CSF Scanning in Achondroplastic Children with Cranial Enlargement

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Introduction

Achondroplasia is an inherited anomaly of endochondrial bone formation and often causes neurological complications in children (Dennis *et al.* 1961). Cranial enlargement is usually found and its evolution may lead to the diagnosis of hydrocephalus. However, typical signs of so-called 'active' hydrocephalus are rarely seen in these cases and further investigations are needed, in particular dynamic

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studies of the cerebrospinal fluid (CSF) pathways and turnover.

Visualisation of CSF dynamics (cisternography) may offer important information for understanding the pathophysiology of such cases and for planning therapy.

Methods

In hydrocephalic children, cisternography is done by injecting 500μCi of 169 Yb-DTPA (diethylenetriamine-pentaacetic acid) into the lumbar thecal space, using a 22 gauge needle (Deland *et al.* 1971). Serial rectilinear, anterior and lateral scans of the head are made two hours, six hours and 24 hours after injection and may be repeated at a 48-hour interval if necessary.

Criteria for a normal transit pattern of 169 Yb-DTPA in children have been given elsewhere (James *et al.* 1971).

At the two-hour scan, basal cisterns are completely visualized and ambiant and Sylvian cisterns can also be seen. The six-hour cisternogram shows how the radionuclide has progressed, centrally in the interhemispheric cistern and laterally in the Sylvian cisterns. At 24 hours, the basal cisterns are cleared and the indicator is found symmetrically concentrated in the frontal parasagittal area. Alternatively, in infants and children the 24-hour cisternogram may show a homogeneous radionuclide distribution around the convex surfaces of the cerebral hemispheres.

In patients with communicating hydrocephalus resulting from an impairment of the transit-resorption process of the CSF around the hemispheric convexities, the deficiency of parasagittal accumulation of the radionuclide corresponds to a constant or a transient entry of the radioactive substance into the ventricular space. In such cases the shape of the lateral ventricles appears on the cisternogram: it may remain for long periods (lasting reflux) or disappear within 24 hours (transient reflux) (Deland *et al.* 1971).

CASE REPORT

An 18-month-old boy with achondroplasia was referred to the paediatric clinic because of an abnormally increasing head circumference. He had been born after 37 weeks gestation. His mother was also achondroplastic but the father had no congenital anomaly. The boy's head circumference had left the 95th percentile at the second month and had then steadily diverged upwards from this line (Fig. 1).

The child showed some degree of cranio-facial disproportion, but there was no evidence of increased intracranial pressure.

Clinical examination of the nervous system was normal except for slight muscular hypotonia. Radiographs of the skull showed typical signs of achondroplasia. A pneumoencephalogram was done under anaesthesia; the ventricles above the tentorium and the basal cisterns were dilated (Fig. 2).

Radioisotope cisternography showed an entirely normal CSF flow pattern. Basal cisterns and subarachnoid spaces of the convexity were symmetrically permeable and no dynamic disorder of CSF was seen (Fig. 3).

No shunting procedure was carried out. At the time of writing, the boy is in good condition, both neurologically and mentally. There are no signs of developing hydrocephalus. The head circumference curve, which was initially divergent, now parallels the upper percentile (Fig. 1).

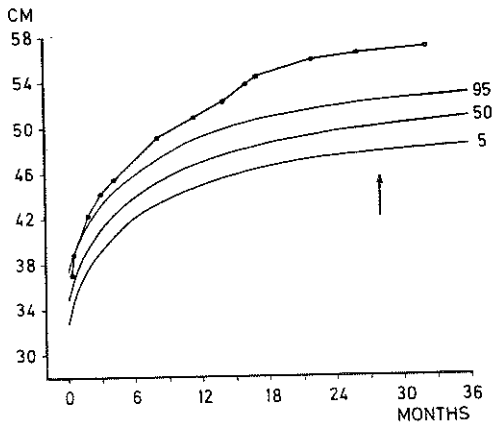

Fig. 1. Chart of head circumference of the patient (upper line) compared with the normal percentile range. After a period of divergence, the curve spontaneously parallels the upper percentile line. (The arrow indicates the time at which the cisternographic study was made.)

