International Journal of Academic Health and Medical Research (IJAHMR)
ISSN: 2643-9824
Vol. 6 Issue 1, January - 2022, Pages:64-67

Trisomy 18 Or Edwards Syndrome in Prenatal Care: A Case Reportla Trisomie 18 Ou Syndrome d'Edwards En Post-Natalla Trisomie 18 Ou Syndrome d'Edwards En Post-Natal a trisomie 18 ou syndrome d'Edwards en post-natal

M. Bendahhou Idrissi, J.Abbou, M. K.Saoud, N. Mamouni, S. Errarhay, C. Bouchikhi, A. Banani

Department of Gynecology and Obstetrics I, Hassan II University Hospital, Fez, Morocco

Abstract: *Trisomy 18 is a chromosomal disorder, due to the presence of a supernumerary chromosome 18. Infants with trisomy 18 have a high mortality rate, secondary to the lethal malformations associated with this syndrome. The prevalence of trisomy 18 is variable and survival is low, with only one in 10 newborns reaching the first year of life. The syndrome can be evoked antenatally in case of presence of abnormalities on obstetrical ultrasound and confirmed by cytogenetic study. We report the case of a 33-year-old parturient, 25 weeks of amenorrhea pregnant, in whom fetal Edwards syndrome was suspected and confirmed by cytogenetic study at birth.*

Keywords: Trisomy 18, facial dysmorphia, prenatal diagnosis

Introduction:

Trisomy 18 is a constitutional chromosomal disorder, defined by the presence of a supernumerary chromosome 18. It is the most common autosomal trisomy after trisomy 21, or Down syndrome [1]. Infants with trisomy 18 have a high mortality rate, secondary to the lethal malformations associated with this syndrome. Only 4% can survive their first year of life [2]. The complexity and severity of the clinical picture at birth and the high rate of neonatal and infant mortality emphasize the interest of prenatal diagnosis of this pathology. We report the case of a parturient admitted to us for pregnancy follow-up at 25 weeks of amenorrhea, in whom the diagnosis of Down syndrome was suspected and confirmed by karyotype after birth.

Case Presentation:

Mrs H.G, 33 years old, consanguineous marriage, mother of 2 children delivered by vaginal route with good psychomotor development, pregnant at 25 weeks of amenorrhea, admitted to the obstetrical emergency room for increased uterine height compared to the gestational age. The physical examination showed a hemodynamically and respiratory stable patient with obstetrical examination: uterine height at 28 cm, active fetal sounds well perceived, and obstetrical ultrasound: evolving pregnancy in cephalic presentation with the presence of an anterior median mass with extra-abdominal development connected to the abdominal wall by a collar, this mass measures 45 mm and it is limited by a peritoneal membrane evoking an omphalocele, The herniated viscera contained in the omphalocele are the intestinal anses, we also note the presence of a microretrognatism with anomalies of the extremities, the biometries were lower than the 3rd percentiles, the amniotic fluid was in hydramnios. (The screening for diabetes was negative). «Figure 1»

In view of these sonographic signs, we suspected Edwards syndrome and indicated an amniocentesis which was refused by the patient.

The evolution was marked by the occurrence of uterine contractions at 26 weeks of amenorrhea, admitted to the emergency room with complete dilatation of the uterine cervix, delivery by vaginal route of a female newborn, birth weight 560 g, The newborn presented signs of craniofacial dysmorphia made of horizontal palpebral clefts, snub nose, wide nasal root, gaping fontanelles, small mouth and important micrognathia, the high forehead and the micrognathia give the face a triangular shape. This is associated with visceral malformations such as omphalocele and limb anomalies such as clenched hands: closed fists, with overlapping fingers and at the level of the external genitalia we note a clitoral hypertrophy «Figure 2». The aftermath was marked by the death of the baby a few hours after its birth.

The cytogenetic study carried out allowed the demonstration of a supernumerary chromosome for the 18th autosomal pair, thus confirming the diagnosis of trisomy 18 in its free and homogeneous form «Figure 3».

