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Hydrocephalus and chronically increased intracranial pressure in achondroplasia

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Abstract Two achondroplastic children with ventriculomegaly are reported. The patients had no signs of increased intracranial pressure, but in one blindness had previously been detected by the parents. Neuroradiological examinations showed ventriculomegaly in both. Intracranial pressure remained at high levels (20–55 mmHg) preoperatively; ventriculo-peritoneal shunting was performed in both cases, and postopera-

tively levels fell to normal (5–16 mmHg). The need for the treatment of ventriculomegaly seen in achondroplastic children is analyzed on the basis of these two patients, with a brief review of literature.

Key words Achondroplasia · Hydrocephalus · Intracranial pressure monitoring

Introduction

There are various ways of managing children with achondroplasia. It results as a secondary phenomenon from abnormal formation of endochondral bone [5]. Macrocephaly is commonly seen in achondroplasia, and such children generally have no clinical evidence of increased intracranial pressure (ICP) but have enlarged and stable ventricles, as in children with compensated hydrocephalus. With ICP measurement, Steinbok et al. [13] demonstrated evidence of active hydrocephalus in five achondroplastic children with documented ventriculomegaly in computerized tomography (CT) scans.

Recently, we have encountered two achondroplastic children with moderate macrocranium, enlarged ventricles but with no clinical evidence of increased ICP. ICP monitoring was performed to help us make a definite decision for treatment. The results of pre- and post-operative recording and the treatment of hydrocephalus in achondroplasia are discussed.

Case reports

Case 1

A 7-month-old girl who was born at term to normal parents was referred to our department for macrocranium. Her head circumference was 36 cm at birth and 47 cm (above 95 percentile) on admission to our service. Neurological examination revealed an enlarged anterior fontanel. Fundus examination was normal. On magnetic resonance imaging (MRI), triventricular hydrocephalus and diffuse subarachnoidal enlargement without periventricular edema were found (Fig. 1a). Sagittal MRI scans showed a small posterior fossa, a patent aqueduct of Sylvius, but tectal beaking, decreased ponto-mammillary distance, stenosis of foramen magnum and compressed IV ventricle (Fig. 1b).

Case 2

This 17-month-old boy was born at 42 weeks of gestation by cesarean section. He came to medical attention after amaurosis was detected by his parents. He was referred to our department with triventricular hydrocephalus. On neurological examination, the head circumference was 47 cm (at the limit of normal), the anterior fontanel was totally closed, and he was totally amaurotic, as was confirmed by means of visual evoked potentials (VEP). During the neuroophthalmological consultation bilateral optic atrophy was seen, and no cause other than hydrocephalus was found to explain the amaurosis. CT and MRI scans revealed triventricular hydrocephalus, a small

Fig. 1 **a** Axial and **b** sagittal T1-weighted MRI scans show triventricular hydrocephalus and diffuse subarachnoidal enlargement. Note on the sagittal MRI scans **(b)** narrowed foramen magnum, compressed IV ventricle and tectal beaking with decreased ponto-mammillary distance

