Agenesis of the corpus callosum

Jaziri D, Hamdi A, Abid C, Cherni A, Boudaya F, Achour E, Chelli D
First Department of Obstetrics and Gynecology, Maternity Center Rabta Tunis, Tunis, Tunisia

Objective

To determine the sensitivity and specificity of ultrasonography in the diagnosis of agenesis of the corpus callosum through anatomical-ultrasound confrontation.

Methods

A retrospective study of 5 fetuses with agenesis of the corpus callosum during three years in service A of CMNT. When the diagnosis of agenesis of the corpus callosum was difficult to affirm (suboptimal views on ultrasound), a control ultrasound was repeated in few days. Some patients received additional ultrasound in the radiology department of Children's Hospital. All patients with suspected agenesis of the corpus callosum received fetal MRI after 23 weeks of gestation.

Results

During the study period, we had suspected by ultrasound an agenesis of corpus callosum in 12 cases. Patients with a diagnosis of suspected agenesis confirmed by MRI were selected. The average age was 31 years. Consanguinity was found in 2 couples. 40% of cases had a history of fetal malformation. The gestational age at which the CCA was suspected ranged from 23-26 weeks. Case 1: Utrasound: Microcephaly, suspicion of partial agenesis of the corpus callosum, facial dysmorphism, MRI: Severe microcephaly, total agenesis of the CC, lissencephaly, Outcome: Medical termination of pregnancy (MTP) at 27 weeks. Autopsy: Partial agenesis of the CC, lissencephaly, microcephaly. Case 2: Ultrasound: Suspicion of partial agenesis of the CC, Vermis agenesis, MRI: Dandy Walker Partial agenesis of the CC, Cerebellar hypotrophy, Outcome: MTP at 25 weeks. Autopsy, Total agenesis of the CC (poor preservation of the brain). Case 3: Ultrasound: Ventriculomegaly, suspicion of partial agenesis of the CC. MRI: Ventriculomegaly, ACC, schizencephaly or proencephalic cavity, vermis agenesis, cerebellar hypotrophy, Outcome: New born: Major hydrocephalus, axial and peripheral hypotonia, semi-lobar holoprosencephaly. Postnatal MRI: Psychomotor retardation, major hydrocephalus. Case 4: Ultrasound: bilateral hydrocephaly, suspicion of CCA, MRI: Absence of CC, triventricular dilatation with ascent of the third ventricle. Outcome: Postnatal MRI: bilateral triventricular dilatation, CCA. Case 5: Ultrasound: Dilatation of the third ventricle, moderate ventriculomegaly at 11 mm, partial agenesis of the corpus callosum, MRI: Complete agenesis of the corpus callosum without any other identifiable brain abnormality, Outcome: cesarean delivery at 35 weeks gestation, axial and peripheral hypotrophy, respiratory distress, neonatal death on day 2.

Conclusion

Agenesis of corpus callosum is a frequent cerebral malformation. It can be total or partial, isolated or most often associated with other malformations. Antenatal discovery by ultrasound needs a morphological assessment including MRI, a family investigation and karyotyping. Findings should be discussed by multidisciplinary medical and fetal staff and with parents leading or not to pregnancy termination according to whether isolated, associated with other anomalies, total or partial.

Case	Ultrasound	MRI	Outcome
1	- Microcephaly - Suspicion of partial agenesis CC - Facial dysmorphism	- Severe Microcephaly - Total agenesis of the CC - lissencephaly	TOP 27w, autopsy: Partial agenesis CC, lissencephaly, microcephaly
2	- Suspicion of partial agenesis CC - Vermis agenesis	-Dandy Walker - Partial agenesis of the CC - Cerebellar hypotrophy	TOP 25w, autopsy: Total agenesis CC (poor preservation of the brain)
3	- Ventriculomegaly - Suspicion of partial agenesis CC	-Ventriculomegaly - ACC - A schizencephaly or proencéphalique cavity - Vermis agenesis - Cerebellar hypotrophy	New Born: Major hydrocephalus Postnatal MRI:, semi lobar holoprosencephaly Action: psychomotor retardation major hydrocephalus, axial and peripheral hypotonia
4	-Bilateral hydrocephaly -Suspicion of agenesis CC	-Absence of CC -Triventricular dilatation with ascent of the third ventricle	Post natal MRI: Bilateral triventricular dilatation, agenesis CC
5	-Dilatation of the third ventricle -Moderate ventriculomegaly 11 mm -Partial agenesis CC	-Complete agenesis of the corpus callosum without other identifiable brain abnormality	CS 35w: Hypotrophy axial and peripheral respiratory distress, died on day 2