Cephaloceles: clinical and neuroradiological appearance

Associated cerebral malformations

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Summary. Cephaloceles are congenital malformations with herniation of intracranial structures through a defect in the cranium. On the basis of a review of the literature and 31 personal observations the authors discuss the clinical and neuroradiological presentation of their various anatomical locations: sphenoidal, ethmoidal, frontal, occipital and parietal.

Key words: Agenesis of corpus callosum – cephalocele (sphenoidal, ethmoidal, nasal, frontal, orbital, occipital, parietal) – Chiari III malformation – Dandy-Walker malformation – heterotopias – lipoma of corpus callosum

Cephaloceles are congenital malformations consisting of a defect in the cranium and the dura mater with extracranial herniation of intracranial structures. The term cranial meningocele is reserved for the forms in which the herniated sac contains only leptomeninges filled with CSF. The term encephalocele designates the forms in which the herniated sac also contains brain tissue. A simple skull defect without prolapse of brain or meninges is referred to as cranium bifidum occultum. Such isolated cranium bifidum occultum is uncommon and is not considered clinically significant [4, 65]. In rare cases, the orifice of the herniated sac may become obliterated secondarily, so the structures contained within it lose their continuity with the analogous intracranial structures. Such a mechanism may account for "nasal gliomas" and for the rests of neuronal tissue occasionally found within the scalp [11, 27, 36].

Cephaloceles occur about once in 4,000 to 5,000 live births. This frequency is grossly equal in regions with low or high incidence of spina bifida and other malformations of the central nervous system [24, 35, 49]. Cephaloceles are less common anomalies than spina bifidas. In series of congenital malformations of the central nervous system, cephaloceles occur six [24] to 16 [55] times less frequently than spina bifida.

In our personal series, cephaloceles are about three times less frequent than agenesis of the corpus callosum, a commonly associated malformation.

The etiology of cephaloceles remains obscure. In most instances, the lesion is sporadic. A genetic influence is sometimes apparent as in the occurrence of cephaloceles (a) in families with spina bifida and other malformations of the nervous system [71] or (b) in the HARD + E syndrome associating hydrocephalus, agyria, retinal dysplasia and (in about 50% of the cases) an encephalocele [52, 73]. Cephaloceles may be produced experimentally by several teratogens, e.g., x-irradiation, trypan blue, and excessive doses of vitamin A [19, 42, 43, 71]. Geographic differences in the distribution of encephaloceles indicate additional racial and perhaps environmental factors in the formation of cephaloceles. Indeed, the cephalocele is most often occipital in location in Europe and America [4, 43] and more frequently frontal in Russia and Southeast Asia [4, 62–64].

Several different mechanisms may be invoked to explain the different locations and natures of the various cephaloceles:

a. Defective tissue induction with malformation early in embryogenesis may explain the concurrence of anterior cephaloceles with cerebral and craniofacial malformations such as prosencephaly, agenesis of the corpus callosum, anomalies of the visual and olfactory structures, and midline facial clefts. The concurrence of these lesions suggests perturbations...