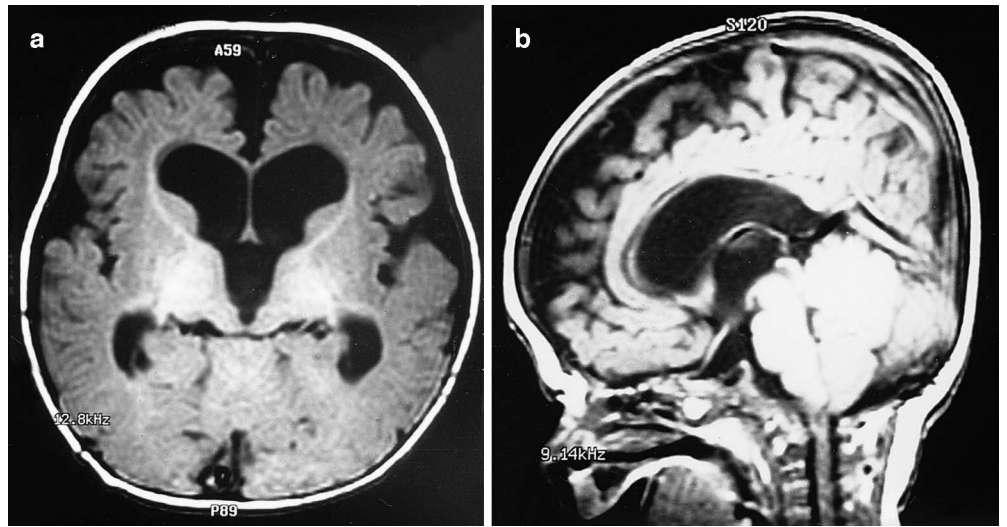
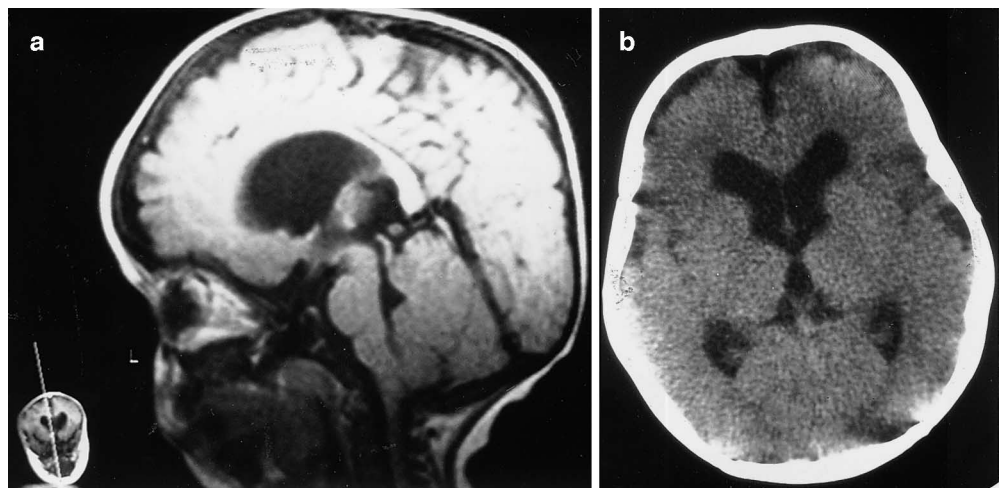


Fig. 2 **a** Sagittal T1-weighted MRI scan shows abnormal vertical deviation of tentorium and enlarged vein of Galen. Elongation of brain stem, tectal beaking, decrease in ponto-mammillary distance, compression of IV ventricle and stenosis of foramen magnum were also seen. **b** Preoperative CT scan shows moderate ventriculomegaly



posterior fossa, anterior and cephalad displacement of cerebellum, an abnormally vertically deviated tentorium, an enlarged vein of Galen, tectal beaking, decreased ponto-mammillary distance and stenosis of foramen magnum with patent aqueduct of Sylvius (Fig. 2a, b).

Method and results

The same protocol was applied to both patients. Before VP shunt insertion, ICP monitoring was performed for 48 h preoperatively and 72 h postoperatively (Spigelberg epidural probe system). Both patients had VP shunts placed (PS Medical Delta valves, performance level 1). In case 1, ICP records range between 25 and 35 mmHg, with some pressure peaks up to 45 mmHg preoperatively. Postoperative records ranged between 7 and 16 mmHg with no peaks. In the second patient preoperative records ranged between 20 and 55 mmHg, with a decrease to 5–13 mmHg postop-

eratively. Control CT scans 5 and 12 months after the surgery showed decrease of ventricular size in both patients, and effacement of subarachnoid enlargement in the first patient was noted.

Discussion

Neurological deficits in achondroplasia result secondary to abnormal formation of endochondral bone, which in extreme cases leads to stenosis of the entire craniospinal axis [5]. In attempts to explain the macrocephaly commonly seen in achondroplastic children [6], a number of possible causes have been proposed. These include hydrocephalus, excessive growth of the calvarium to compensate for the small cranial base and megalencephaly [2, 4, 13]. Because achondroplastic children with ventriculomegaly often

have no symptoms suggestive of increased ICP, it seems doubtful whether treatment is indicated. The ICP records of our two patients, which remained at high levels during preoperative monitoring, suggested evidence of chronically increased ICP in both. Serious neurological complications, such as apneic episodes or sudden death, are also seen in achondroplastic children. They are related to brain stem compression in a small posterior fossa and narrowed, triangular foramen magnum [14]. These findings can be seen on the MRI scans of our patients, but there was no neurological deficit related to brain stem compression, perhaps because it was too early for symptoms to have developed.

