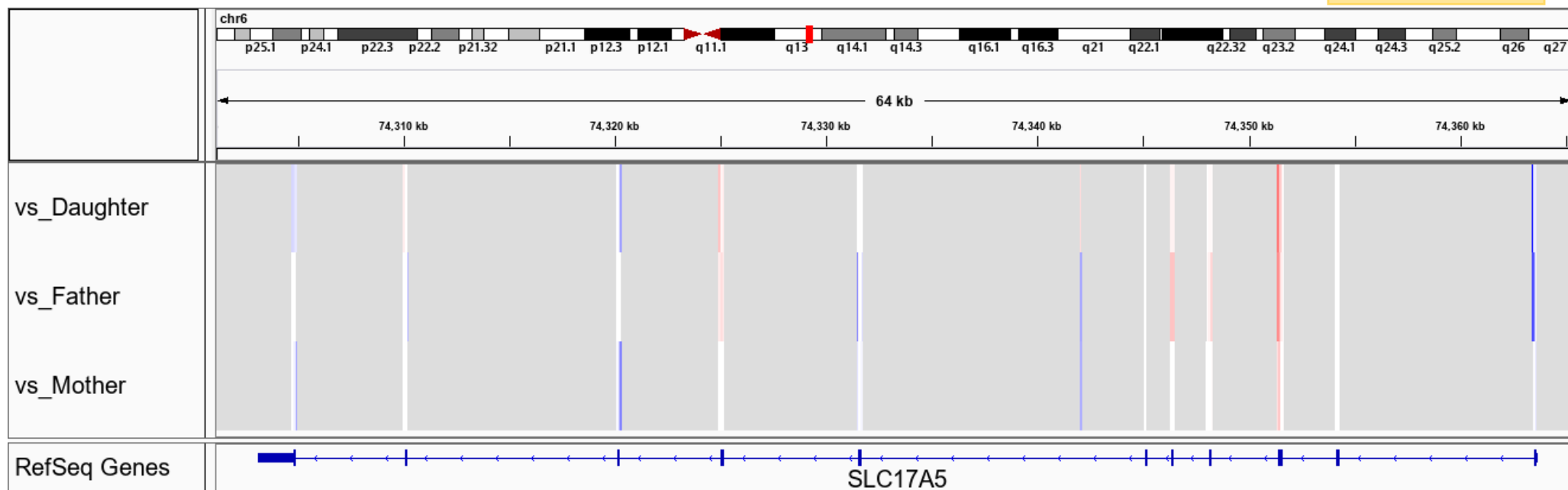
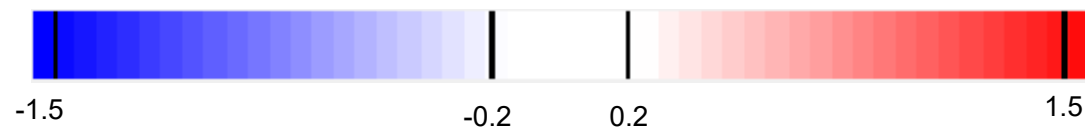


# SLC17A5



VarScan2 estimated copy number, log<sub>2</sub> ratio scale:



**Supplementary Figure S1. CNV from WES studies in the SLC17A5 gene.** CNV alterations (Gains [red] and losses [blue]) retrieved by WES assays through the VarScan2 algorithm for the patient with respect to the other three family members. Graphs were depicted using the integrative genome viewer (IGV) tool for the GRCh37 human genome version.