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# Abnormal cavum septi pellucidi: beyond corpus callosum agenesis

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## Objective

Cavum septi pellucidi (CSP) is one of the milestones on the second trimester morphology scan checklist. It is intimately related to septum pellucidum, therefore tied to not only the development of corpus callosum but also to the anterior and hippocampal commissure. Other issues apart from its presence, such as increased or reduced size, ratio (length/width) or echogenicity are not considered pathological if isolated. Although some conditions such as corpus callosum (CC) partial agenesis, di-George, Patau or Down syndrome are somehow increased and further studies might be advised by several authors.

## Methods

We performed a retrospective review of the cases referred to our Fetal Neurosonography Clinic due to abnormal but present CSP between January 2017 and April 2019. We correlated the size (Falco et al charts), ratio (Karl K et al. reference ranges) and echogenicity characteristics of CSP to the fetal-neonatal outcomes up to the time of submission.

## Results

We had 22 cases referred for suspicious CSP anomaly during the 28 months of study, mostly at 20 weeks' gestation. Abnormal CSP ratio was observed in 63.6% of cases (13 below 5$^{th}$ Percentile (P) and 5 above 95$^{th}$ P). 77% of cases (n=17) were small and just one above P95. Corpus callosum was assessed finding 5 cases with a short CC but just one dysgenesis amongst them. Four more cases of CC dysgenesis were found. Overall, in this cohort we diagnosed in the third trimester: two cases of lissencephaly-subcortical heterotopic band syndrome (L-SBH), three congenital Cytomegalovirus (CMV) and two CC dysgenesis. In the second trimester, within the week of referral, we diagnosed one case of neurofibromatosis type I, one case of hiperprolinemia type I, one case of hemimegalencephaly and one case of syntelencephaly. Postnatally, one Sotos syndrome was also diagnosed. Currently under postnatal work-up are two cases of CC dysgenesis awaiting exome sequencing analysis, a case of bilateral hipoacusia with cervical hypotone awaiting MRI, two ventriculomegaly - developed postnatally, one asymptomatic intrauterine growth restriction (IUGR) and one anterior horn asymmetry with hypospadias, all reaching age milestones. There were 2 completely normal cases with a normal CSP ratio and the remaining two cases have a normal prenatal assessment including fetal MRI awaiting labour and neonatal assesment. Most severe cases had an abnormal CSP ratio with hyperrefrigency (three congenital CMV, two L-SBH and one hyperprolinemia type I)) Nonetheless genetic studies were performed in only 13 patients (59%) and MRI in 7 cases (plus two on waiting list). Final results at the time of submission were 41% of pathological confirmed cases (n=9), 4 patients awaiting for delivery, 6 cases on postnatal workup (CC dysgenesis, postnatal ventriculomegaly and hypotonia) and 3 confirmed normal cases of 5, 7 and 12 months old.

## Conclusion

CSP is considered an important milestone in fetal brain development with a role which is not completely understood. Considering its strong relation to the limbic system due to the septum pellucidum, it is reasonable to consider an anomaly of CSP an indirect sign of brain development. Here in our series, we had 41% clearly pathological cases, diagnosed as a result of an anomalous CSP in morphology scan without agenesis of corpus callosum or lateral ventriculomegaly prenatal ultrasound signs (also 27% and 18% pending postnatal workup due to suspicious or subtle anomalies and labour respectively). Apart from CSP ratio, the hyperechogenicity was also an important variable to promote prenatal and postnatal neurosonographic follow-up.