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Clinical challenges

Down but not out

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1. Case report

A 20-year-old non-obese Hispanic male student with a history of Down syndrome initially presented at an outside institution with intermittent headaches. The headaches had been increasing in frequency and severity and associated with nausea and blurred vision. He had a history of intermittent "red eyes" that had been worsening for the past several months but no diagnosis had been made. He denied any diplopia, pain with eye movement, history of trauma, or recent illnesses. Review of systems was otherwise negative. His past medical and surgical history were unremarkable except for Down syndrome. Family history was positive for hypertension. His medications included acetaminophen, montelukast, and loratadine. He was single and denied tobacco, alcohol, or illicit drug use.

When seen at an outside institution, the patient's best-corrected visual acuity fluctuated between 20/30 and 20/60 in both eyes (OU). The pupils were symmetric without a relative afferent pupillary defect (RAPD). Color vision was normal OU. Confrontational visual fields, extraocular movements, slit-lamp examination, and intraocular pressures were all within normal limits. Dilated funduscopic examination, however, revealed bilateral disk edema.

Laboratory workup at the outside institution for syphilis, *Bartonella henselae*, *Borrelia burgdorferi*, and sarcoidosis gave negative results. Magnetic resonance imaging (MRI) showed no mass lesions and normal-sized ventricles without configuration of hydrocephalus, but a prominent empty sella measuring 15.8 mm anteroposteriorly by 15.0 mm transversely, consistent with increased intracranial pressure (ICP). A subsequent lumbar puncture showed an opening pressure

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>30 cm of water and normal cerebrospinal fluid (CSF) contents.

2. Comments

2.1. *Comments by Neil R. Miller, MD, FACS*

This patient has headaches and papilledema associated with an MRI that shows no mass lesion and normal-sized ventricles and has had a lumbar puncture that reveals an elevated opening pressure with normal CSF content. It would be tempting to diagnose this patient with idiopathic intracranial hypertension (IIH), but the patient does not fit the typical picture of IIH as he is a non-obese male. In this setting, therefore, one must consider other, less common causes of increased ICP, such as venous sinus stenosis. On review of the original outside MRI, one could see a few prominent vessels within the left frontal and parietal convexity on the MRI. This suggests some type of vascular malformation. Thus, it would be appropriate to perform noninvasive angiography before assuming that the patient has "atypical IIH."

3. Case report (continued)

Because of the abnormal vessels seen on MRI, an magnetic resonance angiogram (MRA) and magnetic resonance venogram (MRV) were performed. The MRA showed several prominent cerebral veins in the surface of the left frontal lobe that appeared to be draining to the most anterior aspect of the sagittal sinus. The MRV showed thrombosis of the left transverse and sigmoid sinus, and there was a cortical vein extending into the inferior aspect of the left temporal lobe. A hypercoagulable state workup was positive for cardiolipin antibodies IgG and IgA. Anticoagulation with warfarin was initiated and acetazolamide was given for treatment of "intracranial hypertension secondary to venous sinus thrombosis".

The patient was subsequently referred to The Methodist Hospital at which time he was still experiencing headache. The headache was moderate to severe and diffuse but the Down syndrome in this patient made it difficult to obtain precise details on quality, duration, timing, laterality, and frequency. His visual acuity was 20/30 + OD and 20/40 + OS. The pupils were normally reactive without an RAPD. The slit-lamp examination showed no conjunctival or episcleral vascular arterialization. The intraocular pressure measurements were normal and symmetric. Static perimetry using a Humphrey automated perimeter showed visual fields with a mean deviation of −9.06 dB with mild inferior losses OD, and a mean deviation of −17.88 dB with superior altitudinal and inferior arcuate defects OS (Fig. 1). However, there were some reliability issues with the automated visual field due to the Down syndrome. Extraocular movements were full, and the patient was orthophoric at distance, near, and in the cardinal positions of gaze. Dilated fundus examination revealed that the right optic disk was no longer swollen, but there was persistent left optic disk edema (Fig. 2).

What should be done at this point?

4. Comments (continued)

4.1. *Comments by Dr. Miller*

This patient has bilateral papilledema associated with increased ICP and evidence of cerebral venous sinus thrombosis (CVST). The issue, therefore, is if the sinus thrombosis is a primary or secondary phenomenon. It has become clear in recent years that many patients thought to have "idiopathic intracranial hypertension" actually have primary venous sinus thrombosis or stenosis that is responsible for their elevated ICP and that treatment of the stenosis with stenting results in normalization of the ICP.[21,22] Thus, it would be tempting simply to assume that this is the case in this patient; not all cases of sinus thrombosis are primary, however. In some cases, the thrombosis is a secondary phenomenon related to a pre-existing dural arteriovenous fistula (DAVF).[4,20,25] Thus, before considering further treatment, a catheter angiogram is warranted.

5. Case report (continued)

A catheter angiogram was performed (Figs. 3 and 4) that showed prominence of the transmastoid branch of the right occipital artery and multiple dural branches of the middle meningeal artery crossing the midline and supplying a dural fistula involving the left transverse and sigmoid sinuses. The fistula was fed by a transmastoid branch of the left occipital artery, the posterior auricular artery, the left middle meningeal artery, and the neuromeningeal trunk of the left ascending pharyngeal artery, and it was drained via multiple dilated cortical veins including the vein of Labbé and tentorial veins associated with a venous aneurysm measuring approximately 8 mm × 7 mm.

What should be done at this point?

6. Comments (continued)

6.1. *Comments by Dr. Miller*

Once a DAVF has been found in association with venous sinus thrombosis, the issue is whether to treat the lesion or simply continue to treat the patient's increased ICP with acetazolamide. Although the patient's papilledema has improved on medical therapy, he nevertheless has persistent headache and significant visual field defects. Thus a more aggressive approach would appear to be warranted. The approach would be attempted endovascular closure of the fistula.

7. Case report (concluded)

Although the patient's headache and optic disk edema were resolving with acetazolamide, the decision was made to treat the DAVF because this type of DAVF when associated with CVST has up to a four-fold risk of causing significant neurological manifestations such as intracranial hemorrhage,