

amyloid deposits, including wild-type transthyretin³ as well as other fibrillar proteins.⁴

To our knowledge, this is the first case report of vitreoretinal amyloidosis in the absence of transthyretin mutations. In addition, no signs of systemic amyloidosis suggestive of FAP were evident 7 years after the patient's initial presentation, although we cannot exclude the possibility of a subclinical level of amyloid deposition in other tissues.^{5,6} These findings raise the possibility that isolated vitreoretinal amyloidosis may represent a disorder separate from FAP. We conclude that vitreoretinal amyloidosis encompasses a more heterogeneous group of disorders than has been previously described.

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Optic Nerve Aplasia in an Infant With Congenital Hypopituitarism and Posterior Pituitary Ectopia

Optic nerve aplasia is a rare developmental anomaly characterized by

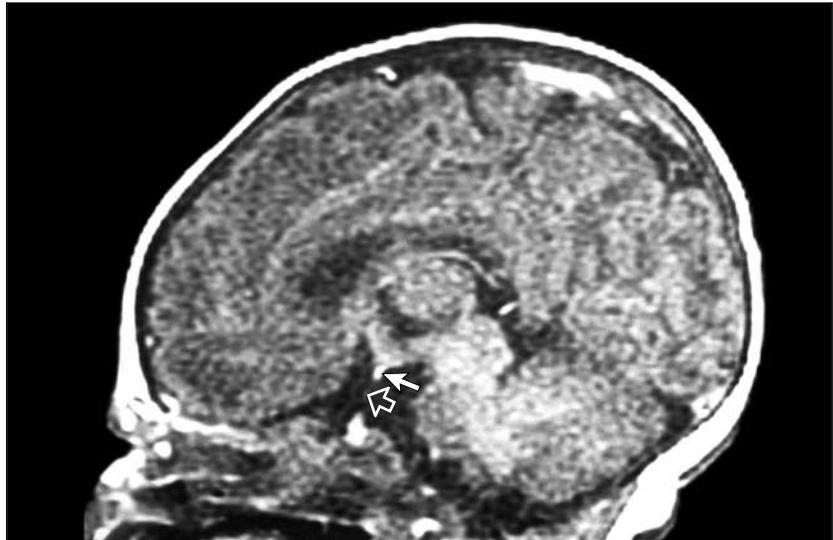


Figure 1. T1-weighted sagittal magnetic resonance image showing ectopic posterior pituitary gland (solid arrow) and absence of the pituitary infundibulum (open arrow denotes normal position of infundibulum).

the congenital absence of the optic nerve, retinal blood vessels, and retinal ganglion cells.¹⁻³ Optic nerve aplasia seems to fall within a malformation complex that is fundamentally distinct from optic nerve hypoplasia, as evidenced by its tendency to occur unilaterally and its frequent association with microphthalmos and other malformations that are confined to the involved eye.^{3,4} Unilateral optic nerve aplasia is generally associated with otherwise normal brain development, while bilateral optic nerve aplasia is usually accompanied by other central nervous system derangement.⁴ To our knowledge, optic nerve aplasia has not been associated with endocrinologic deficiency. This report describes congenital hypopituitarism and posterior pituitary ectopia in an infant with bilateral optic nerve aplasia.

Report of a Case. A male infant was born to nonconsanguineous parents at 38 weeks' gestation with a birth weight of 2750 g. There was no family history of microphthalmos or coloboma. The mother had a history of hyperthyroidism, which was treated in late pregnancy with propothiouracil. Both of the child's eyes were noted to be small at birth, but no other systemic anomalies were observed. The newborn screen showed a total thyroxine level of 4.1 $\mu\text{g/dL}$ (53.0 nmol/L) (normal range, 11-23 $\mu\text{g/dL}$ [142-296 nmol/L]) and an initial serum glucose level of 28 mg/dL (1.6

mmol/L). The patient required intravenous dextrose to maintain a normal serum glucose level. At 4 days of age, magnetic resonance images of the brain disclosed posterior pituitary ectopia with absence of the optic nerves, chiasm, and pituitary infundibulum (**Figure 1**). Endocrinologic testing at 1 week of age showed a free thyroxine level of 0.7 ng/dL (9.0 pmol/L) (normal range, 0.7-1.9 ng/dL [9.0-24.0 pmol/L]), a late afternoon serum cortisol level of 3 $\mu\text{g/dL}$ (77 nmol/L) (normal range, 4-11 $\mu\text{g/dL}$ [110-304 nmol/L]), and a random growth hormone level of 4 ng/mL (185 pmol/L) (normal level > 10 ng/mL [>440 pmol/L]). An insulin-like growth factor I/somatotropin C level obtained 2 months later was 14 ng/mL (normal range, 17-248 ng/mL). Based on the severe hypoglycemia, low anterior pituitary hormone levels, and posterior pituitary ectopia, supplementation with growth hormone, thyroxine, and hydrocortisone was initiated.

