Bilateral femur agenesis in Femoral - Facial syndrome in a fetus of a diabetic mother

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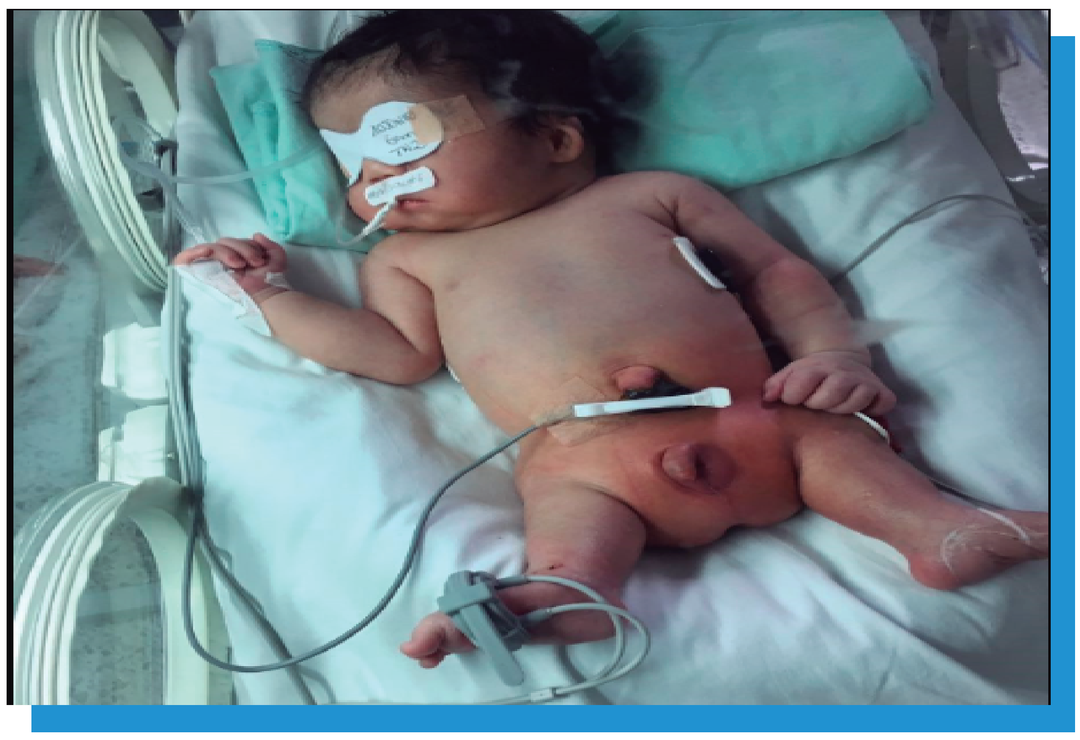
Introduction

Bilateral agenesis of femurs is one of the less common malformations found in the literature, there are 5 reported cases (3-4), this is the first in Colombia and in addition to bilateral femur agenesis we described Face and Cardiac malformations, both findings with strong association to maternal hyperglycemia.



Case

A 27-year-old patient G3P2C2V2 with a pregnancy of 27.1 weeks with 4 years old insulin requesting diabetes and first cesarean section due to fetal macrosomia, who consulted our hospital for obstetric ultrasound with fetal malformations. She had suspended insulin therapy because believing it was risky to find out late that she was pregnant.
Severe hyperglycemia was documented. Obstetric ultrasound (Figure 1): bilateral femur agenesis and cardiac malformation. Fetal echocardiogram finding the outflow tracts identified a single, great vessel containing a single valve and overriding a ventricular septal defect, abnormal cardiac axis deviated to the left: a possible type II truncus arteriosus. TORCH test was negative. Amniotic fluid cell culture failure, so a chromosomal analysis cannot be performed.
At 38 weeks a elective caesarean section was performed, obtaining a male new-born of 2806gr, Apgar 6/8 /10, Ballard 37 weeks with findings of bilateral microtia, right renal hypoplasia, bilateral cryptorchidism, X-ray: bilateral femur agenesis with secondary acetabular dysplasia (figure 2). Echocardiogram confirms type II truncus arteriosus and VSD. Normal karyotype.

