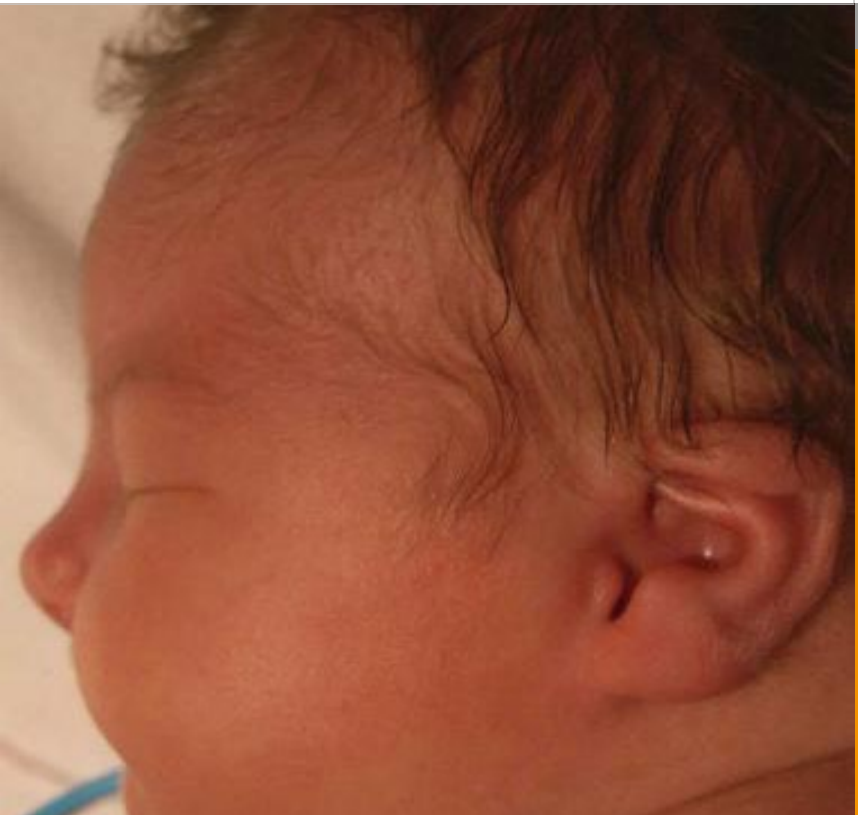
SWISS SOCIETY OF NEONATOLOGY

Chromosome 13q deletion syndrome



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SYNONYMS

Orbeli syndrome, 13q- syndrome, 13q deletion syndrome, chromosome 13q monosomy, del(13q) syndrome, deletion 13q syndrome, monosomy 13q, partial monosomy 13q.

CASE REPORT

This female infant was born at 38 4/7 weeks of gestation following in vitro fertilization and a pregnancy complicated by intrauterine growth restriction. Delivery and adaption were unremarkable. Birth weight was 2370 g, lenghth 43 cm, and head circumference 30 cm. Apart from symmetric growth retardation, the following clinical findings were noted: decreased muscle tone (Fig. 1) and absent suck reflex, microcephaly, small ears, microphthalmos, hypertelorism, short philtrum, prominent nasal bridge, high arched palate, bilateral cataracts (Fig. 2, 3), short metacarpals bilaterally, left hand with 4 fingers, right hand with 5 fingers but a hypoplastic thumb (Fig. 4 – 9).

