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Familial intracranial arachnoid cysts

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Abstract. Three siblings with intracranial arachnoid cysts are described, two males and one female. One of the males has symmetric, bilateral, temporoparietal convexity cysts, and the others have singular, unilateral cysts. Three additional siblings in the family and other known relatives are clinically unaffected. As far as we know, this is the second reported case of familial intracranial arachnoid cysts and the first involving three siblings. The significance of these cysts and a review of the literature are presented.

Key words: Arachnoid cyst – Congenital brain malformation – Cystoperitoneal shunt – Familial malformation – Hydrocephalus

Arachnoid cysts comprise approximately 1% of all intracranial space-occupying lesions [16]. Although the majority of these cysts appear to be developmental and many are diagnosed in the neonatal period [18], only two reports of familial intracranial arachnoid cysts have been published. Handa [7] reported two brothers with bilateral middle cranial fossa cysts, both with increased head circumference, one with convulsions and one with hemiparesis, and normal familial chromosomal studies. Pomeranz [17] has described two of the siblings reporting therein the context of surgical management of these cysts. To our knowledge, this is the first report of intracranial arachnoid cysts in three siblings.

Case reports

Case 1

This male child, the first born to young nonconsanguinous parents, was hospitalized in 1970 at the age of 6 months due to rapidly increasing head size. Ventriculography demonstrated a very large left hemispheric cerebral cyst without communication with the ventricular system. Via a craniotomy a communication was opened between the arachnoid cyst and the lateral ventricle, and part of the cyst wall was resected to form a wide drainage area into the basal cisterns. Postoperatively, the head circumference gradually reached normal limits. The psychomotor development has been slow: borderline intelligence with minimal neurological signs, including clumsiness, jerky nystagmus to the left, and bilateral pyramidal signs. Occasional complex partial seizures are partially controlled medically.

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Case 2

This 5-month-old male, the sixth of six siblings, was hospitalized in 1985 due to rapidly increasing head size. At that time the head circumference was 47 cm (>97%) with a large bulging anterior fontanel and no abnormal clinical findings. Computed tomography of the brain demonstrated symmetric, bilateral, temporoparietal convexity arachnoid cysts with partial erasure and medial displacement of the lateral ventricles. The cysts were shunted to the peritoneum with a "Y"-shaped tubing system having a single abdominal catheter, one-way resistance valves, and antisiphon devices. Tomography of the head 1 week postoperatively (Fig. 1) demonstrated a moderate decrease in the size of the two cysts with compensatory ventricular expansion. Several months later the cysts had decreased further in size, the ventricles had decreased in size, and the head circumference was approaching the upper limits of normal. Neurological and intellectual development of the child has been borderline normal.

Case 3

This 5-year-old female, the fifth of six siblings, was hospitalized in 1986 due to a fever of 40 °C and a single episode of generalized tonic-clonic convulsions. The physical examination and laboratory work-up were compatible with a transient viral upper respiratory tract infection as the source of the fever. Computed tomography of the brain (Fig. 2) demonstrated an arachnoid cyst of the ambient cistern 3.5 cm in diameter; this was the only pathological finding. The administration of phenobarbitone was initiated, and the child has been asymptomatic with no change in the cyst configuration on follow-up tomography.

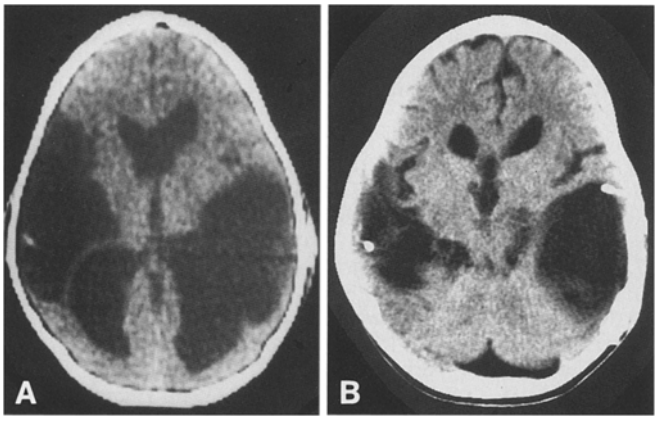

Fig. 1. **A** Axial non-enhanced computerized tomography of the head in case 2 1 week following bilateral arachnoid cyst shunting to the peritoneum. Part of the shunt can be seen within the cyst on the left. The relationship of the cysts to the dilated lateral ventricles can be appreciated. **B** Three months following the shunt placement, the cysts are further decreased in size, as are the ventricles. Both shunt tips can be seen within the cysts. Expansion of the subarachnoid space can be seen relative to **A**

