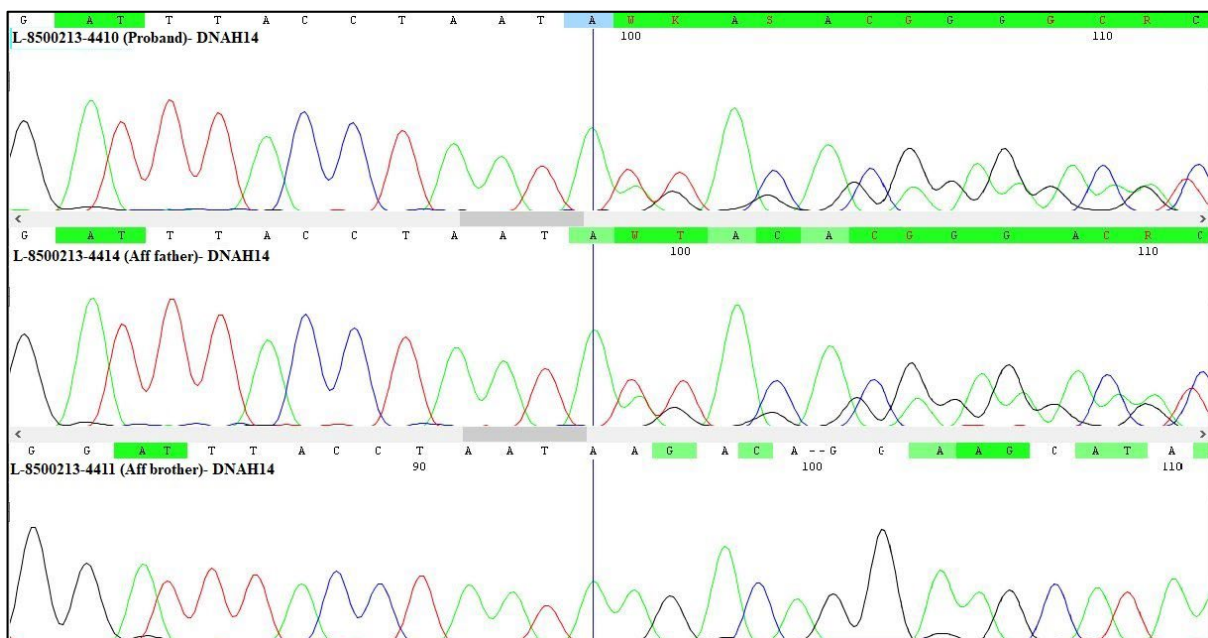
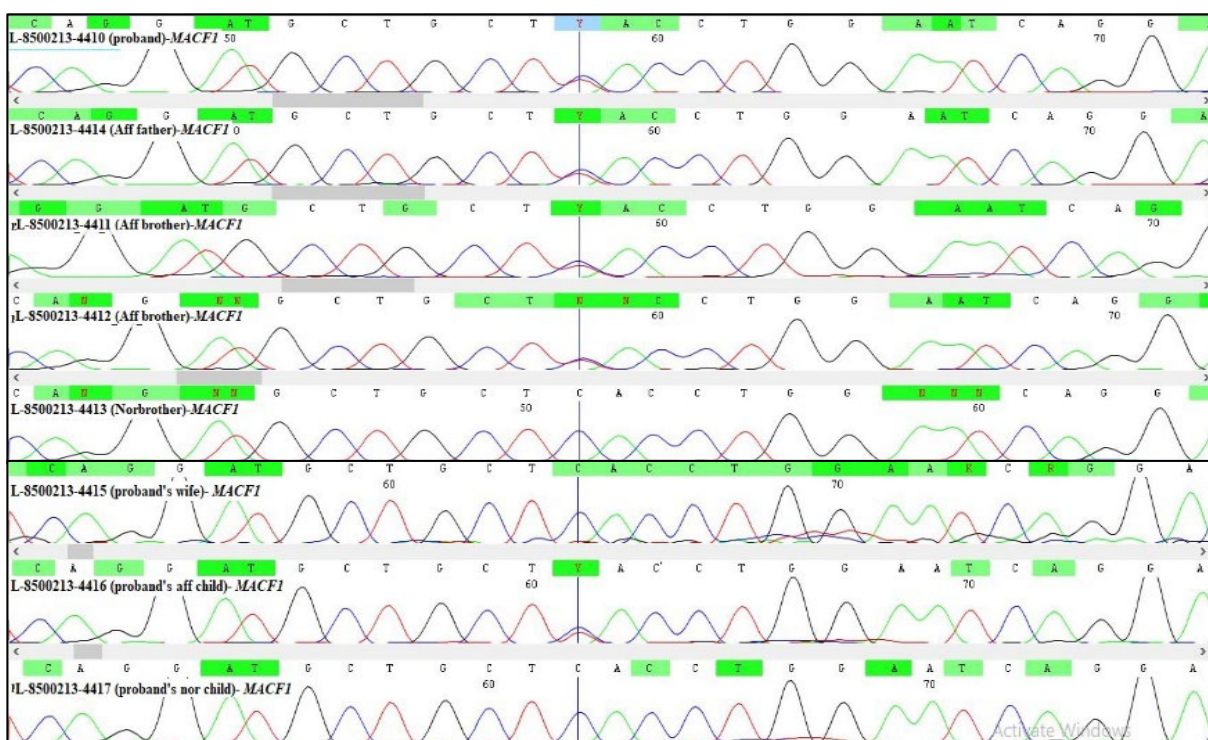


