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CASE REPORT

Teenager with Edwards' syndrome: a rare case report

Igor Soares Trindade[1], Marise Vilas Boas Pescador[1]

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Abstract

Edwards' syndrome is a severe genetic disease (chromosome 18 trisomy) that has a very restrict prognosis, resulting in premature death of the carrier. This study aims to report a case of survival of 12 years old girl diagnosed in the first months of life, her clinical evolution and the treatments performed by it. The multidisciplinary treatment performed by the adolescent since the first months of life shows the importance of this approach to improve the quality of life of patients with this syndrome.

[1] FAG University Center, Medicine - Cascavel - PR - Brazil.

Correspondence to:
Igor Soares Trindade.
Centro Universitário da Fundação Assis Gurgacz. Av. das Torres, 500 - Loteamento Fag, Cascavel, PR, Brazil. CEP: 85806-095. E-mail: iigortrindade@hotmail.com

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INTRODUCTION

Edwards syndrome (ES) is an autosomal trisomy of chromosome 18, with a broad clinical picture, with involvement of multiple organs and systems, being the second most frequently observed autosomal trisomy at birth, second only to Down syndrome. Its estimated prevalence is 1 case for every 3600 to 8500 live births, with the characteristic karyotype 47, XX, +18 or 47, XY, +18[1].

Females are more affected than males, at a rate of 4 girls for every boy born with the disease. Its incidence in pregnancies that do not progress to term is high, but 95% of them progress to miscarriages, most of the time occurring spontaneously[2].

The diagnosis of SE can be hypothesized by fetal ultrasonography and biochemical levels (decreased doses of human chorionic gonadotropin - HCG), but diagnostic confirmation is made by analyzing fetal chromosomes through methods such as amniocentesis and chorionic villus puncture[3].

The clinical manifestations of the syndrome are diverse, with more than 130 different anomalies being described in the literature, which may involve practically all organs and systems. The most commonly present phenotypic characteristics are: severe mental deficiency with delay in neuropsychomotor development; abnormalities of growth, skull and face, thorax and abdomen, extremities, genital organs, in addition to malformations in external organs[4-6].

Around 90% of individuals with trisomy end up dying in the first year of life, with some exceptions for children of non-Caucasoid origin, who end up having a slightly longer life expectancy[7], 30% of those affected by the syndrome die in the first few years. 30 days of life[8].

OBJECTIVE

To report a case of survival of a patient with ES, analyzing her clinical evolution and the treatments performed by her.

CLINICAL CASE REPORT

KVSC, female, 12 years old, born in Capitão Leônidas Marques, presented a borderline preterm birth (37 weeks) by cesarean section, weighing 2310g (small for gestational age - SGA), 42 cm in length and head circumference measuring 32.5 (25th percentile). The Apgar score was 3 in the first minute and 7 in the fifth minute, requiring tactile stimulation and inhaled oxygen in the delivery room. The mother had prenatal care without complications during this period. In the neonatal period, he presented respiratory and cardiac complications with a diagnosis of ventricular septal defect, he needed care in an intensive care unit (ICU) and Edwards syndrome was suspected due to the clinical manifestations presented, such as: weak crying, difficulty in sucking and some other characteristic signs of the syndrome such as clenched hands, nail hypoplasia, short neck; hypertonia and with a characteristic facies (microcephaly, microretrognathia, in addition to a high and arched palate), being referred to a more complex hospital. She remained in the pediatric ICU for 4 months, and the diagnosis of SE was confirmed by performing a karyotype at approximately 30 days of life. The minor had difficulty in weaning from ventilation and swallowing, also having apnea crises and an episode of aspiration pneumonia during the period of hospitalization, being discharged home with tracheostomy and gastrostomy.

After hospital discharge, he remained under treatment and medical follow-up at a university center in the city of Cascavel - Paraná, from 4 to 10 months of age, when he returned to the city of origin with 5,950 kg of body weight. multidisciplinary team, with stimulating treatment through motor and respiratory physiotherapy, speech therapy and occupational therapy.

The child has been followed up with a cardiologist since birth, without any more complex complications. He has S3 on auscultation, a VSD with a closing mechanism of 0.7 mm and an ECG in sinus rhythm, without overload, considered normal for his age. During this entire period of consultations and follow-up at the APAE, he had some hospitalizations in the city of origin, due to low severity conditions, such as bronchitis, pneumonia and gastroenteritis, without major complications.

In February 2017, an orthopedic surgery was performed due to a shortening of the Achilles tendon , which went without complications.

Currently, the minor is starting puberty, with pubertal staging based on the Tanner Criteria with grade 1 hairiness and grade 1 breast development on the left and 2 on the right. Her body weight is 23.6 kg, height is approximately 128 cm (there was technical difficulty in the measurement due to lower limb hypertonia). In addition to medical follow-up, the child undergoes speech therapy once a week and occupational therapy, where stimuli for cognitive ability, proprioceptive training, communicative intention and interaction with the environment are practiced, as well as physical therapy with sensory integration exercises twice a week., showing a good response to these activities, being able to perform a sitting posture without support and crawling seated throughout the environment.