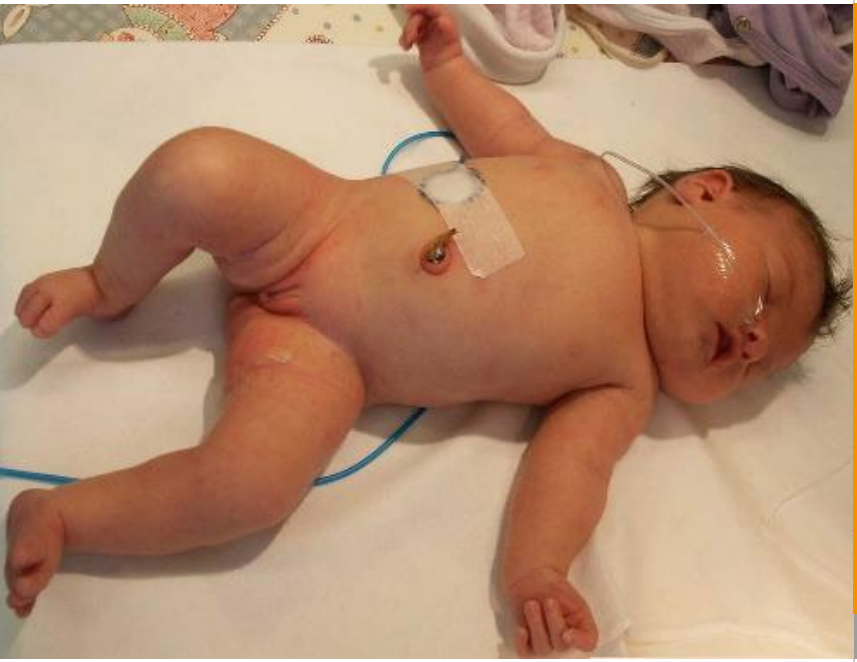

Fig. 1

Muscular hypotonia.

Fig. 2

Small malformed ears.

Fig. 3

Hypertelorism and short filtrum.

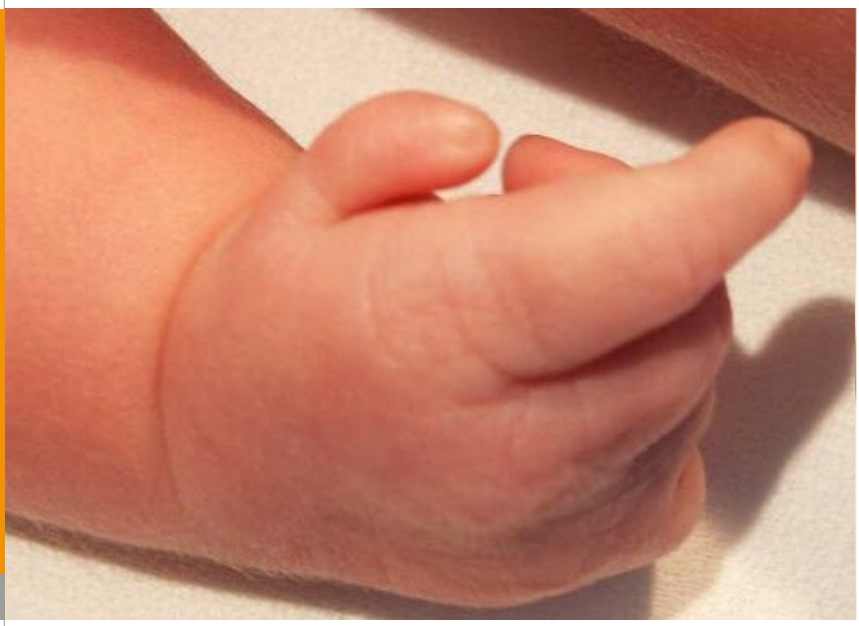

Fig. 4

Hypoplastic thumb.

Fig. 5

Left foot with four toes.

