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Case Report

Prenatal stage diagnosis of large ventricular septal defect in at patient with Edwards Syndrome

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Abstract

Edwards syndrome is the second most common autosomal chromosomal disorder after trisomy 21. It is characterized by abnormalities in multiple organs, with cardiac malformations being predominant. Prenatal diagnosis is complex in centers without maternal-fetal medicine services; however, findings on fetal echocardiography can raise suspicion and broaden the diagnostic approach. We present the case of a newborn with Edwards syndrome, diagnosed at 23 weeks of gestation by Fetal Cardiology, with a large interventricular septal defect, in addition to data of intrauterine growth restriction, with a normal fetal cardiovascular profile, the diagnosis being corroborated at birth with karyotype, and presenting a poor prognosis, with a prolonged hospital stay, dependence on invasive mechanical ventilation, and survival beyond the first month of life, reaching a total of four months. During this period, the patient exhibited inability to feed spontaneously, development of severe neurological sequelae, and final outcome with death.

Introduction

Edwards syndrome, also known as trisomy 18, is a genetic disorder (chromosomal abnormality) characterized by the presence of an extra chromosome in pair 18, which can be complete, partial, or mosaic. The prevalence among live births is estimated to range from 1 in 6,000 to 1 in 8,000. However, the true prevalence may be higher (approximately 1 in 2,500 to 1 in 2,600) owing to the elevated risk of fetal demise and the termination of pregnancy following prenatal diagnosis [2], with a female predominance, affecting all races and geographic regions. It is the second most common autosomal chromosomal abnormality after trisomy 21, and its risk increases significantly in children of older mothers. Approximately 95–96% of cases are complete trisomy caused by meiotic nondisjunction, while the remaining cases are due to translocation, mosaicism, or partial trisomy, which usually present with a less complete phenotype, although in some cases all the typical clinical features are observed. Identified in 1960, trisomy 18 has a serious prognosis with infrequent survival beyond the first year of life [1].

At the cardiovascular level, up to 90% of patients develop congenital heart disease, including ventricular septal defect, multiple valvular disease, patent ductus arteriosus, coarctation of the aorta, pulmonary stenosis, transposition of the great arteries, and tetralogy of Fallot.

We present the case of a 23 weeks fetal patient with a very large interventricular septal defect not associated with critical congenital heart disease diagnosed by echocardiogram. Extracardiac malformations associated with Trisomy 18 were detected at birth, highlighting the importance of the fetal cardiovascular finding as the main fact in order to suspect this type of chromosomal abnormality.

Case Report

A female product of the third gestation, initially assessed at 23 weeks of gestational age, daughter of a 38-year-old mother with gestational diabetes during the second trimester and a 36-year-old father, both of advanced maternal and paternal age. Notable obstetric history includes a spontaneous abortion in the second gestation, with the first gestation resulting in a living, apparently healthy 19-year-old sibling. The patient was referred to Fetal Cardiology for evaluation of possible fetal cardiovascular anomalies.

The first fetal echocardiogram performed, showed a large ventricular septal defect (from the inlet region extending to the perimembranous region). Huhta cardiovascular profile was normal, except for an abnormal pulsatility index of the middle cerebral artery (3rd percentile). Fetal echocardiograms were performed at 25, 28, 31, and 35 weeks of gestation, confirming only the large interventricular septal defect, with normal emergence of great vessels. There were not critical congenital heart defects detected, except an abnormal pulsatility index of the middle cerebral artery and cerebroplacental ratio.

