Table 2. Clinical and Genetic Details of the Cohort.

ID	Age onset (months)	Microcephaly	GMFCS	MACS	CFCS	Gene	Mutation 1	AminoAcid detail mutation 1	Significance	Mutation 2	AminoAcid detail mutation 2	Significance	Age data collection
1	NA	Y	V	V	V	TREX1	c.635delC	p. Thr212fs+2x	P	c.635delC	p. Thr212fs+2x	P	25-48
2	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.592G>A	p.Glu198Lys	P	0-12
3	0-6	N	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.416delC	p.Ala139Valfs*21	P	49-72
4	0-6	Y	V	V	V	TREX1	NA	p.Ala194fs	P	c.506G>A	p.Arg169His (p.Arg114His)	P	0-12
5	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.341G>A	p.Arg114His	P	49-72
6	NA	N	NA	NA	NA	TREX1	c.52G>C	p.Asp18His	P	–	–	_	>120
7	0-6	N	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.667G>A	p.Ala223Thr	LP	25-48
8	0-6	Y	V	V	V	TREX1	c.553G>A	p.Asp185Asn	LP	c.1079A>G	p.Tyr360Cys	LP	13-24
9	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.907A>C	p.Thr303Pro	LP	>120
10	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.341G>A	p.Arg114His	P	0-12
11	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.341G>A	p.Arg114His	P	25-48
12	0-6	N	V	V	V	TREX1	c.655C>T	p.Arg219*	P	c.779C>T	p.Leu260Pro	VUS/LP	49-72
13	0-6	Y	V	V	V	TREX1	c.457_458insA	p.Gln153fs	P	c.457_458insA	p.Gln153fs	P	0-12
14	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.667G>A	p.Ala223Thr	LP	25-48
15	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.341G>A	p.Arg114His	P	0-12
16	0-6	Y	V	V	V	TREX1	c.341G>A	p.Arg114His	P	c.341G>A	p.Arg114His	P	73-96
17	0-6	Y	V	V	V	TREX1	c.262insAG	p.Ser88Lysfs*	P	c.290G>A	p.Arg97His	LP	>120
18	NA	Y	V	V	V	RNASEH2B	c.179T>G	p.Leu60Arg	LP/P	c.529G>A	p.Ala177Thr	P	97-120
19	7-12	N	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	>120
20	7-12	N	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	>120
21	0-6	Y	V	V	V	RNASEH2B	c.136ivs+1delG	–	P	c.529G>A	p.Ala177Thr	P	>120
22	0-6	Y	V	V	V	RNASEH2B	c.136ivs+1delG	–	P	c.529G>A	p.Ala177Thr	P	>120

Table 2. Clinical and Genetic Details of the Cohort.

23	0-6	N	V	II	III	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.128C>A	p.Pro43His	LP	97-120
24	0-6	N	V	NA	NA	RNASEH2B	c.488C>T	p.Thr163Ile	P	c.2T>C (Initiator Codon)	–	P	13-24
25	0-6	N	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	25-48
26	13-24	N	V	III	IV	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	49-72
27	0-6	N	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	25-48
28	13-24	NA	NA	NA	NA	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	25-48
29	0-6	Y	I	I	I	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	13-24
30	13-24	N	IV	IV	IV	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.510+1G>A	–	P	13-24
31	13-24	N	V	III	V	RNASEH2B	c.510+1G>A	–	P	c.529G>A	p.Ala177Thr	P	13-24
32	6-12	N	II	II	II	RNASEH2B	c.132T>A	p.Cys44*	P	c.529G>A	p.Ala177Thr	P	25-48
33	7-12	N	IV	IV	NA	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	13-24
34	0-6	Y	V	NA	NA	RNASEH2B	c.132T>A	p.Cys44*	P	c.529G>A	p.Ala177Thr	P	0-12
35	0-6	Y	V	V	V	RNASEH2B	c.428_434delAGGAAAA	p.Glu144Valfs*5	P	c.529G>A	p.Ala177Thr	P	25-48
36	7-12	N	III	III	IV	RNASEH2B	c.128C>A	p.Pro43His	LP	c.529G>A	p.Ala177Thr	P	25-48
37	0-6	Y	V	V	V	RNASEH2B	c.3G>A	p.Met1?	P	c.412C>T	p.Leu138Phe	LP	25-48
38	0-6	N	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.488C>T	p.Thr163Ile	P	13-24
39	0-6	N	I	I	I	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	>120
40	0-6	Y	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	Exon 2-5 deletion	–	P	97-120
41	0-6	Y	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.488C>T	p.Thr163Ile	P	>120
42	0-6	Y	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.488C>T	p.Thr163Ile	P	>120
43	0-6	N	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	>120

Table 2. Clinical and Genetic Details of the Cohort.

