AJNR

Hartnup disease: MR findings.

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AJNR Am J Neuroradiol 1991, 12 (5) 1026-1027
http://www.ajnr.org/content/12/5/1026.citation

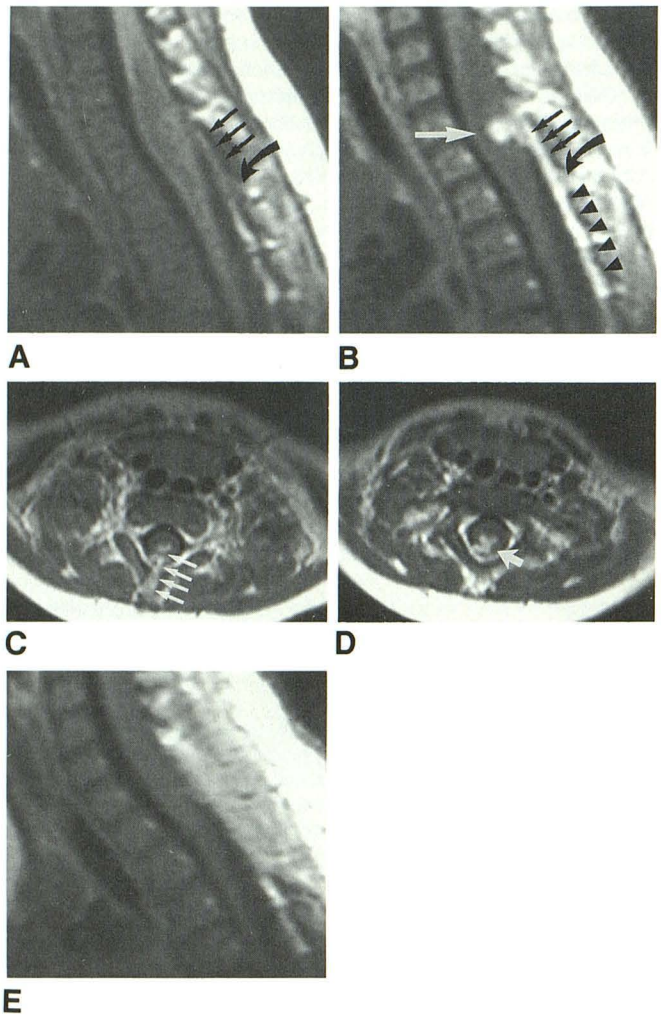

Fig. 1.—Spinal dermal sinus tract.

***A*, Sagittal T1-weighted MR image (500/30) of thoracic spine shows marked enlargement and low signal intensity of cervical cord from C4 to T1. Linear structure of low signal intensity extends cranially (*straight arrows*) from level of entrance of dermal sinus (*curved arrow*).**

***B–D*, Sagittal (*B*) and transverse (*C* and *D*) contrast-enhanced T1-weighted MR images (500/30) show intradural part of dermal sinus tract as high-signal-intensity structure (*B*, *small black arrows*). Linear structure of high signal intensity originating from level of entrance of tract (*B*, *curved arrow*) and descending caudally represents adhesions (*B*, *arrowheads*). Tract ends in intramedullary dermoid tumor (*B*, *white arrow*). Transverse images show tract traversing the dura (*C*, *arrows*) and confirm intramedullary extension of dermal sinus tract into intramedullary dermoid (*D*, *arrow*).**

***E*, Postoperative sagittal contrast-enhanced T1-weighted MR image (500/30) shows subtle residual enlargement of thoracic cord. Hyperintense intradural and intramedullary masses have been resected.**

tract could be followed from the cutaneous lesion across the dura to the thoracic cord between the dorsal columns. The tract was removed totally with some difficulty because of adhesions caused by previous infections. On histologic examination, the diagnoses of sinus tract and dermoid tissue were confirmed. On postoperative MR examination, the diameter of the thoracic cord was normal, and there were no signs of enhancing dura or intramedullary lesions (Fig. 1E).

