Table S3. Genetic features of the individuals with *ZFHX3* variants

Case	**Coordinate**	**cDNA change (NM_006885)**	**Protein change**	**Inheritance**	**MAF**	**SIFT**	**PP2_Var**	**Mutation-Taster**	**CADD**	**GERP++**	**phyloP**	**phastCons**	**SiPhy**
Case 1	chr16:72993731	c.314C>T	p.Pro105Leu	paternal	2.74e-04	T (0.056)	B (0.001)	D (1.000)	T (17.01)	C (5.11)	NC (0.874)	NC (0.534)	C (12.922)
	chr16:72991763	c.2282G>C	p.Gly761Ala	maternal	1.54e-04	D (0.020)	P (0.719)	D (1.000)	T (19.17)	C (4.51)	C (3.856)	NC (0.964)	C (13.864)
Case 2	chr16:72991626	c.2419G>A	p.Glu807Lys	paternal	5.20e-05	T (0.153)	P (0.994)	D (1.000)	D (24.70)	C (5.52)	C (7.858)	C (1.000)	C (19.474)
	chr16:72821322	c.10853C>A	p.Pro3618Gln	maternal	2.39e-04	D (0.001)	P (0.669)	D (1.000)	T (17.82)	C (4.39)	C (6.706)	C (1.000)	C (16.590)
Case 3	chr16:72991374	c.2671T>C	p.Phe891Leu	maternal	-	T (0.064)	P (0.977)	D (1.000)	D (25.30)	C (5.52)	C (9.268)	C (1.000)	C (15.677)
	chr16:72822591	c.9583_9584insT	p.Pro3195LeufsTer44	paternal	3.93e-05	-	-	-	-	-	-	-	-
Case 4	chr16:72991359	c.2686G>A	p.Ala896Thr	paternal	1.52e-04	T (0.352)	B (0.015)	D (1.000)	T (19.79)	NC (-4.08)	NC (0.020)	NC (0.933)	NC (8.169)
	chr16:72821736	c.10439C>T	p.Ala3480Val	maternal	5.38e-05	T (0.114)	P (0.970)	D (1.000)	D (22.80)	C (4.24)	C (7.999)	C (1.000)	C (16.983)
Case 5	chr16:72832453	c.4125_4127del	p.Ala1376del	maternal	3.18e-05	-	-	-	-	-	-	-	-
	chr16:72821322	c.10853C>A	p.Pro3618Gln	paternal	2.39e-04	D (0.001)	P (0.669)	D (1.000)	T (17.82)	C (4.39)	C (6.706)	C (1.000)	C (16.590)
Case 6	chr16:72831429	c.5152A>C	p.Met1718Leu	paternal	1.99e-05	T (0.665)	B (0.009)	D (1.000)	T (13.16)	C (5.74)	C (4.166)	C (1.000)	C (16.331)
	chr16:72821665	c.10510G>C	p.Val3504Leu	maternal	-	D (0.022)	P (0.537)	D (1.000)	D (21.00)	C (4.22)	C (4.744)	C (1.000)	C (16.949)
Case 7	chr16:72830420	c.6161C>T	p.Ala2054Val	paternal	1.85e-05	D (0.000)	P (0.998)	D (1.000)	D (23.10)	C (5.29)	C (9.940)	C (1.000)	C (19.309)
	chr16:72821730	c.10445G>T	p.Ser3482Ile	maternal	2.47e-04	D (0.015)	B (0.052)	D (0.950)	D (20.20)	C (4.24)	NC (1.133)	NC (0.797)	NC (8.770)
Case 8	chr16:72821730	c.10445G>T	p.Ser3482Ile	maternal	2.47e-04	D (0.015)	B (0.052)	D (0.950)	D (20.20)	C (4.24)	NC (1.133)	NC (0.797)	NC (8.770)
	chr16:72821322	c.10853C>A	p.Pro3618Gln	paternal	2.39e-04	D (0.001)	P (0.669)	D (1.000)	T (17.82)	C (4.39)	C (6.706)	C (1.000)	C (16.590)

Abbreviation: B, benign; C, conserved; CADD, Combined Annotation Dependent Depletion; D, damaging; GERP, Genomic Evolutionary Rate Profiling; MAF, minor allele frequency from general population of gnomAD; NC, nonconserved; P, probably_damaging; PP2, Polyphen2_HVAR; phastCons, Phylogenetic Analysis with Space/Time models conservation scoring and identification of conserved elements; phyloP, Computation of p-values for conservation or acceleration, either lineage-specific or across all branches; SIFT, Sorting Intolerant From Tolerant; T, tolerable.