44	0-6	Y	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	>120
45	0-6	N	IV	IV	III	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	>120
46	0-6	N	V	V	IV	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	>120
47	0-6	Y	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	p	c.488C>T	p.Thr163Ile	P	>120
48	0-6	Y	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.3G>A	p.Met1?	P	>120
49	0-6	Y	V	V	V	RNASEH2B	c.554T>G	p.Val185Gly	LP/P	c.554T>G	p.Val185Gly	LP/P	25-48
50	0-6	Y	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	97-120
51	7-12	N	V	IV	III	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	25-48
52	13-24	N	V	V	V	RNASEH2B	c.529G>A	p.Ala177Thr	P	c.529G>A	p.Ala177Thr	P	25-48
53	0-6	N	V	V	V	RNASEH2A	c.703C>T	p.Arg235Trp	VUS/LP	c.869A>G	p.Glu290Gly	VUS/LP	49-72
54	0-6	Y	V	V	V	RNASEH2A	c.622C>T	p.Ser208Pro	VUS/LP	c.622C>T	p.Ser208Pro	VUS/LP	13-24
55	7-12	N	IV	III	IV	RNASEH2A	c.557G>A	p.Arg186Gln	LP	c.557G>A	p.Arg186Gln	LP	25-48
56	0-6	N	III	III	I	RNASEH2A	c.152A>G	p.Tyr51Cys	VUS/LP	c.746C>T	p.Ala249Val	VUS/LP	25-48
57	0-6	Y	III	IV	V	RNASEH2A	c.623C>T	p.Ser208Leu	VUS	c.760T>C	p.Trp254Arg	VUS	25-48
58	0-6	Y	V	V	V	RNASEH2C	c.451C>T	p.Pro151Ser	P	c.450G>A	p.Trp150*	P	25-48
59	0-6	y	NA	NA	NA	RNASEH2C	c.472C>G	p.His158Asp	VUS	c.155T>G	p.Ile52Ser	VUS	0-12
60	0-6	Y	V	V	V	SAMHD1	Exon 1 deletion	–		C.1456A>T	p.Lys186*	P/LP	0-12
61	0-6	Y	V	V	V	SAMHD1	c.490C>T	p.Arg164*	P/LP	5.80kb exon 1 deletion	–		13-24
62	0-6	Y	V	V	V	SAMHD1	chr20:35,578,204-35,583,998	–		chr20:35,578,204-35,583,998	–		>120
63	7-12	N	I	I	I	SAMHD1	chr20:35,578,204-35,583,998	–		chr20:35,578,204-35,583,998	–		>120
64	0-6	Y	V	V	V	SAMHD1	Exon 1 deletion	–		c.676C>G	p.Arg226Gly	VUS/LP	0-12
65	7-12	N	I	I	I	SAMHD1	Exon 1 deletion	–		Exon 1 deletion	–		>120

Table 2. Clinical and Genetic Details of the Cohort.

