KALLMANN SYNDROME: A CASE REPORT OF MRI FINDINGS.

Orasa CHAWALPARIT, Suthisuk SUTHIPONGCHAI

ABSTRACT

The MRI findings of a case of Kallmann syndrome was reported. A 16 years-old female patient came with a history of primary amenorrhea and hyposmia. The hormonal studies were shown as hypogonadotropic hypogonadism. The MRI of olfactory region reveals absent of olfactory bulb and hypoplasia of the olfactory sulci. The imaging was shown and a review of literatures was presented.

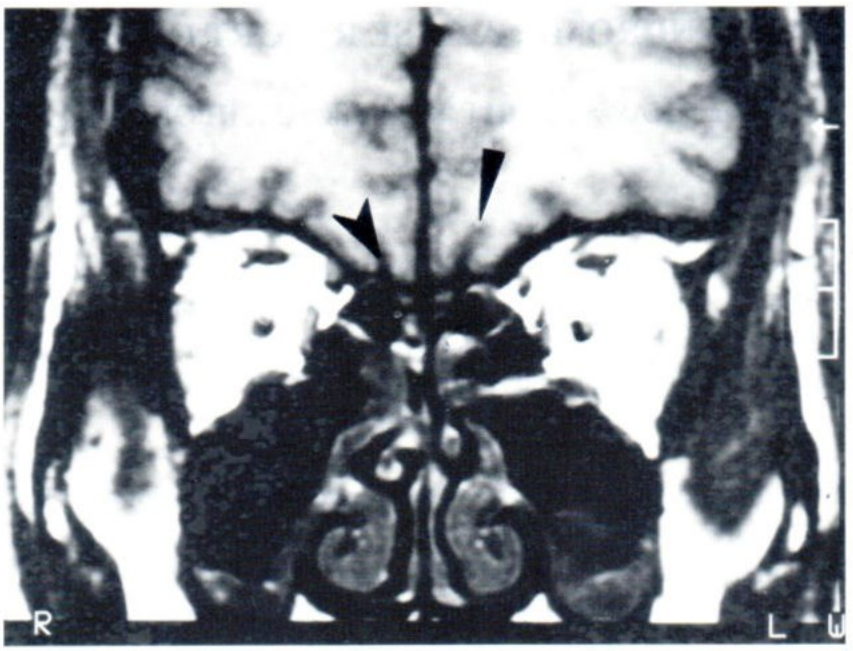
Kallmann syndrome is a form of congenital hypogonadotropic hypogonadism with anosmia or hyposmia. The prevalence has been estimated to be one in 10,000 males and one in 50,000 females.[2] The MR imagings have been reported with absent or hypoplasia of the olfactory bulb and tract as well as hypoplasia of olfactory sulci. We reported one case of MR imaging with clinical suspected to be this disease in a girl, which is a rarer prevalence in the sex-distribution.

A CASE REPORT

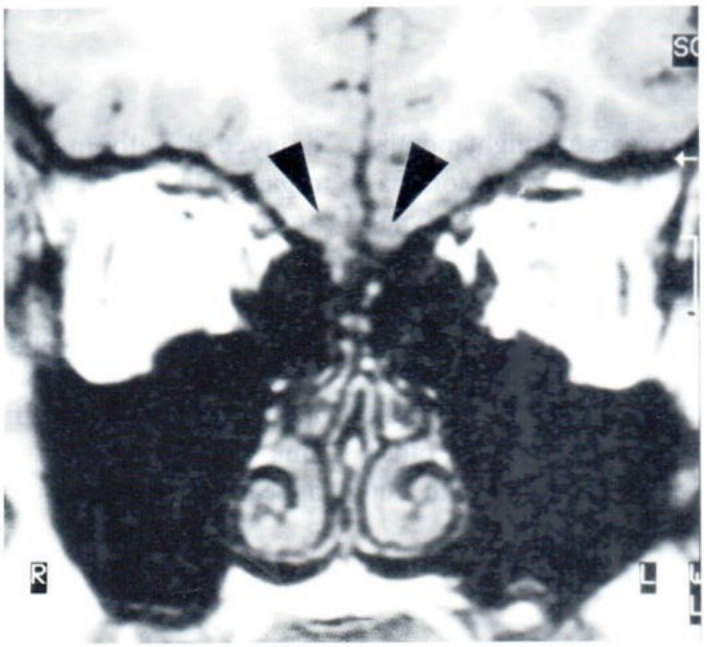
A Thai girl, sixteen years and eight months old came to the hospital with primary amenorrhea. At fifteen years old,she was evaluated by a gynecologist for primary amenorrhea. The serum FSH, LH and E2 were abnormally low and the chromosome study was 46XX. She was referred to the pediatric endocrinologist for further investigation and has been found that she has hyposmia. She was able to smell coffee but not perfume. The other physical examinations were normal growth but delayed development of the external genital organs. The LHRH stimulation test showed to be an early pubertal response. The other hormonal studies were normal.

