

Supplementary Table. List of homozygously shared variants (≥ 20 X) with three affected patients in family (III:1, III:4, III:5) and not in homozygous in healthy brother (III:8). Alteration in the *BEND4* gene (c.1297G>A, p.Gly433Ser, rs779411414) is the only variant with unknown frequency data in populations, shown with red borderline.

ReadDepth	Chr	Position GRCh37/hg19	Ref->Seq	dbSNP	Gene	Transcript	VarSome	Frequency	
								gnomAD- Exoms	gnomAD- Genomes
31	1	90486412	T->G	rs2816881	ZNF326	455342	Benign	0.6300	0.6183
113	1	110086451	G->A	rs17575798	GPR61	404129	Benign	0.2076	0.1807
52	1	110091271	C->T	rs3737182	GPR61	404129	Benign	-	0.1854
113	1	110151395	G->T	rs3738766	GNAT2	351050	Benign	0.05037	0.03229
46	1	110162918	T->C	rs28362580	AMPD2	342115	Benign	0.1100	0.09815
76	1	110741086	C->T	rs41306175	SLC6A17	331565	Benign	0.07288	0.08146
71	1	110766454	G->A	rs59123361	KCNC4	369787	Benign	0.08264	0.07083
71	1	110882444	A->G	rs814771	RBM15	369784	Benign	0.7588	0.7911
45	1	110949720	C->T	rs6671673	HBXIP	483260	Benign	0.5689	0.5894
22	1	111857087	C->T	rs2786151	CHIA	369740	Benign	-	0.5429
28	1	111861714	T->A	rs2786161	CHIA	369740	Benign	0.6219	0.6574
73	1	111861841	A->G	rs2275253	CHIA	353665	Benign	0.6397	0.6950
47	1	111861974	T->C	rs2275254	CHIA	369740	Benign	0.5012	0.4883
57	1	111862952	T->G	rs2256721	CHIA	369740	Benign	0.6394	0.6943
39	1	112251983	C->G	rs3738300	RAP1A	436150	Benign	0.3017	0.3017
66	1	119466275	A->G	rs4659129	TBX15	344218	Benign	0.6621	0.6559
242	1	144994861	C->T	rs2442640	PDE4DIP	530472	VUS	-	0.5336
122	1	150445819	A->G	rs7002	RPRD2	369068	Benign	0.3447	0.3002
72	1	150477061	C->T	rs3738487	ECM1	498579	Benign	0.3405	0.2998
178	1	150484987	G->A	rs13294	ECM1	498579	Benign	0.3521	0.3317
158	1	152538358	G->C	rs17659359	LCE3E	368789	Benign	0.2695	0.2246
133	2	42989994	G->T	rs2278585	OXER1	378661	Benign	-	0.2356
166	2	42990010	C->A	rs12617777	OXER1	378661	Benign	0.2914	0.2942
229	2	42990101	G->C	rs2278586	OXER1	378661	Benign	0.2721	0.2360
93	2	42990522	C->T	rs12712859	OXER1	378661	Benign	0.2753	0.2407
111	2	42991200	C->G	rs34142793	OXER1	378661	Likely Benign	0.02440	0.01810
23	2	45780962	C->G	rs3816107	SRBD1	493649	Benign	-	0.5751
43	2	46739157	A->G	rs3814045	ATP6V1E2	524249	Benign	0.4338	0.4283
33	2	46818992	C->T	rs17818315	PIGF	412717	Benign	-	0.2228
39	2	46987139	G->A	rs17771838	SOC55	306503	Benign	0.08340	0.06805
36	4	37666976	C->G	rs6855306	RELL1	454158	Benign	0.08654	0.1020
72	4	37962133	G->A	rs2290740	PTTG2	504686	Benign	0.1669	0.1804
92	4	42122161	C->T	rs779411414	BEND4	504360	VUS	-	-
53	8	142170884	C->T	rs2289001	DENND3	519811	Benign	0.3357	0.2703
75	8	142445680	A->T	rs11784911	PTP4A3	349124	Benign	-	0.4267
45	8	142458053	A->G	rs6578185	AC100803.1	523857	Benign	0.4316	0.4297
33	8	142505057	G->T	rs2246586	AC100803.1	523857	Benign	0.3373	0.3132
81	8	142517222	A->G	rs2748421	AC100803.1	523857	Benign	0.4157	0.4068
86	8	142517227	C->T	rs2613648	AC100803.1	523857	Benign	0.4156	0.4064
83	8	143399860	G->T	rs56055628	TSNARE1	524325	Benign	0.2075	0.1835

