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Magnetic resonance imaging in Kallmann syndrome: A case report

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Abstract

Kallmann syndrome is a rare genetically inherited condition characterised by hypogonadotrophic hypogonadism and anosmia or hyposmia. It is due to failure of migration of gonadotrophic releasing hormone neuron and olfactory neuron to hypothalamus. This case reports a 39-year-old Maldivian adult with clinical features of Kallmann syndrome and magnetic resonance imaging brain showing absence of olfactory sulcus and bulb.

Key words: Anosmia; Hypogonadotrophic hypogonadism; Kallmann syndrome; Magnetic resonance imaging; Olfactory bulb; Olfactory sulcus.

INTRODUCTION

Kallmann syndrome is a rare condition characterized by features of hypogonadotrophic hypogonadism and anosmia or hyposmia. It is caused due to defect in neuronal migration of olfactory neuron and gonadotrophin releasing hormone neuron. The prevalence of this condition is reported to be one in 10,000 males and one in 50,000 females.[1] In most cases it is diagnosed in the adolescent period as patients present with features of hypogonadism, although it is present since birth. Magnetic resonance imaging (MRI) of brain shows characteristic features of absence of olfactory sulcus and atrophy or absence of olfactory bulb, which adjunct in its diagnosis.

CASE REPORT

A 38-year-old Maldivian male presented to the fertility clinic with small size penis. On further evaluation, loss of smell was also present. There was no significant past medical or surgical history. Ultrasound evaluation showed no renal anomalies, however, small size bilateral testes were noted. His laboratory reports showed decreased level of sex hormones, follicle stimulating hormone (FSH) measuring 0.969 mIU/ml (Normal range: 1.6 to 9 mIU/ml), luteinizing hormone (LH) measuring <0.5 mIU/l (Normal range: 7 to 24 mIU/l) and testosterone measuring 0.69 nmol/L (Normal range: 4.5 to 28 nmol/L). The patient was then referred for MRI brain with contrast for assessment of any intracranial pathology, pituitary gland, olfactory sulcus and bulb particularly, in the view of Kallmann syndrome.

MR imaging of brain T2 coronal section at anterior cranial fossa showed absence of olfactory sulcus and absence of olfactory bulb (Figure 1). T2 sagittal image at the level of sella showed normal size sella with normal size of pituitary gland (Figure 2). Rest of the brain was normal. With reference to these characteristic imaging features of brain, hormonal assay and clinical features, the diagnosis of Kallmann syndrome was confirmed.

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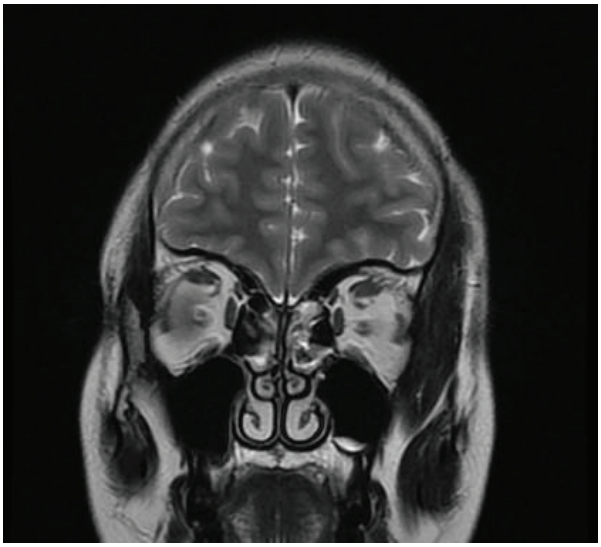

Figure 1: T2 Coronal section image at the level of cribiform plate showing absence of olfactory sulcus and bulb

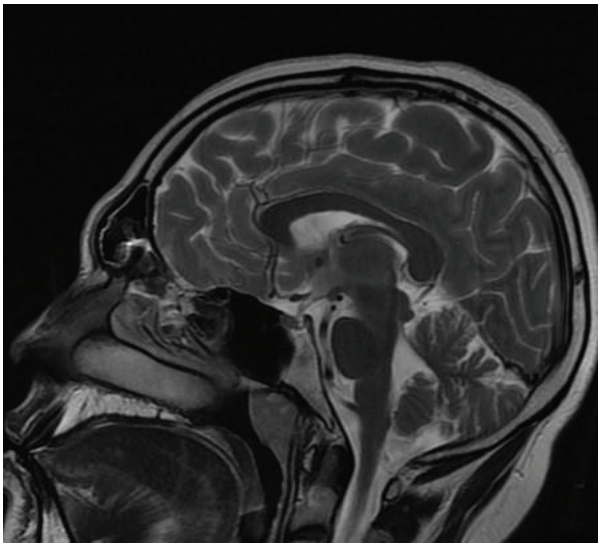

Figure 2: T2 Sagittal section image at the level of sella showing normal size pituitary gland with posterior lobe bright spot

DISCUSSION

Clinically, Kallmann syndrome is characterized by typical features of hypogonadotrophic hypogonadism, anosmia or hyposmia. When anosmia is absent, similar syndrome is referred to as normosmic idiopathic hypogonadotrophic hypogonadism (IHH). It results due to failure of migration of gonadotrophic releasing hormone (GnRH) and olfactory neuron from olfactory placode to the hypothalamus. Genetically, it shows heterogeneous inheritance patterns of X-linked and autosomal, X-linked being more common.[2]

Kallmann syndrome was first described by Maestre de San Juan in 1856 and later characterized as a hereditary condition by Frank Josef Kallmann in 1944.[3] It is associated with various anomalies like renal agenesis, cryptorchidism, midline facial defects, short fourth metacarpal and cardiovascular abnormalitis.[4] In this reported case, such associated anomalies were not noted.

MRI is the imaging of choice for evaluation of olfactory bulb and sulcus. T2 coronal section of anterior cranial fossa helps in evaluation of olfactory sulcus and bulb. In a study conducted by Zaghouani et al. in five patients with clinical features and family history of Kallmann syndrome, olfactory sulcus was absent in three and was hypoplastic in two patients.[3] Similar findings were seen in a study conducted by Madan et al.[4] Both studies are similar to the present case which shows absent olfactory sulcus and bulb. In the study conducted by Allu et al., there was hypoplastic olfactory sulci, which was not seen in this case.[1]

In the study conducted by Vogl et al. to identify morphological difference by MRI between Kallmann syndrome and IHH, they concluded that only Kallmann syndrome has anterior cranial fossa morphological abnormality.[5] Various clinical conditions are associated with hyposmia like Down syndrome, Turner syndrome and holoprosoncephaly. However, absence of olfactory sulcus or bulb was only seen in Kallmann syndrome.[6]

CONCLUSION

The diagnosis of Kallmann syndrome can be made in patients presenting with clinical features of hypogonadotrophic hypogonadism, anosmia with laboratory investigations showing decreased level of sex hormones and characteristic MRI brain findings of absent or hypoplastic olfactory sulcus and bulb.

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