CURRARINO SYNDROME - A PRE AND POST NATAL DIAGNOSIS

CORRELATION: CASE REPORT AND LITERATURE REVIEW

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PROBLEM STATEMENT

- Currarino syndrome is a rare autosomal dominant disorder and affected subjects are at-risk of serious complications, leading to significant morbidity and mortality;
- This disease is formed by a Currarino triad: sacral anomalies, presacral mass and an anorectal malformation; it exhibits variable expressivity and many heterozygotes are asymptomatic;
- Prenatal ultrasonography may identify the presence of a presacral mass, although it is not an accurate or complete investigation for someone at 50% risk.

METHODS

- We discuss a case report of Currarino Syndrome with an early diagnosis through imagiological study during pregnancy for prenatal diagnostic purposes.

RESULTS

24 years-old
Healthy primigravida
Suspicion of spina bifida occulta at 21 weeks

Term vaginal delivery;
Female newborn, 3315gr, Apgar score 9/9/10
Perianal and endorectal polypoid mass

Obstetric ultrasounds and the first fetal magnetic resonance:

- Presented first a closed sacrococcygeal dysraphism without meningocele and cerebral alterations like Arnold-Chiari malformations;
- At 29 weeks, a small meningocele with lowered medullary cone appeared during second fetal magnetic resonance;

Image 1. Fetal magnetic resonance at 29 weeks.

- **Magnetic resonance:** Partial sacral agenesis + anterior myelomeningocele + rectosacral mass + anchored medulla (cone in L4);
- After these results neurosurgical team analysed the fetal resonance and discovered similar neural tube defects in the asymptomatic mother: sacral agenesis and anterior meningocele;
- Afterwards, we had an early **CURRARINO SYNDROME DIAGNOSIS** and performed a neurosurgery to allow the medullar distal release and filum terminal section;
- The child has 1 year and a normal physical and psychomotor development, with symmetric leg movement and a normal intestinal transit;
- She will be monitored by a multidisciplinary team to evaluate her evolution, specially locomotion and sphincter control.

CONCLUSION

- Despite the difficulty of doing a prenatal diagnosis of this syndrome, whenever we detect a presacral mass during the obstetric ultrasounds, we advise the inclusion of Currarino syndrome in the differential diagnosis of neural tube defects;
- We should also expand genetic counselling and promote screenings, allowing identification of asymptomatic heterozygotes;
- The recognition of at-risk subjects should lead to better planning of pregnancies and appropriate management of affected children at birth.