
SEPTO-OPTIC DYSPLASIA (DE MORSIER SYNDROME)

*LELOUP E. *, DELBEKE P.**

ABSTRACT

A 12-year old boy presented with poor vision and nystagmus. Fundus examination revealed bilateral optic nerve hypoplasia. An MRI of the brain demonstrated the absence of the septum pellucidum, which confirmed a diagnosis of septo-optic dysplasia or de Morsier syndrome.

RÉSUMÉ

Un garçon de 12 ans se présentait avec une acuité visuelle diminuée et un nystagmus. L'examen du fond d'oeil révèle une hypoplasie bilatérale du nerf optique. L'RMN démontrait l'absence du septum pellucidum ce qui confirme le diagnostic d'une dysplasie septo-optique ou syndrome de de Morsier.

KEY WORDS

Septo-optic dysplasia, de Morsier syndrome, optic nerve hypoplasia

MOTS-CLÉS

Dysplasie septo-optique, syndrome de de Morsier, hypoplasie du nerf optique

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* *Department of Ophthalmology,
Ghent University Hospital,
Ghent, Belgium*

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INTRODUCTION

Septo-optic dysplasia (SOD) is a rare disorder characterized by optic nerve hypoplasia, dysgenesis of the septum pellucidum and hypothalamic-pituitary dysfunction. Already in 1941, Reeves reported a patient with optic nerve anomalies and absence of the septum pellucidum (6). But it was only in 1956 that de Morsier called attention to the coincident occurrence of these findings. He named this condition septo-optic dysplasia (3).

Ophthalmologically SOD may manifest as strabismus, nystagmus, decreased visual acuity or visual impairment. Neurological and endocrinological abnormalities may be present. Growth hormone deficiency is the most common (8). We report a case of a 12-year old boy immigrated from Afghanistan who presented with decreased visual acuity and congenital nystagmus. The cause was unknown to the patient and his father.

CASE REPORT

A 12-year old boy presented at our department of ophthalmology with (congenital) nystagmus and poor vision. He had immigrated from Afghanistan to Belgium 2 weeks earlier. His par-

ents were consanguinous (first cousins). He was born after a full term pregnancy and a difficult home delivery. He suffered from a left hemiparesis and with a stature of 1.40m he was small for his age.

Visual acuity was 1.5/10 in the right eye and no light perception in the left eye. There was a moderate esotropia and a small hypotropia of the left eye. He had a pendular nystagmus. The left pupil did not respond to light. Ophthalmoscopic examination revealed bilateral optic disc hypoplasia with the typical 'double ring sign' and normal maculae (Figs. 1 & 2). An MRI scan of the brain showed absence of the septum pellucidum (Fig. 3), mild dysplasia of the corpus callosum, schizencephaly, heterotopia, thinning of the pituitary stalk and bilateral hypoplasia of the optic nerve and optic tract. Posterior pituitary ectopia was not seen. The combination of bilateral optic disc hypoplasia and midline CNS abnormalities led to the diagnosis of de Morsier syndrome. He was subsequently referred to our low vision department. He was also referred to the pediatric neurology and endocrinology department for further investigation and treatment. The left hemiparesis is due to the schizencephaly located in the motor cortex. He was referred to a revalidation center.

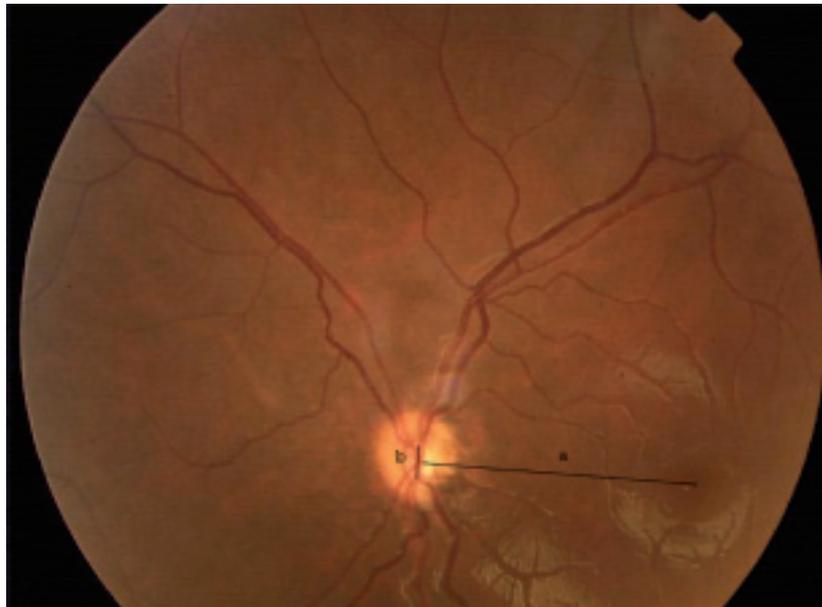


Fig. 1: Optic disc with double ring sign in the left eye. Ratio of a/b is more than 3.