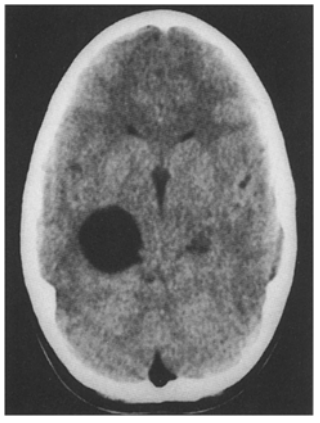

Fig. 2. Axial nonenhanced computerized tomography of the head in case 3. The cerebrospinal fluid density of the ambient cistern arachnoid cyst is evident

Discussion

Intracranial arachnoid cysts are uncommon but not rare [3–6, 8, 9, 12, 15, 18]. These cysts are intra-arachnoidal collections of cerebrospinal fluid whose clinical manifestations are a result of enlargement and pressure on brain tissue and interference with cerebrospinal fluid circulation [8, 18]. Several reports have described series of these cysts in typical locations such as the middle fossa [9], suprasellar region [6], and infratentorially [5]. Singular reports have described cysts that took up most of the intracranial space [17], unusual locations such as within a ventricle [16], and multiple intracranial cysts [7, 17]. The cysts can be diagnosed by their clinical manifestations, combined with the typical computed tomography or magnetic resonance studies [8, 18]. Today surgery is rarely required to differentiate these lesions from other types of cysts or tumors. Controversy exists as to the origin of the cyst fluid [3, 8]: cerebrospinal fluid trapped via a one-way valve mechanism in the cyst wall versus active fluid secretion by the lining. It is generally accepted today that the optimal treatment of symptomatic intracranial arachnoid cysts is drainage by a cystoperitoneal shunt in a fashion similar to ventriculoperitoneal shunting in hydrocephalus [8, 10, 12, 13, 18, 20].

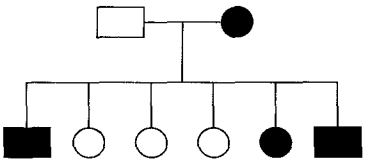

Fig. 3. Family pedigree of cases 1–3. Affected siblings are designated as *blackened shapes*

Although many series of arachnoid cysts have been described, especially in children, with the contention of congenital origin [8, 12, 15, 18], familial occurrence has only been described once. The fact that both of the male siblings that Handa [7] described had bilateral cerebral cysts suggests a familial predisposition.

The existence of three affected siblings with intracranial arachnoid cysts in our report is highly suggestive of a genetic tendency. The uncommon configuration of the symmetric, bilateral convexity cysts that Handa described in his two male siblings also appeared in one of our male patients. If single-gene inheritence is postulated to be in effect within this family, the most probable pattern would be autosomal dominant, given the appropriate phenotype in half the siblings, with two males and a female involved [21] (Fig. 3). The family did not agree to undergo chromosomal studies or have unaffected members undergo computed tomography of the brain. Therefore, both the absolute number of affected family members and a definite genetic pattern can only be conjectured. It is within reason to consider the cyst found in the female sibling as an incidental finding secondary to fever-induced convulsions. It is possible that additional siblings or a parent may have asymptomatic arachnoid cysts, as these are not rare [18].

Arachnoid cysts have been associated with chromosomal and inherited disorders. Masuno [14] described a chromosome 12 trisomy and Allen [1] reported autosomal-dominant polycystic kidney disease with arachnoid cysts. Spinal arachnoid cysts are known to be familial, whether as part of a complex syndrome [11, 19] or as the only pathology [2].

The family we have presented, with three siblings having intracranial arachnoid cysts, is important in that it points to genetic aspects of this pathology. We expect that within this or other families future studies may show clear genetic mechanisms of inheritence and further delineate the etiology of arachnoid cysts.

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