CASE REPORT

A case of 49,XXXXX in which the extra X chromosomes were maternal in origin

Y G Cho, D S Kim, H S Lee, S C Cho, S I Choi

This report describes an 11 month old female baby with features of pentasomy X. A molecular and cytogenetic evaluation revealed that her karyotype was 49,XXXXX and her extra X chromosomes were of maternal origin. She has muscular hypotonia, mental retardation, a cleft palate, mild hydrocephalus as a result of dilatation of both lateral ventricles, hyperextensible elbow joints, proximal radioulnar synostosis, clinodactyly of the fifth finger, valgus of the feet, and small hands and feet. In addition, she has a persistent pupillary membrane and congenital chorioretinal atrophy. The pathogenesis of pentasomy X is not clear at present, but it is thought to be caused by successive maternal nondysjunctions.

Pentasomy X is a rare chromosomal abnormality that affects girls only, and is characterised by the presence of five X chromosomes instead of two. The first case of this chromosomal abnormality was reported in 1963. The condition is typically characterised by severe mental retardation, craniofacial malformation, short stature, and other physical abnormalities. The true incidence of pentasomy X is unknown at present, and the only known risk factor is female sex. We present a case of pentasomy X with maternal origin of the extra X chromosomes.

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The clinical manifestations seen in our case were consistent with those described previously in patients with pentasomy X. They included mental and developmental retardation, craniofacial anomalies, skeletal abnormalities, and cardiovascular anomalies. At first glance, our patient appeared to be normal, but she had multiple malformations and abnormal laboratory findings. The common features found in previously described patients and our patient were mental retardation, hypertelorism, mongoloid slant of palpebral fissures, a flat broad nose, malformed teeth, normal external genitalia, clinodactyly of the fifth finger, small hands and feet, and congenital heart disease. In contrast, features found frequently in previous patients and not in our patient included a short neck, a simian crease, and overlapping toes. A persistent pupillary membrane and congenital chorioretinal atrophy were unique to our patient. In addition, most of the other patients had varus of the feet, whereas our patient had valgus.

“Patients can be misdiagnosed as having Down's syndrome, so that the correct diagnosis requires cytogenetic analysis”

The normal external genitalia seen in our patient have been reported in previous patients examined, although gonadal dysfunction has been seen in many cases, including a postmortem case. These findings suggest that despite the normal appearance of the external genitalia, there is an...
We describe an 11 month old girl with features of pentasomy X: muscular hypotonia, mental retardation, a cleft palate, mild hydrocephalus as a result of dilatation of both lateral ventricles, hyperextensible elbow joints, proximal radioulnar synostosis, clinodactyly of the fifth finger, valgus of the feet, and small hands and feet. She also has a persistent pupillary membrane and congenital chorioretinal atrophy. Molecular and cytogenetic evaluation revealed that her karyotype was 49,XXXXX and her extra X chromosomes were of maternal origin. Pentasomy X may be caused by successive maternal non-dysjunctions.

**Take home messages**

- We describe an 11 month old girl with features of pentasomy X: muscular hypotonia, mental retardation, a cleft palate, mild hydrocephalus as a result of dilatation of both lateral ventricles, hyperextensible elbow joints, proximal radioulnar synostosis, clinodactyly of the fifth finger, valgus of the feet, and small hands and feet.
- She also has a persistent pupillary membrane and congenital chorioretinal atrophy.
- Molecular and cytogenetic evaluation revealed that her karyotype was 49,XXXXX and her extra X chromosomes were of maternal origin.
- Pentasomy X may be caused by successive maternal non-dysjunctions.

Immunoglobulin values in our case were similar to those reported previously in a 49,XXXXX female patient, including reduced serum IgA and IgG2 and normal total IgG. Our patient’s serum IgA and IgM values were inappropriately low compared with age related normal values. However, no history of a greatly increased incidence of recurrent infections was found in our patient, whereas Boeck et al described their patient as having a lifelong history of eczema, recurrent pneumonia, and staphylococcal abscess.

According to a review of pentasomy X, mental retardation was seen in all of the 22 cases reported previously. Delayed psychomotor development from early infancy, as seen in our patient, has also been described in this syndrome.

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