Zellweger Syndrome
A CASE REPORT
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Introduction:
The Zellweger Syndrome is a rare peroxisome biosynthesis disorder (incidence: 1 in 50,000 to 100,000 live births), characterized by a generalized loss of peroxisomal functions. This is a fatal hereditary autosomal recessive disorder, whose features include craniofacial dysmorphism and profound neurologic abnormalities.

Objective:
We report a case of a fetus with brain abnormalities diagnosed with Zellweger Syndrome after birth.

Methods:
Case report of Zellweger Syndrome.

Results:
41 years old pregnant woman; gesta 5/ para 4
Routine prenatal care in our hospital due to maternal age
No relevant personal or family history

12wks+6d  First trimester screening test with low risk for aneuploidies

21wks+3d  Anomaly scan:
  - bilateral mild ventriculomegaly;
  - hyperereogenic bowel;
  - humerus on the 10th centile;
  - right talipes equinovarus.

24 wks  Fetal brain magnetic ressonance imaging (MRI) scan: unspecific findings

25 - 34 wks  Follow up scans: stable findings

30 wks  MRI: fronto-temporo-parietal and perisylvian polymicrogyria, with mild ventriculomegaly

37 weeks  Spontaneous labor with a normal delivery
Newborn: male, apgar score: 5/6/7
Need of resuscitation with positive pressure ventilation and intubation.

Neonatal period

Newborn presented:
  • Profound hypotonia;
  • An episode of seizures;
  • Dysmorphic craniofacial features;
  • Large fontanels;
  • Markedly separated cranial sutures;
  • Glomerulocystic kidney disease;
  • Bilateral cryptorchidism;
  • Bilateral cataracts.

Postnatal neuroimaging - consistent with the cortical abnormalities diagnosed prenatally;

Biochemical studies - increased plasma concentration of very-long-chain fatty-acid

Due to the clinical, biochemical and imaging data the diagnostic hypothesis was Zellweger Syndrome, which awaits confirmation by molecular testing.

Conclusion:
• The lack of family history and nonspecific findings during pregnancy surveillance made the antenatal diagnosis impossible in this case,
• However, the postnatal diagnosis is important as prenatal diagnostic test is available for subsequent pregnancies.

Bibliography:
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3 - "Prenatall genetic evaluation of the anomalous fetus", Neet Vora, MD; Sarah Harris, MS, Uptodate, April 2017