

Table S1 Details about the variants detected in patients

Case number	pmid or doi	Genetic results	zygotic type
1	this study	chr1:1463113:NM_001170535.3:exon:14:c.1492dup(p.T498Nfs*13) (paternal)/ chr1:1463228:NM_001170535.3:exon:14:c.1376T>C(p.F459S)(maternal)	compound heterozygous
2	this study	chr1:1463113:NM_001170535.3:exon:14:c.1492dup(p.T498Nfs*13) (paternal)/ chr1:1463228:NM_001170535.3:exon:14:c.1376T>C(p.F459S)(maternal)	compound heterozygous
3	this study	chr1:1455655:NM_001170535.3:exon 6:c.649G>A(p.E217K)(paternal)/chr1:1447542-1452792:NM_001170535.3:exon 1-3 del(maternal)	compound heterozygous
4	doi:10.3760/cma.j.cmcr.2022.e02143	c.229C>G(p.Leu77Val)(paternal)/c.733_734delGC(p.Ala245fs*98)(maternal)	compound heterozygous
5	doi:10.7499/j.issn.1008-8830.2303077	c.517C>T(p.Q137*,462)(pertanal)	heterozygote
6	27640307	chr1:1464679:NM_001170535.1:c.1582C>T(p.Arg528Trp), Denovo	heterozygote
7	27640307	chr1:1464679:NM_001170535.1:c.1582C>T(p.Arg528Trp), Denovo	heterozygote
8	27640307	chr1:1464679:NM_001170535.1:c.1582C>T(p.Arg528Trp), Denovo	heterozygote
9	27640307	chr1:1464679:NM_001170535.1:c.1582C>T(p.Arg528Trp)(maternal)	heterozygote
10	27640307	chr1:1464679:NM_001170535.1:c.1582C>T (p.Arg528Trp),denovo; karyotype: 47, XXY	heterozygote
11	27640307	chr1:1447806:NM_001170535.1:c.158C>T [p.Thr53Ile] (paternal and maternal)	homozygote
12	27640307	chr1:1447806:NM_001170535.1:c.158C>T(p.Thr53Ile)(paternal and maternal)	homozygote
13	27640307	a heterozygous deletion spanning from ATAD3B to ATAD3A and a heterozygous deletion spanning from ATAD3C to ATAD3A. (~38 kb del ATAD3B-ATAD3A/~68 kb del ATAD3C-ATAD3A)	compound heterozygous
14	28158749	chr1:1459319:NM_001170535.1:c.1064G>A (p.G355D)	heterozygote
15	28158749	chr1:1459319:NM_001170535.1:c.1064G>A (p.G355D)	heterozygote
16	29053797	Chr1:1454346:NM_018188.3:c.634C>T(p.Gln212*)/ Chr1:1451416:NM_018188.4:c.230T>G(p.Leu77Arg)	compound heterozygous
17	29053797	Chr1:1454346:NM_018188.3:c.634C>T(p.Gln212*)/ Chr1:1451416:NM_018188.4:c.230T>G(p.Leu77Arg)	compound heterozygous
18	29053797	Biallelic ATAD3B/ATAD3A deletion	homozygote
19	29053797	Biallelic ATAD3B/ATAD3A deletion	homozygote
20	29053797	Biallelic ATAD3B/ATAD3A deletion	homozygote
21	29053797	Biallelic ATAD3B/ATAD3A deletion	homozygote
22	31239750	chr1:1451437:c.251C>T(p.Thr84Met) (paternal and maternal)	homozygote
23	31727539	NM_001170535.2:c.1217T>G(p.Leu406Arg)(paternal and maternal)	homozygote
24	31727539	NM_001170535.2:c.1217T>G(p.Leu406Arg)(paternal and maternal)	homozygote
25	31727539	NM_001170535.2:c.1217T>G(p.Leu406Arg)(paternal and maternal)	homozygote
26	32933822	NM_001170535:exon 15:c.1609T>A(p.Trp537Arg)(maternal)/NM_001170535:exon 15:c.1614+2_1616+16del(p.Arg503Profs*11)(paternal)	compound heterozygous
27	32933822	NM_001170535:exon 15:c.1609T>A(p.Trp537Arg)(maternal)/NM_001170535:exon 15:c.1614+2_1616+16del(p.Arg503Profs*11)(paternal)	compound heterozygous
28	32607449	NM_018188.4:c.528+3A>G	homozygote
29	28549128	ATAD3B/ATAD3A 38 054 bp deletion(paternal and maternal)	homozygote
30	28549128	ATAD3B/ATAD3A 38 054 bp deletion(paternal and maternal)	homozygote
31	28549128	ATAD3B/ATAD3A 38 667 bp deletion(paternal and maternal)	homozygote
