Case 1584

Eurorad

Multiple sclerosis associated with complete corpus callosum agenesis

Published on 23.07.2002

DOI: 10.1594/EURORAD/CASE.1584
ISSN: 1563-4086
Section: Neuroradiology
Imaging Technique: MR
Imaging Technique: MR
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Case Type: Clinical Cases
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Patient: 32 years, female

Clinical History:

The patient presented with a 4 month history of paraesthesias and numbness of the extremities. She had no past history.

Imaging Findings:

The patient presented with a 4 month history of paraesthesias and numbness of the extremities. Before admission she had no past history. ECG was normal and in neuropschycological examination neither mental retardation nor cognitive disturbance was detected.
An MRI scan was performed for further evaluation. This showed several hyperintense lesions in the periventricular white matter bilaterally (Fig. 1), but also a complete agenesis of corpus callosum with absence of pericallosal and cingulate sulci (Fig. 2). There was also a radial position of sulci and fissures in the parieto-occipital region towards the absent corpus callosum (Fig. 2). A wide separation of the frontal horns from midline structures was noticed and a disproportionate enlargement of the occipital horns (colpocephaly) (Fig. 3).

The diagnosis of a rare case of a complete form of agenesis of the corpus callosum (ACC) coexisting with multiple sclerosis (MS) was established. The patient was proven to have definite MS.

Discussion:

ACC is a rare congenital disorder characterised by a complete or partial defect of the corpus callosum. Although ACC has been described as an isolated entity, more frequently patients with ACC present multiple malformations either of the central nervous system or other systems; agenesis of the corpus callosum may also represent a feature of several craniofacial syndromes, phakomatoses, chromosomal abnormalities, and other rare disorders. Other factors that are not genetically determined but may play a role in aetiology of ACC are intrauterine destructive disorders, such as foetal alcohol syndrome, maternal diabetes, rubella or influenza, and maternal exposure to valproate. Familial cases have also been reported.
ACC is usually detected in childhood. In adults, focal seizures and mental retardation are the most frequent manifestations. Otherwise, asymptomatic patients can be detected by cerebral imagery with specific criteria. ACC may be paucisymptomatic and the clinical findings most often depend on associated cerebral malformations.

This reported association of complete ACC and MS is very unusual. MS is a common demyelinating disease and its prevalence differs at various latitudes, being about 40-60 per 100,000 in high–risk areas. The pathogenesis of ACC is unknown. From postmortem examinations, the incidence of ACC is 1/20,000. However, its real incidence may be greater, because isolated ACC is usually asymptomatic. The coexistence of ACC and MS with no other associated congenital malformation, seizures, or mental retardation, as seen in this patient, is very unusual; although this is probably incidental and may be due to chance, we can not exclude an unknown common underlying aetiological pathophysiological mechanism, for instance genetic, which could explain this rare coincidence. Further identification of the genetics of ACC might be helpful in understanding the aetiological heterogeneity of other associated brain diseases.

Differential Diagnosis List: Corpus callosum agenesis and multiple sclerosis

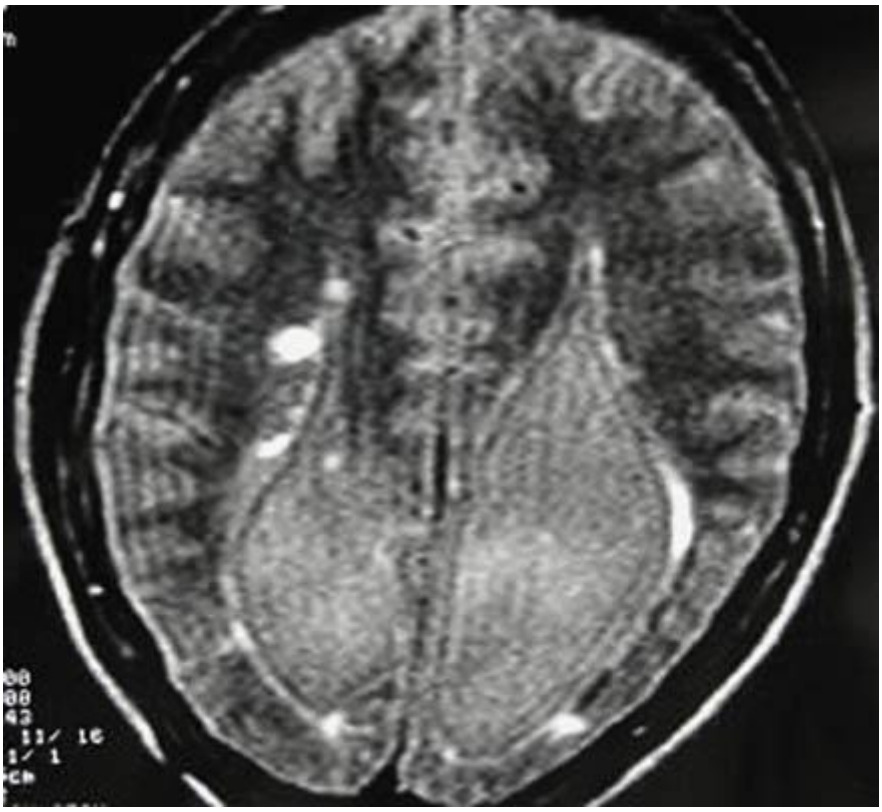
Final Diagnosis: Corpus callosum agenesis and multiple sclerosis

