Title: Charcot-Marie-Tooth Neuropathy Type 4H *GeneReview* Table 3

Author: Delague V

Date: August 2013

Note: The following information is provided by the author listed above and has not been reviewed by *GeneReviews* staff.

Table 2. Clinical, Electrophysiologic, and Morphologic Characteristics of CMT4H in 18 Individuals from 13 Families

Patient	Origin	Consanguinity	Mutations in genomic DNA	Mutations in amino acid sequence	Status	Age at first symptoms (years)	Age at last examination (years)	Age at walking (months)	Distal weakness[a]	Distal muscle atrophy[b]
Ia	Lebanese	yes	c.893 T>G	p.Met298Argfs*8	homozygous	1 to 2 years	15	delayed, 15 to 36 months	+++	++
Ib	Lebanese	yes	c.893 T>G	p.Met298Argfs*8	homozygous	1 to 2 years	18	delayed, 15 to 36 months	+++	++
Ic	Lebanese	yes	c.893 T>G	p.Met298Arg	homozygous	4 years	13	12	+++	++
II	Algerian	yes	c.893 T>C	P.Met298Thr	homozygous	2 years	UN	UN	++	++
III	Turkish	yes	c.670 C>T	p.Arg224*	homozygous	<1 year	30	delayed	+++	++
IV	Turkish	yes	c.1626_1627delAG	p.Glu543Glyfs*5	homozygous	2 years	UN	delayed, 26 months	++	+
V	Tamil	sporadic	c1756 G>T	p.Gly586*	homozygous	9 years	UN	16	+	+
Via	Northern Ireland	yes	c.823 C>T	p.Arg275*	homozygous	Childhood (difficulty to run and poor balance)	58	UN	+	+
VIb	Northern Ireland	yes	c.823 C>T	p.Arg275*	homozygous	Childhood (clumsiness)	50	UN	++	UN
VII	Italian	yes	c.1762-2 A>G	p.Tyr587fs*14	homozygous	<1 year	20	17	+	+
VIII	Lebanese	yes	c.1698 G>A	p.Met566Ile	homozygous	5 years	21	14	+	+
IX	Algerian	yes	c.1325 G>A	p.Arg442His	homozygous	UN	UN	UN	UN	UN
Xa	Tunisian	yes	c.514_515insG	p.Ala172Glyfs*27	homozygous	3 years	6	16	+++	++
Xb	Tunisian	yes	c.514_515insG	p.Ala172Glyfs*27	homozygous	3 years	18	normal	+++	++
Xc	Tunisian	yes	c.514_515insG	p.Ala172Glyfs*27	homozygous	3 years	22	normal	+++	++
XI	Japanese	yes/no	c.1888_1892delAAAGG	p.Lys630Asnfs*5	homozygous	Childhood	UN	UN	UN	UN
XII	Japanese	yes/no	c.[837-2A>G + 1132+1G>A]	p.[Trp279fs* + Tyr355fs*2]	compound heterozygous	Birth	UN	11	UN	UN
XIII	Japanese	No/unknown	c.837-1G>A	p.Glu280Lysfs*23	homozygous	4 years	UN	14	UN	UN

UN = unknown

[a]. - : not affected; + = mild in the lower extremities; ++ = marked in the lower extremities; +++ = also affected the hands and forearms

[b]. - = not affected; + = mild; ++ = severe

[c]. - = no deformities; + = pes cavus and hammer toes; ++ = pes equinus and toes retraction

[d]. - = none; + = mild; ++ = severe; +++ = surgery required

[e]. - = absent; + = present

[f]. - = no deficit; + = decreased sensibility; +++ = no sensibility

[g]. NR = non recordable

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Table 2. Clinical, Electrophysiologic, and Morphologic Characteristics of CMT4H in 18 Individuals from 13 Families (continued)

Patient	Foot deformities[c]	Scoliosis[d]	Tendon reflexes [e]	Distal sensory loss [f]	Progression of disease	Functional impairment	Motor NCVs [g]				Sensory NCVs [g]			References
Ia	++	+++	-	++	Slow	moderate to severe: unsteady gait, walking without aid,	NR	…	NR	NR	NR	NR	…	Delague et al, 2007
Ib	++	-	-	++	Slow	mild: unsteady gait, walking without aid,	NR	NR	NR	…	NR	NR	NR	Delague et al, 2007
Ic	+	++	-	++	Slow	UN	…	7	…	NR	…	…	NR	Stendel et al, 2007
II	+	+	-	UN	Slow	moderate, walking without aid, waddling gait	SR				NR			Delague et al, 2007
III	+	-	-	+	Slow	UN	5	…	…	…	NR	…	NR	Stendel et al, 2007
IV	-	-	-	-	Slow	UN	10	6.6	…	…	NR	…	…	Stendel et al, 2007
V	-	-	-	-	Slow	UN	12	…	…	NR	NR	…	…	Stendel et al, 2007
VIa	+	-	-	++	Slow	moderate : walking without aid at 58	8	13	…	…	NR	NR	…	Houlden et al, 2009
VIb	+	-	-	++	Slow	severe: at 50, walking with two crutches or wheelchair	6	…	…	…	…	…	…	Houlden et al, 2009
VII	+	+	-	+	Slow	moderate : unsteady gait with steppage	6	8	NR	NR	NR	NR	NR	Fabrizi et al, 2009
VIII	+	-	-		Slow	moderate: walking without aid	14	15	NR	NR	23	NR	NR	Baudot et al, 2012
IX	UN	UN	UN	UN	UN	UN	…	…	…	…	…	…	…	Baudot et al, 2012
Xa	+	+	-	-	Slow	walking on her tiptoes	14	…	NR	…	…	…	NR	Boubaker et al, 2013
Xb	+	++	-	+	Slow		…	…	…	…	…	…	…	Boubaker et al, 2013
Xc	+	+++	-	+	Slow	spine surgery at age 16	9	…	NR	…	…	…	NR	Boubaker et al, 2013
XI	+	UN	UN	UN	Slow	Walked without assistance until 65 years. Severe gait disturbance from 68 years	9.6	…	…	…	…	…	…	Hayashi et al, 2013
XII	+	UN	UN	UN	Slow	Abnormal gait from 3 years	NR	…	…	…	…	…	…	Hayashi et al, 2013
XIII	UN	UN	UN	UN	Slow	Frequent falls from age 4 years. Walked limp from 6 years	8.8	…	…	…	…	…	…	Hayashi et al, 2013

UN = unknown

[a]. - : not affected; + = mild in the lower extremities; ++ = marked in the lower extremities; +++ = also affected the hands and forearms

[b]. - = not affected; + = mild; ++ = severe

[c]. - = no deformities; + = pes cavus and hammer toes; ++ = pes equinus and toes retraction

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