Partial agenesis of corpus callosum: case report and review of literature ID: 4764

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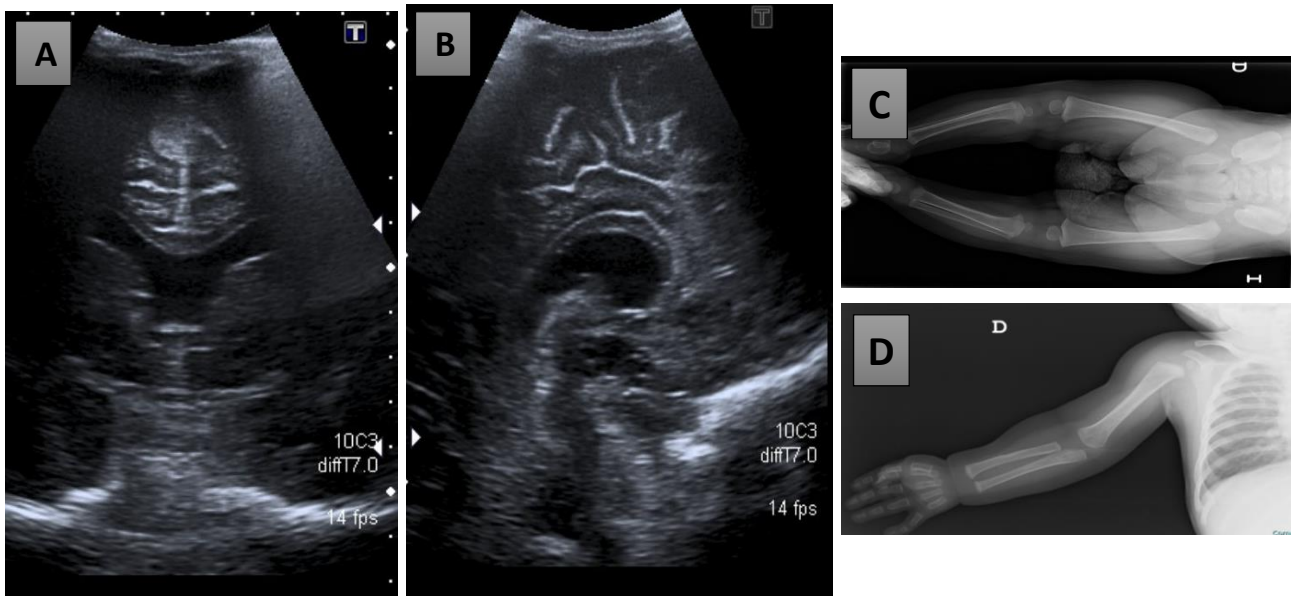
Objective: We present a case report of a disgenesis of corpus callosum (CC) associated with bone dysplasia

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



A-B: Neonatal Transfontanelar US: Absence of septum pellucidum. CC is thin and disgenetic (splenium of CC not visualized)(A: coronal view; B:sagittal view); C-D: X-Ray of IIEE (C) and SSEE (D): shortening of bones

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.

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.**