SEPTO-OPTIC DYSPLASIA (DE MORSIER’S SYNDROME) ASSOCIATED WITH TOTAL CALLOSAL ABSENCE

A new type of the anomaly

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SUMMARY

It is currently believed that there are two subsets of septo-optic dysplasia (de Morsier’s syndrome), one with schizencephaly and the other without schizencephaly. Also, some authors consider septo-optic dysplasia as a mild form of holoprosencephaly. This article describes a third form of the anomaly which is associated with total callosal absence. Two patients similar to this one have previously been reported. None of these three patients had interhemispheric fusion in any form, excluding holoprosencephaly. They had normal facies. All of them presented with seizures, and no hormonal abnormality was found. This new clinicoradiological type of the anomaly is suggested to be labelled as calloso-septo-optic or calloso-optic dysplasia.


INTRODUCTION

In the majority of the previously studied cases of septo-optic dysplasia (De Morsier’s syndrome) by MR imaging the corpus callosum was present, although in some cases it showed diffuse or focal thinning [1, 2]. Recently, two reports have been published in the radiological literature, which documented total [3], and partial [4] absence of the corpus callosum associated with septo-optic dysplasia. In this communication, we describe a further example of this condition.

CASE REPORT

A 6-months-old boy was referred for seizures and very poor visual fixation. An ophthalmological examination revealed bilateral hypoplasia of the optic discs. On a routine cranial MR imaging study the corpus callosum (hence, the septum pellucidum) was totally absent. The optic nerves, chiasm, and the optic tracts were apparently thinned. The temporal horns were dilated in the absence of hydrocephalus, which was due to incomplete inversion of the hippocampal formations. The anterior cerebral artery was azygous. No interhemispheric fusion was evident, and the falx was present. There was no abnormal facies. On MR imaging...
a, b et c) Les images IRM sagittales pondérées en T1 montrent une absence totale du corps calleux, d'où le septum pellucidum. Il n'y a aucun signe de schizencéphalie. La bandelette optique est fine (b) : voir aussi la figure 1d.
Du point de vue ophtalmologique les papilles optiques sont hypoplastiques (dyplasie callo-septo-optique).

d et e) Après injection de contraste, image IRM coronaire pondérée en T1 (obtenue en utilisant la technique inversion-récupération, impulsions de radiofréquence) montre un chiasma optique très fin (flèches). Le chiasma est au moins 3 fois plus fin que celui des sujets normaux. Noter l’absence de fusion inter-hémisphérique, ce qui exclut l’holoprosencéphalie.

a, b et c) T1-weighted sagittal MR images show total callosal absence, hence the septum pellucidum. There is no evidence of schizencephaly. The optic tract is thin (b) (also see fig. 1d). Ophthalmologically the optic discs were hypoplastic (calloso-septo-optic dysplasia).

d and e) Contrast-enhanced, T1-weighted coronal MR image (obtained utilizing the inversion recovery pulse sequence) shows a very thin optic chiasm (arrows). The chiasma is at least 3 times thinner compared to normal individuals. Note that there is no interhemispheric fusion, excluding holoprosencephaly (e).
the cerebellum was normal, and there was no intraorbital anomaly. No hormonal abnormality could be found.

The patient was reexamined by MR imaging at the age of 18 months, mainly directed to the hypothalamic-pituitary region, and callosal absence and thinning of the optic chiasm were well demonstrated (fig. 1a, b, c). The chiasm was at least 3 times thinner compared to normal individuals (fig. 1d). There was no evidence of schizencephaly (fig. 1a, b, c), and holoprosencephaly (fig. 1e). Laboratory tests revealed no hormonal abnormality.

Considering the ophthalmological and radiological evidence of hypoplasia of the optic discs and nerves and the absence of the septum pellucidum a diagnosis of De Morsier’s syndrome (septo-optic dysplasia) was established.

DISCUSSION

According to Barkovich et al. [1] septo-optic dysplasia has two forms. More than half of the patients affected with this condition manifest dysfunction of the hypothalamic-pituitary axis, and half have schizencephaly. Based on these facts, Barkovich et al. [1] have suggested that there are two subsets of patients with septo-optic dysplasia: (a) those with schizencephaly and normal-appearing optic pathways, and (b) those without schizencephaly but with hypoplasia of the optic nerves and hypothalamic-pituitary dysfunction [1].

In the majority of the previously studied cases of septo-optic dysplasia by MR imaging the corpus callosum was present but in some cases it showed diffuse or focal thinning [1, 2]. In 1993, however, we had reported a patient with septo-optic dysplasia associated with total absence of the corpus callosum [3]. Before this, in 1992, Lahat et al. [4] described a patient with septo-optic dysplasia, partial absence of the corpus callosum, and an interhemispheric cyst. The current case is a further example of this condition, and it is suggested that all these cases may represent a rarer third form of the anomaly. Of particular note all of these patients had seizures. No hormonal abnormality was noted. There was no cerebellar or intraorbital lesion. They had normal facies. All these features suggest a new clinicoradiologic type of the anomaly.

It has been previously suggested that the form of septo-optic dysplasia without schizencephaly may indeed represent a mild lobar holoprosencephaly because absence of the septum pellucidum and hypopituitarism are common to both entities [5]. Abnormal facial features may or may not be present in lobar holoprosencephaly. The facial structures of the current patient was normal as well as previously reported patients [3, 4].

In lobar holoprosencephaly which is the least severe form of other holoprosencephalies (alobar and semilobar), there is always varying degrees of fusion of the two cerebral hemispheres usually at the rostral frontal regions and sometimes across the other parts of the interhemispheric fissure. According to Barkovich and Quint this may result from a deficiency of mesenchyme [6]. They suggested that faulty mesenchyme formation accounts for maldevelopment of the interhemispheric fissure, the falx cerebri, and the corpus callosum. In lobar holoprosencephaly the corpus callosum is usually dysgenetic or absent. Also, atypical callosal dysgenesis have been shown in lobar holoprosencephaly, which consisted of presence of merely the splenium but the anterior parts or absence of only the middle parts of the corpus callosum [6, 7]. In the current patient with septo-optic dysplasia the corpus callosum was totally absent, but no interhemispheric fusion was evident. The anterior cerebral artery was azygous, which may be related to callosal absence (and faulty mesenchyme formation).

In conclusion, the author believes that although the current patient and previously reported patients [3, 4] appear to support the previous belief that septo-optic dysplasia may represent a mild form of holoprosencephaly there is a distinct difference between the two, which is readily recognized by MR imaging. That is, in all types of holoprosencephalies including the mildest form there is interhemispheric fusion which is a mandatory feature for the diagnosis [6, 7]. In the current patient and in the previous ones, however, no fusion was seen. For this reason, it seems reasonable to label these patients as a new (third) form of septo-optic dysplasia rather than considering them a mild form of holoprosencephaly. Total callosal absence, a major anomaly, was common to our both patients [3], and the patient reported by Lahat et al. [4] had partial callosal absence. Also, there was no evidence of schizencephaly. Therefore we suggest the term calloso-septo-optic dysplasia (or calloso-optic dysplasia) to describe this new type of the anomaly. It appears that a relationship between the callosal anomaly and the abnormal optic pathways could be considered, which remains to be clarified by further studies.

REFERENCES

