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Original Research Article

A Very Rare Case Report of Septo Optic Dysplasia with Incidental Association of Kallman's Syndrome

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Conflict of interest: Nil

Abstract:

Septo-optic dysplasia with Kallmann's syndrome is a rare condition which includes, visual abnormalities/vision loss, hypogonadotropic hypogonadism, anosmia/hyposmia. We report a case of 12-year old boy with complete vision loss and a poor sense of smell since childhood. Radiological imaging revealed bilateral hypoplastic optic nerves, chiasma and tracts, absent left olfactory bulb and olfactory gyrus, small pituitary gland. He was referred by his primary physician for evaluation.

Keywords: Optic nerves, Olfactory gyrus, Septum pellucidum, Pituitary gland.

Introduction

Septo-optic dysplasia, often known as de Morsier's syndrome [1]. SOD has a 1 in 50,000 incidence rate and is more prevalent in young moms and first-born children. Optic nerve hypoplasia and septum pellucidum absence are the primary pathogenic characteristics.

Septo-Optic Dysplasia Plus (SOD+) is the term used to describe SOD when it is accompanied by schizencephaly or callosal dysgenesis. Numerous suggestions have been put out as potential reasons, including vascular disruption during brain development, subsequent optic nerve fibre degeneration brought on by cerebral injuries, and midline heritable abnormalities. In addition to other brain malformations such as schizencephaly, callosal dysgenesis, and perisylvian cortical dysplasias, hypothalamic pituitary insufficiency affects 2/3 of SOD patients. Homeobox gene HESX1 mutation has also been discovered [2]. Olfactory pathway dysgenesis has not been linked to SOD, despite the discovery of forebrain anomalies. The absence of myelinated fibres in the optic nerve and chiasma, as well as the disorganised layering of tiny neurons, are microscopic characteristics of SOD. A child with small height, endocrine malfunction, vision loss, cerebral incapacity, microcephaly, or anosmia may present clinically.

Kallmann's syndrome is a rare genetic neuronal migration disorder of olfactory axons and gonadotropin releasing hormone producing neurons. Olfactory bulbs and sulci also grow improperly as a result of abnormal olfactory placode development. One in ten thousand men and one in fifty thousand women experience it. The majority of the cases are sporadic. Hyposmia/anosmia, delayed puberty, and low pituitary hormone levels of blood FSH, LH, and Testosterone are all symptoms of Kallmann's syndrome. Agenesis of the kidneys, CVS abnormalities, cryptorchidism, and fascial malformations are possible further related defects [3]. The best way to detect morphological abnormalities is through MRI. Along the cribriform plate, olfactory bulbs are shown in coronal planes. Between the medial orbital gyrus and the rectus gyrus, olfactory sulci are seen. It is preferable to use high resolution coronal fast spin echo T1W and T2W sequences [4]. Abnormal development of olfactory placode also results in improper development of olfactory bulbs and sulci. Hypoplasia of anterior pituitary could be secondary to it.

Case Report

A 12 years old boy presented to Ophthalmology and ENT OPD for complete vision loss and poor sense of smell since childhood. Detailed history was obtained. Parents were married consanguine-

ously. Delivery was normal without any complications, but our patient has an history of developmental delay. Clinical examination revealed short stature, small penis and obesity. Patient was then referred to Neurology OPD for cranial nerve examination and was advised for MRI evaluation of brain with orbits. MR Imaging findings revealed hypoplastic bilateral optic nerves, tracts, chiasma, absence of septum pellucidum, absence of one of the gyriand olfactory bulbon left side with normalmedial rectal and straight gyri, olfactory bulb on right side. Reduced size and volume of pituitary gland. Decreased anteroposterior diameter of skull. Patient's serum Testosterone, FSH, LH levels were low.

Discussion

The etiology of SOD with Kallmann's is not clearly defined. The development of anterior visual and olfactory pathways begins in 1st trimester of gestation. A single insult can cause both visual and olfactory impairment due to a neuronal migration abnormality. The diagnosis goes unnoticed till the teenage and the diagnosis is usually made when the child fails to attain puberty. MRI is the choice of investigation for making the diagnosis. Septo-optic dysplasia which is also called de Morsier's syndrome/septo-optic pituitary dysgenesis/suprasellar dysgenesis is characterized by absence of septum pellucidum and hypoplastic optic nerves, hormonal dysfunction secondary to hypothalamic-pituitary dysfunction.

Multiple etiologic agents such as vascular insults, viral infections, maternal age, and genetic mutations have been associated with the origin. Kallmann's is characterized by hyposmia/anosmia, hypogonadotropic hypogonadism [5].

There are less chances for the association of SOD with kallmann's syndrome, which was seen in our case. Depending upon the severity of associated brain and pituitary malformations, prognosis can be made. There is no definitive treatment. Patient counselling, hormone replacement therapy can be used. Imaging findings of our patient are as follows:

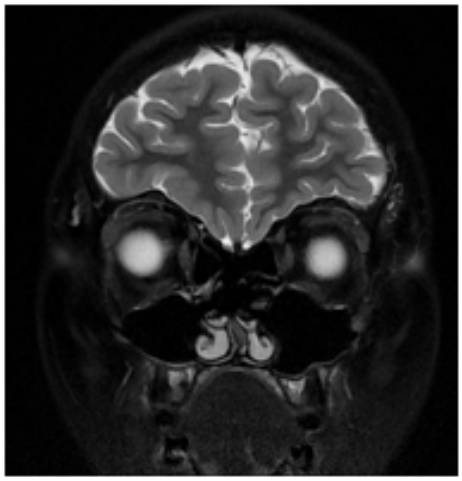


Figure 1: T2 Coronal image showing Absence of left olfactory bulb and one of the gyri on left side.

