

Supplementary Table S1. The conditions for PCR amplification of the *MED13L* gene in gDNA and cDNA samples: primer sequences, location, annealing temperature, and lengths of the amplicons.

Gene (RefSeq transcript)	Sample	Primers (5' → 3')	Location	Annealing temperature	Amplicon length
<i>MED13L</i> (NM_015335.4, NG_023366.1)	cDNA	F: CGGGAATCAAATGGCGTAGG	2 exon	59 °C	953 bp
		R: AGCACTCTGCATACCTCCAC	8 exon	59,5 °C	
		F: CGGGAATCAAATGGCGTAGG	2 exon	59 °C	1262 bp
		R: CCCTGGTTGAGATACTGTGGG	10 exon	59,8 °C	
		F: CGGGAATCAAATGGCGTAGG	2 exon	59 °C	2524 bp
		R: GGTGGAGTGGGAAACATCCT	15 exon	59,3 °C	
	cDNA (Gene expression assay)	F: CGAGCCTGGAGGATTGTCA	1 exon	59,1 °C	118 bp
		R: GCTGGGGCTGAAATTATGGG	2 exon	59 °C	
		TaqMan™ probe: ACCTCTTTTCGCTGGCTGAACTCAC	1–2 exon junction	61,2 °C	
	gDNA (CRISPR-Cas9 assay)	F: CTCCCACCTTAAAATTCGAGAAGA	1 intron	58,5 °C	559 bp
		R: GGGGTGAGAATGTTCTTTTGAG	2 intron	59,5 °C	

F – forward primer; R – reverse primer.

Supplementary Table S2. Clinical features of individuals with pathogenic DNA sequence variants or CNVs of the *MED13L* gene.

Individual	ID	Psychomotor development al delay	Speech delay	Craniofacial deformity	Anomalies of hands and/or feet	Hypotonia	Ophthalmologica l anomalies	Cerebral anomalie s	Congenit al heart defects	Autistic behaviou r
The proband	+	+	+	–	+	–	+	–	–	–
Literature (n = 79) ²⁰⁻²⁷	77/77	79/79	77/78	58/75	34/74	54/74	27/75	27/60	14/70	18/72
12q24.21 deletion (DECIPHER)	#264232	NA	+	NA	+	NA	NA	+	NA	NA
	#260719	NA	+	NA	+	NA	+	NA	NA	NA
	#271765	NA	+	NA	+	NA	NA	NA	NA	NA
	#271595	+	+	NA	+	NA	NA	NA	NA	+
	#285418	+	NA	NA	NA	NA	NA	NA	NA	NA
	#283932	NA	+	+	+	NA	NA	+	NA	NA
	#277527	NA	+	+	+	NA	NA	+	NA	NA
	#272267	NA	+	NA	+	NA	NA	NA	+	NA
	#296332	NA	+	NA	+	+	+	NA	NA	NA
	#328479	NA	+	+	+	NA	NA	NA	NA	NA
	#322949	NA	+	+	+	NA	+	+	NA	NA
	#363856	+	+	+	NA	NA	+	NA	NA	NA
	#307630	NA	+	+	+	NA	+	NA	NA	NA
	#401741	+	+	+	NA	NA	+	NA	NA	+
12q24.21 duplication	#296364	+	+	+	NA	NA	NA	NA	NA	NA
	#296365	NA	+	NA	+	NA	NA	NA	NA	NA
	#272268	Na	+	+	+	NA	NA	NA	NA	NA
	Total number	83/83	97/97	87/88	70/87	41/81	57/78	35/83	28/62	15/72

Total number	100	100 %	98,9 %	80,5 %	50,6 %	73,1 %	42,2 %	45,2 %	20,8 %	26,7 %
(%)	%									

NA - no data available.