Supplementary file 1

Figure S1: Electropherogram of affected and unaffected individuals of family L-8500213 for segregation analysis of *MACF1*, *DNAH14*, *MUC16* and *USP6* genes. In *MACF1*, all affected persons are heterozygote for the c.C1378T and all normal individuals are homozygote for the wild allele, but for three other genes segregation in the family was not confirmed. In *MUC16*, variant was not confirmed in proband too.



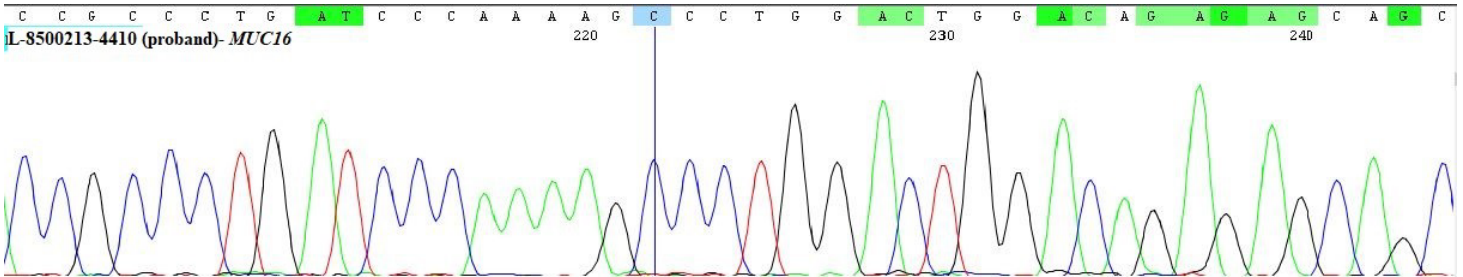
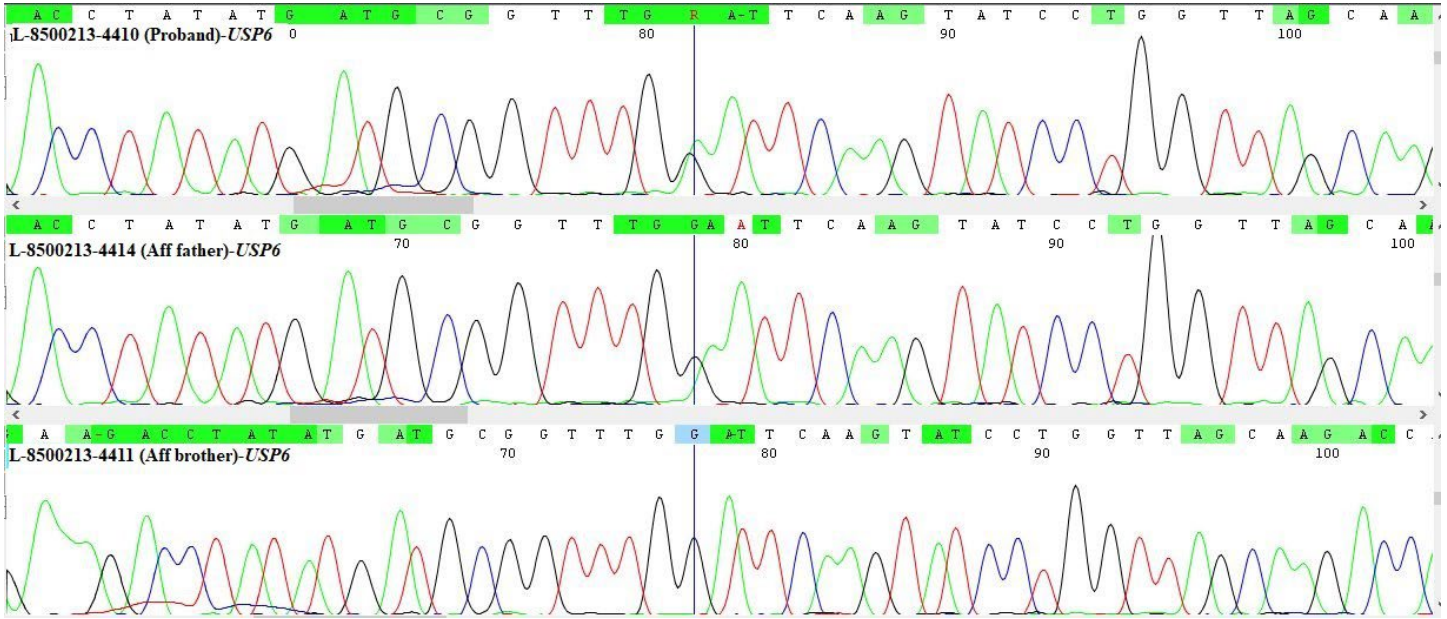


