Tetraploidy in a liveborn infant

I López Pajares, A Delicado, A Diaz de Bustamante, A Pellicer, I Pinel, M Pardo, M Martin

Abstract
We report a 3 month old boy with tetraploidy, found in peripheral blood and skin fibroblast cultures, with severely delayed growth and neurodevelopment, and with a cleft lip; these findings have not been described before. This report brings to seven the total number of liveborn infants with a 92,XXYY karyotype.

Tetraploidy, the existence of four complete sets of chromosomes, has been reported infrequently in liveborn infants. To date, there have been six cases of full tetraploidy published. All these cases have been associated with multiple congenital defects and survival varies from hours to 22 months. We present a 3 month old infant with multiple congenital anomalies and tetraploidy.

Case report
The proband (figure) was the first child of a 19 year old mother and 25 year old father. There was no family history of consanguinity, multiple abortions, congenital malformations, or mental retardation. The mother took medication for headaches during early pregnancy. The patient was delivered vaginally at 37 weeks of gestation, weighing 1900 g.

Physical examination showed a small for dates male infant with a flat occiput and short palpebral fissures. The ears were low set and had a rudimentary preauricular appendage on the right. Bilateral coloboma and left cleft lip with complete cleft palate were also evident. There was arachnodactyly of the hands (with low set thumbs) and feet and cutis marmorata of all the skin. He had normal male genitalia.

Ultrasound examination showed a normal abdomen with kidneys of normal shape and size, although serum creatinine was raised (around 88 μmol/l). EEG at 19 days was normal. Cerebral ultrasound showed a hypoplastic vermis cerebelli and few fissures. A systolic murmur was present from the early perinatal period, with moderate cyanosis that worsened with crying, but without signs of cardiac insufficiency. Echocardiography confirmed the presence of severe tetralogy of Fallot. With the results of the chromosome studies and with parental consent, we adopted a conservative approach to the cardiopathy. He is growing poorly.

CYTOGENETIC STUDIES
The patient’s chromosomes were studied in peripheral blood lymphocytes. All of the 200 metaphases...
analyzed had 92 chromosomes. GTE banded metaphases showed a 92,XXYY karyotype A fibroblast culture was established from a skin biopsy; 100 metaphases were analyzed and all of them had 92 chromosomes. The patient’s parents have not been studied.

Discussion
Tetraploidy has been reported infrequently in liveborn infants. Most of these patients have been mosaics with a tetraploid and a normal cell line. This patient is the seventh case of full tetraploidy to be reported so far. The six other patients with ‘pure’ tetraploidy have been reported by Golbus et al.,2 Pitt et al.,2 Scarbrough et al2 (three patients), and Lafer and Neu.4

Our patient is compared with these other cases in the table. The most common manifestations are microcephaly, prominent, narrow forehead, high arched/cleft palate, short palpebral fissures, microphthalmia, low set and dysplastic ears, and positional and structural limbs defects. Our patient had a cleft lip and cutis marmorata, which have not been described before.

It is interesting to note the survival of these patients, from a few hours to 22 months. Our patient is alive at 3 months.

As observed by Golbus et al.,1 these patients with 46 extra chromosomes had abnormalities comparable to those seen in patients with a single extra chromosome, such as trisomy 13 or trisomy 18. They suggested that the balance between chromosomes and the ratio between different portions of the chromatin is more important than the absolute number of chromosomes present, but they did not deny the fact that even ‘balanced’ polyploidy is developmentally deleterious.

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