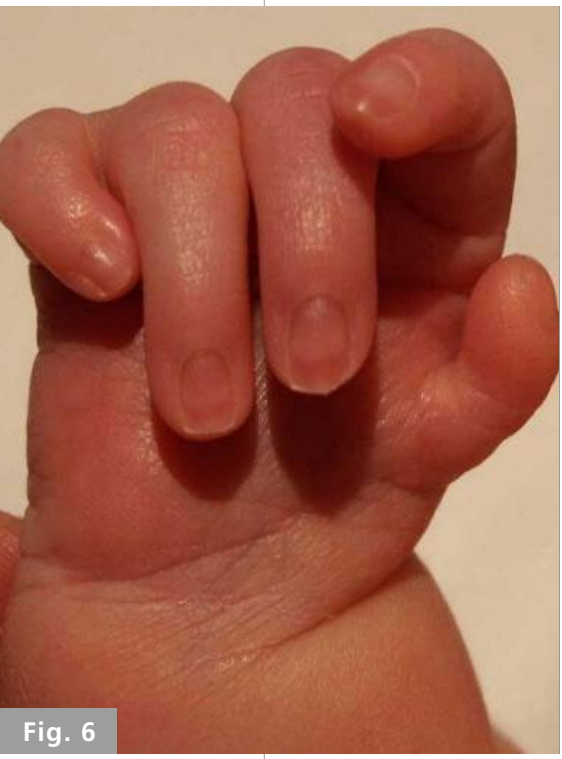

Fig. 6

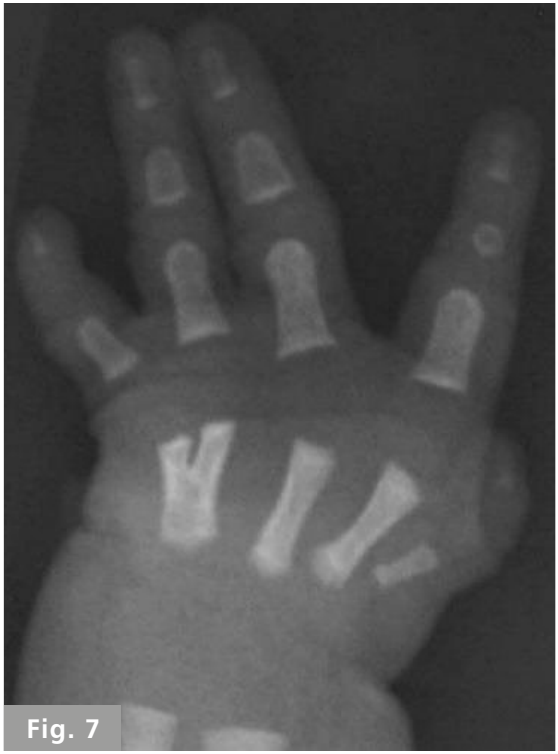

Fig. 7

Hypoplastic thumb. *X-ray right hand*

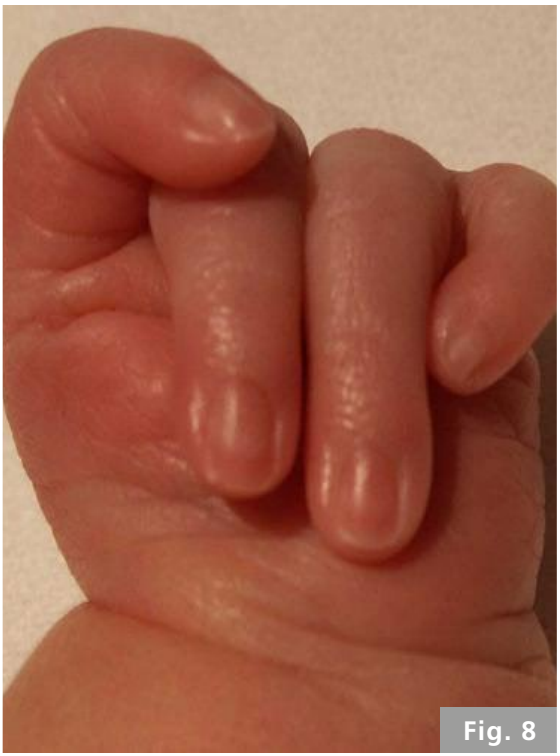

Fig. 8

Four digits.

