

Supplementary Table. Distribution of the genetic variations in the *ABCD1* gene in our study

S.No	Phenotype	Variant identified	Chromosomal position	Transcript ID	dbSNP ID	Gene region	Type of variation	ACMG Criteria	Novel/ Reported
1	ccALD	c.320 [T>C]; p.Leu107Pro	chrX-152991041 T>C	NM_000033.4; ENST00000218104	rs1569540688	Exon 1	Missense Variation	Pathogenic [PM3, PP3, PM2, PM1, PP2 PP5]	Reported
2	ccALD	c.796 [G>A]; p.Gly266Arg	chrX-152991517 G>A	NM_000033.4; ENST00000218104	rs128624218	Exon 1	Missense Variation	Pathogenic [PM3, PP3, PM2, PM5, PM1, PP2 PP5]	Reported
3	ccALD	c.874 [G>A]; p.Glu292Lys	chrX-152991595 G>A	NM_000033.4; ENST00000218104	--	Exon 1	Missense Variation	Likely Pathogenic [PP3, PM2, PM5, PM1, PP2, PP5]	Reported
4	ccALD-	c.893 [G>T]; p.Gly298Val	chrX-152991614 G>T	NM_000033.4; ENST00000218104	rs868911300	Exon 1	Missense Variation	Likely Pathogenic [PP3, PM2, PM5, PM1, PP2]	Reported
5	ccALD	c.904 [G>T]; p.Glu302*	chrX-152994690 G>T	NM_000033.4	--	Exon 1	Nonsense Variation	Likely Pathogenic [PV51, PM2]	Novel Variation
6	ccALD	c.901-1 [G>A]	chrX-152994686 G>A	NM_000033.4	--	IVS 1	Nonsense Variation	Pathogenic [PV51, PM3, PM2, PP5]	Reported
7	Adolescent ALD	c.252 dup C; p.Arg85Pro fs*110	chrX-152990969T>TC	NM_000033.4	rs713993050	Exon 1	Nonsense Variation	Pathogenic [PV51, PM3, PM2, PP5]	Reported
8	Adolescent ALD	c.252 dup C; sp.Arg85P fs*110	chrX-152990969T>TC	NM_000033.4	rs713993050	Exon 1	Nonsense Variation	Pathogenic [PV51, PM3, PM2, PP5]	Reported
9	Adolescent ALD	c.323 [C>T]; p.Ser108Leu	chrX-152991044 C>T	NM_000033.4	--	Exon 1	Missense Variation	Pathogenic [PS2, PM3, PP3, PM2, PM1, PP2, PS3, PP5]	Reported
10	Adolescent ALD	c.323 [C>T]; p.Ser108Leu	chrX-152991044 C>T	NM_000033.4	--	Exon 1	Missense Variation	Pathogenic [PS2, PM3, PP3, PM2, PM1, PP2, PS3, PP5]	Reported
11	Adolescent ALD	c.347 [G>A]; p.Gly116Glu	chrX-152991068 G>A	NM_000033.4	-	Exon 1	Missense Variation	Likely Pathogenic [PP3, PM2, PM5, PM1, PP2]	Reported
12	Adolescent ALD	c.443 [A>G]; p.Asn148Ser	chrX-152991164 G>A	NM_000033.4	rs128624216	Exon 1	Missense Variation	Pathogenic [PM3, PM2, PM5, PM1, PP3, PP2, PP5]	Reported
13	Adolescent ALD	c.649 [A>G]; p.Lys217Glu	chrX-152991370 A>G	NM_000033.4	--	Exon 1	Missense Variation	Likely Pathogenic [PP3, PM2, PM5, PM1, PP2]	Reported
14	Adolescent ALD	c.847 [C>T]; p.His283Tyr	chrX-152991568 C>T	NM_000033.4	--	Exon 1	Missense Variation	Likely Pathogenic [PP3, PM2, PM5, PM1, PP2, PP5]	Reported
15	Adolescent ALD	c.1186 [G>A]; p.Ala396Thr	chrX-153001670 G>A	NM_000033.4	rs1569541006	Exon 3	Missense Variation	Likely Pathogenic [PM3, PM2, PP3, PM1, PP2, PP5]	Reported
16	Adolescent ALD	c.1202 [G>A]; p.Arg401Gln	chrX-153001686 G>A	NM_000033.4	rs128624219	Exon 3	Missense Variation	Pathogenic [PM3, PP3, PM2, PM5, PP2, PP5]	Reported
17	Adolescent ALD	c.1225-1 [G>A]	chrX-153001798 G>A	NM_000033.4	--	IVS 3	Nonsense Variation	Likely Pathogenic [PV51, PM2]	Novel Variation
18	Adolescent ALD	c.1505_06 [ins C]; p.Leu503Ser fs*53	chrX-153005562 A>AC	NM_000033.4	--	Exon 6	Nonsense Variation	Likely Pathogenic [PV51, PM2]	Reported