This case illustrates the advantages of gadopentetate dimeglumine–enhanced images in the detection of concomitant infection in this type of dysraphy. As infection often occurs as a complication of spinal dermal sinus tracts, and these infections might occur without clinical signs and symptoms of meningitis, we suggest routine administration of gadopentetate dimeglumine in preoperative evaluation of spinal dermal sinus tracts.

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REFERENCE

1. Barkovich AJ, Edwards MSB, Cogen PH. MR evaluation of spinal dermal sinus tracts in children. *AJNR* **1991**;12:123–129, *AJR* **1991**;156:791–797

Reply

I would like to compliment Drs. Algra and Hageman on their enlightened use of a paramagnetic contrast agent in the detection of a dermal sinus tract with associated infection. Indeed, it is well known that inflammation, such as that from repeated infections, induces the formation of granulation tissue [1]. It is equally well known that granulation tissue enhances significantly after administration of IV contrast material, because of considerable vascularity and lack of a blood-tissue barrier [2, 3]. Therefore, an infected dermal sinus tract, particularly a repeatedly infected one, will enhance dramatically after infusion of IV contrast material, as shown in the case reported by Drs. Algra and Hageman. However, I think it is premature to suggest that IV paramagnetic contrast agent should be administered to all patients with suspected dermal sinus tracts. Those patients who have not had infections as a result of bacteria seeping through the dermal sinus tract will not necessarily have contrast enhancement. Ideally, someone should perform a blinded study in which some patients with dermal sinus tracts are given paramagnetic contrast material and others are not. Only after a controlled study will we know whether the added expense of the paramagnetic contrast material is justified in patients with dermal sinus tracts.

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REFERENCES

1. Harriman D. Bacterial infections of the central nervous system. In: Adams J, Corsellis J, Duchen L, eds. *Greenfield's neuropathology,* 4th ed. New York: Wiley, **1984**:236–259
2. Enzmann D, Britt R, Yeager A. Experimental brain abscess evolution: CT and neuropathologic correlation. *Radiology* **1979**;133:113–120
3. Enzmann D, Britt R, Placone R. Staging of human brain abscess by computed tomography. *Radiology* **1983**;146:703–708

Hartnup Disease: MR Findings

Recently, we have examined a 6-year-old boy who 3 years earlier had had growth failure, hyperactivity, chronic diarrhea, intermittent ataxia, and weakness. Height and weight at that time were below the third percentile for his age, and developmental delay was present. The results of a muscle biopsy and a urine amino acid profile were consistent with Hartnup disease. MR of the brain (Figs. 1A and 1B) when the patient was 3 years old showed atrophy, delayed myelination, and dysgenesis of the corpus callosum. Follow-up MR when he

