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Case Report

Are Amniocentesis Results 100% Reliable?

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Abstract

Beckwith-Wiedemann syndrome (BWS) is characterized by neonatal hypoglycemia, macrosomia, macroglossia, omphalocele, visceromegaly and ear creases.

We presented a case of a female newborn with phenotype of BWS.

The prenatal diagnosis showed normal karyotype. The postnatal high resolution band karyotype confirmed the diagnosis with change in the chromosome 11. The father and paternal grandfather showed equilibrated translocation of terminal part of short arm of chromosome 3 and 11.

The reason why the karyotype was normal is because during the amniocentesis, the high resolution band is more difficult to obtain in culture and normally this technique is not use. To avoid repeat this situation in the next pregnancy, we need to do FISH of chromosome 11 to exclude BWS. The risk of recurrence is 50%.

Keywords: *Beckwith-Wiedemann Syndrome; Amniocentesis; FISH*

Background

Beckwith-Wiedemann syndrome (BWS) is characterized by neonatal hypoglycemia, macrosomia, macroglossia, omphalocele, visceromegaly and ear creases.

The incidence is 1/12000 newborns, with female predominance.

The inheritance is complex.

Possible patterns include autosomal dominant transmission with variable expression and disomy.

In case of duplication of 11p15.5, the origin is mainly the father side.

In case of equilibrated translocation of 11p15.5, or insertion, the responsible is maternal side in most of the cases.

The gene CDKN1C confirm the diagnosis in those cases with normal chromosome test.

The BWS is associated with more risk of cancer, particularly hepatoblastoma, Wilms tumor and neuroblastoma.

Case Report

Female newborn, 40 weeks of gestational age, born by suction. Birth weight: 3875g (P90), length: 53 cm (P90), head circumference: 33 cm (P50). Apgar 7/10. IIGIP, one spontaneous abortion at 5 weeks of gestational age.

The parents are unrelated and healthy. Mother: 24y old. Father: 28y of age.

Prenatal diagnosis was performed because the fetus showed bilateral ecstasy of the kidneys. The karyotype showed 46XX with normal alpha-fetoprotein (AFP) in the amniotic fluid.

On physical examination after born, was detected hypotonia, macrosomia, macroglossia, creases in the ears (Figure 1).

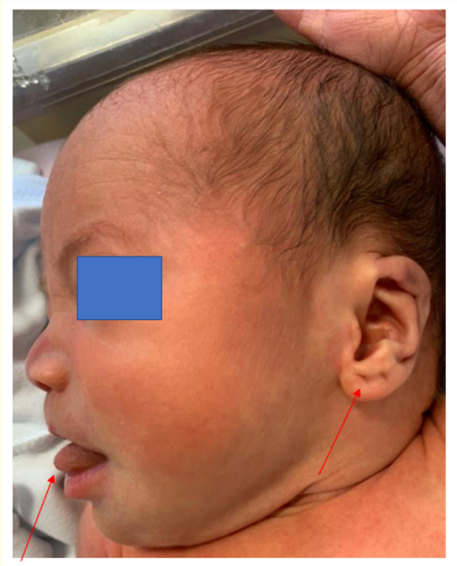

Figure 1: *Macroglossia and ear crease.*

Because the phenotype was compatible with BWS, karyotype was repeated and the result showed terminal deletion of short arm of chromosome 11: 46 XX, del(3), t(3,11), (p26.2:p15.4) compatible with the suspected syndrome. The father and paternal grandfather showed equilibrated translocation of terminal part of short arm of chromosome 3 and 11.

The AFP was high at birth: 1654 UI/ml (N: 0.0 - 5.0) with normalization at 6 months of age.

Chest x-ray and abdominal ultrasound for screening of tumor showed so far normal results.

The evolution of the patient revealed delay of motor skill at 16 months and at 17 months was performed surgery for reducing the size of the enlarged tongue.

Discussion

Our index case was suspected because the newborn showed phenotype of BWS.

The patient didn't presented with neonatal hypoglycemia, that is frequent in 61% of cases or omphalocele.

In the prenatal period, the presence of hydramnios, a large placenta and increase length of the umbilical cord, are signs of BWS.

It is important to do the screening of AFP and abdominal/renal ultrasound (risk of hepatoblastoma/Wilms tumor respectively) every 3 months until 3 years of age and after every 6 months until the age of 5 years and finally annually until 18 years old, because 25% of Wilms tumor appear after 5 years of age.

Yearly, we need to check chest x-ray for screening of neuroblastoma, although is rare.

Around 15% of cases the patient with BWS showed mental retardation. Our case we found development delay, particularly delay motor skills. Early stimulation is the most important treatment in this situation.

Tongue surgery is controversy but when we have a patient with sleep apnea, the reducing of the enlarged tongue is the best solution.

In our index case, the diagnosis was done after birth with change in the chromosome 11. The karyotype with high resolution band confirmed the BWS. During the amniocentesis, the high resolution band is more difficult to obtain in culture and normally this technique is not use. This explain why the karyotype was normal in the prenatal diagnosis and later after birth showed an abnormal result.

For future prenatal diagnosis and genetic counselling, we need to inform the parents that the risk of recurrence is 50% in the next pregnancy. To exclude an affected case, we need to do FISH of chromosome 11 as additional test of conventional karyotype during the amniocentesis [1-12].

Conclusion

Amniocentesis still an important prenatal examination but is not a 100% reliable test.

If for any reason, (with karyotype or microarray normal in the amniocentesis), the phenotype of the patient remind us a syndrome or the patient showed polymalformation or dysmorphia, we must repeat the above test or study the exome or genome of the child.

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