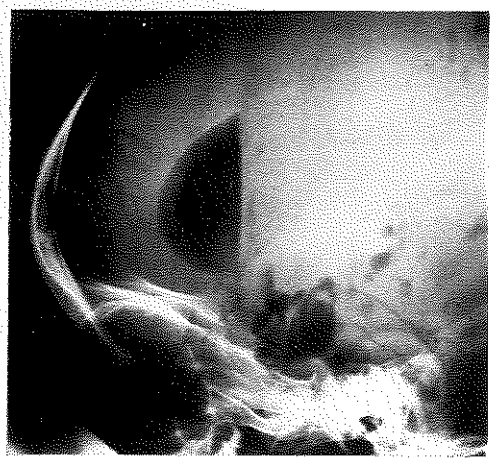

Fig. 2. Air contrast study showing a mild enlargement of the ventricular system and of the basal cisterns.

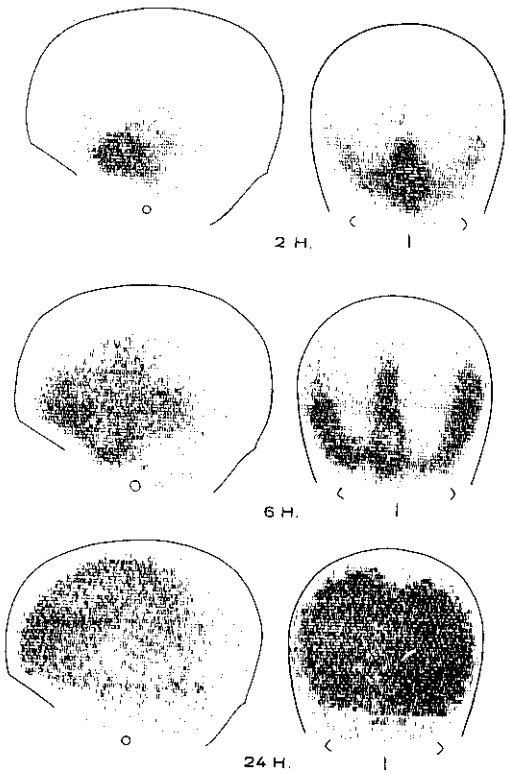

Fig. 3. Anterior and lateral cerebrospinal fluid scans performed two hours, six hours and 24 hours after injection of 169 Yb-DTPA. The CSF dynamics appear entirely normal; the flow in the basal cisterns and around the cerebral convexities is symmetrical and normally delayed and there is no ventricular reflux.

Discussion

Achondroplastic dysgenesis is associated with cranial enlargement and with some degree of cranio-facial disproportion. Usually there are no typical signs of increased intracranial pressure. Neurological and EEG examinations are normal in the great majority of cases. The child's psychomotor performance remains within normal limits. However, because the growth curve of the cranial circumference is outside the 95th percentile, and as its slope sometimes diverges upwards from the normal growth curve, an impairment in CSF circulation is always a possibility (Cohen *et al.* 1967).

Radiographs of the skull confirm the head increase, showing a large calvarium and a shortening of the basilar bones (Caffey 1961). It remains an open argument whether the widening of the cranial sutures in achondroplasia implies an underlying increased intracranial pressure.

Pneumoencephalographic studies usually show mild communicating hydrocephalus with dilated lateral ventricles (Dandy 1921, Langer *et al.* 1967). Some authors have demonstrated an enlargement of the whole ventricular system (Spillane 1952, Cohen *et al.* 1967). Wise *et al.* (1971) showed involvement of the basal cisterns. Pneumoencephalography does not usually yield a definite conclusion as to the morphology of the subarachnoidal spaces of the convexity, whether they are visualized or not.