The patient had no behavioral response to light shined into either eye during an ophthalmologic examination at 2 months of age. Both palpebral fissures were small, and there was bilateral microphthalmos. The corneas were clear, with a corneal diameter of 4 mm OD and 9 mm OS. The pupils were fixed, round, and nonreactive to light. The retina of the right eye could not be visualized. The left eye showed absence of the left optic nerve and cen-

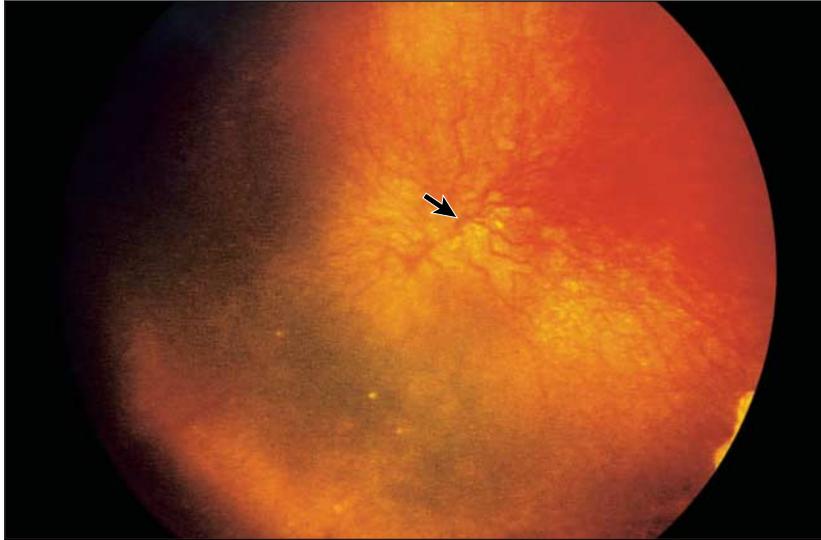


Figure 2. Fundus photograph of the patient's left eye showing absence of the optic nerve and retinal vascular circulation, with a cluster of aberrant vessels occupying the normal position of the optic disc (arrow).

tral retinal vessels during indirect ophthalmoscopy (**Figure 2**). Numerous large, round, circumscribed, whitish areas of retinochoroidal depigmentation were dispersed throughout the retinal periphery. Examination of both parents disclosed normal-sized eyes without evidence of colobomas.

Peripheral blood chromosomes were normal at 450-band resolution. Polymerase chain reaction amplification and sequencing of the coding regions and nearby flanking sequences of the *SIX6* (formerly *OPTX2*) homeobox gene, a developmental regulatory gene that is a candidate gene for oculopituitary disorders,³ showed no mutations or amino acid sequence variants.

Comment. Posterior pituitary ectopia is a neurohypophyseal malformation that is visible on T1-weighted magnetic resonance images as a hyperintense midline nodule localized at or near the tuber cinereum.⁶ It is usually accompanied by absence of the pituitary infundibulum and absence of the normal posterior pituitary bright spot.⁶ While this ectopic cluster of posterior pituitary cells still functions as a normal posterior pituitary gland, the associated absence of the pituitary infundibulum portends a potentially life-threatening anterior pituitary hormone deficiency.⁶ Posterior pituitary ectopia can be seen in patients with optic nerve hypoplasia or as an iso-

lated malformation.⁶ In a recent study of children with optic nerve hypoplasia, Phillips et al⁶ found posterior pituitary ectopia or absence of the pituitary infundibulum in 23 of 26 patients with congenital hypopituitarism vs none of the 41 patients with normal endocrinologic function.

To our knowledge, isolated optic nerve aplasia with hypopituitarism has not been described. However, the association of optic nerve aplasia and hypopituitarism is well recognized in the clinical context of severe microphthalmos and anophthalmos (a condition in which the optic nerves are absent or rudimentary).^{7,8} Keppen et al⁷ found hypogonadotropic hypogonadism in 5 of 13 patients with mental retardation and anophthalmos or severe microphthalmos, suggesting a generalized defect in forebrain development. Brodsky and Frindick⁸ documented a neurohypophyseal malformation closely resembling posterior pituitary ectopia in a male infant with bilateral anophthalmos and low serum cortisol and pituitary gonadotropin levels. Neuroimaging results showed our patient's central nervous system abnormalities were confined to the anterior visual pathways and pituitary infundibulum. The risk of sudden death from Addisonian crisis in children with corticotropin deficiency mandates early recognition of congenital hypopituitarism in high-risk patients.⁹ Posterior pituitary ectopia may prove to be

a neuroimaging marker for congenital hypopituitarism in children with optic nerve aplasia.

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Visual Improvement in an Adult Amblyopic Eye Following Radiation-Induced Visual Loss in the Contralateral Eye

Amblyopia is a visual deficit defined as decreased best-corrected visual acuity of at least a 2-line difference between the two eyes that is not