19th World Congress in Fetal Medicine

Advanced screening of congenital malformations. Prevalence of the corpus callosum anomalies

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Objective

After including direct observation and measurement of corpus callosum we present the prevalence of corpus callosum pathology (agenesis / dysgenesis) in our center over a ten years period.

Methods

Since 2011, direct visualization and measurement of the corpus callosum have been incorporated into our morphological screening ultrasounds. Retrospective review of data collected prospectively from cases of corpus callosum pathology. It is classified into two categories: a) Complete agenesis of the corpus callosum (total absence) and b) dysgenesis of the corpus callosum (which includes, corpus callosum with partial agenesis, short, narrow or thickened), suspected by ultrasound in the fetal medicine unit between June 2011 and June 2021 and postnatal follow-up of cases that did not perform Termination of pregnancy (TOP).

Results

During that period, 44 cases of corpus callosum pathology were identified in our center, 9 patients who attended a second opinion were excluded, so the remaining 35 were studied. During those ten years, 31736 obstetric processes were performed in our center, which means a prevalence of 1.1/1000. Of the 35 cases, 34 were detected prenatally and one was a postnatal diagnosis of corpus callosum hypoplasia in the study of a microcephaly. 11 cases were complete agenesis (CCA) of which 10 performed I and 23 cases of corpus callosum dysgenesis (DCC), including short, thinned or thickened corpus callosum, of which 11 elected TOP, 1 was a neonatal exitus of 25 weeks due to prematurity and 11 were full-term deliveries. The mean age at diagnosis was 22 weeks. Chromosomal/genetic anomalies were found in 5 cases (14%). The percentage of other associated abnormalities was 28% (10 cases). MRI added additional information in 3 cases (8.5%). A telephone/mail follow-up (6 months- 10 years) was attempted and 3 children with learning disabilities were identified out of the 10 we achieved follow-up (30%).

Conclusion

The incorporation of direct visualization of the corpus callosum allows to improve diagnostic capacity of the pathology of this structure. The usual indirect signs are not useful for dysgenesis. The prevalence of corpus callosum pathology is 1/1000 and due to its high association with other CNS and extracerebral anomalies, and genetic syndromes, it is important to perform array and MRI to all patients who wish to continue with the pregnancy to offer good counselling.