Discussion:

The prevalence of trisomy 18 is variable. Worldwide, it is estimated at 1/6000 live births, the most affected being female [3]. The complexity and severity of the clinical picture at birth and the high rate of neonatal and infant mortality underline the interest of prenatal diagnosis of this pathology. In chromosomal pathology, in the case of a history of a child with a chromosomal anomaly or the presence of a chromosomal disorder in one of the parents, a fetal karyotype is proposed to search for a chromosomal anomaly in the fetus. In the absence of this history, trisomy 18 is evoked antenatally thanks to obstetrical morphological ultrasound. Thus, the main ultrasound signs of appeal are intrauterine growth retardation, increased nuchal translucency and absence of nasal bone (also used in Down syndrome and Patau syndrome) which are observed in 66% of fetuses with trisomy 18 [10], as well as signs in favor

of visceral and extremity malformations, including permanently closed fists. In its conventional approach, prenatal diagnosis of a genetic, chromosomal disease, such as trisomy 18, is based on cytogenetic analyses of samples of fetal origin, obtained by invasive procedures such as choriocentesis (chorionic villi), amniocentesis (amniotic fluid) or cordocentesis (fetal blood). These analyses are performed by conventional cytogenetics (karyotype) or molecular cytogenetics (fluorescence in situ hybridization) [11]. Molecular biology tools can also provide information on the number of chromosomes 18 by analysis of DNA through genetic polymorphisms or by relative genome quantification techniques [11]. In our case, the diagnosis of trisomy 18 was suspected during the morphological ultrasound in front of the presence of a polymalformative syndrome made of microretrognatism, a visceral malformation such as omphalocele and extremity anomalies.

In postnatal, the diagnosis of Edwards syndrome is clinically evoked in front of a hypertonic newborn with a poly-malformative syndrome. Like most autosomal chromosomal disorders, it is also accompanied by growth retardation (trisomy 21 for the best known) [4]. The craniofacial dysmorphism characteristic of this syndrome associates microcephaly with protrusion of the occiput, a receding forehead, characteristic low set and pointed ears, a small mouth with an ogival palate and microretrognathy. Extremity anomalies are represented by the "supplicant" position of the forearms, closed fists and fingers in permanent flexion. Thus, the index finger overlaps the 3rd finger and the little finger overlaps the 4th finger. The pelvis is narrow with varus equinus club feet.
There are several visceral malformations associated with trisomy 18, including cardiac, pulmonary, renal and digestive, with omphalocele and diaphragmatic hernia [5,6]. In our case the fetus had characteristic malformations with dysmorphic facies with omphalocele and extremity anomalies. Furthermore, survival is poor and only one in 10 newborns reaches the first year of life, with female infants having the longest survival time [3]. The main causes of death are cardiomyopathy, heart failure and respiratory failure [7-9]. Thus, survival is 42% in the first week; 29% in the first month, 12% at 3 months and 8% at 6 months [2].
Genetic counseling should be performed, in which it is explained that for a couple with a child with free and homogeneous trisomy 18, the probability of recurrence in the next pregnancy is 1% [3, 8]. In cases where trisomy 18 is partial, it is necessary to perform a karyotype in the parents to eliminate carriers with a balanced translocation, including the trisomic segment, because in these cases the probability of recurrence is greater [3]. In addition, the incidence of trisomy 18 increases with advanced maternal age. Eighty percent of cases are the result of maternal meiotic non-disjunction, and 5% of cases are due to paternal meiotic non-disjunction. Exceptionally, trisomy 18 is secondary to a chromosomal translocation [1].

Conclusion:

Trisomy 18, being a pathology of poor prognosis, its diagnosis must be made antenatally, its management is limited to comfort care, since surgical management of associated visceral malformations does not improve the prognosis [2].

