

On-line Table: Clinical and genetic data on patients with CHARGE

Patient No.	Major				Minor				Final Clinical Typology ^a	CHD7 Gene Testing	Protein Variant	Mutation Type	Clinical vs Genetic Diagnosis
	Ocular	Choanal	Cranial Nerve	Ear	Genital	DD	CHD	GD	Orofacial Clefing	TEF	Facies		
1	+	+	+	+	+	+	+	+	Definite	NA			Clinical
2	+		+	+	+		+	+	Definite	NA			Clinical
3	+		+	+	+	+	+	+	Probable	c.632G>A	p.Gly2108Arg	Missense	Genetic
4	+	+	+	+	+	+	+		Definite	ISCN array result: -arr[hg19] 8q12.2(61,757,599–61,805,579)x1		Interstitial deletion	Genetic
5	+	+	+	+	+	+	+		Definite	c.3523-2A>C		Splice site	Genetic
6	+		+	+	+	+		+	Definite	NA			Clinical
7			+	+	+		+		Probable	NA			Clinical
8	+	+	+	+	+		+		Definite	Negative			Clinical
9	+		+	+	+	+	+		Definite	c.7957C>T	p.Arg2653X	Stop-gain	Genetic
10	+		+	+	+		+	+	Definite	NA			Clinical
11	+		+	+	+		+	+	Definite	c.799G>T	p.Glu267X	Stop-gain	Genetic
12	+	+	+	+	+	+	+	+	Definite	c.4105C>T	p.Arg139X	Stop-gain	Genetic
13			+	+	+	+	+	+	Probable	c.5405-7G>A		Splice site	Genetic
14			+	+	+	+	+	+	Probable	Negative			Clinical
15			+	+	+	+	+	+	Probable	Positive		NOS	Genetic
16	+		+	+	+	+	+	+	Definite	c.760C>T	p.Gln254X	Stop-gain	Genetic
17	+		+	+	+		+	+	Definite	Positive		NOS	Genetic
18	+	+	+	+	+	+	+	+	Definite	c.3728dupA	p.Asni243LysfsX20	Frameshift	Genetic
19	+		+	+	+	+	+	+	Definite	NA			Clinical
20	+		+	+	+		+		Definite	Negative			Clinical
21			+	+	+		+		Negative	Positive		NOS	Genetic
22	+		+	+	+	+	+	+	Definite	c.2066delC	p.Thr689SerfsX22	Frameshift	Genetic
23	+		+	+	+	+	+	+	Probable	NA			Clinical
24		+	+	+	+	+	+	+	Definite	NA			Clinical
25	+		+	+	+	+	+		Definite	NA			Clinical
26	+	+	+	+	+	+	+		Definite	ISCN array result: arr[hg19] 8q12.1q12.2(61,585,541–61,613,066)x1		Interstitial deletion	Genetic
27	+		+	+	+	+	+	+	Definite	c.5350_5374dup25	p.Lys1792ArgfsX5	Frameshift	Genetic
28			+	+	+	+	+	+	Probable	Negative			Clinical
29			+	+	+		+	+	Probable	NA			Clinical
30	+		+	+	+		+	+	Definite	c.469 C>T	p.Arg157X	Stop-gain	Genetic
31	+		+	+	+	+	+	+	Definite	c.5607 + 1G>A		Splice site	Genetic
32	+		+	+	+	+	+		Definite	NA			Clinical
33	+	+	+	+	+	+	+	+	Definite	NA			Clinical
34		+	+	+	+	+	+	+	Probable	c.4247C>G	p.Thr1416Arg	Missense	Genetic
35	+	+	+	+			+		Probable	NA			Clinical

Note:—DD indicates developmental delay; CHD, congenital heart disease; GD, growth deficiency; TEF, tracheoesophageal fistula; NOS, not otherwise specified; NA, not applicable; +, present; ISCN, international system for human cytogenetic nomenclature.

^a Final clinical typology (after Lalani, et al¹³). Definite: 4 major or 3 major/3 minor criteria; Probable/Possible: 1–2 major/3+ minor criteria.