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DIABETIC EMBRYOPATHY OR A NEW GENETIC SYNDROME

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ABSTRACT

We hereby present the case of a pregnant woman, acknowledged with type I diabetes mellitus, aged 25 years old, who during the 23rd week of pregnancy aborted a polimalformed fetus, with severe facial dysmorphia, multiple congenital anomalies of the vital bodies, and polydactyly of the feet.

KEYWORDS: polimalformed fetus, facial dysmorphia, ultrasound examination, prenatal diagnosis, genetic syndrome, diabetes mellitus.

INTRODUCTION

The genetic syndrome is represented by all the signs and symptoms that together make up the clinical picture of genetic disorders.[1]

The prenatal detection of the congenital malformations can be done through noninvasive prenatal diagnosis methods (ultrasound examination) and/or invasive methods (amniocentesis, chorial villosity biopsy), supplemented by genetic investigation and genetic advice.[2-5]

MATERIALS AND METHODS

Pregnant woman, acknowledged with type I diabetes mellitus, aged 25 years old, during the 23rd week of pregnancy aborted a plurimalformed fetus, due to the fact that the mother requested the termination of the pregnancy for medical reasons, because the fetal morphology ultrasound examination performed a week before, pointed out the presence of a fetus with severe and multiple congenital malformations.

The ultrasound investigation was performed in a private clinic, from Bucharest, Romania, with a Voluson E10 Ultrasound machine, by a maternal fetal medicine specialist.

RESULTS

The ultrasound examination pointed out a mono-fetal pregnancy, 22 weeks advanced, in evolution with dysmorphic facies, micrognathia, low-set ears, flat face, very tall and flattened forehead, hypoplastic fetal nose and cheiloschisis (Fig. 1 and Fig. 2).

At the level of the thorax, anomalies of the vertebras and malformed heart were noticed, with hypoplasia of the sigmoid valve of the pulmonary artery and the pulmonary trunk.

The ultrasound examination of the abdomen and pelvis have shown: bilateral renal agenesis, hypoplasia of the urinary bladder and male external genital organs, but hypoplastic, with testicles that did not descend into the scrotum.

At the level of the lower extremities, the ultrasound examination pointed out: equine leg, polydactyly and large fingers.

Considering the result of the ultrasound examination, during the genetic counselling, the parents were explained the severity of the fetal malformations. They understood the fact that they had a genetically affected fetus, reasons why they chose to terminate the pregnancy.

The anatomopathological examination of the fetus confirmed the results of the ultrasound examination. The anatomical pathology of the fetal skull also pointed out the presence of a 30/40 mm hamartoma at the level of the septum pellucidum. The fetal karyotype performed post-abortum indicated a normal male chromosomal formula: 46, XY (Fig. 3).