References:

Jeret JS, Serur D, Wisniewski KE, Lubin RA. Clinicopathological findings associated with agenesis of the corpus callosum. Brain Dev 1987;9:255-64. (PMID: 3310713)
Lemesle M, Giroud M, Madinier G, Martin D, Baudouin N, Binnert D, Dumas R. Agenesie du corps calleux: les modes de revelation chez l'adulte. Rev Neurol Paris 1997;153:256-61. (PMID: 9296144)
Nyul LG, Udupe JK. MR image analysis in multiple sclerosis. Neuroimaging Clin N Am 2000;50(4):79 -816. (PMID: 11359726)

Figure 1

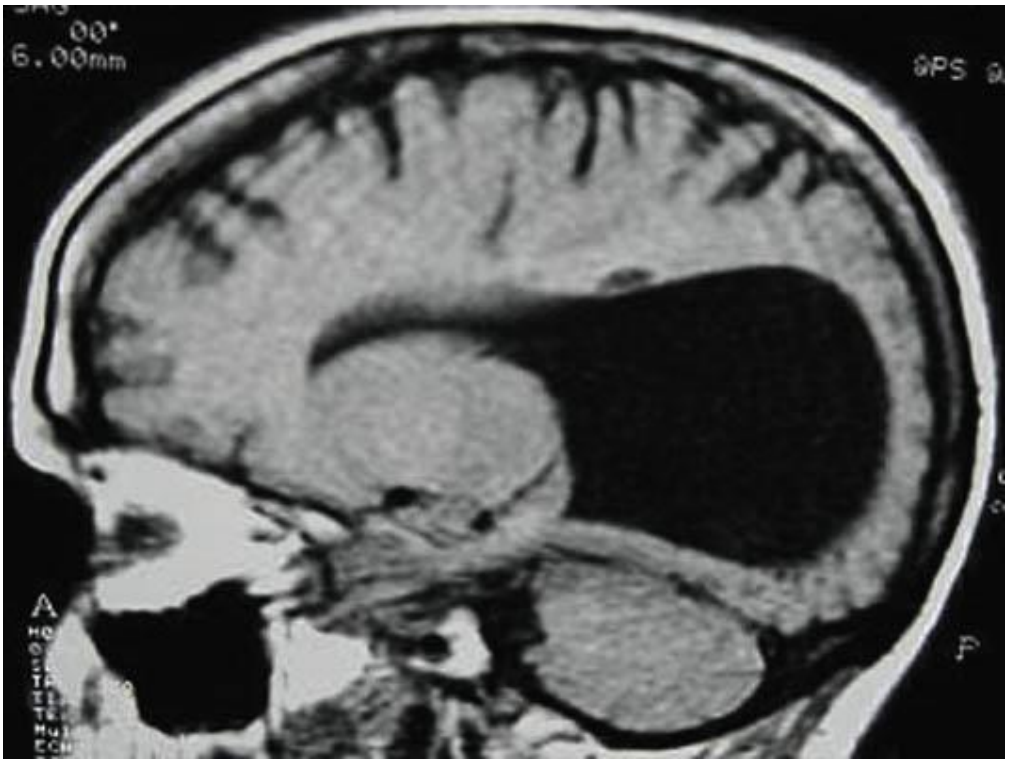
a



Description: Bilateral demyelinating hyperintense lesions in the periventricular underdeveloped white matter, in PD–weighted axial image. **Origin:**

