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RESEARCH ARTICLE

AN INTERESTING PRESENTATION OF HYPOGONADOTROPIC HYPOGONADISM -KALLMANN SYNDROME

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ABSTRACT

Kallmann syndrome is a rare sporadic and inherited chromosomal disorder, resulting in olfactogenital dysplasia. Embryonic olfactory epithelium gives rise to cells that differentiate into GnRH secreting neurons. These GnRH neurons which originate in the olfactory placode (i.e. the early developing nose); then migrate along the fetal olfactory neurons that also originate in the nose; and eventually enter the brain ultimately finding their way to the hypothalamus. In KS, there is Failed neuronal migration from olfactory placode to the hypothalamus. An 18-year-old male presented to the OPD with complaints of not attaining secondary sexual characteristics along with absence of sense of smell. Further investigations (USG, hormone panel, MRI brain) were performed and he was diagnosed to have kallmann syndrome and treatment with monthly intramuscular testosterone was started.

INTRODUCTION

Kallmann syndrome is a sporadic and inherited chromosomal disorder, resulting in olfactogenital dysplasia. It is a congenital form of hypogonadotropic hypogonadism (HH) that manifests with hypo- or anosmia. Embryonic olfactory epithelium gives rise to cells that differentiate into GnRH secreting neurons. These GnRH neurons which originate in the olfactory placode (i.e. the early developing nose); then migrate along the fetal olfactory neurons that also originate in the nose; and eventually enter the brain ultimately wending their way to the hypothalamus, their ultimate residence during early gestation. In KS, there is Failed neuronal migration from olfactory placode to the hypothalamus. Abnormal migration of olfactory neurons and gonadotropin-releasing hormone producing neurons causes GnRH deficiency which results in decreased levels of sex steroids leading to a lack of sexual maturity and the absence of secondary sexual characteristic and variable degrees of loss of smell. The cause of this condition is genetic but can be the result of a multitude of different genetic mutations.

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It can be sporadic (majority) as well as X linked, autosomal recessive and autosomal dominant. Kal1 gene was the first mutated gene identified on the X chromosome in many inherited forms. Kal1 gene encodes for anosmin-1, a protein with neuronal cell adhesion properties, involved in neuronal migration and outgrowth of axons. Speculated failure of anosmin-1 to direct olfactory nerves and GnRH producing neurons during development. Mutations in other genes identified involved in neuronal migration are - fibroblast growth receptor 1 (FGFR1), prokineticin-2 (PROK2) and prokineticin receptor-2(PROK2R).

CASE REPORT

An 18-year-old male presented to the OPD with complaints of not attaining secondary sexual characteristics along with absence of sense of smell. On examination – patient is well built with no body / facial and pubic hair; Small sized phallus; Small sized testis; Anosmia present. Serum testosterone -<2.5 ng/dl; FSH- 1.25mIU/ml; LH- 0.75mIU/ml. TFT- WNL; USG-B/L small sized testis(1.5cc) MRI brain revealed – B/L absent olfactory bulb with normal pituitary gland and sella. Along with the endocrinologist, a decision to start Inj.

TESTOSTERONE 100mg deep IM /monthly was taken. The patient is currently on the sixth month of treatment. There is visible development of facial and pubic hair with increase in size of the phallus.

