Suspected Syndrome of Chromosome 22 Deletion in a Fetal Autopsy

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Abstract: Malformation syndrome caused by genetic mutation of chromosome 22 was described in 1965 by Angelo DiGeorge, an italo-american doctor. He observed a common clinical picture in a group of children characterized by cardiac malformation, recurrent infection due to absence of thymus and a typical phenotypic aspect of these children. Only in 1992 the chromosomal anomaly was found as a result of studies based on Fluorescence In Situ Hybridation technique (F.I.S.H.). In this report it is described a case of a stillbirth during the second gestational trimester with multiple malformations that are suspected for syndrome of chromosome 22 deletion. The present case is an example of what careful macroscopic and microscopic examinations can be able to identify syndromic defects attributable to chromosome 22 mutations.

Keywords: DiGeorge Syndrome, Chromosomal Deletion, Fetal Autopsy

1. Introduction

DiGeorge Syndrome is caused by a defect in the embryological development of the third, the fourth gill pocket and fourth gill arch. It is characterized by abnormalities of the heart, thymus, parathyroid and specific facial dysmorphic features.

The incidence is estimated to be 1/5000 live births. Males and females are afflicted in equal proportion.

It is caused by a partial deletion of the long arm of chromosome 22. The deletion can occur random in the affected individual (80/85% of cases) or transmitted by a parent, which in turn can be affected in a nuanced form (15/20% of cases). In these last cases the risk of recurrence of the disease in future pregnancies is 50% at each conception, regardless of the sex of the infant [1].

We reported a case of female fetal autopsy with peculiar malformations that can be attributable to DiGeorge Syndrome.

2. Case Presentation

In this report a fetal autopsy on a female dead at 15 week of gestation for placental failure is described.

At macroscopic reply, we didn't find thymus, in absence of other salient reports. Organs were taken as a whole, paraffin embedded and Alcian-hematoxilyn coloured.

Microscopic examination revealed the presence of a hypoplastic residual thymic on atrial pericardium (figure 1). Also, we have observed abnormal dilatation of the brain lateral hemispherical ventricles, with diffuse cortical atrophy (figure 2), and papillary hyperplasia of the choroid plexus. No alterations were detected at cardio-pulmonary or gastroenteric level, except for pancreas, that presented a partial parenchymal atrophy with decreased both of Langerhans islands than of exocrine portion (figure 3). Remarkable bilateral cortical atrophy was found in the kidneys with dilation of the caliceal spaces facilities from possible clogging downstream (figure 4). As regards the reproductive system, there were female gonads with aspects of uterine hypoplasia.

Placental microscopic evaluation revealed acute decidual infection identified as the cause of stillbirth. We also have noticed the presence of many including stromal syncytiotrophoblast in staminal chorionic villi (figure 5).
3. Discussion

In cases of diagnosis or diagnostic suspect of stillbirth for 22q11 deletion, it would be important a cytogenetic and molecular analysis of the parents to rule out a possible vertical transmission of the disease.

The survey is carried out using the F.I.S.H. method to detect and locate the absence of specific DNA sequences in chromosomes. Chromosomal microarray is also increasingly utilized for genetic testing of individuals with unexplained developmental intellectual disability, autism spectrum disorders, or multiple congenital anomalies. The International Standard Cytogenomic Array Consortium held two international workshops and conducted a literature review of 33 studies, including 21,698 patients tested by microarrays. They agreed on an international consensus statement about low cost and clinical utility in using chromosomal microarrays to detect genetic anomaly in suspected cases [1].

The symptoms and signs of 22q11.2 deletion configure a wide range of clinical conditions. So they have been described as many syndromes. These include the velocardiofacial syndrome (also called Shprintzen syndrome), the DiGeorge Syndrome and other [2]. The acronym CATCH-22 is what best summarizes the anomalies found, where C means cardiac defects, A means abnormal facies, T is for thymic hypoplasia, C is for cleft palate and H means hypocalcemia. The syndrome is caused by deletion of many thousands of bases on the long arm of chromosome 22. For this reason, the term “22q11.2 deletion syndrome” is the most frequently used today.

Microdeletions in 22q11 region are described too. They are associated with a risk 30 times higher than normal for developing schizophrenia. [3, 4].

The syndrome is caused by a new mutation, however it can also be an autosomal dominant familial transmission in a minority of cases. Sometimes the syndromic phenotype described is due to partial deletion on the short arm of chromosome 10. The deletion regards about 30 genes, not well known.

The key gene for the onset of the symptoms of the
The peculiarity of our described case consists in having a complete autopsy finding including full examination of all organs must be considered necessary in the event of stillbirth, especially in the absence of clinical or obstetric reasons. It would ultimately be desirable that all the cases of perinatal death were treated in a multidisciplinary context, especially when a malformative phenotype is detected.

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**References**