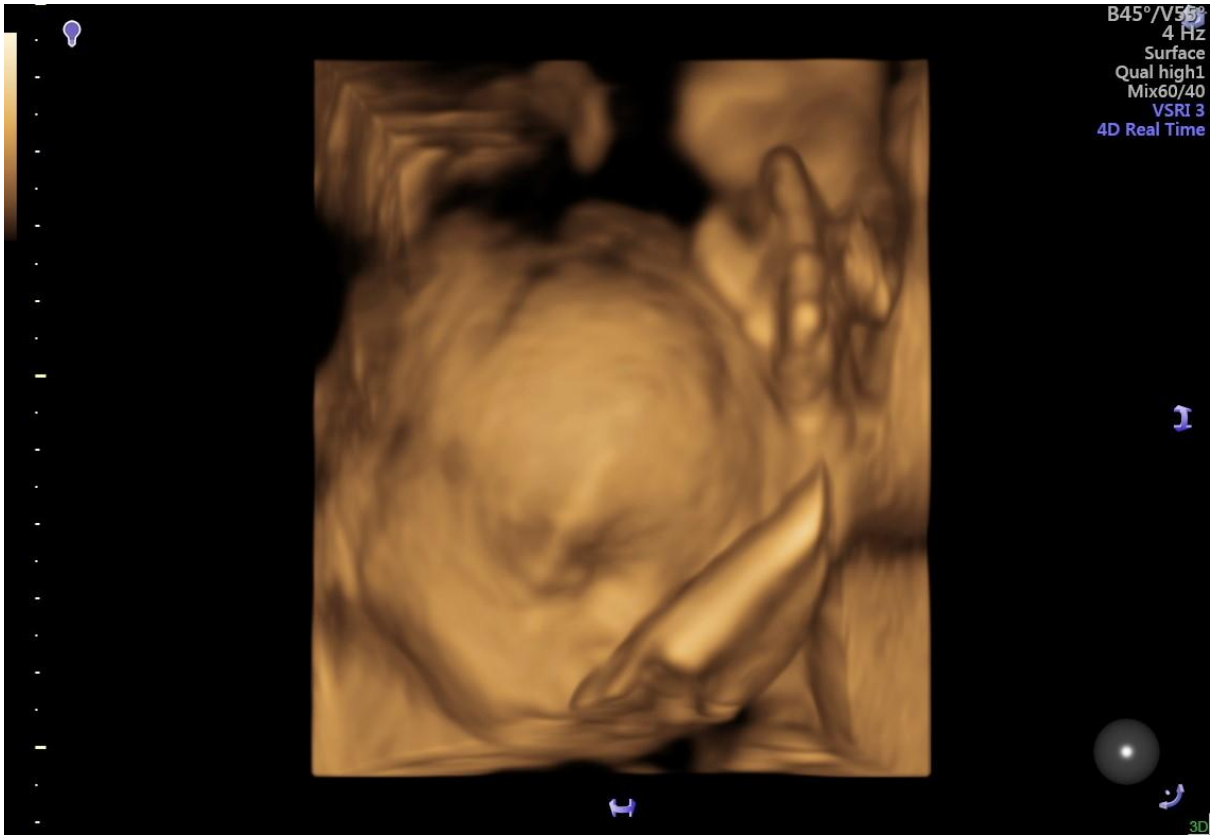

Figure 1 – 4D Real Time ultrasound examination: Fetal facial morphology.

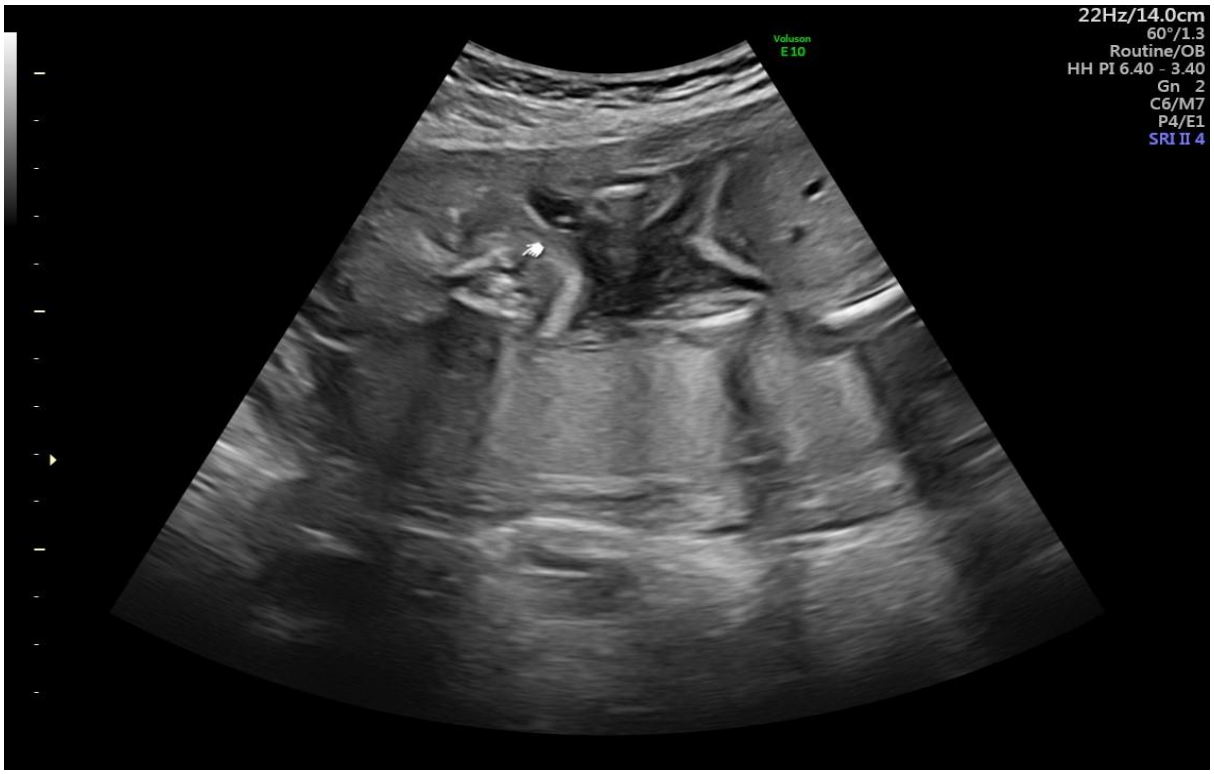

Figure 2 - 2D ultrasound examination: Cheiloschisis.

