

Magnetic Resonance Imaging Findings of a Patient with Kallmann Syndrome

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Abstract: We present a case of Kallmann syndrome in a 33-years-old man. He was unable to sense smell since birth. He showed scale-out results on T&T olfactogram. He displayed non-development of secondary sexual characteristics for the past 20 years. On urological examination, he had sparse pubic hair, right undescended testis, and small penis. Blood investigations showed low basal levels of serum testosterone, follicle stimulating hormone and luteinizing hormone. Brain magnetic resonance imaging (MRI) detected absence of the olfactory bulbs, tracts, and hypoplasia of the olfactory sulci. An apparently normal appearing pituitary gland was detected. MRI findings were useful for diagnosing and confirming Kallmann syndrome.

Keywords: Kallmann syndrome, Anosmia, Hypogonadotropic hypogonadism, Magnetic resonance imaging, Olfactory bulb, Olfactory sulcus.

INTRODUCTION

Idiopathic hypogonadotropic hypogonadism combined with complete or incomplete olfaction disturbance has been termed Kallmann syndrome [1-6]. Kallmann syndrome is due to abnormal migration of gonadotropin-releasing hormone (GnRH) and olfactory neurons [1-6]. Abnormal development of the olfactory placode leads to improper development of the olfactory bulb and olfactory sulcus. Recently, several reports have been published concerning the possibility of diagnosing morphological abnormalities of the olfactory bulbs, tracts, and sulci by using magnetic resonance imaging (MRI) to examine Kallmann syndrome patients [1-9]. MRI findings in Kallmann syndrome include absent or hypoplastic olfactory bulbs and olfactory sulci [1-9]. Herein, we describe the MRI findings of a patient with Kallmann syndrome.

CASE REPORT

A 33-years-old Japanese man was referred to Jichi Medical University Hospital with an olfactory complaint. He was unable to experience a sense of smell since birth. He had surgical history of orchiopexy for right cryptorchidism at the age of 6. Examination by T&T olfactogram revealed total anosmia. Otolaryngological endoscopic examination was unremarkable in both nasal cavities. He suffered non-development of secondary sexual characteristics resulting in an underdeveloped penis for the past 20 years. His family histories were unremarkable. On

physical examination, his weight was 52 Kg, height was 170 cm. He has high pitched voice. On urological examination, he had sparse pubic hair, right undescended testis, and small penis.

His blood investigations revealed very low serum testosterone, follicle stimulating hormones (FSH), and luteinizing hormones (LH) (testosterone 0.27 ng/mL, FSH 1.1 mU/mL, LH < 0.2 mU/mL; normal range 1.3 ~ 8.71, 2.1 ~ 18.6, 1.7 ~ 11.2, respectively). On a GnRH challenge test, there was a slight increase in FSH and LH (FSH 4.4 mU/mL, LH 2.4 mU/mL). After a human chorionic gonadotropin (HCG) challenge test, his testosterone level mild increased to 3.2 times that of his basal level. Karyotyping was normal male (46XY). Thus, these above hormonal assay results revealed hypogonadotropic hypogonadism in this patient.

The coronal T2-weighted MRI showed absence of the olfactory bulbs, grooves (Fig. 1A and B arrows), and decreased olfactory sulci (Fig. 1C and D arrows) on both sides. An apparently normal pituitary gland was detected.

Hence, a diagnosis of Kallmann syndrome was given. The patient is now on treatment with intramuscular injection of testosterone enanthate 125 mg once a month.

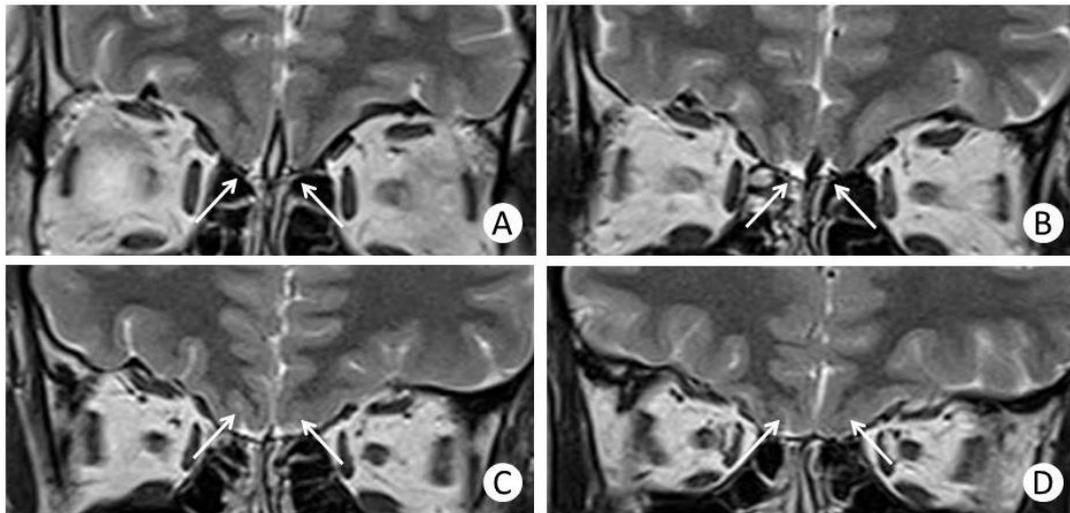


Fig. 1: Coronal T2-weighted MRI shows absence of the olfactory bulbs (A, B) and decreased olfactory sulci (C, D)

DISCUSSION

Kallmann syndrome is a rare genetic disorder with an estimated prevalence of 1 in 10,000 males and 1 in 50,000 females [1, 7, 8]. Clinical diagnosis of Kallmann syndrome in adults is dependent on the coexistence of anosmia with signs of hypogonadotropic hypogonadism. The main biochemical parameters in men are low serum testosterone and low levels of LH and FSH, and in women low serum estrogen and low levels of LH and FSH [6-9]. When the GnRH and HCG challenge tests are performed, there is an increase in serum FSH, LH values and testosterone values, respectively, which gives the biochemical diagnosis of Kallmann syndrome [6-9].

Recently, several reports have been published concerning the possibility of diagnosing morphological abnormalities of the olfactory bulbs, tracts, and sulci by using MRI to examine Kallmann syndrome patients [1-9]. MRI findings in Kallmann syndrome include absent or hypoplastic olfactory bulbs and olfactory sulci [1-9]. Koenigkam-Santos *et al.* [1] evaluated the MRI findings of 21 patients with Kallmann syndrome patients. According to their report, macroscopic findings on MRI are as follows: (1) 18 patients (85%) presented altered rhinencephalon structures, (2) 16 patients (76%) presented olfactory bulb aplasia (14/16 bilaterally), and these patients presented a total of 16 aplastic sulci. In addition, Jagtap *et al.* [2] evaluated the MRI findings of 25 patients with Kallmann syndrome patients compared with 16 patients with normosmic hypogonadotropic hypogonadism. According to their report, MRI revealed abnormalities of the olfactory apparatus in 68% of Kallmann syndrome and 37.5% of normosmic hypogonadotropic hypogonadism patients. Moreover, the MRI abnormalities of the olfactory apparatus in

Kallmann syndrome patients were aplasia (56%) and hypoplasia (44%).

Similarly, our patient showed absence of the olfactory bulbs, tracts, and hypoplasia of the olfactory sulci.

CONCLUSION

Although this case lacks novel findings and genetic screening was not available, we emphasize that MRI findings are useful for diagnosing and confirming Kallmann syndrome.

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