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47,XX + 21: A CASE REPORT

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Abstract: Down Syndrome (DS), it is one of the most frequent chromosomal abnormalities in humans congenital type, being free trisomy 21 (+21), the most important in the clinical diagnosis of this pathology. This article describes and analyzes a case study of a Venezuelan female patient 24 hours old, who was referred to consultation genetics submit to physical examination: oblique fissures, led nostrils upward, protrusion of tongue, and small ears. Underwent chromosomal mapping (karyotype) through G banding technique, cardiology studies, hearing and vision. The results showed 47,XX +21 in all metaphase cells analyzed. Also, the cardiac evaluation revealed no congenital heart disease. Similarly, neonatal hearing screening and ophthalmologic examination were within normal parameters. We conclude that the patient has trisomy 21, no findings of consequences resulting from this chromosome disorder. It is recommended that a neurological stimulation and early monitoring to evaluate psychomotor activity.
Key words: Down syndrome, trisomy 21, congenital chromosome disorder.

INTRODUCTION

The SD is the most common cause of chromosome disorder in humans, being trisomy 21 the most important (95%), usually due to meiotic non disjunction in the ovum (female sex cell). This is a chromosomal abnormality that has an incidence of 1 in 800 live births and their frequency increases with maternal and paternal age (Altamirano et al., 2000).

Dysmorphic features of this syndrome differ from patient to patient. Therefore, the realization of the karyotype is essential for proper diagnosis and genetic counseling, since the risk of recurrence in successive generations depends on the karyotype of the patient. However, some common features present in most infants with Down syndrome are: short stature, slanted eyes, hands and short fingers, single palmar crease and muscular hypotonia. It is also associated with other congenital diseases, among which are: heart disease, disorders of the digestive tract and eye diseases, so the necessary is long-term monitoring of these patients (Gómez-Valencia et al., 2011; Tartaglia et al., 2010).

In this paper, a case of a child with DS (47, XX + 21), which does not present all common phenotypic characteristics for this condition is reported.

MATERIALS AND METHODS

Case Presentation

Female patient 24 hours old, natural and from Maracaibo, referring to genetic consultation to present suspicion of SD. The girl is the product of a union outbred, the second daughter of parents (first child 8 years old, without chromosomal abnormalities). The gestation period was 38 weeks without significant medical complications. Both of them parents of 32 year old at the time of caesarean section. Dad is a native of San Jose de Perija, and mother, a native of Maracaibo, Zulia state. There are no similar cases in both families.

Physical exam

A comprehensive physical evaluation was performed to identify phenotypic characteristics associated SD in hands, feet, cephalic area, muscle, oral cavity, nose, ears, eyes, height and weight.

Karyotype

20 lymphocyte cells from peripheral venous puncture with heparin as an anticoagulant were studied to evaluate their chromosomes through the technique Banding G (Gallego, 2011; Rojas-Atencio et al., 2012) in the laboratory of cytogenetics Cendilamú, CA in Maracaibo. To this, they were sown about 10 drops of blood in an enriched medium and incubated at 37C for 72 hours. Stimulation of cell division was achieved with the addition of a mitogenic factor (phytohemagglutinin). After that, a solution of colchicine was added to stop cell division and prevent cells complete mitosis. This substance acts by inhibiting the mitotic spindle formation and cells only reach metaphase and accumulate in the culture. Then I was added to hypotonic solution That causes the cells to swell and burst with a drip technique on a slide and then chromosomes are released

Cardiological evaluation

Cardiological status of the patient was assessed by an electrocardiogram and echocardiography in the cardiovascular research unit CESCARDIN in city of Maracaibo.

Hearing and ophthalmologic evaluation

In the unit of otolaryngology and pediatric ophthalmology Cuatricentenario Hospital of Maracaibo, it was evaluated hearing health (neonatal hearing screening) and vision of the patient for the discarding of ocular diseases as: Cataracts, blindness, strabismus and Brushfild spots.

RESULTS

Physical exam

Physical examination revealed facial dysmorphic features such as: oblique fissures, upwards directed nostrils, protrusion tongue and small ears with narrow ear hole (Figure 1). Birth weight was 3100 kg and 50 cm size. He did not show excess skin in the cervical area,

flat feet, clinodactyly, dysplasia phalanges in hands and feet, simian crease and dysplastic pelvis.

Figure 1. Dysmorphic facial features

karyotype

The results of the 20 cells studied showed a karyotype 47, XY + 21, representing 100% SD in all of metaphases analyzed (Figure 2).