A 34 weeks female newborn was delivered, weighing 1,460g, measuring 38 cm, with a 28 cm head circumference, 24 cm chest circumference, 25 cm abdominal circumference, 6.5 cm foot length, and APGAR scores of 6/8. Physical examination showed multiple malformations: microcephaly, dolichocephaly, with biparietal narrowing, low hair implantation in the posterior region, triangular face, short palpebral fissures directed upwards, microphthalmia, a nose with a hypoplastic root and upturned tip, apparent microstomia, microretrognathia, low-set and dysplastic auricles, a short neck, and bilateral telethelia. Upper extremities with agenesis of the radius and ulna in the left arm, agenesis of the left thumb, right arm with arthrogryposis, radial defect, long fingers clenched with the second over the third and the fifth over the fourth. Lower extremities symmetrical with both feet in equinovarus. External genitalia were phenotypically female. As part of the diagnostic approach, a postnatal echocardiogram was performed which demonstrated a large interventricular septal defect extending from the posterior region to the membranous septum region measuring 8.6 x 9.4 mm, in addition to a patent ductus arteriosus, both with bidirectional shunting due to still elevated pulmonary vascular resistance (10 hours of life). Head CT scan showed bilateral retrocerebellar cysts. Abdominal CT scan was performed due to an image suggestive of diaphragmatic hernia on the thoracoabdominal X-ray, revealing hepatomegaly and a right diaphragmatic eventration caused by liver protrusion secondary to

weakness or elevation of the right hemidiaphragm. Due to the findings on the head CT scan, an MRI was performed, which showed cerebellar asymmetry with greater amplitude in the right cerebellar hemisphere, secondary to images suggestive of left parasagittal retrocerebral cysts versus megacisterna magna.

A karyotype was performed on peripheral blood at 12 days of life, confirming a cytogenetic diagnosis characterized by a regular trisomy of chromosome 18 in 20 analyzed metaphases, 47,XX,+18. Genetic counseling was provided to the parents, indicating that in cases of regular and homogeneous trisomy, the recurrence risk in a subsequent pregnancy is generally estimated to be around 1% and increases with maternal age.

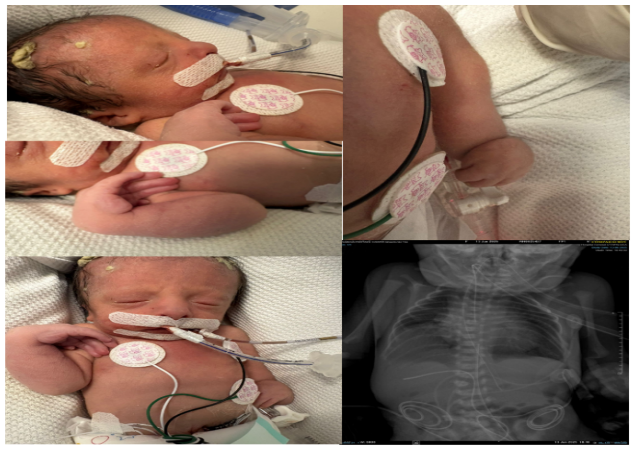

Fig1: Newborn Whit Edwards syndrome. Physical Examination and Xray

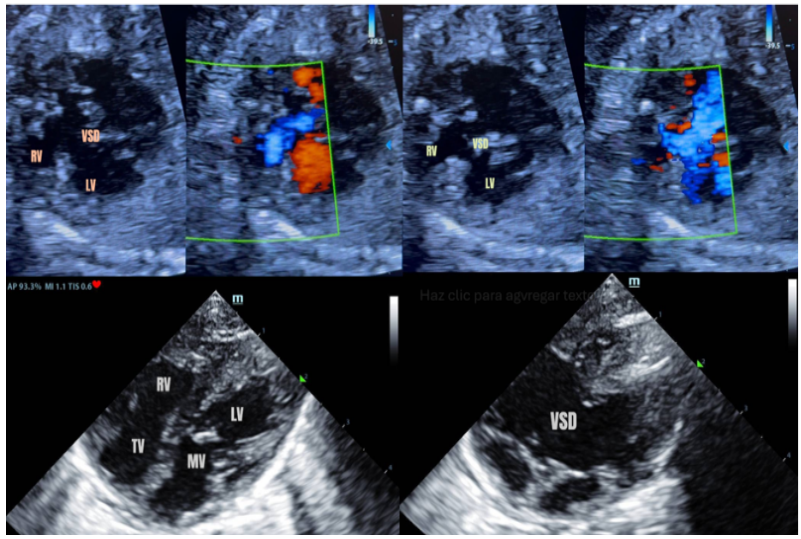

Fig 2: Prenatal (23 weeks) and postnatal Echocardiograms whit a large ventricular Septal Defect

