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Bilateral Optic Disc Pits With Posterior Pituitary Ectopia

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Abstract

Posterior pituitary ectopia has been reported previously in association with optic nerve hypoplasia, as a variant of septo-optic dysplasia. We describe a 14-year-old boy with posterior pituitary ectopia and bilateral optic disc pits. He had hypopituitarism and a reduction in visual acuity to 20/40 in each eye, owing to loss of foveal ganglion cells. Optic pits and posterior pituitary ectopia may have occurred together in the same subject by chance, but the rarity of both conditions suggests a possible association.

A 14-year-old boy was brought to the eye clinic because of unexplained vision loss. For many years, he had lagged behind his peers in his school work. He received a diagnosis of attention deficit disorder and was treated with methylphenidate. Two years ago, a concussion occurred in a football game, followed by chronic headache. Amitriptyline was prescribed, and he was banned from all physical exertion including running. He was also being treated with levothyroxine, somatropin, and hydrocortisone for chronic pituitary insufficiency of unknown cause.

On examination, the visual acuity was 20/40 in each eye with a refraction of plano in the right eye; −1.00 in both eyes. The pupils were normal. He was too distractible to perform computerized perimetry. There was a prominent gray optic pit at the temporal edge of each optic disc (Fig. 1). A subtle gap in the nerve fiber layer extending from each optic pit to the fovea was visible in each macula with an ophthalmoscope. There were peripapillary pigment rings, a finding often associated with optic nerve pits (1).

Optical coherence tomography (Optovue Avanti RTVue XR) confirmed the presence of optic disc pits, with no subretinal fluid in the macula. Analysis of the ganglion cell complex (nerve fiber layer, ganglion cell layer, and inner plexiform layer) revealed a distinct trough running from the optic pits to the fovea in each eye (Fig. 2). These defects in the ganglion cell complex were about 400 μm wide and 70 μm deep. The patient fixated eccentrically, presumably because the population of ganglion cells serving the fovea in each eye was reduced by the optic pits.

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Brain magnetic resonance imaging showed a slightly diminutive optic chiasm and corpus callosum (Fig. 3A). The most striking finding was the ectopic location of the posterior lobe of the pituitary gland, just beneath the optic chiasm. There was a shortened infundibulum, and the anterior lobe was not visible. The septum pellucidum was preserved. A coronal image through the orbits showed small optic nerves (Fig. 3B).

Brodsky and Glasier (2) have described a variety of central nervous system abnormalities in association with optic nerve hypoplasia. In their report, 6 of 40 patients had absence of the pituitary infundibulum and posterior pituitary ectopia, with resulting hypopituitarism. The term “optoinfundibular hypoplasia” has been proposed for this variant of septo-optic dysplasia, although in fact, the septum pellucidum may be preserved (3).

The finding of bilateral optic nerve pits in a patient with posterior pituitary ectopia is unique. Our subject had no evidence of a serous maculopathy, but loss of foveal ganglion cells due to optic pits caused a reduction in acuity, bringing him to our attention. Optic nerve pits themselves rarely are associated with central nervous system findings (4,5). It is possible that the pits and posterior pituitary ectopia in this child were unrelated, and occurred merely as a coincidence. Alternatively, their occurrence in the same individual could signify a real but rare association, adding to the remarkably diverse range of structural developmental anomalies encountered in patients with pituitary dysgenesis.

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FIG. 1.

Bilateral optic nerve pits are present. The arrows denote pigments surrounding each optic disc, which is organized into 2 distinct, concentric rings nasally. Ophthalmoscopy revealed a gap in the nerve fiber layer, which extended from each pit to the fovea.

FIG. 2.

Ganglion cell complex maps demonstrate a trough of reduced thickness (see scale in microns) extending from each fovea toward the optic pit. Red crosshairs mark patient's fixation, which was eccentric in each eye. Scale denotes microns.

FIG. 3.

Brain magnetic resonance imaging. **A.** Sagittal fluid-attenuated inversion recovery image shows ectopic posterior lobe of the pituitary gland (*arrow*), and apparently absent anterior lobe. The corpus callosum and optic chiasm are slightly thin. **B.** Coronal T2 scan reveals small optic nerves (*arrows*).