



Supplementary Figure S2. Copy number assay of the *KITLG* gene. Diploidy of the mutant allele of *KITLG* gene was confirmed in the patient's genome by TaqMan[®] Copy Number Assay (QIAGEN). In short, the real-time PCR measurement of *KITLG* using *RNase P* as the standard reference was performed for the genome of the patient and the family members. Signals were normalized to that of the patient. The results of the assay are not affected by the c.94G>A mutation. The ratio of 1.0 shows that all family members have the same copy number of the *KITLG* gene.