Venous congestion secondary to stenosis at the level of the jugular foramen or to obstruction at the level of the thoracic inlet and obstruction of the basal cisterns due to brain stem distortion and obstruction of the foramina of Luschka and Magendie as a result of foramen magnum stenosis have also been proposed as mechanisms of the ventriculomegaly seen in achondroplastic patients [9, 11, 13]. Sainte-Rose et al. [12] have affirmed that venous sinus hypertension is caused by the hydrocephalus in most cases, but in achondroplasia and craniostenosis the hydrocephalus is secondary to increased intracranial venous sinus pressure caused by sigmoid sinus obstruction at the level of jugular foramen. Steinbok [13] added another possible proximal obstruction at the level of thoracic inlet, demonstrated by venous angiograms in two of his five patients. He attributed this event to small bony thorax. All these proposed mechanisms define a communicating hydrocephalus, but the type of hydrocephalus in achondroplasia is triventricular rather than tetraventricular. Furthermore, in a recent study, DiMario et al. [3] have documented dynamic changes in brain morphometry in achondroplastic patients, which result in either an intermittent or a progressive obstructive hydrocephalus superimposed on the existing communicating hydrocephalus typically encountered. All these mechanisms seem to provide reasonable explanations of hydrocephalus in achondroplasia. The enlarged vein of Galen in our second patient is probably an indirect sign of venous congestion, and the small IV ventricles in both must be related to the dynamic changes in brain morphometry described by DiMario et al. [3]. The resultant tectal beaking and decreased ponto-mamillary distance are potential causes of obstruction to cerebrospinal fluid outflow in the aqueduct of Sylvius.

The treatment of hydrocephalus in achondroplasia is controversial. Ryken et al. [11] state that ventricular shunting is generally reserved for demonstrated symptomatic hydrocephalus, as the ventriculomegaly is generally arrested with time. Thomas et al. [14] affirm that the degree of ventricular dilatation is variable and many children have arrested hydrocephalus that may not need a shunt. Yamada et al. [15] reserve shunting procedures for symptomatic patients only. McLone and Partington [10] emphasize that children 3 years old or younger with moderate or greater

ventriculomegaly should be shunted. Neither patient in this article had any symptoms suggestive of increased ICP on admission to neurosurgical unit, but the high ICP levels recorded pointed to a chronic hydrocephalic state in both, and this state is not arrested hydrocephalus, but compensated hydrocephalus [10]. Thus, we suggest that ICP monitoring is indicated in achondroplastic children with moderate ventriculomegaly and that treatment is necessary when the ICP exceeds 15 mmHg, especially with episodic high pressure waves, even if there are no signs of increased ICP. The blindness in the second patient may be due to this chronic elevated ICP and/or to optic nerve compression at the optic foramen level. There are few data in the literature on achondroplasia and visual disturbances [7, 8]. In a case recently reported by Landau and Gloor [8], bilateral blindness, which developed in a previously shunted achondroplastic dwarf over a short period of time, was attributed to increased ICP and shunt malfunction, rather than to stenosis at the optic foramen level. According to Landau and Gloor [8], increased ICP in achondroplasia is secondary to impaired venous outflow, as discussed above, e.g. as a part of a pseudotumor cerebri syndrome. The failure of the narrowed spinal canal to act as an elastic reservoir for balancing cerebrospinal fluid pressure fluctuations, as seen in some patients with spinal cord tumors [1], is another mechanism proposed by the same authors [8] to explain papilloedema in achondroplasia. Lunder et al. [9] reported a 10-month-old child with angiographically demonstrated bilateral venous outflow obstruction caused by stenosis of the jugular foramen. The child was successfully treated with surgical decompression by opening the right jugular foramen, which relieved the clinical signs of intracranial hypertension, but the ventricular size was only slightly reduced. In our two patients treated with low-pressure VP shunting, an immediate decrease in ICP levels was documented. On control CT scans, ventricular sizes were found to be decreased in both. The risks of subdural hematomas can be minimized by using shunt systems with an antisyphon device. We think that endoscopic III ventriculostomy may not be an alternative to VP shunting, as it is technically difficult owing to decreased ponto-mamillary distance and because of the communicating and noncommunicating nature of hydrocephalus in these patients.

Ventriculomegaly in achondroplastic children represents hydrocephalus. The clinical course may be asymptomatic, but the chronically increased ICP levels may also cause progressive, invisible neurological deficits. However, treatment is necessary in selected cases, though decompressive posterior fossa surgery must be reserved for patients with signs of brain stem compression. Simple VP shunting can be the treatment of choice for achondroplastic patients with ventriculomegaly.

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