

Table 1.

Characteristic	Present case	Ray et al. (1981)	Lobo et al. (1992)	Holden et al. (1985)	Zenger-Hain et al. (1993)	Shapiro et al.(1985)A	Davis et al.1988)	Mori et.(1988)	Glover et al. (1987)	Farell et al. (1993)	Shapiro et al. (1985)B	Van de Vooren et al.(1984)	Rooney et al. (1989)
Breakpoint	q11.23q22.11	q11q12	q11.1q22.1	q11.2q21	q11.2q22.1	q11.2q22.1	q21q22	q22q24	q22q24	q22.3q23.2	q22.4q24	q24.2q25.3	q25.2q26.1
Growth retardation	-	-	-	-	-	-	-	-	-	-	-	-	-
Psychomotor retardation	+	-	+	+	-	+	+	+	-	-	-	+	+
Hypotonia	+	+	+	+	+	+	+	+	+	+	-	-	+
Cranial shape	Plagiocephaly	Plagiocephaly	-	-	-	-	-	Macrocephaly	Plagiocephaly	Dolichocephaly	-	-	Microcephaly
Broad forehead/ Frontal bossing	+	-	+	-	+	+	-	+	-	-	+	-	-
Hypertelorism/ Telecanthus	-	-	+	-	+	-	+	+	+	-	+	-	+
Strabismus	+	-	+	-	-	+	+	-	-	-	-	+	-
Upslant palpebral fissures	-	+	-	-	+	-	-	-	-	+	+	-	-
Broad/Flat nasal bridge	+	-	+	-	-	+	+	+	+	-	+	-	+
Anteverted nares	-	-	-	-	-	-	-	+	+	+	+	-	-
Prominent/ Long philtrum	+	-	+	-	+	-	-	-	+	-	-	-	+
Lowset/ Malformed ears	+	+	+	+	+	+	+	+	+	-	+	-	-
Thin/Cupid bow upper lip	+	+	+	-	-	?	-	-	-	-	-	-	+
High arched palate	-	-	+	-	-	-	+	+	-	-	-	-	-
Cleft palate	-	-	-	+	-	-	-	-	-	-	-	-	-
Microgenitalia	+	-	-	-	-	-	-	-	-	-	-	-	-
Widespaced/ Hypoplastic nipples	-	-	+	-	-	-	-	-	+	+	+	-	-
Spinal deformity	-	-	-	-	-	-	-	+	-	-	+	+	-

Pectus excavatum	-	-	-	-	-	-	-	-	-	-	+	+	-
Sacral dimple	-	+	+	-	-	-	-	+	-	-	+	-	-
Microgenitalia	+	+	-	-	-	-	-	-	-	+	-	-	-
Cryptorchidism	-	-	-	-	-	-	-	-	-	-	-	+	-
Hypoplastic labia majora	-	-	-	-	-	-	-	-	-	-	-	-	+
Bifid uvula	-	-	-	-	-	+	-	+	-	-	-	-	-
Vesicourethral reflex	-	-	-	-	-	-	-	+	-	-	-	-	-
Rectal prolapse	-	-	-	-	-	-	-	-	-	-	+	-	-
Umbilical/Hiatal hernia	-	-	-	-	-	-	-	-	-	-	+	-	-
Clinodactyly	-	-	-	-	-	-	-	-	-	-	-	+	-
Equinovarus/Planovalgus	-	-	-	-	-	-	-	+	-	-	-	+	-
Joint limitation	-	-	-	-	-	-	-	-	-	-	-	-	-
Joint deformity	-	-	-	-	-	+	-	-	-	-	+	-	-
Heart defect	VSD	Murmur	VDS	-	Murmur	VSD	Murmur	Murmur	Murmur	Murmur	VSD	-	-