Partial agenesis of corpus callosum: case report and review of literature


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Objective: We present a case report of a disgenesis of corpus callosum (CC) associated with bone dysplasia

Methods: Review of clinical reports of pregnancy and pediatric follow up of the newborn

CASE REPORT:

41 year G1 with rhizomelic bone dysplasia

- 1st trimester US:
  - TN: 1.74 mm (normal), ßhCG: 0.432 (MoM); PAPPA: 0.596 (MoM)
  - Risk of Trisomy 21: 1/1056, Trisomy 13 or Trisomy 18: 1/2463

- 2nd trimester US: disgenetic CC + bilateral mild ventriculomegaly (10-12 mm) + cerebellar hypoplasia + limb shortening (femur length p3)
  - TORCH, parvovirus infection: negative
  - Karyotype and arrays: normal
  - MRI (22 and 31 weeks): partial agenesis of corpus callosum (absence of splenium) + cerebellar cystic encephalomalacia + moderate ventriculomegaly (12.8 mm)
  - Evolution of pregnancy: IGR + mild polyhidramnios in 3rd trimester
  - Cesarean section at 39 weeks (non-progression of labor)
  - Newborn: girl, 2495 g , Apgar 7/10
    - Post-natal MRI: confirmed previous findings
    - Bone X-Ray: shortening and delay of ossification of calcaneum
    - Arrays panels specific for bone dysplasia: negative
    - EEG: excess of beta activity (diffuse brain pathology)

- 3 years: need reinforcement of early neurologic intervention and rehabilitation (walk at 3 years). Mild psychomotor delay persists at the moment.

Conclusion: Most CC/CSP anomalies can be detected before 22 weeks of gestation. Related and non-related CNS anomalies must be ruled out. MRI can confirm the diagnosis and add new information (neuronal migration). Evolution of the newborns depend on associated anomalies.