Radioisotope cisternography remains the most valuable method of tracing the progress of the CSF through the hemispheric subarachnoidal spaces and the Pachonian corpuscles, the occlusion or dysfunction of which induces active communicating hydrocephalus. All the cases of true communicating hydrocephalus in the literature showed transient or lasting ventricular refluxes of the CSF indicator (Tator *et al.* 1968, Glasauer *et al.* 1970, Deland *et al.*

1971). Radioisotope cisternography is of prognostic value in such cases because only patients who demonstrate lasting cisterno-ventricular refluxes are likely to benefit from surgical intervention (McCullough *et al.* 1970). Large ventricular dilatation of atrophic origin may explain some cases of transient ventricular radioactive contamination with a normal or a slightly slower flow of the CSF indicator around the convexities.

Cisternographic investigation of achondroplastic children with dilated ventricles is an important prerequisite; if the CSF flow pattern is shown to be normal, surgery may not be necessary.

Cisternography in three achondroplastic children with communicating hydrocephalus did not show true hydrocephalus and these patients did well without surgical shunting procedures, even though progressive cranial enlargement and some degree of widening of sutures had been present initially (Dennis *et al.* 1961, Glasauer *et al.* 1970). In our patient, too, we felt that the normal cisternographic findings justified surgical abstention and this child's clinical course has shown that, with time, the anomalies associated with achondroplasia can become stable. CSF scanning by this method should be repeated during the follow-up period.

The process of communicating hydrocephalus in achondroplastic patients is highly controversial (Dandy 1921, Dennis *et al.* 1961, Cohen *et al.* 1967, Wise *et al.* 1971, James *et al.* 1972). In our opinion one general consideration should be whether the basilar and peripheral cerebral subarachnoid spaces show a normal dynamic pattern. If they do, the hypothesis of the existence of a subarachnoidal block or of a perturbed CSF resorption is refuted.

It is reasonable to assume that the ventricular enlargement seen in achondroplasia is, in most instances, the result of a morphogenetic expansion of the telencephalic ependymal cavities in relation to the cranial dysgenesis, and not of active hydrocephalic or atrophic processes. However, in cases where increased intracranial pressure is present the bone anomalies may be responsible for an additional active hydrocephalic process.

AUTHORS' APPOINTMENTS

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SUMMARY

This paper reports on an 18-month-old achondroplastic child with dilated lateral ventricles. Results of a cisternographic investigation were normal. No shunting procedure was carried out, and subsequent clinical progress was satisfactory. Some patho-physiological aspects of achondroplastic dysgenesis are discussed.

RÉSUMÉ

Examen du LCR *chez des enfants achondroplastes avec dilatation crânienne*

L'article rapporte le cas d'un jeune achondroplaste de 18 mois avec des ventricules latéraux dilatés. L'exploration cisternographique était normale. Aucun processus de shunt n'a été entrepris et l'évolution clinique ultérieure a été satisfaisante. Les auteurs discutent quelques aspects physio-pathologiques de la dysgénésie achondroplastique.

ZUSAMMENFASSUNG

Liquorszintigraphie bei Kindern mit Achondroplasie und Makrocephalus

Diese Arbeit berichtet über ein 18 Monate altes Kind mit Achondroplasie und Seitenventrikelerweiterung. Die Zisternographie war normal. Es wurde keine Ventiloperation durchgeführt und der weitere klinische Verlauf war zufriedenstellend. Die Autoren diskutieren einige physiopathologische Aspekte der achondroplastischen Dysgenesie.

RESUMEN

Scanning del l.c.r. en niños acondroplásicos con agrandamiento craneaí

Esta comunicación trata de un niño acondroplásico de 18 meses de edad con dilataciones de los ventriculos laterales. Una cisternografia fue normal. No se procedió a ninguna derivación y el curso clinico posterir fue satisfactorio. Los autores algunon aspectos fisiopatológicos de la disgenesia acondroplásica.

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