Short report

Journal of Medical Genetics 1989, **26**, 280–281

Gastrointestinal abnormalities in the syndrome of mosaic trisomy 9

The proband, an 18 month old child, was delivered by caesarian section after an uneventful 38 week gestation, birth weight 2400 g. She was the fourth daughter of healthy, non-consanguineous parents aged 43 and 40 years at the time of birth. The family history was unremarkable.

From the age of six months, the child has been examined repeatedly for clinical and radiological findings of upper and lower respiratory tract infections. At the age of 18 months she was admitted to our department because of massive right lower lobe pneumonia. Physical examination showed a child with severe failure to thrive and moderate psychomotor retardation whose weight was 7500 g (below the 3rd centile), height 71 cm (below the 3rd centile), and head circumference 45 cm (25th centile). The sagittal suture and anterior fontanelle were widely open. She had bilateral microphthalmia, deep set eyes, short palpebral fissures, broad nasal bridge, and bulbous nose. The palate was highly arched and the ears were low set and posteriorly rotated. No heart murmurs were audible. She also had acrocyanosis with distal mottling and non-pitting oedema of both legs. Adduction of both hips was limited to 20°. Neurological assessment showed generalised marked hypotonia and hyporeflexia of the deep tendon reflexes. She could neither sit by herself nor stand or crawl. Her fine motor performance and speech capability were retarded.

Skull x ray showed a widely open fontanelle and sagittal suture. Chest x ray showed right sided aortic arch with infiltrates of the right lower lobe and thickening of the bronchial tree. Pelvic x ray showed bilateral coxa valga. CT scan of the brain and renal ultrasound were normal. Echocardiography showed right aortic arch with no other cardiac anomalies.

A barium swallow was regurgitated up to the oral cavity and aspirated, indicating that the reflux and malfunctioning of the swallowing mechanism could be the underlying cause of the repeated pulmonary infections. Kidney and liver function tests, as well as endocrinological and sweat tests, were within normal limits.

Cytogenetic studies performed on peripheral blood lymphocytes, cultured with phytohaemagglutinin for 72 hours, indicated that 70 out of 100 cells (70%) had a normal female karyotype (46,XX) and 30 cells (30%) were trisomic for chromosome 9, 47,XX,+9 (figure). Mosaic trisomy 9 is a rare chromosomal aberration. Only 23 cases of this trisomy have been previously reported, some of them complete, and some of them with mosaic trisomy 9. The affected children show failure to thrive, multiple craniofacial anomalies, musculoskeletal, cardiac,

renal, and central nervous system malformations, as well as psychomotor retardation and early death.^{1–3} The clinical features of our patient and those previously described are summarised in the table.

There are two major anomalies in the present case which we believe to be previously unreported: severe velopharyngeal insufficiency and gastro-oesophageal reflux leading to recurrent pulmonary infections. We do not know, at this point, if the gastrointestinal anomalies are part of the syndrome or merely coincided with the trisomy

TABLE Features of reported cases of trisomy 9 compared with present case.

	Present case	No of reported cases
Craniofacial anomalies		
Microcephaly	–	8/23
Wide fontanelles and sutures	+	12/23
Microphthalmia	+	9/23
Deep set eyes	+	8/23
Short palpebral fissures	+	12/23
Bulbous nose	+	12/23
Wide nasal bridge	+	6/23
High arched palate	+	6/23
Cleft palate	–	5/23
Low set, posteriorly rotated ears	+	16/23
Micrognathia	–	10/23
Short neck	–	7/23
CNS anomalies		
Subependymal cysts of lateral ventricle	–	2/23
Hydrocephalus	–	2/23
Congenital heart anomalies		
Atrial septal defect	–	6/23
Ventricular septal defect	–	10/23
Patent ductus arteriosus	–	5/23
Right aortic arch	+	1/23
Genitourinary malformations		
Hypoplastic external genitalia	–	3/23
Cryptorchidism	–	4/23
Cystic dilatation of renal tubules	–	2/23
Diverticulae of the bladder	–	2/23
Hydronephrosis and hydroureters	–	6/23
Musculoskeletal anomalies		
Congenital dislocation of the hip	–	11/23
Hypoplastic pelvic bones	–	5/23
Anomalous rib cage	–	4/23
Flexion contractures of joints	–	7/23
Bilateral coxa valga	+	1/23
Club foot	–	4/23
Short stature	+	4/23
Punctate mineralisation of joints	–	1/23
Skin and adnexa		
Non-pitting oedema of legs	+	3/23
Acrocyanosis	+	1/23
Simian crease	–	8/23
Distal mottling of the skin	+	1/23
Nail hypoplasia	–	2/23
Gastrointestinal tract		
Gastro-oesophageal reflux	+	0/23

Received for publication 9 August 1988.

Revised version accepted for publication 16 September 1988.

FIGURE G banded karyotype of a cell with mosaic trisomy 9.

9, since these anomalies have also been noted in otherwise apparently healthy children. To elucidate this point more cases need to be reported.

I LEVY, Y LEVY, Z MAMMON,
M NITZAN, AND R STEINHERZ

Department of Paediatrics A, Beilinson Medical Centre
and Tel Aviv University Sackler
School of Medicine,
Israel.

References

- ¹ Feingold M, Atkins L. A case of trisomy 9. *J Med Genet* 1973; **10**:184-7.
- ² Lewandowski RC, Yunis J. Trisomy 9 mosaicism. *Clin Genet* 1977; **11**:306-10.
- ³ Kaminker CP, Dain L, Lamas MA. Mosaic trisomy 9 syndrome with unusual phenotype. *Am J Med Genet* 1985; **22**:237-41.

Correspondence to Dr Itzhak Levy, Department of Paediatrics A, Beilinson Medical Centre, Petah Tiqva 49 100, Israel.