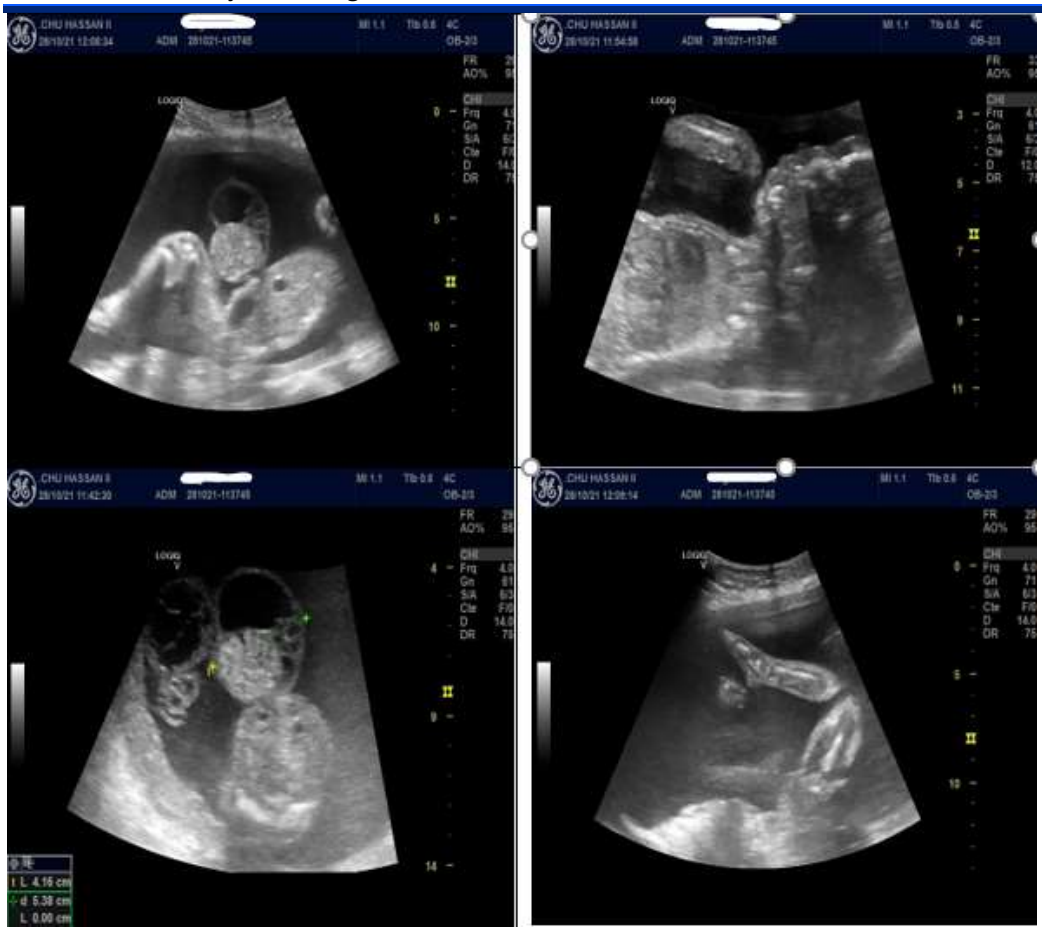

Figure 1: ultrasound images showing the malformations described above: omphalocele, retrognatism with amniotic fluid in hydramnios

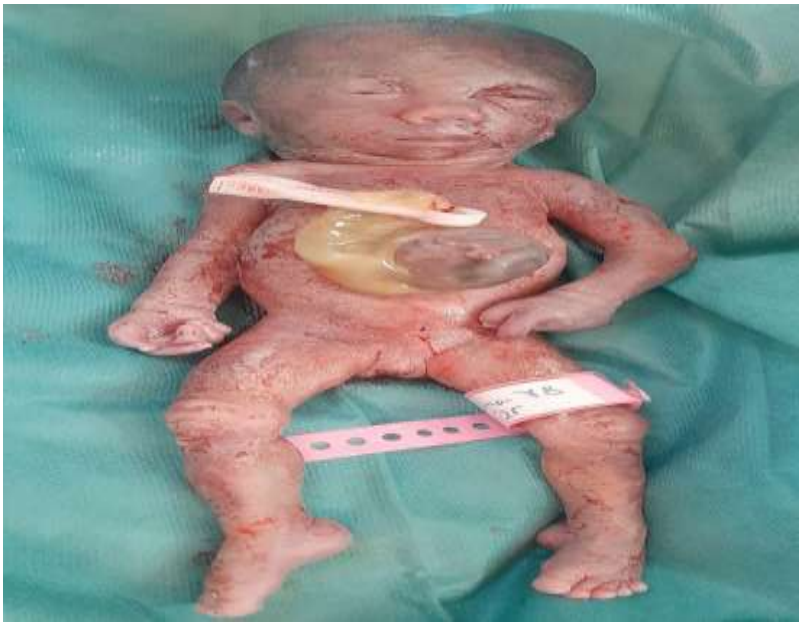

Figure 2: phenotype of the newborn at H1 of life showing facial dysmorphia, retrognatism, omphalocele and closed fists