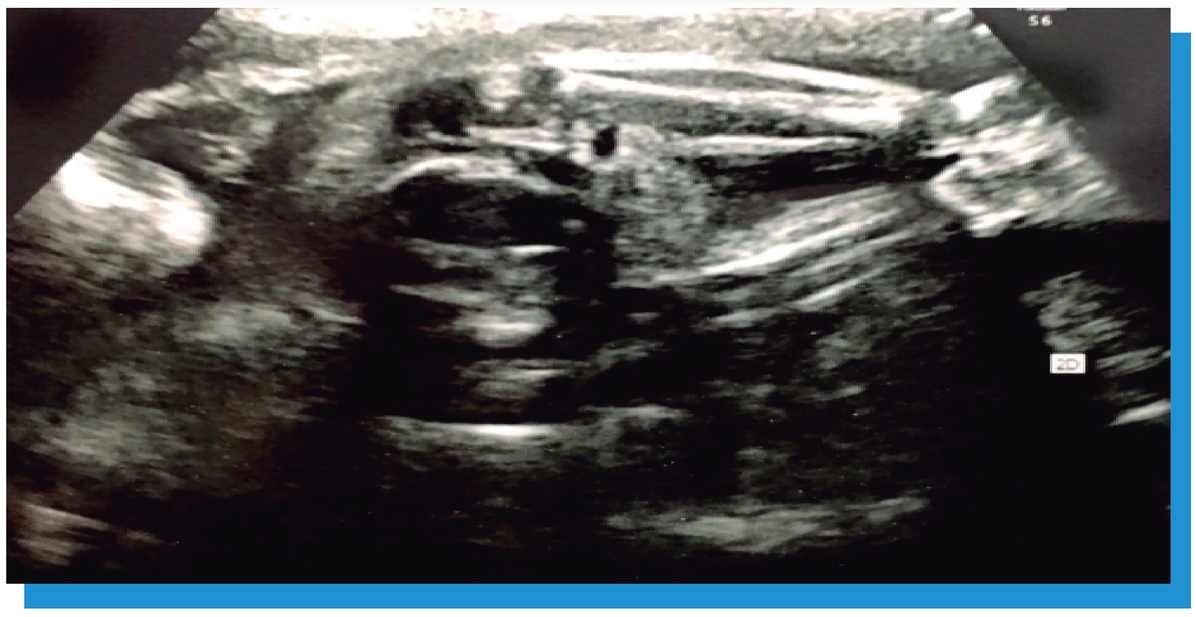


Fig 1: se ilustra agenesia bilateral de fémur , nótese tibia y peroné bilateral presentes en comunicación con acetábulo.

Discussion

Although femoral hypoplasia have been described in relationship with the facial femoral syndrome since 1975 (2-5) bilateral femoral agenesis is a variant in sporadic cases (1-7). In addition to bilateral femur agenesis, it is related to cardiac, facial and genitalia malformations, all of them have been strongly related to maternal hyperglycemia, infectious factors and genetic factors.
In the specific case of our patient, the most important infectious and genetic factors were ruled out. Maternal hyperglycemia is the most possible etiological factor associated in this case. There are several situations in which there is a genetic expression problem in which there is DNA damage associated with hyperketonemia. Oxidative stress and hyperglycemia produce fetal malformations.
Maternal diabetes as an entity associated with fetal malformation. The classical caudal regression syndrome is a disorder that impairs the development of the lower half of the body. Affected areas can include the lower spine and limbs, the genitourinary tract, and the gastrointestinal tract (6 -8) Femoral – facial syndrome has been described associate as well with severe maternal hyperglycemia in diabetic patients.

Conclusion

Although the teratogenic mechanisms leading to perturbed organogenesis are still incompletely understood in diabetes, we know the spectrum of malformations related to maternal diabetes is wide, and can be associated with different parts of the body, including the cardiac and outflow tract, craniofacial structures, as well as the central nervous, gastrointestinal, musculoskeletal, and urogenital system. But although maternal diabetes is a risk factor for fetal malformations, the real risk factor is hyperglycemia. So an adequate control of blood glucose levels prevents fetal anomalies. Pre-Pregnancy Counselling for women with diabetes considering pregnancy and early prenatal control should become in a public health policy in order to reduce fetal malformations.

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