

Olfactory Sulcus Hypoplasia Images of a Case With Kallmann Syndrome

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Abstract

Kallmann syndrome is a form of hypogonadotropic hypogonadism accompanied by anosmia or hyposmia. MR images of midline defects in Kallmann syndrome were rarely seen in the literature. We wanted to put emphasis on the midline developmental defects accompanied to Kallmann syndrome shown by MR images of olfactory sulcus hypoplasia.

Keywords: Olfactory sulcus hypoplasia; Kallmann syndrome; Hypogonadotropic hypogonadism

Introduction

Kallmann syndrome is a form of hypogonadotropic hypogonadism accompanied by anosmia or hyposmia. It is transmitted genetically as autosomal dominant, autosomal recessive and X-linked. Several gene mutations are identified. Herein, we present our case of Kallmann syndrome.

Case Report

A 22-year-old male patient was admitted to the outpatient clinic with the complaint of absence of facial hair. He also had complaints of erectile dysfunction, decrease in libido and disability to smell. Physical examination results were as follows: height: 177 cm, weight: 60.5 kg, BMI: 19.15 kg/m², arm length: 187 cm, and penile length: 3 - 4 cm. Right testicular volume was 1

cc, and left testicular volume was 0.6 cc. His epiphyseal plates were currently open and his bone age was 15 years. Laboratory examination results were as follows: FSH: 0.27 mIU/mL, LH: 0.1 mIU/mL, t. testosterone: 0.25 ng/dL, and TSH: 1.63 μ IU/mL. Kallmann syndrome was diagnosed with regard to hypogonadism together with anosmia. KAL1 gene mutation was not detected. On craniofacial MRI, left olfactory sulcus was significantly shallow and gyrus rectus was hypoplastic. Bilateral olfactory sulci in a case without Kallmann syndrome were thicker (Fig. 1) compared to those of our case with the syndrome (Fig. 2). Human chorionic gonadotrophin was given for 6 months; after that, testosterone hormone replacement was started. His symptoms were relieved with hormone replacement.

Discussion

Co-occurrence of hypogonadotropic hypogonadism with anosmia was first defined in 1944 by Kallmann and his colleagues. Prevalence of Kallmann syndrome was 1/10,000 in females



Figure 1. Coronal T2 image of a subject without Kallmann syndrome: bilateral olfactory sulci (light arrows), olfactory groove (circle), and olfactory bulbi inside olfactory groove (arrow).

Manuscript accepted for publication June 24, 2015

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doi: <http://dx.doi.org/10.14740/jem279w>

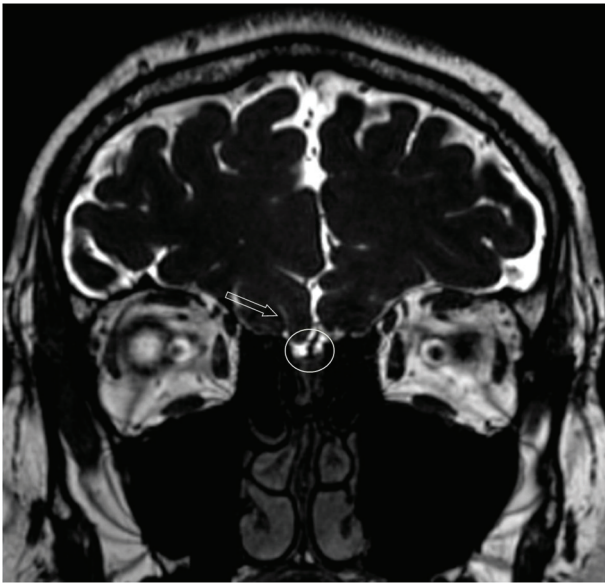


Figure 2. Coronal T2 image of the case with Kallmann syndrome. Normal olfactory sulcus was at the right (light arrow), olfactory sulcus was not seen on the left. Olfactory grooves were narrower on the left (circle), and olfactory bulb were thinner compared to the MRI of the subject without Kallmann syndrome.

and 1/15,000 in males [1, 2]. Association of hypogonadism with anosmia is due to developmental defects in rhinencephalon. In addition to KAL-1 gene mutation, FGFR-1, FGF8, prokineticin (PROK2) and its receptor (PROK2R) mutations were also defined. MR images of midline defects in Kallmann syndrome were rarely seen in the literature. We wanted to put emphasis on the midline developmental defects accompanied to Kallmann syndrome shown by MR images of olfactory sulcus hypoplasia.

Conflict of Interest

The authors have no conflict of interest.

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