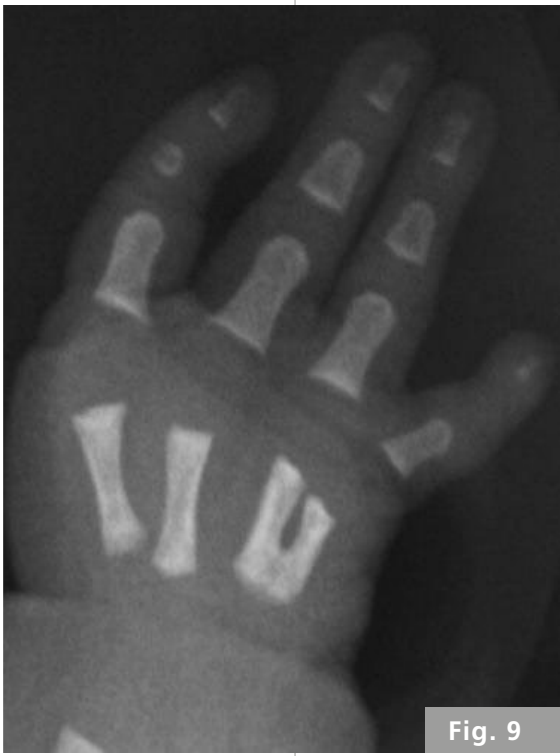

Fig. 9

X-ray left hand

Ambiguous genitalia with partially non-attached labia minora and non-patent introitus (Fig. 10). On chest X-ray non-union of the third thoracic vertebra was noted (Fig. 11). Ultrasound revealed hypoplasia of the corpus callosum (Fig. 12, 13), hypoplastic kidneys (Fig. 14), and absent uterus and ovaries. An atrial septal defect, ventricular septal defect, and patent ductus arteriosus were demonstrated on echocardiography. Chromosome analysis of the patient showed 46, XX, del(13)(q31), while chromosome analyses of the parents were normal.

