

Table S1: Detailed clinical evaluation of CD patients included in the study.

Clinical findings	Patients								
	1	2	3	5	5	6	7	8	9
Prominent frontal bone	+			+					
Prominent occipital bone			+	+	+	+		+	
Flat face				+	+		+	+	+
Low-set ears	+			+	+				
Epicanthal folds			+	+					
Midface hypoplasia	+						+		
Arched/high palate			+	+					
Cleft palate		+			+	+		+	
Bulbous nose			+	+					
Flat nasal bridge	+	+	+						
Inverted nares	+					+	+		
Long philtrum			+	+	+				
Micro/retrognathia	+	+	+	+	+	+	+	+	
Pierre-Robin sequence					+				
Short/wide neck	+			+	+	+	+		
Excessive neck skin			+						+
Short trunk	+					+			+
Wide thorax				+			+		
Mammillary hypertelorism				+		+	+		
Inverted nipples				+			+		

	Patients [continued]								
Clinical findings	1	2	3	4	5	6	7	8	9
Short limbs	+	+	+	+	+	+	+	+	+
Bowing of lower limbs	+	+	+	+	+	+	+	+	+
Pretibial skin dimples	+		+	+	+	+	+	+	+
Nail hypoplasia	+							+	
Club feet	+	+	+	+	+		+	+	