

Table S1. VHL gene mutation status of the analyzed pedigrees with the Human Genome Variation Society nomenclature on complementary and genomic DNA level.

Pedigrees	gDNA Level	Expected gene change (cDNA)
1	Chr3(GRCh37): g.10182301_10188764del	c.-1231_463+444del6464
2	Unknown	c.(?_1)_463+?del
3	Chr3(GRCh37): g.10183758_10183760del	c.227_229del
4	Chr3(GRCh37): g.10183764A>G	c.233 A>G
5	Chr3(GRCh37): g.10184154_1019013delins14	c.340+283_464-1335delins14
6	Chr3(GRCh37): g.10184170_10189331del	c.341-4028_463+1011del5162
7	Chr3(GRCh37): g.10188245G>T	c.388 G>T
8	Chr3(GRCh37): g.10191460T>A	c.464-11 T>A
9	Chr3(GRCh37): g.10191480dup	c.473dup
10	Unknown	<i>VHL</i> full length deletion
11	Chr3(GRCh37): g.10191507G>A	c.500 G>A
12	Chr3(GRCh37): g.10188309T>G	c.452 T>G
13	Chr3(GRCh37): g.10183797T>C	c.266 T>C
14	Chr3(GRCh37): g.10183722G>C	c.191 G>C
15	Unknown	<i>VHL</i> full length deletion

cDNA = complementary DNA; gDNA = genomic DNA; del = deletion; dup = duplication; VHL = von Hippel-Lindau.