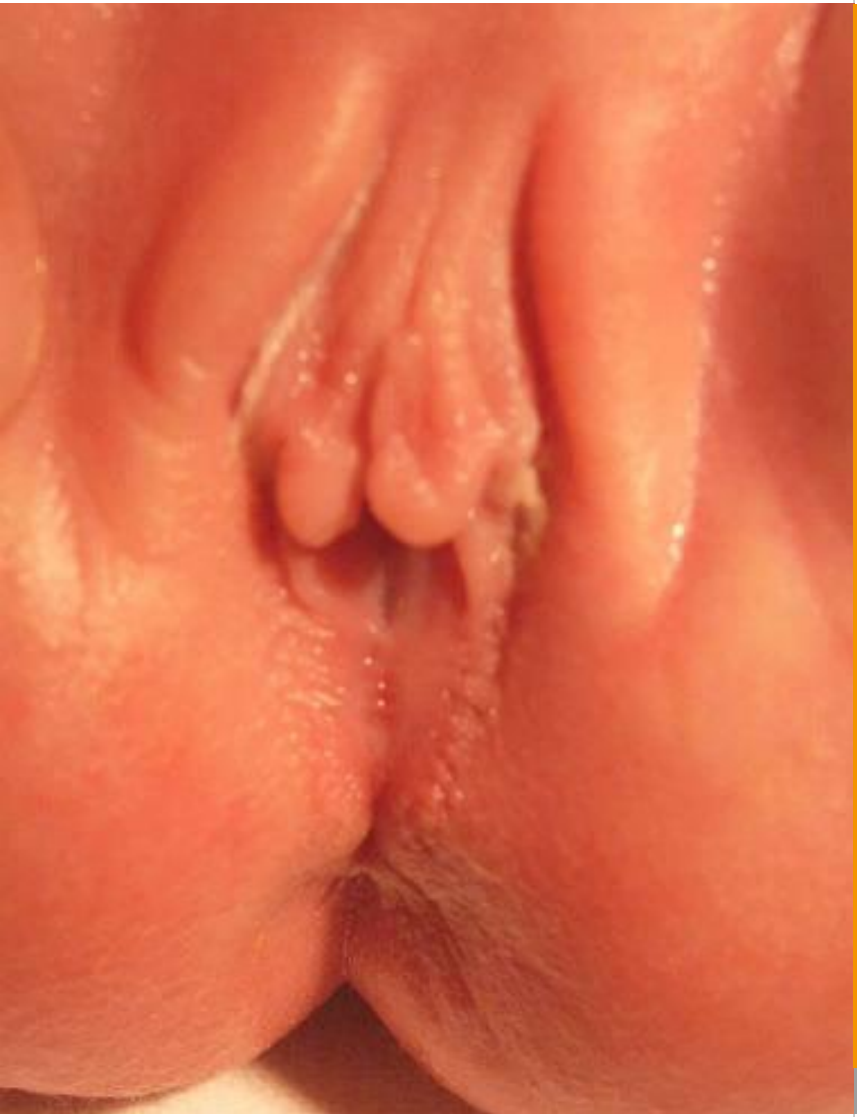

Fig. 10

Non-patent introitus.

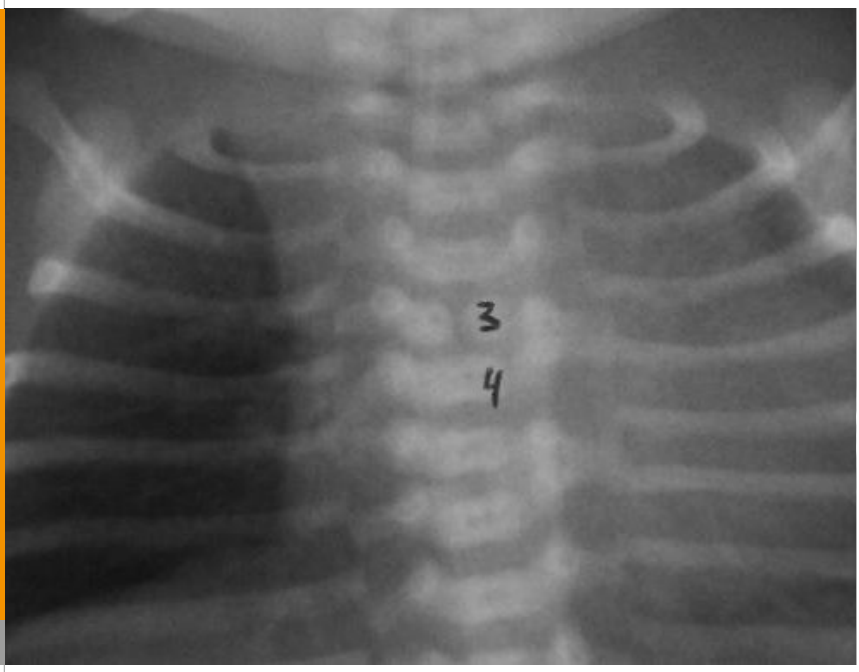

Fig. 11

Chest X-ray: non-union of 3rd thoracic vertebral body.

