Atroventricular Canal Associated with Trisomy 9

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The features of a newborn with the full clinical aspect of trisomy 9 presenting with an atroventricular canal is described. This association of anomalies has never been reported before. Interestingly, the patient also had a left-sided obstruction which is known to be more characteristically associated with atrioventricular canal without Down's syndrome.

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AVC = atroventricular canal

The genetic anomaly most frequently associated with AVC is Down's syndrome. In order to establish the prevalence of chromosomal anomalies in this congenital heart defect we performed a prospective cytogenetic study on a consecutive series of 172 patients with AVC. Less than half of these patients (78 = 45.3 percent) had normal karyotypes with or without phenotypic anomalies, while 94 patients (54.6 percent) had Down's syndrome with homogenous or mosaic trisomy 21. Two additional chromosomal anomalies were detected in this sample. One was a pericentric inversion of chromosome 1, which was segregating from a healthy father, the other was a trisomy 9, which is reported here in detail.

CASE REPORT

A male infant was admitted to our hospital at 2 days of age because of multiple congenital anomalies and congestive heart failure. He was the product of a 38-week gestation of a normal 28-year-old mother and a 30-year-old father. Both parents were phenotypically normal and unrelated. The patient had a normal sister. Family history was unremarkable. Pregnancy was uncomplicated and delivery was by cesarian section because of an early detachment of placenta. Birth weight was 2,600 g, length, 42 cm; head circumference, 33 cm (all below the third centile). Physical examination (Fig 1) revealed microbrachycephaly, frontal bossing, with a depressed medial part of frontal bone which was covered with a fine lanugo, asymmetric face, thin, poorly furnished eyebrows, small palpebral fissures, enophthalmos, bulbous nasal tip, well-defined philtrum, microstomia, with a small inverted upper lip overriding the lower, which was retracted, because of a severely hypoplastic and receding mandible. Ears were low set and mildly slanted with an incompletely overfolded scapha helix, a prominent and tortuous antihelix, hypoplastic tragus and lobe. External genitalia were severely hypoplastic and testes were not palpable.

Humeri and femora were rather short; hands were broad with clinodactyly of fifth fingers. There was bilateral pes cavus with retracted toes.

Cardiac Assessment

A grade 2 systolic murmur was heard at the left sternal border. There was mild cyanosis, tachycardia (180/m), tachypnea (60/m), liver enlargement and poor peripheral pulses. The chest x-ray film showed cardiac enlargement with increased pulmonary blood flow and the ECG showed left axis deviation and right ventricular hypertrophy. With two-dimensional echocardiography the definitive diagnosis of complete AVC type e' was made. In addition, there was right ventricular dominance with a hypoplastic left ventricle and ascending aorta (Fig 2). The clinical condition rapidly worsened in spite of vigorous medical support, and the patient died at 3 days of age. Permission for autopsy was not granted by the parents.

Cytogenetic Study

Chromosome analysis was performed on short-term lymphocyte cultures. The modal number was 47 in all 65 examined cells. Giemsa-banded cells showed that the extra chromosome was a number 9. Thus, the karyotype was 47,XY,+9 (Fig 3). The parents had normal chromosome constitutions.
Our patient is the 12th observation of an apparent nonmosaic trisomy 9 newborn. However, since only lymphocytes were examined, a mosaic trisomy 9 aneuploidy cannot be ruled out. On clinical examination our patient had most of the outstanding features currently found in association with this aneuploidy, including facial dysmorphia, genital anomalies, heart malformations and distal limb abnormalities.

Congenital heart defects have been reported in all patients with complete trisomy 9. However, the association between AVC and trisomy 9 had not been recognized so far.

In general, AVC is very common in trisomy 21 patients, but it is rare in subjects with different types of chromosomal aberrations. In addition, we have previously reported that the non-Down’s syndrome patients with AVC have a prevalence of left-sided obstructive lesion as associated cardiac malformation. This pattern is opposite the one currently found in Down’s syndrome patients that rarely presented left-sided obstruction.

Furthermore, in the former group of patients, which in general have normal karyotype, we demonstrated an increased frequency of minor anomalies, compared with a group of patients with different types of congenital heart defects. Interestingly, the patient reported here, had an AVC with a left-sided obstruction, which further supports the assumption that the AVC pattern found in trisomy 21 is rather peculiar. However, additional prospective studies are needed to improve current understanding of the association between AVC and chromosomal imbalances.

REFERENCES

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