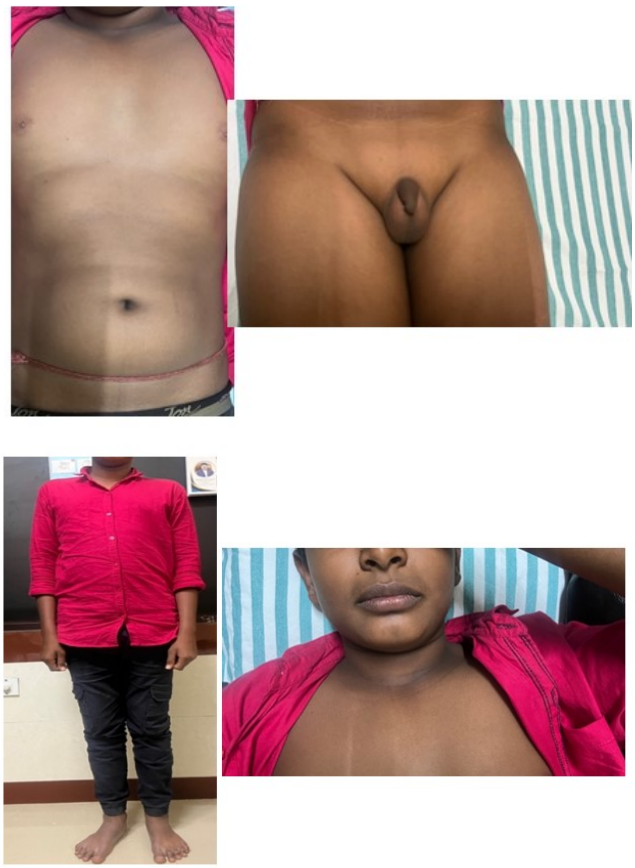

Figure 1 4. pre treatment images

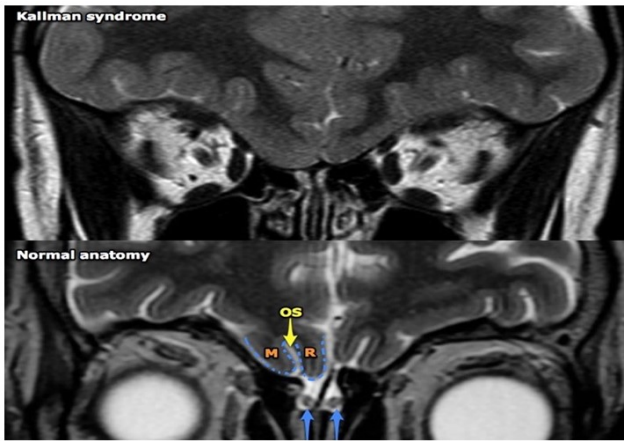

Figure 5. MRI brain

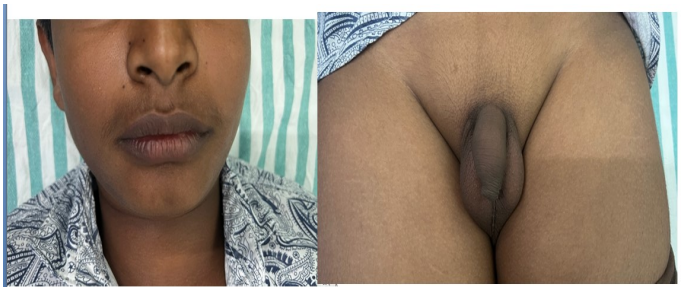

Figure 6 7. (with) 6 months treatment images

DISCUSSION

Typical diagnosis of KS occurs when a child fails to attain puberty(lack of testicular development and amenorrhea) associated with anosmia. These signs can include a lack of testicular development determined by testicular volume in men and amenorrhea in women. Other tests to be done include a Gene testing for mutations; MRI and hormone panel. Once the diagnosis is made, in both sexes, treatment is aimed at inducing puberty and maintaining normal hormonal levels. Testosterone in males and oestrogen /progesterone in females is given to induce puberty and maintain hormone levels and when fertility is desired hCG and hMG or recombinant FSH [rFSH]) can be given in males and gonadotropins or pulsatile GnRH therapy in females.

Currently there is no treatment for anosmia

CONCLUSION

KS is a rare presentation of hypogonadotropic hypogonadism with anosmia. Diagnosis depends on clinical examination, USG scrotum, MRI Brain and hormone panel. Treatment is by replacing testosterone/ oestrogen and progesterone for attaining secondary sexual characteristics and changing to gonadotropins for spermatogenesis/ folliculogenesis.

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