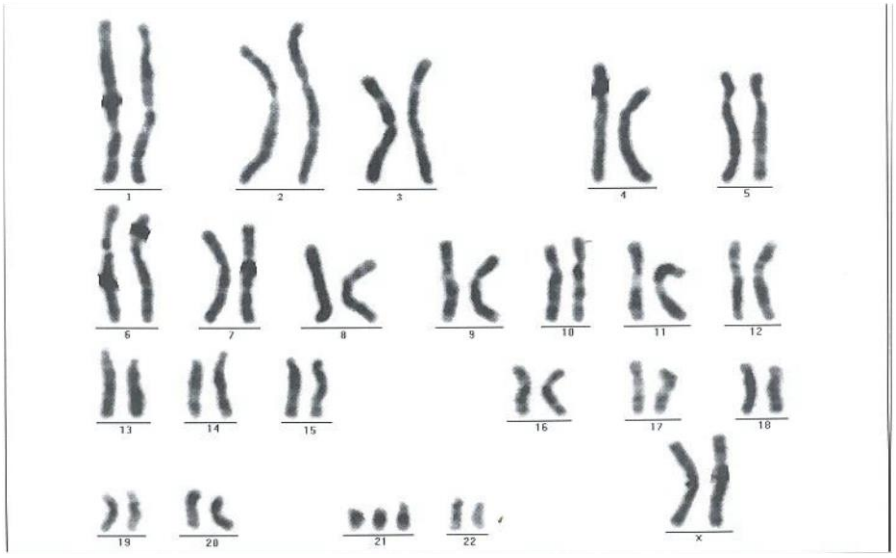

Figure 2. Karyotype 47, XX + 21. XXX presence in pair 21 is clearly evident, corresponding to nondisjunction of chromosome these

Hearing and ophthalmologic evaluation

Audiometry test showed no abnormalities, nor showed cracks in the eardrum. Furthermore, the ophthalmologic evaluation was within normal parameters without evidence of cataracts, blindness, strabismus and Brushfield spots.

Cardiological evaluation

The electrocardiogram was normal, with correct pulse, rhythmic heart sounds. While the transthoracic echocardiogram showed a structurally normal heart, with patent foramen ovale and functional light source pulmonary stenosis branches.

DISCUSSION

The DS by Trisomy 21 free in the (non-germ) somatic cells in the body, is due to an error during the first meiotic division of gametes (95% frequency in eggs) that leads to incomplete separation of genetic material. Approximately 5% of cases, the extra chromosome 21 is paternal origin (Kaminker and Armando, 2008; Pérez, 2014). During formation sex cells of parent, chromosomes is separated, so that each maternal and paternal line, transmits information only one chromosome of each pair. When the disjunction does not occur both chromosomes are transmitted. They are not precisely known etiology that causes the wrong disjunction. However, there have been proposed some multifactorial hypothesis involving (environmental exposure, cellular aging, age of parents, among others), without having been able to establish any direct causal link between any agent and the appearance of trisomy (Basile, 2008).

The findings of this study are consistent with the phenotypic manifestations reported by De La Torre and Ochoa (2015) in a child with SD and Klinifelter. Similarly, Sanchez *et al*. (2001) published an article from a patient with SD and Achondroplasia, whose physical characteristics correspond to those found in the patient of this study. In both studies the etiology was of maternal origin. In this work we not were performed karyotype to the parents.

In Venezuela, according to reports from the Unit of Medical Genetics at the University of the Andes, trisomy 21 is the most frequent with 84.07% of all samples analyzed in the period 2005-2012 (Araque et al., 2013). Similarly, Garduño-Zarazúa *et al*. (2013) reported that between 1986 and 2010 in the Department of Genetics of the General Hospital of Mexico, 87.3% of patients karyotype SD corresponded to trisomy 21 regularly. Both studies correspond to this case.

According Kaminker and Armando (2008), the risk of occurrence increases with maternal age: 15-24 years: 1/1300, 25-29 years: 1 / 1,100, 1/350 at 35, at 40 years: 45 years 1/100 and 1/25. However, the incidence of chromosomal abnormalities in infants is changing due to environmental changes, exposure to chemicals and de novo chromosomal alterations.

Regarding the missing features in the patient of this study: heart disease, flatfoot, clinodactyly, dysplasia phalanges in hands and feet, simian crease and dysplastic pelvis, was also published in the country in two cases of trisomy 21. One by translocation tandem, where two chromosomes 21 were attached to the long arm of chromosome 10 (45, XX + tan (10; 21; 21)) and the other by translocation inverted tandem between two chromosomes 21 united through their long arms (46, December XY + (21q: 21q)) (Sanchez, Marade and Guerra, 2001).

This demonstrates the importance of cytogenetic studies in the differential diagnosis of chromosomal abnormalities, for better monitoring of patients a medium and long term. Regarding the phenotype, have been described more than 100 dysmorphic features associated with SD, able to present phenotypic variations from one individual to another. Therefore, none of the signs and symptoms is considered constant for diagnosis.

CONCLUSION

According to clinical evidence and cytogenetic results, it is concluded that the patient has SD with trisomy (47, XX + 21), with no effect on the heart, hearing and vision. continuous monitoring in the service neurology and genetics is recommended, with accompaniment of early stimulation exercises for the development of motor and cognitive skills.

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