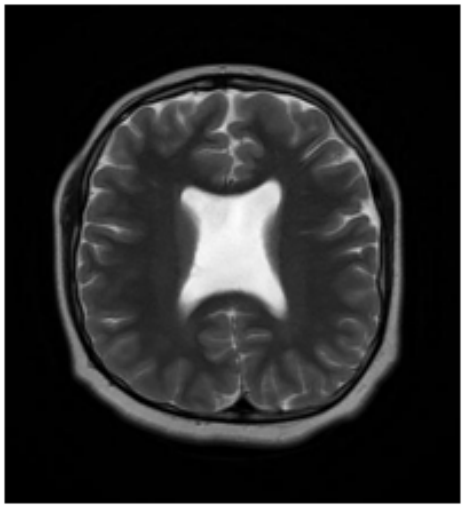
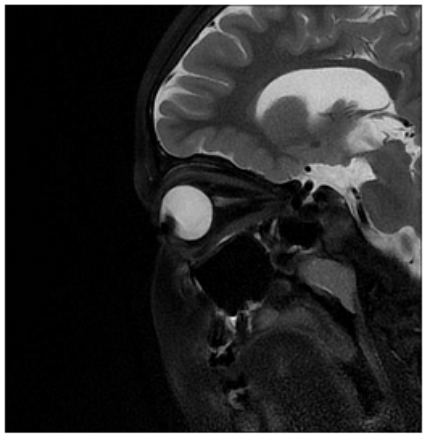
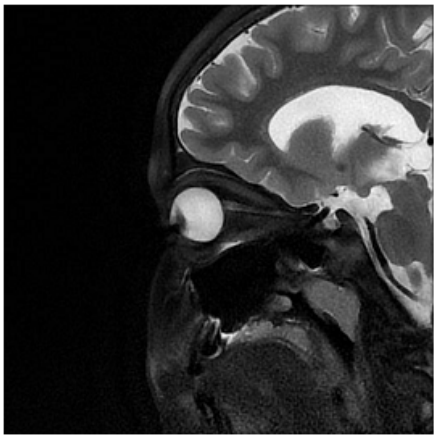


Figure 2: T2 Axial image showing Absence of septum pellucidum.



(Right)



(Left)

Figure 3 and 4: T2 Sag image showing hypoplastic right and left optic nerves.

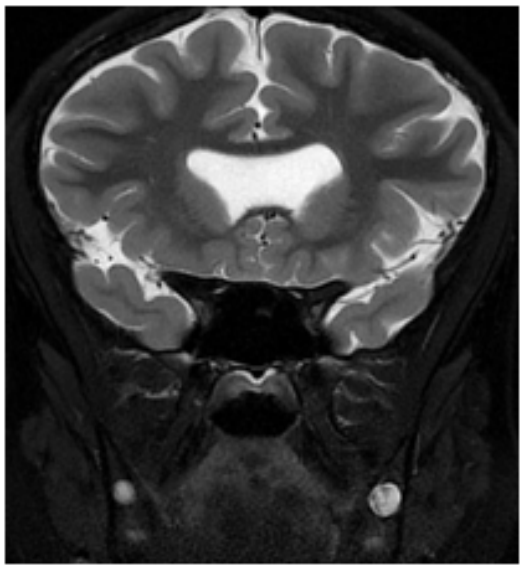


Figure 5: T2 coronal image showing point down appearance of the anterior horns of the bilateral lateral ventricles.

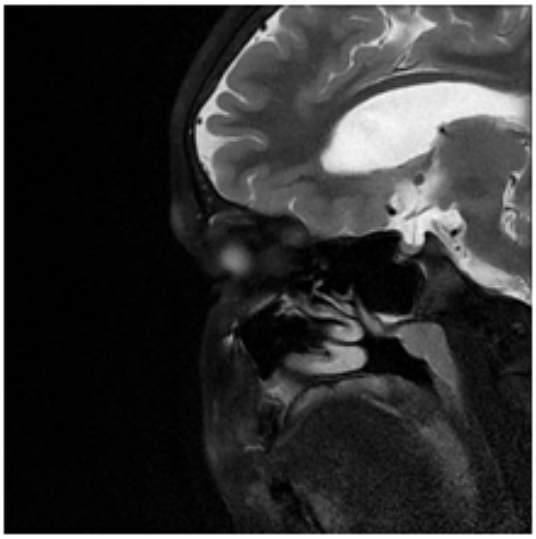


Figure 6: T2 Sag image showing reduced volume of pituitary gland.

Conclusion

Most of the patients present with Septo-optic dysplasia and Kallmann's syndrome individually but were less reported together in the literature, we are seeing both the diseases in our case which is a very rare phenomenon and confirms that both can occur together in one case. MRI is the gold standard diagnostic test to confirm the diagnosis.

Finding: Nil

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