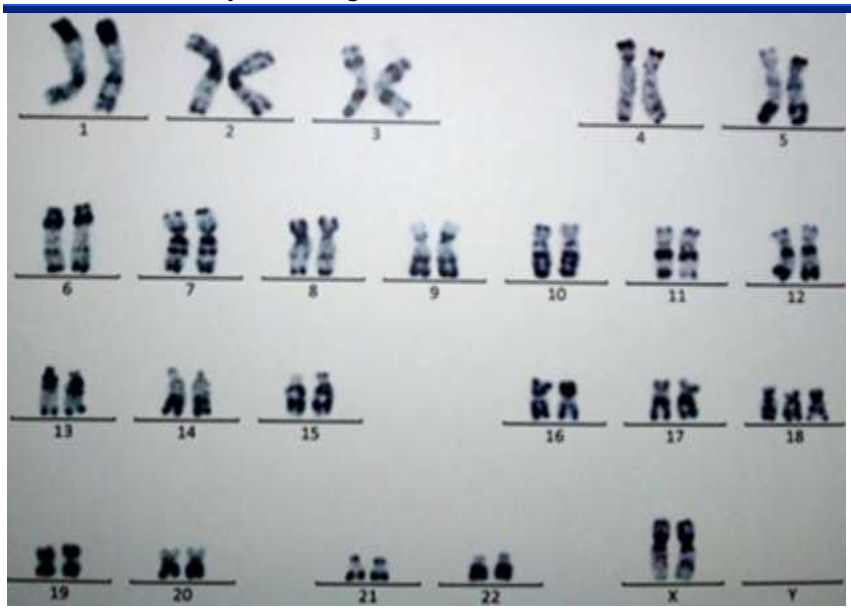

Figure 3: cytogenetic study in RHG banding, resolution 400 bands, highlights a female karyotype, and the presence of a supernumerary chromosome 18, confirming the diagnosis of free and homogeneous trisomy 18: 47,XX+18

Conflit d'intérêt

Les auteurs ne déclarent aucun conflit d'intérêt.

Figures

Figure 1: ultrasound images showing the malformations described above: omphalocele, retrognatism with amniotic fluid in hydramnios

Figure 2: phenotype of the newborn at H1 of life showing facial dysmorphia, retrognatism, omphalocele and closed fists

Figure 3: cytogenetic study in RHG banding, resolution 400 bands, highlights a female karyotype, and the presence of a supernumerary chromosome 18, confirming the diagnosis of free and homogeneous trisomy 18: 47,XX+18

Références :

1. Jones KL. Philadelphia: Saunders; 1997. Smith´s Recognizable Patterns of Human Malformation. [Google Scholar]
2. Lorenz JM, Hardart GE. Evolving medical and surgical management of infants with trisomy 18. *Curr Opin Pediatr.* 2014 Apr;26(2):169–76. [PubMed] [Google Scholar]
3. Cereda A, Carey JC. The trisomy 18 syndrome. *J Rare Dis.* 2012;7(1):81. [Google Scholar]
4. Bouhours-Nouet N, Donzeau A, Coutant R. Conduite pratique devant une anomalie de la croissance. *J Pédiatrie Puériculture.* 2017 Avr;30(2):67–97. [Google Scholar]
5. Jean de Grouchy, Catherine Turleau. «Atlas des maladies chromosomiques» *Expansion Scientifique Française; 2e édition-1982.*
6. Dubois M. Découverte d´un nouveau-né malformé en salle de travail: L´«annonce» de l´anomalie. *J Pédiatrie Puériculture.* 2004 Juin;17(4):229–32. [Google Scholar]
7. Carey JC. Trisomy 18 and trisomy 13 syndromes. *Wiley Online Library.* 2010:807–23. [Google Scholar]
8. Kosho T, Nakamura T, Kawame H, Baba A, Tamura M, Fukus-hima Y. Neonatal management of trisomy 18: Clinical detailsof 24 patients receiving intensive treatment. *Am J Med Genet.* 2006 May 1;140(9):937–44. [PubMed] [Google Scholar]
9. Niedrist D, Riegel M, Achermann J, Schinzel A. Survival with trisomy 18--data from Switzerland. *Am J Med Genet.* 2006 May 1;140(9):952–9. [PubMed] [Google Scholar]
10. Geipel A, Willruth A, Vieten J, Gembruch U, Berg C. Nuchal fold thickness, nasal bone absence or hypoplasia, ductus venosus reversed flow and tricuspid valve regurgitation in screening for trisomies 21, 18 and 13 in the early second trimester. *Ultrasound Obstet Gynecol.* 2010 May;35(5):535–9. [PubMed] [Google Scholar]
11. Costa J-M, Bazin A. Approche non invasive du diagnostic prénatal biologique: du génotypage rhésus D à la trisomie 21. *La Revue Sage-Femme.* 2008 Juill;7(3):130–3. [Google Scholar]