Fig. 2: Optic nerve hypoplasia in the right eye

DISCUSSION

In many cases the etiology of SOD is unknown. The association of SOD with maternal diabetes and the use of alcohol, LSD, quinine, phenytoine and anti-convulsive medication during pregnancy has been described (4,5). In our case, the cerebral hemispheric abnormalities are highly predictive of neurodevelopmental deficits. Pathologic studies suggest that schizencephaly results from severe hemispheric injury early in the second trimester of pregnancy. The specific mechanism of injury may differ from case to case. In our case no periventricular leukomalacia was seen. Periventricular leukomalacia usually results from hypoxic-ischemic injury to watershed zones of arterial supply in the periventricular white matter of the immature brain (2). The absence of such abnormalities in our patient suggests that the difficult home delivery of our patient is probably not related to his septo-optic dysplasia and hemiparesis. The consanguinity of our patient's parents, a genetic cause cannot be excluded. Optic nerve hypoplasia may have various clinical manifestations. The visual acuity, endocrinological status, and CNS anatomy vary wide-

ly, ranging from a child with functional vision without endocrine or CNS defects, to the blind child with panhypopituitarism (8). Since the ocular abnormality is detected first in most of the cases, it is essential for ophthalmologists to recognize the optic disc hypoplasia, to look for midline CNS abnormalities and to evaluate pituitary function (7).

Children with corticotropin deficiency are at risk for sudden death during febrile illness, which may be caused by an impaired ability to increase corticotropin secretion to maintain blood pressure and blood sugar in response to physical stress. Because this corticotropin deficiency represents an imminent threat to life, a complete pituitary evaluation should be performed in children with clinical symptoms and/or signs of pituitary abnormalities on neuroimaging. Because growth hormone deficiency is the most common abnormality, and all patients who manifest with endocrinopathy have growth hormone deficiency, this may be used as a screening test (8).

Our case was specifically interesting because it nicely illustrates the pathognomonic 'double ring sign'. This sign has been found histopathologically to consist of a normal junction be-

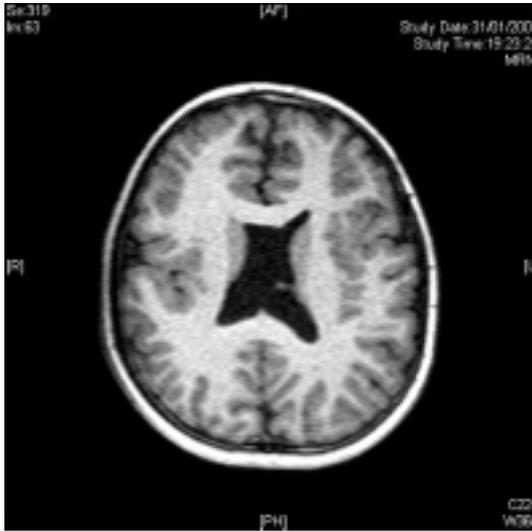


Fig. 3: MRI: absence of septum pellucidum.

tween the sclera and the lamina cribrosa, which corresponds to the outer ring, and an abnormal extension of retina and pigment epithelium over the outer portion of the lamina cribrosa, which corresponds to the inner ring. This ring sign helps the clinician to differentiate optic disc hypoplasia from optic atrophy. However, recognizing optic disc hypoplasia often remains challenging, with a correct diagnosis sometimes missed on indirect ophthalmoscopy due to nystagmus. A useful tool is the ratio of the distance between the fovea and the center of the optic disc to the optic disc diameter. If this ratio is more than 3, a diagnosis of optic disc hypoplasia is confirmed (1) (Fig 1). Regular ophthalmological follow-up is important. Optimal refraction and, if necessary occlusion therapy, is necessary for maximum visual maturation.

CONCLUSION

We have presented a case of septo-optic dysplasia with bilateral optic nerve hypoplasia. The pathognomonic double ring sign is seen in the left eye. The diagnosis of bilateral hypoplasia seen on fundoscopy was the feature which led to the diagnosis. A brain MRI scan confirmed the diagnosis. It is important to realise that further pediatric screening, including endocrinological and neurological evaluation is required in such patients.

REFERENCES

- (1) ALVAREZ E., WAKAKURA M., KHAN Z., DUTTON G.N. – The disc-macula distance to disc diameter ratio: a new test for confirming optic nerve hypoplasia in young children. *J Pediatr Ophthalmol Strabismus* 1988;25:151-4.
- (2) BRODSKY M.C., GLASIER C.M. – Optic nerve hypoplasia. Clinical significance of associated central nervous system abnormalities on magnetic resonance imaging. *Arch Ophthalmol* 1993;111:66-74.
- (3) DE MORSIER G. – Etudes sur les dysraphies crânio-encéphaliques. III. Agénésie du septum lucidum avec malformation du tractus optique; la dysplasie septo-optique. *Schweitz Arch Neurol Psychiatr* 1956;77:267-92.
- (4) HOYT C.S., BILLION F.A. – Maternal anti-convulsants and optic nerve hypoplasia. *Brit J Ophthalmol* 1978;62:3-6.
- (5) NELSON M., LESSELL S., SADUN A.A. – Optic nerve hypoplasia and maternal diabetes mellitus. *Arch Neurol* 1986;43:20-25.
- (6) REEVES D.L. – Congenital absence of the septum pellucidum: a case diagnosed encephalographically and associated with congenital amaurosis. *Bull Johns Hopkins Hosp* 1941; 69:61-71.
- (7) ROESSMANN U. – Septo-optic dysplasia or de Morsier syndrome. *J Clin Neuro-ophthalmol* 1989;9:156-9.
- (8) SIATKOWSKI R.M., SANCHEZ J.C., ANDRADE R., ALVAREZ A. – The clinical, neuroradiographic, and endocrinological profile of patients with bilateral optic nerve hypoplasia. *Ophthalmology* 1997;104:493-6.

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Correspondence and reprints:
 Eva LELOUP
 UZ Gent Oogheekunde (poli 1)
 De Pintelaan 185
 B-9000 Gent
 email: eva.leloup@telenet.be