SEPTO-OPTIC DYSPLASIA

Gina Necula, Florina Antochi
Neurology Department, Emergency University Hospital, Bucharest, Romania

ABSTRACT

Septo-optic dysplasia is a developmental disorder manifested mainly by hypoplasia of the optic nerve, hypopituitarism and absence of septum pellucidum. We present the case of a 58-year-old male with congenital blindness in both eyes, with a MRI scan suggestive for this disorder.

Key words: septo-optic dysplasia, de Morsier syndrome, HESX1, corpus callosum, septum pellucidum, schizencephaly.

Septo-optic dysplasia (SOD), also known as de Morsier syndrome, is a developmental disorder resulting from a defect of normal embryological development. The incidence of septo-optic dysplasia is unknown, but rare.

Also, the cause of septo-optic dysplasia is not known. Rare familial recurrence has been reported, suggesting at least one genetic form (HESX1) (1), but in most cases it is a sporadic birth defect of unknown cause and does not recur again with subsequent pregnancies. Septo-optic dysplasia is linked to young maternal age and nulliparity, one third of septo-optic births resulting from teenage pregnancies (2).

SOD is manifested by hypoplasia of the optic nerve, hypopituitarism and absence of the septum pellucidum. Other midline abnormalities may affect the corpus callosum and cerebellum. However, there are milder degrees of each of the three problems, and some children only have one or two of the three.

Clinically, these result in blindness in one or both eyes, nystagmus, pupil dilation in response to light, inward and outward deviation of the eyes, deficiencies in pituitary hormones (from panhypopituitarism to isolated GH, ACTH or ADH insufficiency), intellectual problems (some children with SOD have normal intelligence, others have learning disabilities and mental retardation), seizures, hypotonia.

MRI or CT findings associated with SOD include: hypoplasia of the optic nerve, optic chiasm and of the optic tracts, absence of the septum pellucidum, abnormal hypothalamic-pituitary axis, agenesia/hypoplasia of the corpus callosum, schizencephaly.

Barkovich et al. (3) classified SOD into two distinct anatomic subsets according to the embryogenesis and the neuropathological findings. One subset included patients with schizencephaly, normal-size ventricles, a remnant of the septum pellucidum and normal-appearing optic radiations. The second group of patients had no schizencephaly, but did exhibit complete absence of the septum pellucidum and diffuse white matter hypoplasia that resulted in ventriculomegaly. Miller et al. (4) suggested the term SOD-Plus to describe SOD associated with malformation of cortical organization, which clinically manifests as global developmental delay and/or spastic motor deficits.

CASE STUDY

A 58-year-old male, with congenital blindness to both eyes (left eye – complete blindness, right eye – distinguishing only the light), with a right hemisphere ischemic stroke in his past, admitted to our clinic for neurological reevaluation.

He was normal born, after a normal pregnancy and labor. The mother was 43-year-old at delivery and he was the third child.

The physical examination revealed a conscious and cooperative patient, with a high value of the arterial tension (200/90 mmHg), a cardiac rhythm of 96/min, a postural tremor, mild motor deficit and superficial hypoesthesia on his left limbs, with muscle

Author for correspondence:
Florina Antochi, Neurology Department, Emergency University Hospital, 169 Splaiul Independentei, Bucharest, Zip Code 050098, Romania
email: **flrant@yahoo.com**

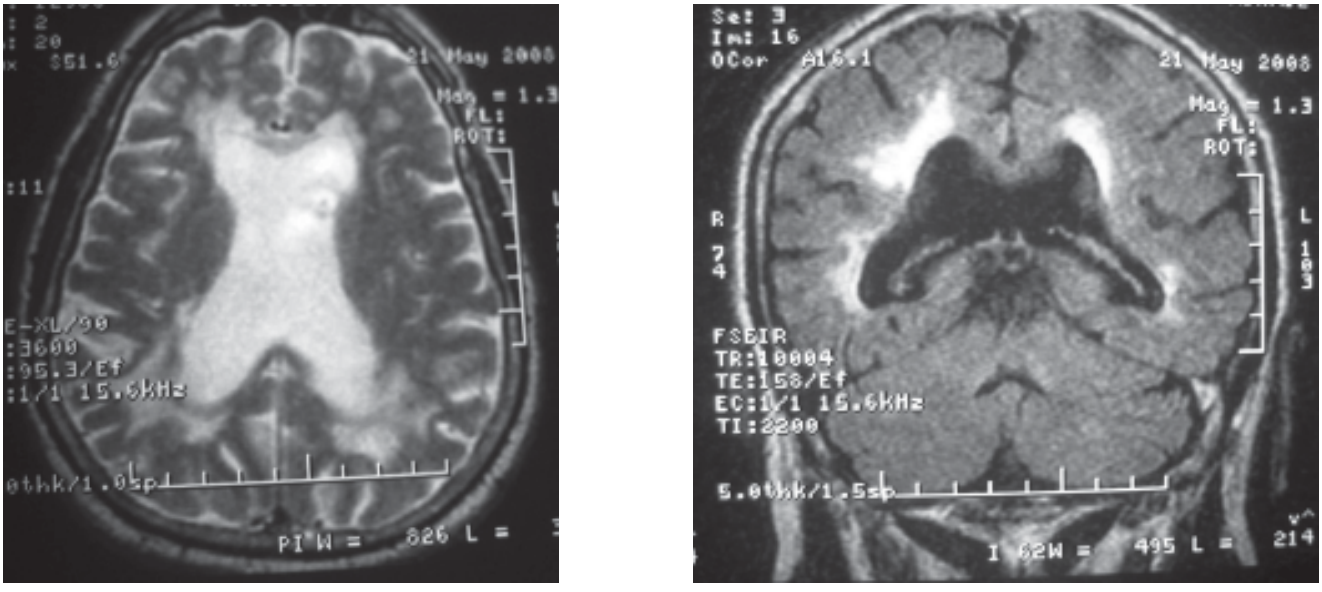
stretch reflexes exaggerated on the left, with the right eye deviated outward and the left one deviated inward, with rotatory nystagmus; both pupils responded normally to direct and consensual light. Physically and mentally, he was normal for his age.