DISCUSSION

Edwards syndrome is a serious genetic disease that has many systems affected. Manifestations such as low birth weight, weak crying, weak sucking, high palate, micrognathia and small nipples are some of the peculiarities present in newborns with the disease[8-10]. In the present study, the patient presents most of these characteristics which, added to the child's syndromic facies , allowed the hypothesis of SE to be raised, and this diagnosis was confirmed in the first 30 days of life. (Figure 1)

Figure 1. Knee valgus, feet turned inwards (in-toeing) and a characteristic fascia of the syndrome are noted.

Due to the poor prognosis of the syndrome, early diagnostic confirmation is important not only for the adequate clinical management of these individuals, but also for the correct genetic counseling to be given to the family. The diagnostic suspicion of SE in the prenatal period can be raised by fetal ultrasound (including measurement of translucency nucal) by biochemical measurements (such as reduced levels of human chorionic gonadotropin, alpha-fetoprotein , and unconjugated estriol in maternal serum during the first and second trimester of pregnancy), and confirmed by fetal chromosome analysis through procedures such as villi puncture chorionic and amniocentesis[3]. In Brazil, the identification of patients with trisomy 18 during prenatal care is important, especially for planning the birth, since termination of pregnancy is not permitted by law[11].

The survival of affected individuals is low, most fetuses with the syndrome do not reach term, evolving to death in the fetal period. Of the affected newborns, 90% will die within 6 months of life[14]. The child reported in this study has an age well above the age considered as the limit of life expectancy for individuals affected by the syndrome.

Patients with ES rarely reach the stage of adolescence, mainly due to the seriousness of the cardiorespiratory malformations that occur in these individuals[12]. Children over 30 days old often die as a result of heart problems[13]. In the present case, the child is already 12 years old, is in the initial phase of pubertal development, thus being considered an adolescent and, unlike the study reported by Torres et al.[12] and a minimal outflow perimembranous VSD, without the presence of more serious respiratory complications.

Due to the few cases reported in the literature with a prolonged life expectancy, when considering the treatment for patients with the syndrome, there are still no protocols that make its benefit clear. The smallest mentioned in this study, presents services aimed at improving their motor and cognitive deficits, interaction with the environment and also the strengthening of their oral motricity, being accompanied by a physical therapist and speech therapist, showing an improvement in their development when compared to the period prior to more intensive treatment. Therefore, it is considered that the multidisciplinary treatment could help patients with ES to present an improvement in their quality of life.

REFERENCES

1. Souza JCM, Solarewicz MM, Mordaski RYM, et al. Síndromes cromossômicas: uma revisão. Cad Esc de Sal. 2010[acesso em 2019 Out 05];3:1-12.
2. Sugayama MMS, Chong AK, Leone CR, Diniz EMA, Koiffman CP, Gonzalez CH. História natural de 24 pacientes com trissomia 18 (síndrome de Edwards) e de 20 pacientes com trissomia 13 (síndrome de Patau). Rev Pediatr. 1999 Jan/Mar;21(1):69-77.
3. Kagan KO, Wright D, Maiz N, Pandeva I, Nicolaides KH. Screening for trisomy 18 by maternal age, fetal nuchal translucency, free beta-human chorionic gonadotropin and pregnancy-associated plasma protein-A. Ultrasound Obstet Gynecol. 2008 Set;32(4):488-92.
4. Smith DW. Autosomal abnormalities. Am J Obstet Gynecol. 1964 Dez;90(Supl 1):S1055-S77.
5. Marion RW, Chitayat D, Hutcheon RG, Neidich JA, Zackai EH, Singer LP, et al. Trisomy 18 score: a rapid, reliable diagnostic test for trisomy 18. J Pediatr. 1988 Jul;113(1 Pt 1):45-8.
6. Zen PR, Rosa RF, Rosa RC, Mulle LD, Graziadio C, Paskulin GA. Unusual clinical presentations of patients with Patau and Edwards syndromes: a diagnostic challenge? Rev Paul Pediatr. 2008 Set;26(3):295-9.
7. Rasmussen SA, Wong LY, Yang Q, May KM, Friedman JM. Population-based analyses of mortality in trisomy 13 and trisomy 18. Pediatrics. 2003 Abr;111(4 Pt 1):777-84.
8. Koiffmann C, Gonzalez HC. Trissomia do 18 ou síndrome de Edwards. São Paulo: Instituto da criança "Professor Pedro de Alcantara" do Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo; 1992.
9. Jolmie IE. Down syndrome and other autossomal trissomies. In: Rimoin D, Pyeritz R, Korf B, eds. Emery and Rimoin's principles and practice of medical genetics. 3rd ed. New York: Churchill Livingstone; 1997.
10. Nelson WE. Princípios de pediatria. 4ª ed. Rio de Janeiro: Guanabara Koogan; 2004.
11. Diniz D. Selective abortion in Brazil: the anencephaly case. Dev World Bioeth. 2007 Ago;7(2):64-7.
12. Hinojal MCT, Miguelsanz JMM, Fernandez LMR. Fourteen-year survival in a patient with Edwards syndrome. An Pediatr (Barc). 2005 Nov;63(5):458-9.

13. Kaneko Y, Kobayashi J, Achiwa I, Yoda H, Tsuchiya K, Nakajima Y, et al. Cardiac surgery in patients with trisomy 18. Pediatr Cardiol. 2009;30:729-34.

14. Rosa RFM, Rosa RCM, Zen PRG, Graziadio C, Paskulin GA. Trissomia 18: revisão dos aspectos clínicos, etiológicos, prognósticos e éticos. Rev Paul Pediatr. 2013 Mar;31(1):111-20.