66	>24	N	I	I	I	SAMHD1	Exon 1 deletion	–		Exon 1 deletion	–		>120
67	0-6	N	I	I	I	SAMHD1	5 prime end deletion	–		5 prime end deletion	–		>120
68	0-6	N	IV	II	II	SAMHD1	c.602T>A	p.Ile201Asn	LP/P	c.1293A>T	p.Leu431Phe	VUS	73-96
69	0-6	Y	V	V	V	SAMHD1	c.1321dupG	p.Ala441Glyfs*9	LP	c.1343T>C	p.Ile448Thr	LP	13-24
70	7-12	Y	IV	III	IV	SAMHD1	c.1657T>G	p.Tyr553Asp	VUS	c.464A>G	p.Tyr155Cys	VUS	49-72
71	0-6	Y	V	V	IV	SAMHD1	c.1153A>G	p.Met385Val	P	c.1153A>G	p.Met385Val	P	73-96
72	NA	Y	V	V	V	ADAR1	c.3019G>A	p.Gly1007Arg	P	–	–	_	97-120
73	7-12	NA	NA	NA	NA	ADAR1	c.556C>T	p.Gln186*	P	c.577C>G	p.Pro193Ala	P	73-96
74	13-24	Y	V	IV	V	ADAR1	c.982C>T	p.Arg328*	P	c.577C>G	p.Pro193Ala	P	49-72
75	13-24	N	IV	III	III	ADAR1	c.577 C>G	p.Pro193Ala	P	c.3020-3C>G	IVS11-3C>G	LP	>120
76	0-6	Y	IV	IV	IV	ADAR1	c.3019G>A	p.Gly1007Arg	P	–	–	_	25-48
77	13-24	N	III	III	II	ADAR1	c.3019G>C	p.Gly1007Arg	P	–	–	_	49-72
78	0-6	N	V	V	IV	ADAR1	c.3577G>A	p.Glu1193Lys	LP	c.1493_1494delAG	p.Glu498Valfs*18	P	49-72
79	13-24	N	V	V	V	ADAR1	c.3019G>A	p.Gly1007Arg	P	–	–	_	>120
80	0-6	Y	V	V	V	ADAR1	c.3019G>A	p.Gly1007Arg	P	–	–	_	97-120
81	>24	N	I	I	I	ADAR1	c.3443+1G>C	–	LP	c.577C>G	p.Pro193Ala	P	73-96
82	0-6	Y	V	V	V	ADAR1	c.577C>G	p.Pro193Ala	P	c.2675G>A	p.Arg892His	P/LP	0-12
83	7-12	N	III	II	II	ADAR1	c.3019G>A	p.Gly1007Arg	P	c.2653G>A	p.Val885Ile	LP	25-48
84	0-6	N	I	NA	NA	ADAR1	c.3019G>A	p.Gly1007Arg	P	c.2653G>A	p.Val885Ile	LP	0-12
85	0-6	Y	V	V	V	ADAR1	c.577C>G	p.Pro193Ala	P	c. 2608 G>A	p.Ala870Thr	P/LP	97-120
86	7-12	Y	V	V	V	ADAR1	c.577G>T	p.Pro193Ala	P	c.1076_1080delAGCGA	p.Lys359Argfs	P/LP	97-120
87	7-12	Y	V	V	V	IFIH1	c.1009A>G	p.Arg337Gly	P	–	–	_	>120
88	0-6	N	V	V	IV	IFIH1	c.2342G>A	p.Gly781Glu	VUS/LP	–	–	_	49-72
89	7-12	Y	V	V	V	IFIH1	c.2159G>A	p.Arg720Gln	P	–	–	_	97-120

Table 2. Clinical and Genetic Details of the Cohort.

90	0-6	Y	IV	V	V	IFIH1	c.2335C>T	p.Arg779Cys	P	–	–	_	49-72
91	0-6	N	V	V	V	IFIH1	c.2336G>T	p.Arg779Leu	LP	–	–	_	49-72
92	0-6	Y	V	V	V	IFIH1	c.2159 G>A	p.Arg270Gln	VUS/LP	–	–	_	97-120
93	0-6	N	III	II	III	IFIH1	c.2336G>A	p.Arg779His	P	–	–	_	25-48
94	>24	N	I	I	I	IFIH1	c.2335C>T	p.Arg779Cys	P	–	–	_	97-120
95	0-6	Y	V	V	V	IFIH1	c.2936T>G	p.Leu979Trp	LP	–	–	_	0-12
96	13-24	N	I	I	I	IFIH1	c.2336G>A	p.Arg779His	P	–	–	_	49-72
97	13-24	N	I	I	I	IFIH1	c.2336G>A	p.Arg779His	P	–	–	_	25-48
98	0-6	N	V	V	V	IFIH1	c.2407A>T	p.Ile803Phe	VUS	–	–	_	13-24
99	7-12	N	V	IV	II	IFIH1	c.2471G>A	p.Arg824Lys	VUS/LP	–	–	_	>120
100	13-24	N	VI	III	I	IFIH1	c.2561T>A	p.Met854Lys	VUS/LP	–	–	_	>120

Abbreviations: CFCS, Communication Function Classification System; GMFCS, Gross Motor Function Classification System; LP, likely pathogenic; MACS, Manual Ability Classification System; P, pathogenic; VUS, variant of unknown significance.