Supplementary Table 2

Homozygous or compound heterozygous	Country	Sex	Age at onset, yrs	Age of death (*) or latest report	Mutation genomic DNA	Mutation cDNA	Mutation Protein	Growth failure	Low birth weight	Cahexia/bird-like facies	Mental retardation	Microcephaly	Cataracts	Retinal degeneration	Hearing Loss	Clinical photosensitivity	Dental abnormalities	Arthogryposis	Reference
CSB																			
Hom	China	M		16	c.526C>T	r.(526c>u)	p.(Arg176*)	+		+	+			+	+				[1]
Het	USA	M	4	10	c.[543+4delA];[2008C>T]	r.[423_543del (del exon 3)];[2008c>u]	p.[p.Ser142Asnfs*4;[*Arg670Trp*]	+			M	+		+	+				[2], patient 1
Het	USA	M	1.8	7	c.[543+4delA];[2008C>T]	r.[423_543del (del exon 3)];[2008c>u]	p.[p.Ser142Asnfs*4;[*Arg670Trp*]	+	-			+		+	+				[2], patient 2
Het	USA	F		6	c.[543+4delA];[2008C>T]	r.[423_543del (del exon 3)];[2008c>u]	p.[p.Ser142Asnfs*4;[*Arg670Trp*]	+	-			+		+	+				[2], patient 3
Hom	Amish, Ohio	F		2	c.2709+1G>T	r.?	p.?	+		+	+	+	+			+			[3] (family 1)
Het	Amish, Ohio	F			c.[1293_1320del];[2709+1G>T]	r.[(1293_1320del)];[?]	p.[(Glu432Lysfs*24)];[?]												[3] (family 2). No precise clinical description
Het	China	F			c.[1595A>G];[1607T>G]	r.[(1595a>g)];[(1607t>g)]	*p. [(Asp532Gly)] ; [(p.Leu536Trp)]*	+	-	+	S					+			[4]
Hom	Turkey	F		12.7	c.1992+3A>G	r.1911_1992del	p.Arg637Serfs*34	+		+	M					+			[5] (6 patients from the same family, intrafamilial variability)
Het	China?	F		12	c.[643G>T];[3776C>A]	r.[(643g>t)];[(3776c>a)]	p.[(Glu215*)];[(Ser1259*)]							+					[6]
Hom	?	M	2.5	22	c.2800C>A	r.(2800c>a)	p.*(Pro934Thr)*	+	-	+	+	+		-	+	+			[7], patient 1
Hom	South Africa	F	0	4	c.2808G>C	r.(2808g>c)	*p. (Trp936Cys)*		+	+	+	+	+		+		+	+	[7], patient 2
Het	China	M	0.25	8	c.[1834C>T];[2923C>T]	r.[(1834c>u)];[(2923c>u)]	*p.[(Arg612*)];[(Arg975Trp)]*		NA	+	+	+	+		-	-			[11], patient III.1
Het	China	M	<1	9	c.[1834C>T];[2923C>T]	r.[(1834c>u)];[(2923c>u)]	*p.[(Arg612*)];[(Arg975Trp)]*	NA	+	+	+	-	+						[11], patient III.2
Het	Canada	F	<0.5	4.5*	c.[del];[2779_2782delAACA]	r.[0];[(2779_2782delaaca)]	p.[0];[(Asn927Glufs*24)]	+	+	+	+	+	+	+			+		[12]
Het	China	F	1		c.[1387C>T];[*de novo* del]	r.[(1387c>t)];[0]	*p.[(Gln463*)];* [0]	+	-	+		+	-	-		+			[13]
CSA																			
Het	China	M	1	7	c.[394_398del];[551-2A>C]	r.[(394-398del)];[(?)]	p.[(Leu132Asnfs*6)];[(?)]	+	-	+	+	+			+	+	+		[8]
Het	China	F	1	14	c.[394_398del];[842_843+1delinsCTA]	r.[394-398del];[719_843del (exon 9 del)]	p.[Leu132Asnfs*6];[Ala240Glyfs*8]	+		+	+	+			+		+		[9]
Het	China	M		11	c.[394_398del];[842_843+1delinsCTA]	r.[394-398del];[719_843del (exon 9 del)]	p.[Leu132Asnfs*6];[Ala240Glyfs*8]												[9] No precise clinical description.
Het	China	M	0	13	c.[618-2A>G];[complex deletion/inversion/deletion]	r.[618_626del (del first 9 nt of exon 8)];[276_399del (exon 4 del)]	p.[Ala207_Ser209del];[Asp93Leufs*26]	+			+	+		+	+	+	+		[10], patient II.6
Het	China	M		3	c.[618-2A>G];[complex deletion/inversion/deletion]	r.[618_626del (del first 9 nt of exon 8)];[276_399del (exon 4 del)]	p.[Ala207_Ser209del];[Asp93Leufs*26]	+			+	+			+	+	+		[10], patient II.10

The Table shows mutations reported by other groups since 2011. Previously reported cases are listed in [14]

References for Supplementary Table 2

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