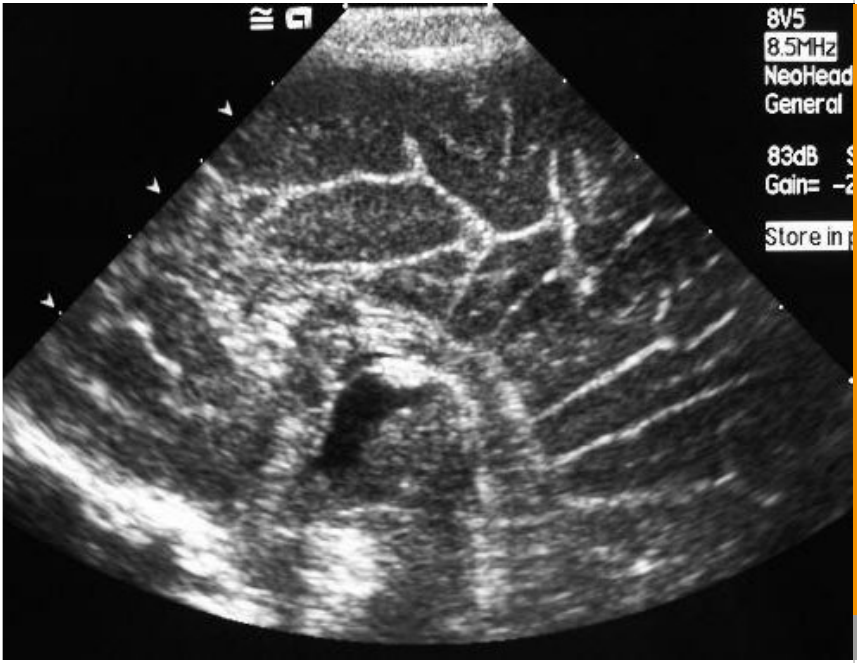

Fig. 12

Cerebral ultrasound: hypoplastic corpus callosum.

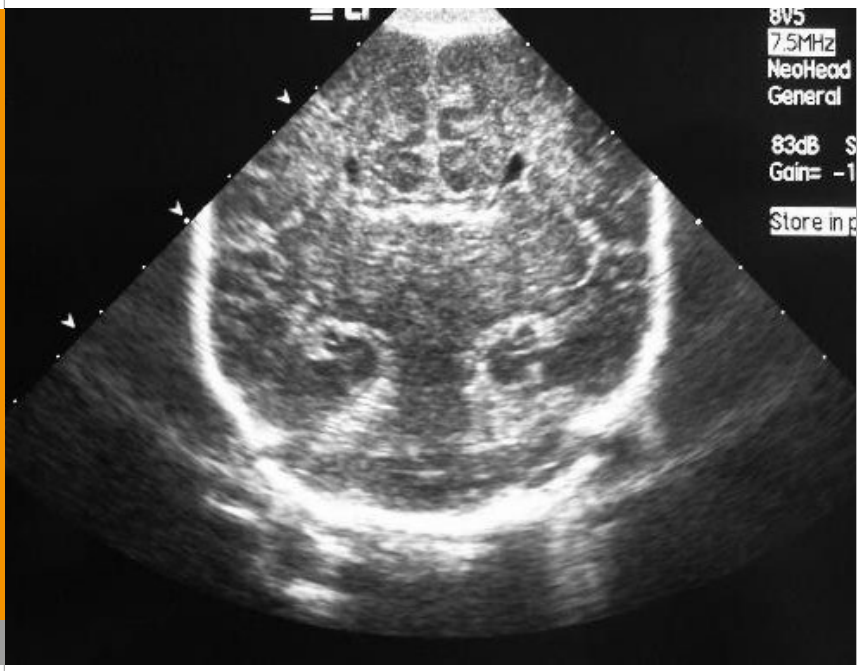

Fig. 13

Cerebral ultrasound: hypoplastic corpus callosum.