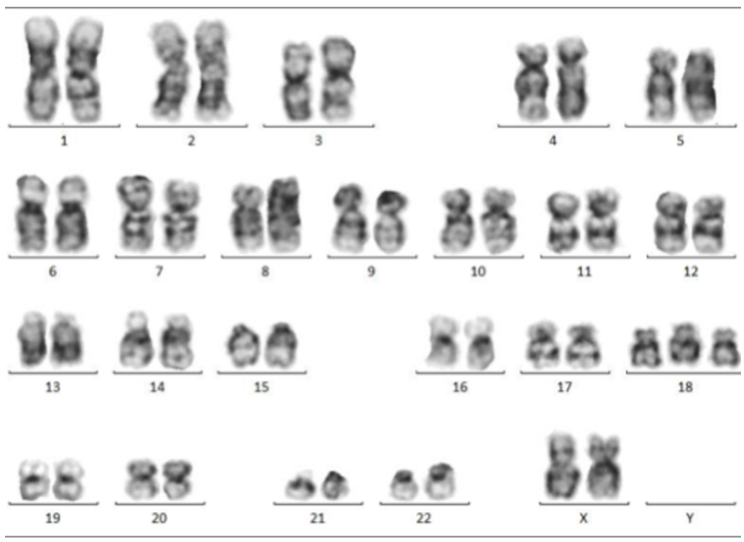

Fig 3: Cytogenetic Analysis

Discussion

The clinical manifestations of Edwards syndrome are numerous and affect various systems. Growth is characterized by pre- and postnatal delay, low birth weight, poor muscle mass, and reduced subcutaneous fat. From a neuromuscular standpoint, newborns initially exhibit hypotonia that progresses to hypertonia. Craniofacial abnormalities include microcephaly, prominent occiput, micrognathia, low-set ears, small mouth, high-arched palate, cleft lip and/or palate, short palpebral fissures, and ocular anomalies such as cataracts, microphthalmia, or iris coloboma. The limbs present with hands with closed fists and overlapping fingers, hypoplastic nails, limited hip mobility, prominent heel, and clubfoot. Other malformations include urogenital abnormalities (cryptorchidism, genital hypoplasia, hypospadias), renal abnormalities (horseshoe kidney, ureteral duplication, hydronephrosis), and digestive abnormalities (anal atresia, Meckel's diverticulum, intestinal malrotation). Additionally, central nervous system anomalies may be present, such as hypoplasia or agenesis of the corpus callosum, hydrocephalus, and spina bifida, as well as cutis marmorata, hirsutism, short sternum, and small pelvis.

The diagnosis of trisomía 18 is confirmed through cytogenetic analysis, which reveals the presence of an additional chromosome 18. During the prenatal stage, the diagnosis can be suspected through detailed ultrasound, showing suggestive signs such as intrauterine growth restriction, cystic hygroma, shortening of long bones, neural tube defects, as well as cardiac malformations and abnormalities of the central nervous system. For genetic confirmation, invasive studies like amniocentesis or cordocentesis are employed, allowing for samples to be obtained for conventional karyotyping and fluorescence in situ hybridization (FISH) techniques, facilitating the precise identification of aneuploidies or possible structural chromosomal rearrangements. After birth, the definitive diagnosis is established through karyotyping, usually from

peripheral blood.

The prognosis for Edwards syndrome is poor. Mortality is very high, with approximately 50% of newborns dying within the first week of life and only 5-10% survive at one year. Only a minority survive beyond the first year, and longer survival is more common in female infants. The most frequent causes of death are congenital heart disease, apnea, and pneumonia. Survivors have severe chronic complications, such as feeding difficulties requiring tube feeding or gastrostomy, scoliosis and other orthopedic abnormalities, as well as profound intellectual disability, which significantly reduces their quality of life. This situation underscores the importance of prenatal diagnosis, genetic counseling, and family planning, as well as preparation for the complex medical care of surviving patients.

Conclusion

In hospitals where maternal-fetal medicine is not available to assess common fetal abnormalities in Trisomy 18 (cerebral malformation, musculoskeletal malformation, facial dysmorphisms), the detection of large ventricular septal defects not associated with other critical congenital heart diseases may guide the diagnosis of Edwards syndrome or other aneuploidies during the fetal stage.

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