

Letter to the Editor

Ocular manifestations of de-Morsiers syndrome

Sir,

A two-year old male child presented to the ophthalmology outpatient department (OPD) with parents complaining of child's inability to make an eye contact. On examination, the patient had horizontal manifest nystagmus with no fixing and following. The pupillary reactions and the anterior segment examination were normal. Fundus examination in both eyes revealed small optic disc with 360 degree hyperpigmented ring in both the eyes (Figure 1 A and B). The macula was >2 disc diameter (DD) in distance from optic disc, suggestive of optic nerve hypoplasia. Magnetic resonance imaging (MRI) brain showed absent septum pellucidum (Figure 1 C and D). Serum growth hormone levels were significantly high. Based on the examination and investigations, diagnosis of de Morsier's syndrome was made.

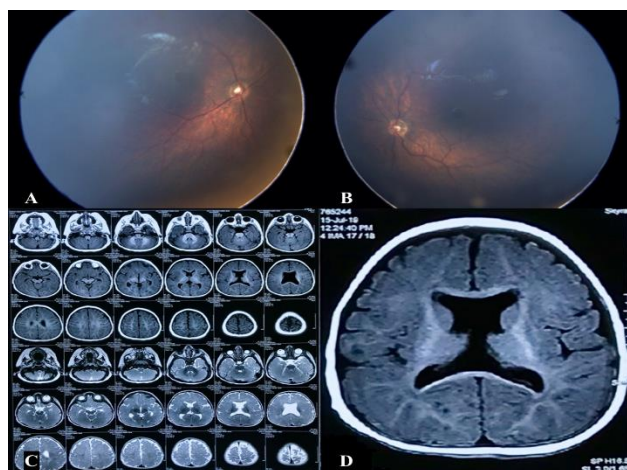


Figure 1: Retcam images showing (A) and (B) optic disc hypoplasia with double ring sign, and (C) and (D) MRI brain showing absent septum pellucidum.

DISCUSSION

De Morsier's syndrome, also known as septo-optic dysplasia is characterized by optic nerve hypoplasia, pituitary endocrine dysfunction and midline brain abnormalities as hypoplasia or absence of septum or corpus callosum or both.¹ Optic nerve hypoplasia (ONH) is one of the leading causes of childhood blindness worldwide.² ONH is a non-progressive congenital disc anomaly characterized by a small optic disc with a double-ring sign which may result from excessive apoptosis of retinal ganglion cells during visual pathway

development.^{3,4} Visual acuity may range from no light perception to normal vision. Associated ophthalmological features include nystagmus (26-86%), strabismus (42-52%), axial myopia, and astigmatism.⁵ Variable pituitary-hypothalamic dysfunction ranging from isolated deficit of pituitary hormones to pan-hypopituitarism is commonly seen.¹ The most prevalent findings on neuroimaging included absent septum pellucidum (53%), corpus callosum hypoplasia and abnormalities of the pituitary gland (13-34%), such as ectopic pituitary gland, empty sella, and infundibular hypoplasia.⁶

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