Ophtalmologic examination revealed an optic disc hypoplasia on the left eye, while for the right eye the examination was difficult.

MRI scan revealed the absence of septum pellucidum, hypoplasia of the optic nerves, optic chiasm, optic tracts and hypophyse, and a hypoplasic corpus callosum, suggesting the SOD. A cerebral and a cerebellar atrophy, with a secondary dilation of the ventricular system, were also revealed.

Endocrinological tests weren't performed, but, physically and mentally, the patient was normal for his age, with no other problems to suggest an endocrinological dysfunction.

Our patient had a good prognostic because, except his blindness, he had no other dysfunction (like psychomotor retardation, or seizures, or deficiencies of pituitary hormones). He could be included in the second group of patients from the classification established by Barkovich and al. Also, he didn't have environmental risk factors (young maternal age, nulliparity).



a) b)

Figure 1. *Axial FLAIR (a) and coronal T1 weighted MRI (b) revealing complete absence of the septum pellucidum*

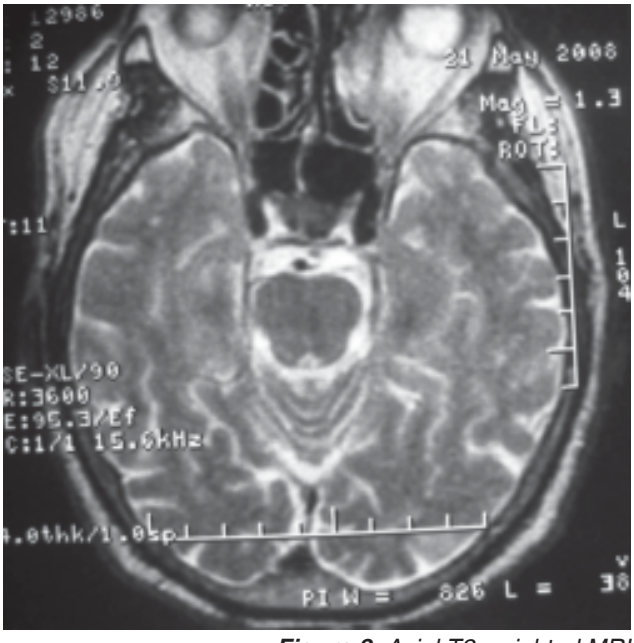


Figure 2. *Axial T2 weighted MRI revealing optic nerve and optic chiasm hypoplasia*

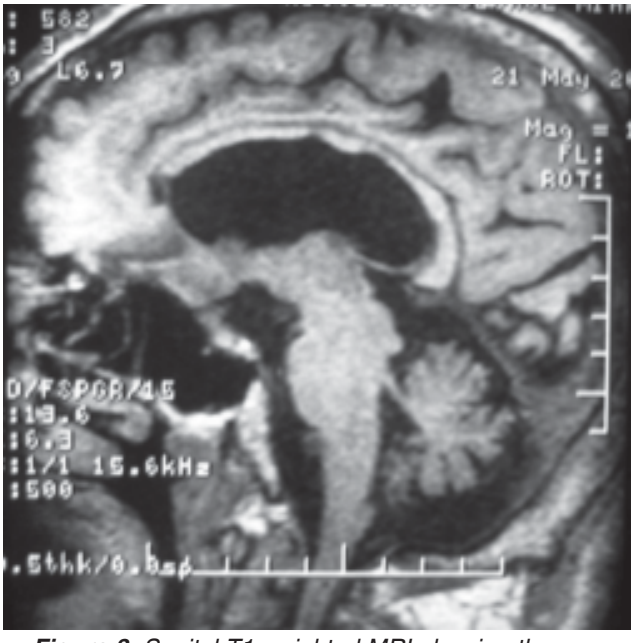


Figure 3. *Sagital T1 weighted MRI showing the corpus callosum hypoplasia*

REFERENCES

1.**Dattani MT, Martinez-Barbera JP, Thomas PQ, Brickman JM, Gupta R, Wales JK, Hindmarsh PC, Beddington RS, Robinson IC** – HESX1: a novel gene implicated in a familial form of septo-optic dysplasia. *Acta Paediatr Suppl.* 1999 Dec;88(433):49-54.
2.**Lippe B, Kaplan SA, La Franchi S.** – Septo-optic dysplasia and maternal age. *Lancet*. 1979;2(8133):92-3.
3.**Barkovich AJ, Fram EK, Norman D.** – Septo-optic dysplasia: MR imaging. *Radiology* 1989; 171: 189-192.
4.**Miller SP, Shevell MI, Patenaude Y, et al.** – Septo-optic dysplasia plus: spectrum of malformations of cortical development. *Neurology* 2000; 54: 1701-1703.