The MR imagings of brain revealed hypoplasia of the olfactory sulci. The olfactory tract is indistinguishably identified (Fig.1). The pneumosinus dilatans was also shown. The pituitary gland appears normal.

Department of Radiology, Siriraj Hospital, Faculty of Medicine, Mahidol University, Bangkok, Thailand.



1A



1B

Fig.1. Coronal MRI. of frontal region in A: normal and B: Kallmann syndrome. The arrow indicates the olfactory bulb/nerve and the olfactory groove in the normal case. In B, the olfactory nerve cannot be well identified and the groove is hypoplasia.

DISCUSSION

In 1944, Kallmann[3] described the disease characterized by hypogonadotropic hypogonadism with hyposmia. It was postulated that a failure of neuronal migration from the olfactory placodes results in both olfactory aplasia or hypoplasia and faulty development of the hypothalamic-pituitary axis for reproductive hormones.[5] Schwanzel-Fukuda et al have confirmed (by immunohistochemical method) such a failed neuronal migration. They demonstrated that the terminalis and possibly the vomerunasal nerves act as the embryonic scaffolding for the LHRH-releasing cells to migrate into the forebrain to the medial septal nucleus and hypothalamus in normal development. In Kallmann syndrome, the olfactory, terminalis and vomerunasal nerves never reach the brain and the olfactory bulbs are not induced. The LHRH-releasing cells are also arrested and are found in the dural layers of the meninges beneath the forebrain. The arrested LHRH-releasing cells result in the inability of the hypothalamus to control adenohypophyseal synthesis and release of FSH and LH. Clinically, the patients have anosmia and hypogonadism, which is reversible.

Transmission of the Kallmann syndrome can occur according to the autosomal dominant, autosomal recessive or X-linked patterns.[1] It is most commonly found in men. Occasional cases in women have been reported, as in our patient. At least in the X-linked cases, recently the KALIG-1 gene has been identified.

Many reports have shown the efficacy of the MRI in demonstrating the olfactory bulb and tract.[4,6] The coronal MRI seems to be the best modality to identify the nerve. In Kallmann syndrome, the olfactory nerve may be hypoplasia or absent. The associated hypoplasia of the olfactory groove of the frontal lobe has also been demonstrated. Truwit et al[6] demonstrated patients with abnormal soft tissue presented in the region between the upper nasal vault and the forebrain. They suspected the possibility of the tissue to represent the dysplastic "tangle" noted by Schwanzel-Fukuda.

In conclusion,we have reported a case of female Kallmann syndrome with MRI study.

REFERENCES

1. Franco B., Guioli S, Pragliola A, et al. A gene deleted in Kallmann syndrome shares homology with neural cell adhesion and axonal path-finding molecules. Nature 1991;353:529-536.
2. Jones JR, Kemmann E. Olfactogenital dysplasia in the female. Obstet Gynecol Annu 1976;5:443-466.
3. Kallmann FJ, Schoenfeld WA, Barrera SE. The genetic aspects of primary eunuchoidism. Am J Ment Defic 1944;48:203-206.
4. Klingmuller D, Dewes W, Krahe T, et al Magnetic resonance imaging of the brain in patients with anosmia and hypothalamic hypogonadism (Kallmann syndrome). J Clini Endocrinol Metab 1987;65:581-584.
5. Schwanzel-Fukuda M, Bick D, Pfaff DW, Luteinizing hormone-releasing hormone (LHRH) expressing cells do not migrate normally in an inherited hypogonadal (Kallmann) syndrome. Mol Brain Res 1989;6:311-326.
6. Truwit CL, Barkovich AJ, Melvin M, et al. MR imaging of Kallmann syndrome, a genetic disorder of neuronal migration affecting the olfactory and genital systems. AJNR 1993;14:827-838.