TABLE I. Clinical Features and Mutations Identified Among 15 Patients With Cornelia de Lange Syndrome

Clinical data	Cases														
	CdL1	CdL2	CdL3	CdL4	CdL5	CdL6	CdL7	CdL8	CdL9	CdL10	CdL11	CdL12	CdL13	CdL14	CdL15
IUGR		+		+	+	+		-	+	+	-	+	-		+
Postnatal growth retardation		+		+	+	+		+	+	+	+	+	+		+
Mental retardation		+		+	+	+		+	+	+	+	+	+		+
Hirsutism		+		+	+	+		-	+	+	+	+	+		-
Synophrys		+		+	+	+		+	+	+	+	+	+		+
Long, curly eyelashes		+		+	+	+		+	+	+	+	+	-		+
Microbrachycephaly		+		+	+	+		+	+	+	+	+	+		+
Down turned corners of mouth		+		+	+	+		+	+	+	+	+	+		+
Phocomelia		+		+	-	-		-	-	-	-	-	-		-
Oligodactyly		+		+	-	-		-	+	-	-	-	-		+
Ulnar deficiency		+		+	-	-		-	-	-	-	-	-		+
Cutis marmorata		-		-	-	+		-	+	-	-	-	-		-
Gastrointestinal malformation		-		GER	-	-		-	-	+*	-	+**	-		-
Cardiovascular malformation		+		-	-	-		-	-	-	ASD	-	ASD,TR		+
Hearing impairment		+		+	+	+		-	+	+	-	nd	nd		nd
Parental age at birth															
Paternal age (years)		27		34		33		32	42	42	27				29
Maternal age (years)		22		31		32		33	40	31	22				26
Deletion by FISH	-	nd	-	nd	nd	-	-	-	-	-	-	-	-	nd	nd
NIPBL mutation	-	1921G>T (E641X)	-	1885C>T (R629X)	-	-	-	-	-	5483G>A (R1828Q)	-	-	-	-	3346G>T (E1116X)

IUGR: intrauterine growth retardation; GER: gastroesophageal reflux; ASD: atrial septal defect; TR: tricuspid regurgitation; nd: not determined; * : pyloric stenosis,; ** : diaphragmatic hernia; +: positive; -: negative. Detailed clinical information was not available for CdL1, CdL3, CdL7, or CdL14.