

# Septo-optic Dysplasia

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## Summary

The term septo-optic dysplasia was coined in 1956 by de Morsier, who pointed out the association of optic nerve hypoplasia and absence of the septum pellucidum. Patients with this condition may present with clinical features of hypopituitarism, decreased visual acuity and neurodevelopmental disabilities that lead to this diagnosis. The case that is presented here is unusual in that this patient was initially diagnosed as having low tension glaucoma during a routine screening examination and was treated for glaucoma for over a year before he was discovered to have septo-optic dysplasia, also known as de Morsier's syndrome.

**Key Words:** Septo-optic dysplasia, de Morsier's syndrome, Glaucoma, Schizencephaly

## Introduction

The term septo-optic dysplasia denotes the association of optic nerve hypoplasia with an absent septum pellucidum and a thin corpus callosum, a condition first recognized in neuropathological specimens by de Morsier in 1956<sup>1</sup>. Often this diagnosis is brought to light when patients present with hormonal deficiencies, reduced vision or neurodevelopmental delay. The case that is presented here is unusual in its presentation because the patient was diagnosed initially to have glaucoma when he underwent a routine eye screening examination. As a result he was treated for glaucoma for more than one year before the true diagnosis came to light.

## Case Report

A 20-year-old Malay man was referred to us from Tawau Hospital in January 2001 for glaucoma treatment. He had been diagnosed as having glaucoma after he went for an eye screening examination in February 2000 in Tawau. The ophthalmologist there had detected asymmetrically raised cup disc ratios (right eye 0.6, left eye 0.4). However, the intraocular pressures in both eyes were normal. Perimetry done in Tawau showed "early glaucomatous changes" according to the referral letter. He was therefore diagnosed to have low tension glaucoma and was on Timolol eye drops 0.5% twice a day. Vision in both eyes was 6/6, unaided.

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## CASE REPORT

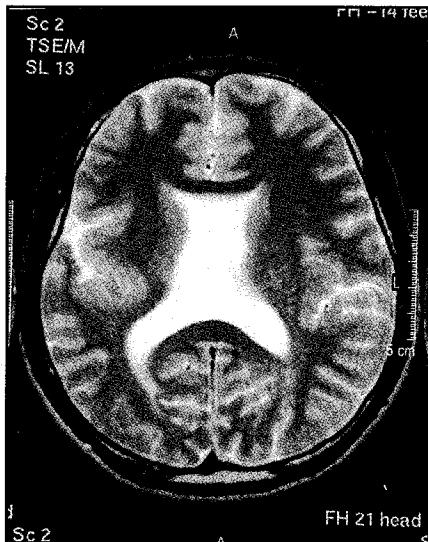
By the time he presented us at the ophthalmology outpatient clinic in Kajang Hospital he had run out of timolol eye drops for one week's duration. On ocular examination, right eye vision was 6/9, left eye 6/6. He had a right divergent squint of 15 degrees for near and distance. There was no relative afferent pupillary defect. (Marcus Gunn pupil).

Anterior segment findings were unremarkable. The intraocular pressure was 18mmHg in both eyes and gonioscopy revealed grade four angles in bilaterally. The cup disc ratios were right eye 0.6 and left eye 0.4. (However the optic discs did not have a typical glaucomatous appearance).

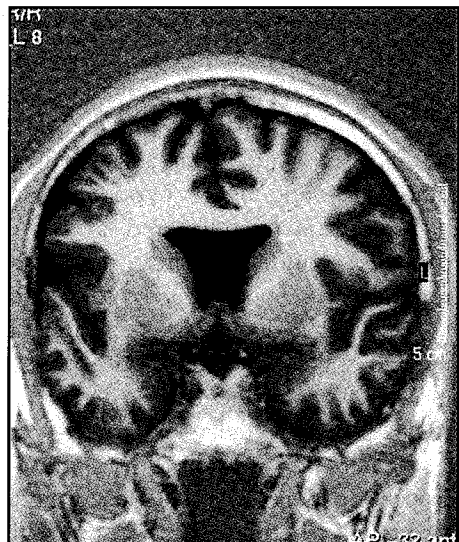
An automated visual fields was ordered for this patient. Visual fields however did not show visual field loss that was consistent with the diagnosis of glaucoma. The patient had bitemporal hemianopia which meant a space

occupying lesion at the optic chiasm had to be excluded. A CT scan of the orbit and brain ordered urgently showed a normal pituitary gland. However there was absence of the septum pellucidum, resulting in a large midline cavity. There was also a cleft in the right parieto-temporal region which was lined by a thick mantle of grey matter.

It was concluded that the patient had septo-optic dysplasia with schizencephaly. The optic nerve head appearance in both eyes was thus due to septo-optic dysplasia rather than glaucoma. An elective MRI scan also revealed a normal pituitary gland, absence of septum pellucidum and schizencephaly (Figure 1 and 2). Over a 6 month follow-up with us the patient's intraocular pressures were not raised even after stopping Timolol eye drops once the diagnosis of septo-optic dysplasia was confirmed by CT scan.



**Fig. 1: T2 weighted MRI film showing large midline cavity and right parietotemporal cleft**



**Fig. 2: T1 weighted MRI film showing absence of septum pellucidum and right schizencephaly**

## Discussion

The term septo optic dysplasia was coined by De Morsier in 1956, who pointed out the association of optic nerve hypoplasia and absence of the septum pellicidum. Often, patients with this condition are diagnosed at childhood as they have pituitary dysfunction, most commonly growth hormone deficiency. The patient we reported was diagnosed to have septo optic dysplasia in adulthood. He did not have any developmental delays as a child and grew to a height of 170 cm. His physical appearance did not suggest he had hypopituitarism. It is also interesting that this diagnosis was only brought to light when the patient reached adulthood, after the patient was first diagnosed as having glaucoma during a routine eye screening examination.

Schizencephaly is a cerebral hemisphere migration anomaly in which an abnormal gray matter lined cleft extends through the cerebral hemisphere from the lateral ventricle to the cortical surface. Schizencephaly may accompany septo-optic dysplasia and can lead to seizure disorder and mental retardation<sup>2</sup>. This patient was however seizure free and was pursuing an information technology course in a local institution. A small optic disc is expected in patients with septo-optic dysplasia, as this

diagnosis is usually associated with a small optic disc area and a reduced number of nerve fibers. However a small optic disc is not a prerequisite for the diagnosis<sup>3</sup>. This patient did not have small optic discs, however the reduced number of nerve fibers gave rise to the appearance of a raised cup disc ratio in both optic discs, which led to the misdiagnosis of glaucoma initially.

Rarely is a nonneoplastic disorder responsible for a bitemporal visual field defect. Fortunately for this patient, the bitemporal visual field loss was secondary to optic nerve hypoplasia rather than a neoplastic lesion.

We need to maintain a high index of suspicion with regards to the diagnosis of glaucoma if the patient has a visual field defect that is not consistent with the diagnosis of glaucoma and also does not have recorded pretreatment raised intraocular pressures. The patient did not have raised intraocular pressures at any stage even when he was not on Timolol eyedrops. It would have been unfair to subject the patient to a lifelong treatment of glaucoma, which he did not have. Also if the cause of the bitemporal visual field defect had been a neoplastic lesion, the patient's proper treatment would have been delayed by a year which could have led to potentially unwarranted complications.

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