

Table S1. Craniofacial anomalies of 13 cases in this cohort

	P1	P2	P3	P4	P5	P6	P7	P8	P9	P10	P11	P12	P13	
Craniofacial anomalies														12/12
Microcephaly	-	-	+	-	-	-	-	-	-	-	-	-	-	1/13
Triangular face	+	-	-	-	-	-	-	-	-	-	-	+	+	3/13
Abnormality of the hairline	-	-	-	-	+	-	+	+	-	-	-	-	-	3/13
Highly arched eyebrow/Broad eyebrow	+, broad eyebrow	-	-	-	+	-	+	+	+	+	-	-	+	7/13
Thick eyebrow/synophrys	-	-	-	-	+	+	+	+	+	-	-	-	-	6/13
Thick eyelashes	+	-	-	-	-	-	+	+	+	-	-	-	-	4/13
Hypertelorism	-	-	-	-	+	-	-	+	-	+	+	-	-	4/13
Epicanthus	-	-	-	-	+	-	-	+	+	+	+	-	-	5/13
Ptosis	+	+	+	+	-	-	+	-	-	-	-	-	-	5/13
Wide nasal bridge	-	+	-	-	-	-	+	+	-	-	-	-	+	4/13
Anteverted nares	-	+	-	-	-	+	+	+	-	-	-	-	+	5/13
Bulbous nose	-	-	-	-	-	-	-	+	-	-	-	-	+	2/13
Underdeveloped nasal alae	-	-	-	-	-	-	-	+		-	-	-	-	1/13
Short columella	-	-	-	-	+	-	-	-	-	-	-	-	-	1/13
Long/Smooth philtrum	+, long philtrum	-	-	-	-	-	-	+, Smooth philtrum	+	-	-	+	+	5/13
Thin upper lip	-	-	-	-	-	-	-	+	-	-	-	+	-	2/13
Cupid bow upper lip	-	-	-	-	-	-	-	+	-	-	-	-	+	2/13
Micrognathia	-	+	-	+	-	+	-	-	+	-	-	+	+	5/13
Preauricular skin tag	+	-	-	-	-	-	-	-	+	-	-	-	-	1/13
Preauricular pit	+	-	-	-	-	-	-	+	-	-	-	-	-	2/13
Abnormal location of the ear	-	-	-	+, Auricle supination	+, Asymmetry of the ears	-	-	-	-	+, Asymmetry of the ears	-	-	-	3/13

+, Positive Phenotype; -, Negative Phenotype; NA, Data non-available.