32	28549128	ATAD3B/ATAD3A 38 667 bp deletion(paternal and maternal)	homozygote
33	28549128	ATAD3B/ATAD3A 38 054 and 38 667 bp deletions(paternal and maternal)	compound heterozygous
34	32004445	NC_000001.11:1456616_1524663dup,67 kb duplications in ATAD3A exon 11 and ATAD3C exon 7,de novo	heterozygote
35	32004445	NC_000001.11:1456616_1524663dup,67 kb duplications in ATAD3A exon 11 and ATAD3C exon 7,de novo	heterozygote
36	32004445	NC_000001.11:1456616_1524663dup,67 kb duplications in ATAD3A exon 11 and ATAD3C exon 7,de novo	heterozygote
37	32004445	NC_000001.11:1456616_1524663dup,67 kb duplications in ATAD3A exon 11 and ATAD3C exon 7,de novo	heterozygote
38	32004445	NC_000001.11:1456616_1524663dup,67 kb duplications in ATAD3A exon 11 and ATAD3C exon 7,de novo	heterozygote
39	33845882	Chr1:(1391729_1394541)_(1459777_1461841):NM_001170535.1:ATAD3C-ATAD3A del (~67 KB)(maternal)/Chr1:(1413926_1414584)_(1452593_1453251),NM_001170535.1:ATAD3B-ATAD3A del (~38 kb)(paternal)	compound heterozygous
40	33845882	Chr1:(1416206_1416369)_(1454260_1454423):NM_001170535.1:ATAD3B-ATAD3A del(~ 38kb)/Chr1:(1420558_1420661)_(1458286_1458389):NM_001170535.1:ATAD3B-ATAD3A del(~ 38kb)	compound heterozygous
41	33845882	Chr1:(1416206_1416369)_(1454260_1454423):NM_001170535.1:ATAD3B-ATAD3A del(~ 38 kb)(maternal)/NM_001170535.1:c.229C>G(p.Leu77Val) (paternal)	compound heterozygous
42	33845882	NM_001170535.1:c.150C>G(p.Phe50Leu)(maternal)/chr1:1451952_1453386:NM_001170535.1:exon 3-4:c.(282+1_283-1)_(444+1_445-1)del(paternal)	compound heterozygous
43	33845882	NM_001170535.1:c.150C>G(p.Phe50Leu)(maternal)/chr1:1451952_1453386:NM_001170535.1:exon 3-4:c.(282+1_283-1)_(444+1_445-1)del(paternal)	compound heterozygous
44	33845882	NM_001170535.1:c.1141dup(p.Val381Glyfs*17)(maternal)/NM_001170535.1:c.508C>T(p.Arg170Trp)(not maternal; paternal DNA not available)	compound heterozygous
45	33845882	NM_001170535.1:c.1141dup(p.Val381Glyfs*17)(maternal)/NM_001170535.1:c.508C>T(p.Arg170Trp)(not maternal; paternal DNA not available)	compound heterozygous
46	33845882	NM_001170535.1:c.1141dup(p.Val381Glyfs*17)(maternal)/NM_001170535.1:c.508C>T(p.Arg170Trp)(not maternal; paternal DNA not available)	compound heterozygous
47	33845882	NM_001170535.1:c.1414del(p.His472fs) (maternal)/NM_001170535.1:c.707G>T(p.Gly236Val) (paternal).	compound heterozygous
48	33845882	NM_001170535.1:c.1703_1705delAGA (p.Lys568del)(maternal) /NM_001170535.1:c.150C>G:(p.Phe50Leu) (de novo).	compound heterozygous
49	33845882	NM_001170535.1:c.980G>C(p.Arg327Pro)(paternal and maternal)	homozygote
50	33845882	NM_001170535.1:c.980G>C(p.Arg327Pro)(paternal and maternal)	homozygote
51	33845882	NM_001170535.1:c.980G>C(p.Arg327Pro)(paternal and maternal)	homozygote
52	34387651	c.1582C>T(p.Arg528Trp),de novo	heterozygote
53	34387651	c.1064G>A(p.Gly355Asp),de novo	heterozygote
54	34387651	c.1582C>T(p.Arg528Trp),de novo	heterozygote
55	34387651	c.1582C>T(p.Arg528Trp),de novo	heterozygote
56	36061954	Chr1:(1416465_1417219)_(1454520_1455274):NC_000001.10:ATAD3B/3A del(38 kbp ATAD3B/3A del)(paternal)/ NM_001170535:exon 6:c.646_664del(p.Ala216Serfs*235)(19 bp del in ATAD3A exon 6) (maternal)	compound heterozygous