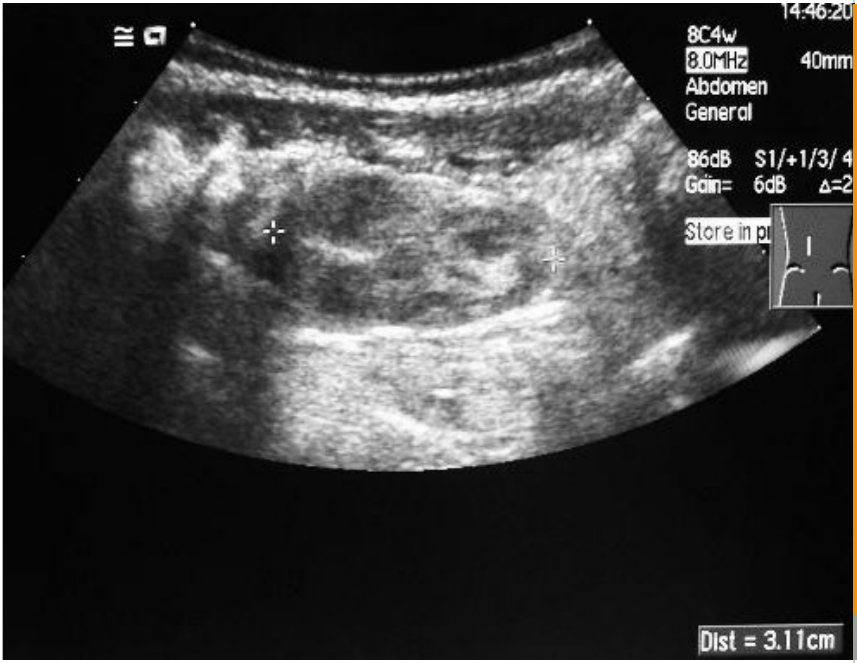

Fig. 14

Abdominal ultrasound: hypoplastic kidneys.

DISCUSSION

Deletion of the long arm of chromosome 13 is associated with a wide spectrum of abnormalities, including retinoblastoma, mental and growth retardation, brain malformations, heart defects, distal limb deformities, digestive, urogenital, and other abnormalities. Deletions limited to proximal bands (q13-q31) are characterized mainly by growth retardation but no major deformities, those involving band 32q are usually associated with numerous major malformations, and distal deletions are usually complicated by severe mental retardation with comparatively minor abnormalities. Garcia-Lurie syndrome and this disorder share many common clinical features.

Deletions include: q13-qter (mat), q14-q32, q14-ter, q14.3-q31.2, q21.2-q32, q22-q31, q22-q32, q22-q34, q22-qter, q31.2-q33, q31.2q34 q31.3-qter, q31-qter, q32.3-q33.2, q32-qter.

The most common features of this syndrome and their frequencies are listed in the table. Only cases with accurate banding were included in this table. Column A refers to cases involving deletion of 13q3, column B refers to cases of ring chromosome 13.

Table. Features of chromosome 13 deletion syndrome (Adapted from Mary Louise Buyse. Birth Defects Encyclopedia 1990, p 367-368)	A	B
General		
Low birth rate (< 2500g)	5/12	11/14
Growth failure	6/9	11/11
Psychomotor retardation	12/12	15/15
Craniofacial		
microcephaly	8/12	15/15
brachycephaly	6/8	-
trigonocephaly	1/5	3/14
facial asymmetry	3/9	1/11
hypertelorism	5/12	8/11
upslanting palpebral fissures	9/12	4/10
coloboma	2/11	3/10
broad, abnormal or prominent nasal bridge	8/10	15/15
large or abnormally formed ears	9/11	11/14
short philtrum, protruding maxilla/upper incisors	5/9	5/9
Congenital heart disease	2/10	3/10
Renal abnormalities (renal agenesis or hypoplasia)	-	3/7
Genital abnormalities (cryptorchidism, hypospadia)	4/9	6/9
Anal atresia	-	1/15

Monosomy 13q3 may arise as a result of a deletion or may occur as a result of abnormal segregation of a parental translocation. Of the cases in which parental karyotypes are reported, it appears that approximately 60% result from parental translocations. Rings are generally de novo events, however cases have been described in association with translocations. The male to female ratio is 1:1. There is no specific treatment. Psychomotor retardation, usually moderate to severe, is always present. The exact life span is unknown, the oldest patient included in the above survey is 8 years old.

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