

Case Report: Septo-Optic Dysplasia

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Patient history

A 14-year-old boy was admitted to the hospital with severe, chronic headaches. MRI shows absence of the septum pellucidum is consistent with his clinical diagnosis of septo-optic dysplasia.

Sequence details

Imaging was performed at 3 Tesla MAGNETOM Trio, A Tim System, using 12-channel head coils. A single shot spin-echo echo-planar imaging (EPI) was used for DTI (diffusion tensor imaging) acquisition with the following parameters: 60 slices without a gap, FOV = 190 mm, phase FOV = 100 %, slice thickness = 2 mm, base resolution = 96, phase resolution = 100 (that makes voxel size = 2 x 2 x 2 mm), phase partial Fourier = 6/8, TR = 9900 msec, TE = 102 msec, average

= 1, b-value = 1400 sec/mm², bandwidth = 1080 Hz, EPI factor = 96, echo spacing = 1 msec. Average ADC map, trace weighted map, FA map, and tensor data were created inline.

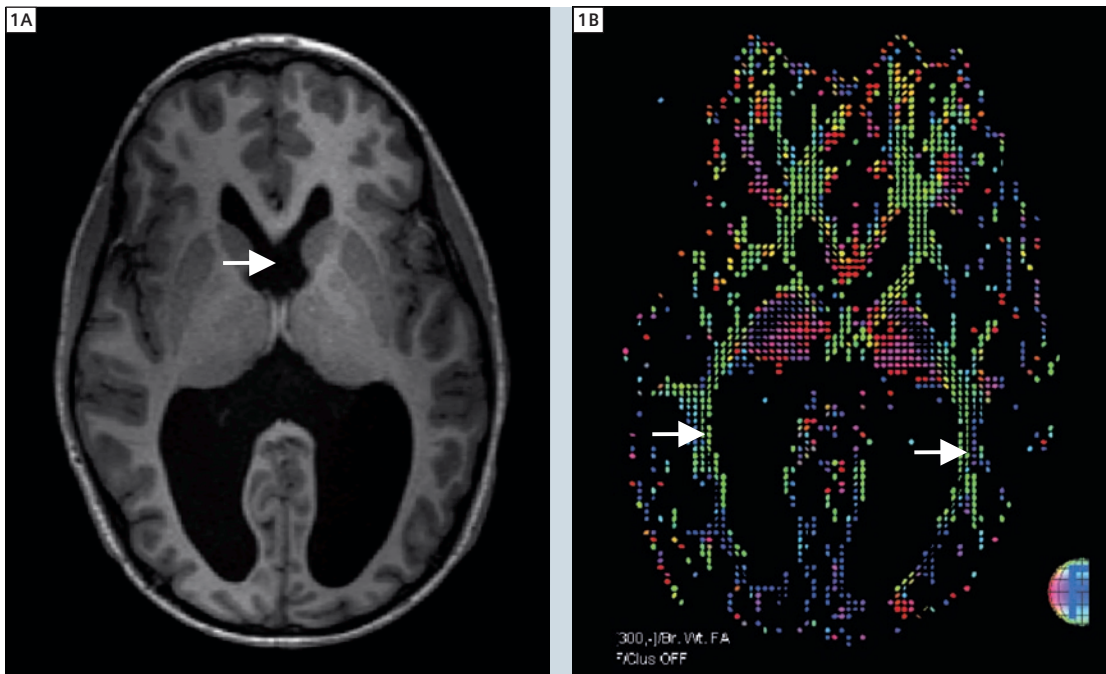
Results and discussion

Moderate hydrocephalus and thinning of the corpus callosum were found. DTI show marked diminution, with significant reduction in the visual fiber tracts of the optic radiations but with persistent anisotropy. These findings are concordant with his clinical presentation of intact visual function and are also in accordance with reports in the literature. Septo-optic dysplasia consists of a heterogeneous deficit of midline brain struc-

tures which include absence or dysplasia of septum pellucidum, optic nerve and pituitary-hypothalamic dysfunction. In addition, the lesions may associate cerebral development malformations such as schizencephaly and polymicrogyria. A HESX1 gene mutation has been identified in familial septo-optic dysplasia. Clinical manifestations include diminished visual acuity, color blindness, nystagmus, microphthalmia, mental retardation, and endocrine disturbance.

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1 (A) T1 (MPRAGE) anatomic image demonstrating enlargement of the ventricles and absence of the septum pellucidum (arrow). (B) Tensor diagram computed from a 4 minute DTI scan performed with isotropic 2 mm voxels at 3 Tesla. The shape and color of the ellipsoids corresponds to the orientation of the white matter tract (anterior-posterior is green, transverse is red, and cranio-caudal is blue). The optic radiations (arrowheads) adjacent to the lateral ventricles are thin but intact.