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Septo-optic dysplasia with lissencephaly

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ABSTRACT

Septo –optic dysplasia with lissencephaly rarely occurs simultaneously. We describe such a patient presenting with absent visual fixation, mild developmental delay, ophthalmological examination revealed small pale optic disc with typical double rim and B/L optic nerve hypoplasia with CT scan finding of lissencephaly and absence of septum pellucidum. The above finding leads to the diagnosis of septo-optic dysplasia with lissencephaly.

Keywords: septo-optic dysplasia, lissencephaly, optic –nerve, septum pellucidum.

INTRODUCTION

Septo-optic dysplasia (SOD) is a syndrome of midline abnormalities of central nervous system resulting in absence of septum pellucidum and dysplasia of optic chiasm or optic nerves [1]. Often diagnosis is brought to light when patient presents with abnormal hormonal deficiencies, reduced vision or neurodevelopmental delay [2]. Cases of septo-optic dysplasia with classical features or its variants have been described in western literature [3]. We present a case of SOD associated with lissencephaly. We are not aware of any report from Indian workers and this prompted the current description.

CASE REPORT

A 9 months old female patient was seen for absent visual fixation since last 4 months .At the age of 5 months, child developed fever, for which he had been treated as a case of meningitis. Fever subsided on the third day of treatment.

She was born normally after a term uncomplicated delivery. She cried immediately after birth with no postnatal complication. There was mild developmental delay, as social smile was observed with 3 month of age and head control at 4 months .Ophthalmological examination revealed absence of response to light, small pale optic disc with typical double rim, B/L optic nerve hypoplasia. Endocrinological function tests were done but did not show any abnormalities.

CT scan of head showed malformation of cortical development (lissencephaly) and absence of septum pellucidum (Fig.1). TORCH and MRI was suggested but the party was not willing for the tests. The diagnosis was clenched with the help of CT scan. Later on counseling was given and physiotherapy was advised.

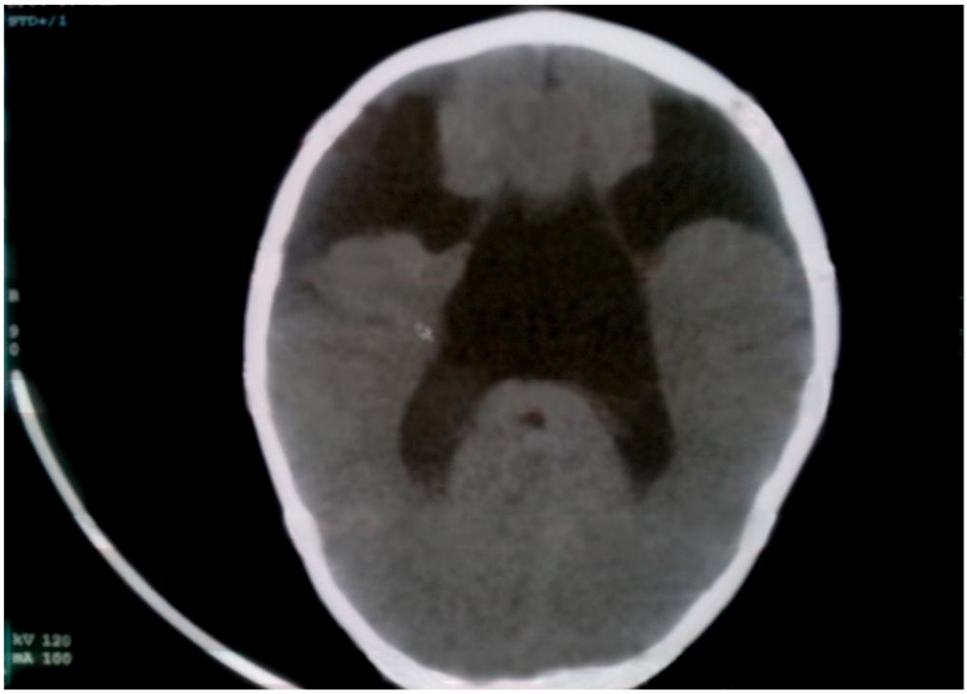

Fig.01- septo-optic dysplasia with lissencephaly

DISCUSSION

Lissencephaly , which literally means "smooth brain", is a rare ,gene-linked brain malformation characterized by the absence of normal convolution (folds)in the cerebral cortex and an abnormally small head (microcephaly) [4]. Lissencephaly is caused by defective neuronal migration during embryonic development, the process in which nerve cells move from their place of origin to their permanent location within the cerebral cortex grey matter[5]. Septo –optic dysplasia was first described by De Morsier in 1956[6]. There is wide variety of the clinical picture usually, patient with SOD presents with hormonal deficiency, reduced vision or neurodevelopmental delay .In some cases, intellectual and neurological development as well as language and behavior of SOD patient have been described as normal. Williams et al [7] studied a group with SOD and noted that an absent septum pellucidum was not associated with important neurological and intellectual deficiencies.

Approximately 2/3rd of SOD patients suffer from hypothalamic pituitary dysfunction .The most commonly affected hypothalamic pituitary hormone is growth hormone. Hypopituitarism may become apparent at any age. Although early onset is most common, absence of endocrine deficiency does not preclude development of endocrine deficiency in later [8].

Brako EK et al described a patient with lissencephaly, agenesis of corpus callosum and septo- optic dysplasia. This was unique CNS finding in a patient with proximal chromosome 14q deletion [9].Peripheral retinal non-perfusion associated with optic nerve hypoplasia and lissencephaly had also been reported [10].

The prognosis of SOD varies according to the presence and severity of symptoms. Neurological research which focus on identifying and studying the genes involved in normal brain development are required .The knowledge gained from these

fundamental studies provides the foundation for understanding how the process can go awry and thus may eventually gives clues to understanding disorders such as SOD.

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