

Supplemental Table S1. Overview of ABHD12 variants found in this study

Nucleotide change	Protein effect	Mutation type	Frequency (n =)	SIFT	Align GVG D	Polyphen-2	References
c.193C>T	p.(Arg65*)	Nonsense	2	-	-	-	Eisenberger <i>et al.</i> 2012, Igelman <i>et al.</i> 2021
c.337_338delinsTTT	p.(Asp113Phefs*15)	Frameshift	13	-	-	-	Fiskerstrand <i>et al.</i> 2010
c.341dup	p.(Leu114Phefs*14)	Frameshift	1	-		-	This study
c.374C>T	p.(Thr125Met)	Missense	1	Affects protein function	C65	Probably damaging	Igelman <i>et al.</i> 2021
c.423-1_425del	p.(?)	Splice site	3	-	-	-	This study
c.477G>A	p.(Trp159*)	Nonsense	1	-	-	-	Nishiguchi <i>et al.</i> 2014
c.557G>C	p.(Arg186Pro)	Missense	1	Affects protein function	C65	Probably damaging	Nishiguchi <i>et al.</i> 2014
c.620-2A>G	p.(?)	Splice site	2	-	-	-	Igelman <i>et al.</i> 2021
c.784C>T	p.(Arg262*)	Nonsense	1	-	-	-	Thimm <i>et al.</i> 2020, Igelman <i>et al.</i> 2021
c.867+5G >A	p.(?)	Splice site	1	-	-	-	Ellingford <i>et al.</i> 2019
c.1063C>T	p.(Arg355*)	Nonsense	2	-	-	-	This study
c.1075del	p.(Val359Phefs*27)	Frameshift	1	-	-	-	This study
c.1154T>C	p.(Leu385Pro)	Missense	1	Affects protein function	C65	Probably damaging	Igelman <i>et al.</i> 2021

Nucleotide numbering is based on the reference sequence NM_001042472.3. The Human Gene Mutation Database was accessed on July 5th, 2021.