Olfactory Sulcus Hypoplasia Images in a Case of Kallmann Syndrome

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

Case: A twenty-two-year-old male patient was admitted to the outpatient clinic with the complaint of absence of facial hair. He had also complaints of erectile dysfunction, decrease in libido, and loss of smell. Physical examination results were as follows: height 177 cm, weight 60.5 kg, body mass index 19.15 kg/m^2, arm length 187 cm, and penile length 3-4 cm. Right testicular volume was 1cc, left testicular volume was 0.6cc. His epiphyseal plates were currently open and his bone age was 15 years. Laboratory examination results revealed: follicle-stimulating hormone 0.27 mIU/mL, luteinizing hormone 0.1 mIU/mL, total testosterone 0.25 ng/dL, thyroid-stimulating hormone 1.63 µIU/mL. Kallman syndrome was diagnosed as regarding to hypogonadism together with anosmia. *KAL1* gene mutation was not detected. On craniofacial magnetic resonance imaging (MRI), left olfactory sulcus was significantly shallow and gyrus rectus was hypoplastic. Olfactory sulci bilaterally were thinner . Human chorionic gonadotropin 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 and anosmia was first defined in 1944 by Kallmann and his colleagues. Prevalence of Kallmann syndrome was 1/10 000 in females and 1/15 000 in males. Association of hypogonadism with anosmia is due to developmental defects in rhinencephalon. In addition to *KAL1* gene mutation, *FGFR-1, FGF*8, prokineticin (*PROK2*) and its receptor (*PROK2R*) mutations were also defined. MRI of midline defects in Kallmann syndrome were rarely seen in the literature. We wanted to put emphasis on the midline developmental defects accompanying Kallmann syndrome shown by MRI as olfactory sulcus hypoplasia.

Key words: Kallmann syndrome, hypogonadotropic hypogonadism, anosmia, MRI of midline defects, olfactory sulcus hypoplasia