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Letters to the Editor

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Congenital heart disease in the main aneuploidy syndromes

Cardiopatías congénitas en los principales síndromes causados por aneuploidías

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To the Editor:

In a world where there is a rise in the socio-economic development and progress in the control of infectious diseases and malnutrition, congenital malformations are presented as a health problem. These malformations, in general, are defined as inadequate tissue formation due to genetic defects themselves, where genetic abnormality affects genes involved in the development[1]. According to Acosta and Mullings[1], the frequency of congenital malformations in humans is estimated in one out of 16 newborns and they are responsible for 15% of deaths that take place in the first year of life, and therefore, they represent the leading cause of mortality in infants.

Meanwhile, congenital heart diseases are disorders present at the time of birth due to alterations in the form and function of the heart, the circulatory system and great vessels, that appear during the cardiac embryogenesis[2]. Tassinari *et al*[2] suggest that they are among the most frequent congenital anomalies with the highest mortality in the world, with a prevalence that can vary between 0.04-0.19, and reach up to 80 cases per 10.000 live births.

Being demonstrated the effectiveness of preventive medicine, at current times, the efforts must be aimed at the search of risk factors for congenital malformations, since the study of their main causes is of vital importance.

Aneuploidies are chromosomal defects in which the normal component of 46 chromosomes is altered, by excess or defect. They are due to a failure in the segregation of chromosomes in gametes during the meiotic division, or also in the first mitotic division of the zygote[3]. The most frequent are trisomies 21, 13, 18, the Klinefelter syndrome (XXY trisomy) and Turner syndrome or monosomy of the X chromosome[3]. These aneuploidies bring multiple congenital malformations, including cardiac, which are some of the most frequent.

Patau or Bartholin-Patau syndrome, also known as trisomy 13, is a genetic disease resulting from the presence of an extra chromosome 13. Díaz-Véliz *et al*[4] state that it was first observed by Thomas Bartholin in 1657, but it was not until three centuries later, in 1960, when Patau described it. The average survival time of affected children is seven days, and 90% die during the first year of life. Nearly 80% of cases with this syndrome have congenital heart disease, including dextrocardia and transposition of the great vessels, both with high prevalence[5].

Edwards syndrome or trisomy 18 is a rare autosomal disease, as a result of chromosomal imbalance. According to Romero and Atobe[6], 95% die in the first year of life, mainly due to congenital heart disease, apnea and pneumonia, or complications secondary to pulmonary hypertension. Almost all patients with this syndrome (almost 100%) have congenital heart disease; the most common are ventricular and atrial septal defects, and persistent ductus arteriosus[5].

Down syndrome is the most frequent chromosomal alteration and the leading cause of intellectual disability worldwide[7]. It was described by John

Langdon Down in 1866, within his proposal to classify patients with intellectual disabilities. It is characterized by complete trisomy of chromosome 21 in 95% of cases, and it is present in approximately one out of 650-680 live births. Between 35 and 60% of children with this syndrome have congenital heart disease, a higher frequency than the general population[9]. According to Díaz-Cuéllar *et al*[7], in one of the most important population studies about the topic, the most frequent malformations were the complete atrioventricular canal, the interventricular and interatrial communications, the tetralogy of Fallot and the persistent ductus arteriosus.

Klinefelter syndrome was described in 1942 by Klinefelter, Reifenstein and Albright, and in 1959, the presence of an additional X chromosome was described in men, as a cause of the syndrome[10], thus, the most common chromosomal formula is 47, XXY, present in about 80% of patients[10]. The most frequently associated congenital heart anomaly is the tetralogy of Fallot. Others, like the ventricular and atrial septal defects, the Ebstein anomaly, the aortic stenosis and the double emergence of the right ventricle followed in importance. Despite this, in general, congenital heart diseases are not frequent[5].

Turner syndrome is a consequence of the partial or complete absence of the second sex chromosome, which appears as a female phenotype[11]. The characteristic clinical findings are: short stature (100%), infertility (99%), absence of pubertal maturation (96%) and congenital heart disease (55%)[12].

Syndromes caused by trisomy in chromosome 13, 18 and 21, as well as monosomy of chromosome X may involve, in a significant percentage, a congenital heart disease; but this does not happen in the XXY trisomy, where they are not usually frequent.

CONFLICTS OF INTERESTS

None.

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