Case 7099

Agenesis of Corpus Callosum
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Section: Neuroradiology
Case Type: Clinical Cases
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Patient: 35 years, male

Clinical History:
A 35 year old male patient presented with severe headaches.

Imaging Findings:
The patient had headaches since two years ago, that became severe and disabling (interfering with daily activities). He went to medical attention because of more frequent periodic attacks of severe left-sided and pulsating headaches, without preceding aura. Accompanying complaints of nausea, vomiting, and extreme sensitivity to bright lights (photophobia) and sound (phonophobia). Migraine headaches were diagnosed and medical therapy was introduced. Family history was positive with the mother having migraine. The neurological examination was normal. Neuroimaging studies were done due to significant change in headache pattern, to exclude possible causes. Incidental finding is reported - agenesis of corpus callosum (without known cases of proved association).

Discussion:
The corpus callosum is the most prominent commissura of the human brain, connecting almost the entire neocortex of both hemispheres. The formation of the corpus callosum and its precursors occurs between about 8 and 20 weeks of gestational age, although it continues growing. Its size and thickness evolve also in the first months of life, depending mostly on myelin content. It normally forms from anterior to posterior (except for the rostrum, which forms last) and myelinates from posterior to anterior. It sweeps from the anterior commissura anteriorly to hippocampal commissura posteriorly. Most of the cerebrum and cerebellum form also at the same time.

Hypogenesis of the corpus callosum can be complete (i.e. agenesis) or partial and probably results from a vascular or inflammatory insult occurred before the 12th week of gestation. It frequently is associated with other malformations, chromosomal abnormalities and genetic syndromes. Agenesis of the corpus callosum (ACC) is among the most frequent human brain malformations. It is a heterogeneous condition, for which several different genetic causes are known - ACC as part of monogenic syndromes (such as Mowat-Wilson syndrome, Walker-Warburg syndrome, oro-facial-digital syndrome type 1) or complex chromosomal rearrangements. A variety of genetic causes of disorders of the corpus callosum (callosal agenesis/ dysgenesis) were identified with cytogenetic anomalies representing the most common underlying aetiology. Isolated commissural anomalies are rare; most cases are associated with complex telencephalic, diencephalic, or rhombencephalic malformations. Reduced cerebral hemispheric white matter volume and malformations of cortical development are seen in more than half of the patients, suggesting that many commissural anomalies are part of an overall cerebral dysgenesis. ACC and HCC appear to lie along a dysgenetic spectrum, as opposed to representing distinct disorders. In that cases, the inter-hemispheric cerebral axonal fibres do not migrate across the midline; the residual commissural fibres are distributed longitudinally as Probst bundles - they are located along the superomedial margins of the lateral ventricles and can protrude into the frontal horns.

Complete ACC is accompanied by absent cingulate gyrus and sulcus and dorsal or occasionally rostral
interhemispheric arachnoid cyst. Colpocephaly (dilatation of atria and occipital horns of lateral ventricles) is often seen and the 3rd ventricle extends superiorly (high-riding). Agenesis may be accompanied by isolated dilatation of the temporal horns of the lateral ventricles (does not imply early hydrocephalus) and is associated with abnormally shaped hippocampi due to their incomplete rotation. Associated anomalies are Chiari II, neuronal migration disorders, Dandy-Walker complex, holoprosencephaly, interhemispheric lipoma, migration anomalies, azygous anterior cerebral artery, and abnormalities of optic chiasm and pituitary gland. Although it is reported that isolated callosal agenesis may be asymptomatic, incidental detection of callosal agenesis is very rare. More typically, affected patients present with seizures, delayed development, mental retardation, macrocephaly and/or hypothalamic dysfunction. So, it is associated with a broad functional spectrum ranging from normal to severe cognitive impairment (that is often due to additional brain abnormalities).

Differential Diagnosis List: Agenesis of Corpus Callosum

Final Diagnosis: Agenesis of Corpus Callosum

References:

Description: It shows complete absence of corpus callosum (no crossing fibers above the ventricles) and the abnormal appearance of the medial hemispheric sulci (one of the hallmarks of ACC). The formation of the CC is associated with inversion (rotation) of the cingulate gyri, resulting in the formation of the cingulate sulci, superior to the gyri. When the CC doesn’t form, the cingulate gyri remain everted (unrotated) and tend to be small; thus, the cingulate sulci remain unformed. Persistent eversion of the cingulate gyri results in extension of the sulci of the medial hemispheres all the way into the third ventricle in a radial pattern. The medial hemispheric sulci has also a rather disorganized appearance, probably secondary to disorganization of white matter tracts in and near the midline. There is no evidence of anomalies in the posterior fossa. Cerebellar vermis seems normal. Origin:
Description: Interhemispheric fissure is normal, without cystic ... (associated)... Origin:
Description: Images demonstrate parallel, straight and vertical orientation of the lateral ventricles. Lack of callosal tissue crossing the midline. There is disproportionate prominence of the atria and occipital horns of the lateral ventricles - Colpocephaly. Origin:
Description: Coronal image shows high third ventricle, crescentic frontal horns (LV) compressed by Probst bundles. The caudate heads and lentiform nuclei keep the size of the frontal horns relatively small, even in the absence of the CC. However, the frontal horns are convex laterally (instead of concave) as in normal cases. Origin:
Figure 5

Description: It shows the typical ventricular configuration known as colpocephaly (dilatation of trigones and occipital horns of the lateral ventricles), present in ACC. The temporal horns also frequently enlarge, mainly inferomedially into the location in which the cingulum (cingulate fasciculus) is usually located. Origin:
Description: CT scan shows parallel and straight orientation of the lateral ventricles. There is disproportionate prominence of the atria and occipital horns of the lateral ventricles - Colpocephaly. No other lesion is identified (neoplasm, vascular malformation) to explain symptoms. Because of these findings, suggestive of dysgenesis of corpus callosum MR was done. Origin:
Description: On axial imaging of the brain, the Corpus Callosum is visible as a narrow band of tissue in the shape of "I" between the lateral ventricles (LVs). On sagittal and coronal imaging it appears as a curved shape separating the cingulum superiorly from the LVs inferiorly. Origin: