

Supplemental material. Description of the craniofacial features of the 31 POLR3-HDL subjects in this study's cohort																		
Subject	Gene	High anterior hairline	High forehead	Bitemporal narrowing	Hypertelorism	Telecanthus	Long palpebral fissures	Low-set ears	Flat midface	Pinched nose	Bulbous tip of the nose	Short philtrum	Smooth philtrum	Thin upper lip	Full lower lip	Short chin	Pointed chin	Other dysmorphic features (observed in <10%)
1	POLR3A	+			+	+	+					+	+				+	Large mouth, bilateral syndactyly 2-3 toes
2	POLR3A			+	+		+		+	+				+				
3	POLR3A								+	+		+						+
4	POLR3A				+			+									+	Chin dimple
5	POLR3A						+	+	+		+	+					+	
6	POLR3A	+	+						+	+	+						+	Sparing of lateral eyebrows
7	POLR3A	+	+		+				+			+					+	Upturned nose
8	POLR3A								+				+				+	
9	POLR3A	+	+						+				+		+			Deviating nasal root, retrognathia
10	POLR3A			+	+							+				+		Short neck
11	POLR3A	+	+							+		+					+	Triangular face
12	POLR3A								+		+		+				+	
13	POLR3A	+								+			+				+	Ptosis, bifid tip of the nose
14	POLR3A	+	+	+					+			+	+					Ptosis, downslanting palpebral fissure, mildly overfolded helixes, prognathism
15	POLR3A								+	+		+		+	+			Hypotelorism, broad nasal tip, low-hanging columella
16	POLR3A	+	+		+								+		+		+	Spaced medial eyebrows, broad nasal bridge
17	POLR3B	+					+		+			+	+	+	+	+		Broad and flat nasal bridge
18	POLR3B									+			+					Depressed nasal bridge, retrognathia
19	POLR3B								+	+	+		+	+			+	
20	POLR3B							+	+	+								Deviating nasal root
21	POLR3B	+				+						+	+	+	+		+	Large ears
22	POLR3B				+		+		+				+	+	+		+	Short nose
23	POLR3B	+	+			+			+						+	+		Inferior epicanthus
24	POLR3B	+	+				+		+				+	+				Hockey-stick palmar creases
25	POLR3B	+	+				+			+			+	+				Hockey-stick palmar creases
26	POLR3B		+		+			+				+	+				+	Copped ears, low hanging columella
27	POLRIC			+		+		+	+				+		+		+	Long eyelashes, long philtrum, inferior epicanthus
28	POLRIC			+	+							+						Downslanted palpebral fissure, large mouth, short neck, abnormal external ears
29	POLRIC										+	+		+			+	Peri-orbital fullness, low-hanging columella
30	POLRIC								+	+			+				+	Broad nasal bridge, retrognathia
31	POLR3A (+/- POLR3B)	+	+				+	+	+	+	+	+	+	+	+	+	+	Epicanthal fold
		14/31 (45.2%)	11/31 (35.4%)	5/31 (16.1%)	9/31 (29.0%)	4/31 (12.9%)	7/31 (22.6%)	6/31 (19.4%)	19/31 (61.3%)	11/31 (35.5%)	6/31 (22.6%)	12/31 (38.7%)	18/31 (58.0%)	9/31 (29.0%)	8/31 (25.8%)	6/31 (19.4%)	16/31 (51.6%)	
		15/31 with an anomaly of the forehead (48.4%)			16/31 with an anomaly of the eyes (51.6%)					15/31 with an anomaly of the nose (48.4%)		26/31 with an anomaly of the philtrum (83.9%)		13/31 with an anomaly of the lip (41.9%)		22/31 with an anomaly of the chin (71.0%)		