19	Adolescent ALD	c.1534 [G>A]; p.Gly512Ser	chrX-153005591 G>A	NM_000033.4	rs1569541088	Exon 6	Missense Variation	Pathogenic [PM3, PP3, PM2, PM5, PM1, PP2, PP5]	Reported
20	Addison type ALD	c.453_454 [ins CGTT]; p.Tyr153Ser fs*43	chrX-152991174 C>CT	NM_000033.4	--	Exon 1	Nonsense Variation	Likely Pathogenic [PV51, PM2]	Novel Variant
21	AMN	c.488 [G>T] ;p.Arg163Leu	chrX-152991209 G>T	NM_000033.4	--	Exon 1	Missense Variation	Pathogenic [PP3, PM2, PM5, PP2, PP5]	Reported
22	AMN	c.493_494 [ins CAC]; p.165Pro	chrX-152991213 C>CCCCA	NM_000033.4	--	Exon 1	Nonsense Variation	Likely Pathogenic [PM2, PM4, PM2]	Novel Variation
23	AMN	c.901-1[G>A]	chrX-152994686 G>A	NM_000033.4	--	IVS 1	Nonsense Variation	Pathogenic [PV51, PM3, PM2, PP5]	Reported
24	AMN	c.1415_1416 [del AG]; p.Gln472Arg fs*83	chrX-153002631 CAG>C	NM_000033.4	rs387906494	Exon 5	Nonsense Variation	Pathogenic [PV51, PM3, PM2, PP5]	Reported
25	AMN	c.1415_1416 [del AG]; p.Gln472Arg fs*83	chrX-153002631 CAG>C	NM_000033.4	rs387906494	Exon 5	Nonsense Variation	Pathogenic [PV51, PM3, PM2, PP5]	Reported
26	AMN	c.1451[C>G] ;p.Pro484Arg	chrX-153002688 C>G	NM_000033.4	rs128624214	Exon 5	Missense Variation	Likely Pathogenic [PP3, PM5, PM1, PP2, PP5]	Reported
27	AMN	c.1488+1 [G>A] ;p.Val497fs*	chrX-153002706 G>A	NM_000033.4	--	Exon 5	Nonsense Variation	Pathogenic [PV51, PM2, PP5]	Reported
28	AMN	c.1553. [G>A] ;p.Arg518Gln	chrX-153005160 G>A	NM_000033.4	rs398123102	Exon 6	Missense Variation	Pathogenic [PM3, PP3, PM2, PM5, PM1, PS2, PP2, PP5]	Reported
29	AMN	c.1771. [C>T] ;p.Arg591Trp	chrX-153006164 C+E11>T	NM_000033.4	rs398123106	Exon 7	Missense Variation	Pathogenic [PM3, PM2, PM5, PM1, PP3, PP2, PP5]	Reported
30	Asymptomatic Sibling	c.252 dup C; p. Arg85Pro fs*110	chrX-152990969T>TC	NM_000033.4	rs713993050	Exon 1	Nonsense Variation	Pathogenic [PV51, PM3, PM2, PP5]	Reported
31	Asymptomatic Sibling	c.443 [A>G] ;p.Asn148Ser	chrX-152991164 G>A	NM_000033.4	rs128624216	Exon 1	Missense Variation	Pathogenic [PM3, PM2, PM5, PM1, PP3, PP2, PP5]	Reported
32	Asymptomatic Sibling	c.649 [A>G] ;p.Lys217Glu	chrX-152991370 A>G	NM_000033.4	--	Exon 1	Missense Variation	Likely Pathogenic [PP3, PM2, PM5, PM1, PP2]	Reported
33	Asymptomatic Sibling	c.1488+1 [G>A] ;p.Val497fs*	chrX-153002706 G>A	NM_000033.4	--	Exon 5	Nonsense Variation	Pathogenic [PV51, PM2, PP5]	Reported
34	Asymptomatic Sibling	c.1488+1 [G>A] ;p.Val497fs*	chrX-153002706 G>A	NM_000033.4	--	Exon 5	Nonsense Variation	Pathogenic [PV51, PM2, PP5]	Reported
35	Asymptomatic Sibling	c.1488+1 [G>A] ;p.Val497fs*	chrX-153002706 G>A	NM_000033.4	--	Exon 5	Nonsense Variation	Pathogenic [PV51, PM2, PP5]	Reported

ALD, adrenoleukodystrophy; cc.ALD, childhood cerebral ALD; AMN, adrenomyeloneuropathy