48	8	144732026	G -> A	rs3750203	ZNF623	328466	Benign	0.5275	0.4842
31	8	146156247	C -> A	rs3735789	ZNF16	532351	Benign	0.2587	0.2956
47	8	146157339	T -> C	rs3735787	ZNF16	532351	Benign	0.2407	0.2250
50	8	146171285	G -> A	rs3824110	ZNF16	532351	Benign	0.2322	0.2436
44	8	146171437	A -> G	rs3735785	ZNF16	532351	Benign	0.2426	0.2380
58	8	146203412	A -> G	rs2979109	ZNF252	426361	Benign	0.4616	0.5452
95	11	70118251	T -> G	rs670012	PPFIA1	253925	Benign	0.3829	0.4725
75	11	70208311	G -> A	rs7130178	PPFIA1	253925	Benign	0.3810	0.4721
35	11	70222604	G -> A	rs664130	AP000487.5	530690	Benign	0.2193	0.2086
66	11	70229171	G -> A	rs552282	AP000487.5	530690	Benign	0.3353	0.4313
103	11	70253586	T -> C	rs569732	CTTN	301843	Benign	0.4332	0.5296
117	11	70279766	C -> T	rs643301	CTTN	301843	Benign	0.1994	0.2144
28	13	110435231	C -> T	rs1805097	IRS2	375856	Benign	0.3500	0.2964
43	13	110435953	A -> G	rs4773092	IRS2	375856	Benign	0.5869	0.6106
98	13	110436232	G -> A	rs3742210	IRS2	375856	Benign	0.5843	0.6034
29	13	111090909	T -> C	rs7984937	COL4A2	257309	Benign	0.2842	0.3449
37	13	111090924	G -> A	rs7984100	COL4A2	257309	Benign	0.2818	0.3377
67	13	111114554	G -> A	rs41275110	COL4A2-AS2	458403	Benign	0.1646	0.1818
23	13	111121620	C -> T	rs9583500	COL4A2-AS2	458403	Benign	0.1751	0.2193
40	13	111145676	G -> C	rs2391833	COL4A2	360467	Benign	0.5156	0.4433
65	16	4714639	T -> C	rs1659479	MGRN1	536343	Benign	-	0.5035
91	16	4718388	A -> G	rs34069734	MGRN1	536343	Benign	-	0.4183
71	16	4721468	T -> C	rs715109	MGRN1	536343	Benign	0.5738	0.5890
60	16	4723451	A -> G	rs2289314	MGRN1	536343	Benign	0.4991	0.5114
99	16	4796393	A -> G	rs737700	ZNF500	545009	Benign	0.6184	0.6208
41	16	4797457	C -> T	rs17853375	ZNF500	545009	Benign	0.5441	0.4748
30	16	4812209	G -> A	rs12599765	ZNF500	545009	Benign	-	0.5462
84	19	804456	G -> A	rs351980	PTBP1	349038	Benign	0.1290	0.1662
109	19	885818	A -> G	rs1060442	MED16	395808	Benign	0.5962	0.6122
35	19	984533	T -> C	rs2301810	WDR18	251289	Benign	0.4845	0.4958
51	19	990394	T -> C	rs2240148	WDR18	251289	Benign	0.4854	0.5132
135	19	1095336	C -> T	rs1046041	POLR2E	215587	Benign	0.5645	0.6171
110	19	1391059	G -> T	rs2074896	NDUFS7	538662	Benign	0.07837	0.04933
70	19	1397504	C -> T	rs2074899	NDUFS7	538662	Benign	0.07685	0.03497
140	19	1507483	T -> C	rs791460	ADAMTSL5	330475	Benign	-	0.3798
25	19	1534042	T -> C	rs10853954	PLK5	454744	Benign	0.3034	0.2507
106	19	2040061	T -> C	rs56197957	MKMK2	541165	Benign	-	0.2788
41	19	2717256	C -> G	rs3745812	DIRAS1	323469	Benign	0.1872	0.1479
56	19	2732986	T -> C	rs759073	SLC39A3	455372	Benign	0.5095	0.5868
41	19	2761603	C -> T	rs3745815	SGTA	221566	Benign	-	0.1958
39	19	2762585	A -> C	rs2302491	SGTA	221566	Benign	0.3237	0.3539
127	19	2790394	G -> A	rs1736176	THOP1	307741	Benign	0.3295	0.3416
55	19	2796021	G -> A	rs1736180	THOP1	307741	Benign	0.3222	0.3389
82	19	2812153	A -> G	rs941407	THOP1	307741	Benign	0.3471	0.4201
35	19	3186085	G -> T	rs11551095	NCLN	246117	Benign	0.5279	0.5317
57	19	8999453	C -> T	rs11085765	MUC16	380947	Likely Benign	0.5344	0.4688