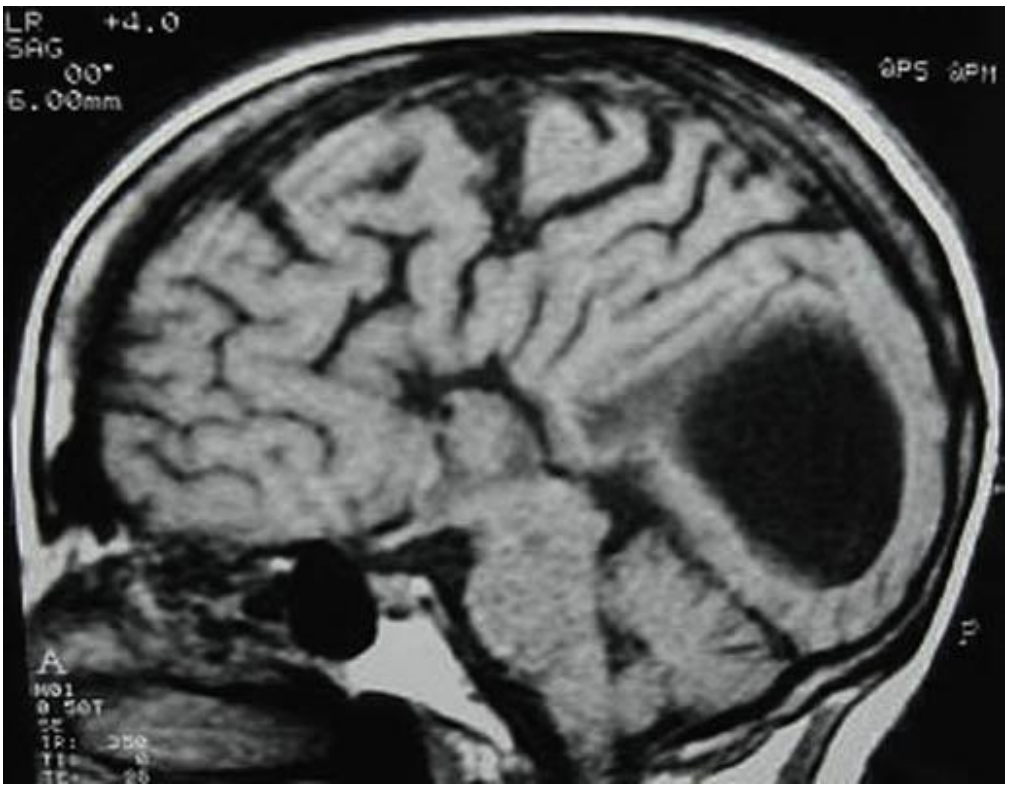
Figure 2

a



Description: Complete agenesis of the corpus callosum and absence of pericallosal and cingulated sulci. The sulci and fissures of the parieto–occipital region are distributed in a radial position, in T1–weighted mid-sagittal image. **Origin:**

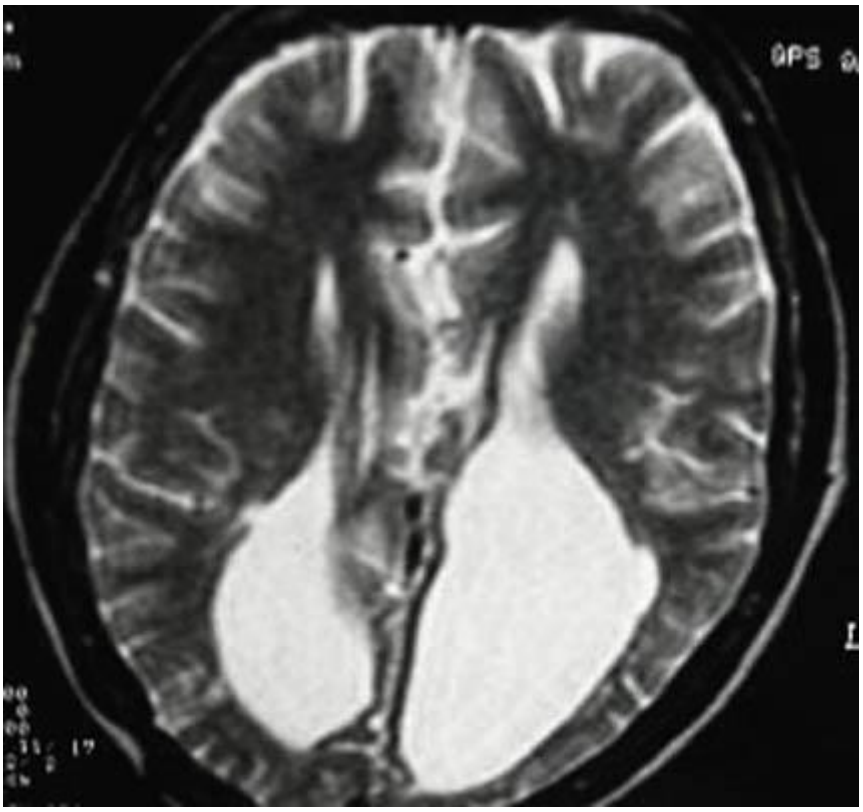
b



Description: Complete agenesis of the corpus callosum and absence of pericallosal and cingulated sulci. The sulci and fissures of the parieto–occipital region are distributed in a radial position, in T1-weighted para-sagittal image. **Origin:**

Figure 3

a



Description: MRI scan with separation of the frontal horns and dilatation of the occipital horns (colpocephaly), in T2–weighted axial image. **Origin:**