57	36061954	Chr1:(1416465_1417219)_(1454520_1455274):NC_000001.10:ATAD3B/3A del(38 kbp ATAD3B/3A del)(paternal)/ NM_001170535:exon 6:c.646_664del(p.Ala216Serfs*235)(19 bp del in ATAD3A exon 6) (maternal)	compound heterozygous
58	36061954	Chr1:(1416465_1417219)_(1454520_1455274):NC_000001.10:ATAD3B/3A del(38 kbp ATAD3B/3A del)(paternal)/ NM_001170535:exon 6:c.646_664del(p.Ala216Serfs*235)(19 bp del in ATAD3A exon 6) (maternal)	compound heterozygous
59	33575671	chr1:1392270_1460317dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7;de novo	heterozygote
60	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
61	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
62	33575671	Chr1:1392294_1460341dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
63	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C: intron11/intron7	heterozygote
64	33575671	Chr1:1392560_1460670dup:NC_000001.10:ATAD3A/ATAD3C: exon12/exon8	heterozygote
65	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
66	33575671	Chr1:1395419_1462718dup:NC_000001.10:ATAD3A/ATAD3C:intron13/intron9	heterozygote
67	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
68	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
69	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
70	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
71	33575671	Chr1:1391576_1459623dup:NC_000001.10:ATAD3A/ATAD3C:intron10/intron6	heterozygote
72	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
73	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
74	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
75	33575671	Chr1:1391996_1460043dup:NC_000001.10:ATAD3A/ATAD3C:intron11/intron7	heterozygote
76	this study	Chr1p36.33:[(1386018-1414136)dup;(1417507-1422059)del;(1447638-1452803)del;(1455510-1459788)dup](73.7kb)(paternal)/Chr1p36.33:(1417507-1452803)del(35.3kb)(maternal)	compound heterozygous
77	this study	Chr1p36.33:[(1386018-1414136)dup;(1417507-1422059)del;(1447638-1452803)del;(1455510-1459788)dup](73.7kb)(paternal)/Chr1p36.33:(1417507-1452803)del(35.3kb)(maternal)	compound heterozygous
78	35012852	c.368G>A; de novo	heterozygote
79	36856321	NM_018188.5:c.624_644del(p.Glu209_Glu215del)(paternal and maternal)	homozygote
80	37095554	NM_001170535.3:exon 2:c.229C>G(p.Leu77Val)(paternal)/a deletion of exons 3 and 4 in the ATAD3A gene(maternal)	compound heterozygous
81	37095554	NM_001170535.3:exon 2:c.229C>G(p.Leu77Val)(paternal)/a deletion of exons 3 and 4 in the ATAD3A gene(maternal)	compound heterozygous
82	37095554	NM_001170535.3:exon 2:c.229C>G(p.Leu77Val)(paternal)/a deletion of exons 3 and 4 in the ATAD3A gene(maternal)	compound heterozygous
83	37095554	NM_001170535.3:exon 2:c.229C>G(p.Leu77Val)(paternal)/a deletion of exons 3 and 4 in the ATAD3A gene(maternal)	compound heterozygous
84	37031571	c.251T>C(p.Thr84Met)(paternal)/chr1:1417480-1451489:exon 1-2:a 3.6 kb deletion on chromosome 1p36.33(maternal)	compound heterozygous
85	38173481	NM_018188.4:exon 14:c.1517A>C(p.Q506P)(maternal)	heterozygote
86	38173481	NM_018188.4:exon 14:c.1517A>C(p.Q506P)	heterozygote
87	33675208	Chr1p36.33:1404883_1451417:duplication of at least 47 kb within 1p36.33 involving the ATAD3 gene cluster;de novo	heterozygote
88	28549128	ATAD3C/ATAD3B deletion(paternal); ATAD3B/ATAD3A genomic rearrangement(maternal)	compound heterozygous