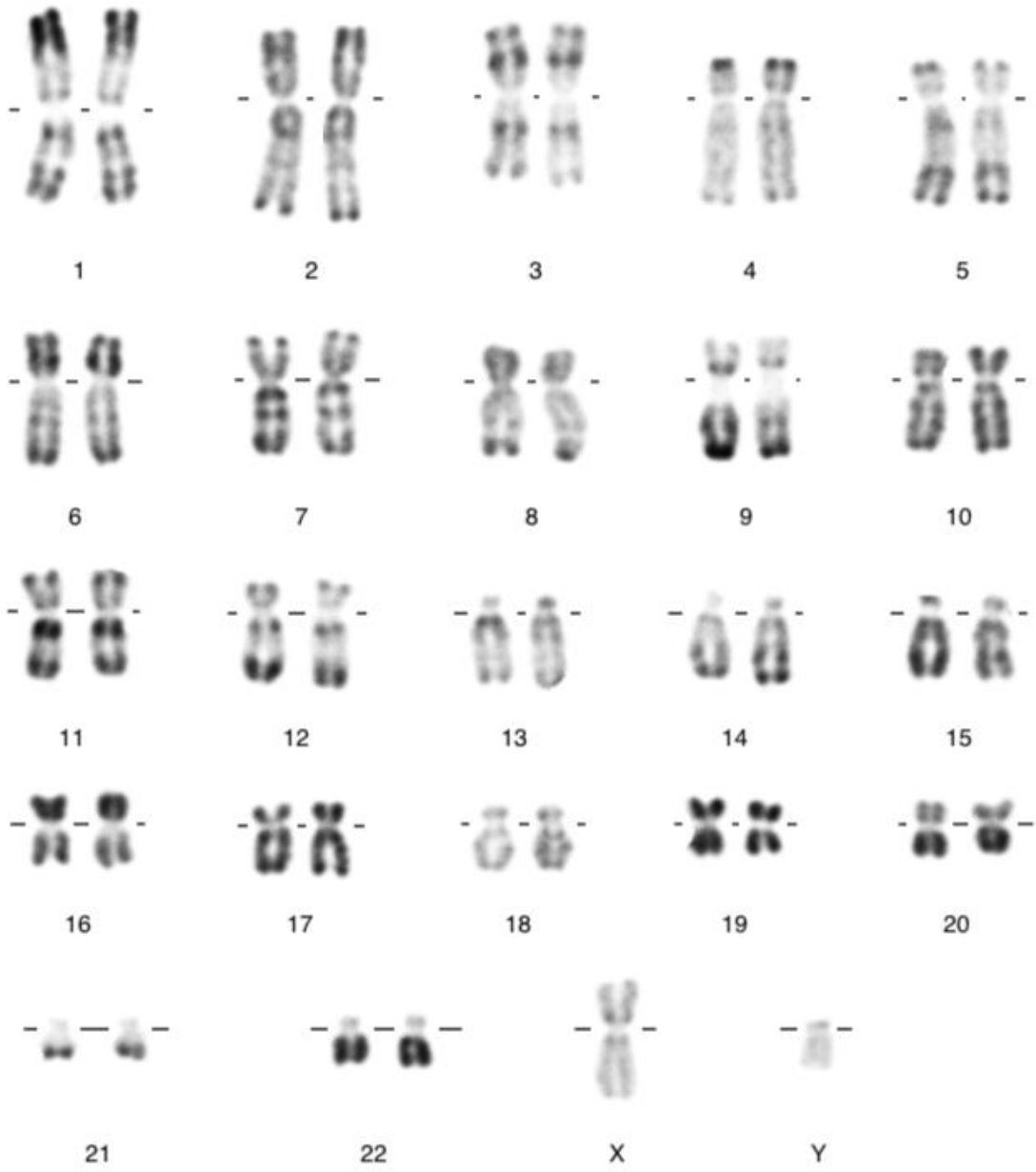

Figure 3 - Fetal karyotype: 46, XY.

DISCUSSION

The presented features are suggestive for the association of the malformations encountered in VACTERL, characterized by congenital vertebral anomalies, anal atresia, cardiac defects, tracheoesophageal fistula, anomalies of the kidneys and malformed limbs, but the presence of the polydactyly and of the facial dysmorphias infirm this diagnosis.[6-11]

Polydactyly, renal agenesis, vertebral and cardiac malformations are characteristics known to be those of diabetic embryopathy.[12]

However, this child also displays a series of different malformations and dysmorphic traits, which suggest another syndrome not described until present.

The differential diagnosis includes the acrocallosal syndrome, the oral-facial-digital syndrome, the campomelic syndrome, the Pallister-Hall syndrome, the Patau syndrome, the Edwards syndrome, the Down syndrome and even maternal diabetic embryopathy.[13-15]

CONCLUSION

The ultrasound prenatal screening for congenital malformations is indispensable in the early prenatal diagnose of severe congenital malformations, incompatible with postnatal life, and the personalized genetic examination is of real help to the mother/couple, and also to the society as a whole, allowing the personalized management of the affected cases, which will limit the incidence of the genetic defects in newborns.

Authors' contributions

All authors contributed equally with the first-author, in the preparing, review and editing of the article. All authors read and approved the final version of the manuscript.

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