Table S1: All of candidate variants and ones which have been performed segregation analysis

Gene	Chromosome position	Variant	Segregation
<i>DNAH14</i>	Chr1: 225237989	c.1990-1991insTT	No
<i>USP6</i>	Chr17: 5058836	c.G2763A	No
<i>MUC16</i>	Chr19: 8999498	c.40674-40677del	No
<i>MACF1</i>	Chr1: 39751285	c.C1378T	Yes
<i>NLRC4</i>	Chr2: 32476071	c.C862T	-
<i>NCOA5</i>	Chr20: 44699165	c.G49A	-
<i>SLC25A5</i>	ChrX: 118604428	c.T691C	-
<i>MYH6</i>	Chr14: 23858093	c.C4150T	-
<i>INPP1</i>	Chr2: 191235908	c.T980C	-
<i>ANK3</i>	Chr10: 61834991	c.C5648T	-
<i>INTS1</i>	Chr10: 1536799	c.C1577T	-
<i>PABPC3</i>	Chr13: 25671168	c.C832T	-
<i>GTF3C2</i>	Chr2: 27559149	c.A1271G	-
<i>CRB2</i>	Chr9: 126125419	c.C370T	-
<i>WASHC2A</i>	Chr10: 51829421	c.A241G	-
<i>ITGA7</i>	Chr12: 56087806	c.C1190T	-
<i>SLC25A5</i>	ChrX: 118604444	c.G707C	-

<i>AGAP2</i>	Chr12: 58125741	c.G1804A	-
<i>CCDC47</i>	Chr17: 61829377	c.C1294T	-
<i>NDUFS2</i>	Chr1: 161182257	c.G1103A	-
<i>ALG1L</i>	Chr3: 125648332	c.C367T	-
<i>ADAMTS2</i>	Chr5: 178772229	c.99_101del	-
<i>PABPC3</i>	Chr13: 25671150	c.814_816del	-
<i>PABPC3</i>	Chr13: 25671155	c.819_827del	-
<i>LVRN</i>	Chr5: 115351067	c.2569_2570insTCACAGATGCTGGCTA	-
<i>NBPF9</i>	Chr1: 145368473	unknown	-