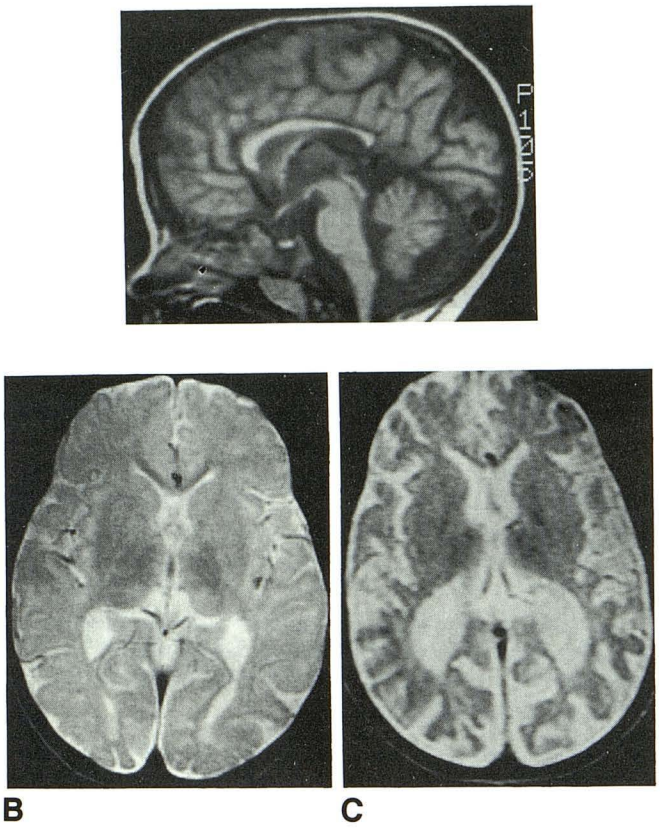

Fig. 1.—Hartnup disease.

***A*, Midline sagittal MR image (800/25/2) obtained when patient was 3 years old shows generalized atrophy. Corpus callosum is dysgenetic, rostrum is not seen, and splenium is thinned.**

***B*, Axial MR image (2000/80/1) obtained at same time as *A* shows prominent extraxial spaces throughout brain. On this image, mature myelin is present in posterior limbs of internal capsules and in rostrum of corpus callosum. However, signal intensity of subcortical white matter is slightly increased with respect to gray matter, indicating delayed myelination. Fine arborization of subcortical white matter normally should be present by 24 months of age. Although cortical sulci appear somewhat flat in both posterior temporal regions, these findings were not present in any other image, and therefore a neuronal migration anomaly was not considered likely.**

***C*, Axial MR image (2000/80/1) obtained 3 years after *A* and *B* shows progressive diffuse atrophy, which is slightly more prominent in both occipital regions. Lack of fine arborization of subcortical white matter is seen again, indicating delayed myelination.**

was 6 years old showed progressive atrophy (Fig. 1C) despite adequate clinical response to treatment (oral tryptophan ethyl ester).

Hartnup disease occurs in approximately 1 in 30,000 live births [1]. Increased excretion of serin, alanine, valine, leucine, isoleucine, phenylalanine, tyrosine, tryptophan, and histidine in urine and feces is characteristic [2]. The affected persons may be asymptomatic (Hartnup disorder) or may have a wide range of signs and symptoms, including chronic diarrhea, ataxia, and generalized neuropsychiatric dysfunctions (Hartnup disease) [1]. Most patients with Hartnup disease also have a pellagralike illness. Because of this similarity, it has been postulated that the signs and symptoms of Hartnup disease are due mainly to tryptophan deficiency [1]. The reasons for the variability in the expression of the Hartnup genotype are unknown. Except for the absence of the pellagralike skin disorder, our patient's signs and symptoms fit the standard description. Pathologic examination in Hartnup disease shows brain atrophy, most prominent in the occipital lobes and cerebellum. The lateral geniculate ganglia may show gliosis, and wallerian degeneration can involve the optic nerves and tracts [2].

According to anecdotal reports [3], in one case of Hartnup disease, MR of the brain was normal. In our case, the late MR findings, although nonspecific, correlated well with the pathologic descriptions of this disease. However, to our knowledge, delayed myelination in Hartnup disease has not been reported. We think that differentiation from other metabolic and mitochondrial disorders is not possible on the basis of the MR findings alone. Despite satisfactory clinical response to treatment, our patient showed progressive atrophy on follow-up MR.

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REFERENCES

1. Scriver CR. Hartnup disease: a genetic modification of intestinal and renal transport of certain neutral amino acids. *N Engl J Med* **1965**;273:530–535
2. Tahmoush AJ, Alpers DH, Feigin RD, et al. Hartnup disease: clinical, pathological, and biochemical observations. *Arch Neurol* **1976**;33: 797–807
3. Pomeranz SJ. *Craniospinal magnetic resonance imaging.* Philadelphia: Saunders, **1989**:479