Septo-optic dysplasia with cerebellar hemiagenesis-a rare congenital malformation

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ABSTRACT

Septo-optic dysplasia (De morsier's syndrome) is a rare congenital malformation of the central nervous system and represents a clinical spectrum rather than a specific entity. It is defined by any combination of pituitary hypoplasia with or without consequent panhypopituitarism, optic nerve hypoplasia and midline neurological abnormalities such as absent septum pellucidum and corpus callosum agenesis or thinning. Septo-optic dysplasia with associated cerebellar hemiagenesis is a much rarer congenital anomaly and may represent one end of this spectrum.

Keywords: Septo-optic dysplasia, cerebellar hemiagenesis, De morsier syndrome.

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INTRODUCTION

Septo-optic dysplasia (De morsier's syndrome) is a rare congenital malformation of the central nervous system and represents a clinical spectrum rather than a specific entity. It is defined by any combination of pituitary hypoplasia with or without consequent panhypopituitarism, optic nerve hypoplasia and midline neurological abnormalities such as absent septum pellucidum and corpus callosum agenesis or thinning¹. The overall incidence of septo-optic dysplasia is 1 in 10 000 live births. There is no specific gender predilection with males being affected as frequently as females. There appears to be some correlation with maternal age as infants born to younger mothers seem to be affected more commonly than others^{2, 3}. A number of risk factors have been identified including maternal diabetes, medications like quinidine ingestion and antiepileptics, drug and alcohol abuse and cytomegalovirus infection^{1,4}.

CASE HISTORY

We report a case of a 2 month old girl child, first in birth order and a product of non-consanguineous marriage, who

was diagnosed as having hydrocephalus on antenatal ultrasound. The patient was followed up postnatally and a noncontrast CT head was done at age of 1 month which also revealed hydrocephalus. MRI brain revealed absence of septum pellucidum and thinning of corpus callosum, box shaped dilated lateral ventricles, down pointing of frontal horns of lateral ventricles (Figure 1), cerebellar hemiagenesis and rudimentary vermis on left side (Figure 2), cerebral volume loss and hypoplastic left optic nerve with normal optic chiasma (Figure 3). General physical examination revealed lethargic baby with normal vitals. No cutaneous markers of any neurocutaneous syndrome were found. Ophthalmological examination revealed normal anterior segment of both eyes. Fundus examination revealed hypoplastic left optic disc. Hormonal assays for

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pituitary function were performed which revealed reduced levels of multiple hormones. ACTH was 2pg/dl, cortisol (8 A.M) was 2.1µg/dl, growth hormone was 0.3ng/ml and TSH was 0.35µIU/ml. Based on radiological and ophthalmological evidence of absence of septum pellucidum, hypoplasia of optic disc and nerves, thinning of corpus callosum diagnosis of De morsier's syndrome (septo-optic dysplasia) was made.

FIGURE 1: Coronal T2W (1A) and Sagittal T1W (1B) MR images of brain reveal dilated lateral ventricles with absent septum pellucidum, down pointing of frontal horns and thinning of corpus callosum (arrow). Cerebral volume loss is also noted.

FIGURE 2: Axial T2W (2A) and T1W (2B) MR images of brain reveal agenesis of left cerebellar hemisphere (black arrow) with rudimentary vermis (white arrow).

FIGURE 3: Coronal CISS MR Images of brain reveal hypoplastic left optic nerve (3A) with normal optic chiasma (3B).

DISCUSSION

Michaud J et al reported a rare case of agenesis of the

cerebellar vermis with fusion of the dentate nuclei and cerebellar hemispheres (rhombencephalosynapsis) associated with absence of septum pellucidum, hypoplasia of the commissural system, optic nerves, chiasm and tracts, moderate hydrocephalus, and agenesis of the posterior lobe of the pituitary in a seven hour old neonate⁵. Our patient was diagnosed as having hydrocephalus on antenatal ultrasound during level II scan. Subsequent MR brain revealed absent septum pellucidum, hydrocephalus, hypoplastic left optic nerve and left cerebellar hemisphere agenesis with rudimentary vermis, however, no rhombencephalosynapsis was present. Barkovich et al reported two subsets of septo-optic dysplasia, half of the patients had dysfunction of hypothalamic-pituitary axis with hypoplasia of the optic nerves, while remaining half had schizencephaly with normal optic nerves. Our case had hypoplastic left optic disc, however, neither schizencephaly nor hypothalamic - pituitary axis dysfunction was present at time of scan. Both olfactory bulbs were normal. Septo-optic dysplasia with schizencephaly is a disorder distinct from those cases without schizencephaly, because hypopituitary dwarfism is not present in the schizencephalic group⁶. On MR imaging, corpus callosum may be normal in majority of cases but some cases may demonstrate diffuse or focal thinning^{6,7}. Sener RN reported a case with septo-optic dysplasia associated with complete absence of the corpus callosum in the year 1993⁸. In 1996, Sener RN et al reported another case with total corpus callosal absence⁹. Our case had thinned out corpus callosum. In 1992, Lahat et al described a patient with septo-optic dysplasia, partial absence of the corpus callosum, and an interhemispheric cyst¹⁰. Among the major reported clinical findings, hypopituitarism is seen in 62-80% of the cases with growth hormone deficiency being the commonest endocrine abnormality. Significant visual impairment is found in 23% of the cases. Developmental delay is more common in children with bilateral optic nerve hypoplasia (57%) as compared to those with unilateral optic nerve hypoplasia (32%). The most common features of septo-optic dysplasia include mental subnormality, learning disorders, epilepsy (including infantile spasms), hypotonia, hemiparesis (particularly in cases with schizencephaly or porencephaly), spastic diparesis (especially those cases with periventricular leukomalacia), and neurosensory hearing loss. Social communication difficulties, at times evolving into a frank autism spectrum disorder are

frequently observed in children with septo-optic-pituitary dysplasia complex as well as in children with isolated bilateral optic nerve hypoplasia and blindness. The most frequent neurological features are seizures, developmental delay and cerebral palsy. Our case had no developmental delay, seizures, spastic diparesis, hypotonia, hearing loss or cerebral palsy. There is ongoing research in the field to understand the etiology of this syndrome as the exact etiology is still unknown.

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