Kallmann Syndrome and Chronic Myeloid Leukemia: A Rare Occurrence

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A 31 year unmarried male presented with loss of appetite, malaise, left upper quadrant abdominal discomfort since 3 months. Patient was found to have massive splenomegaly. Patient also had gynecomastia, lack of facial hair and sparse axillary and pubic hair (Figure 1A, 1B). He gave history of absent puberty and lack of development of secondary sexual characters. On genital examination, patient had micropenis (3 cm), poorly developed scrotal sac, bilateral undescended testes with left testis palpable at base of scrotum (Figure 2).

Endocrine work-up revealed isolated hypogonadotropic hypogonadism (FSH-1.1 mIU, LH-0.08 mIU, Serum testosterone not detectable, T3, T4, TSH, cortisol and prolactin levels normal). Patient also had impaired olfactory sensation and two of his maternal uncles had history of similar complaints, suggestive of X-linked transmission. MRI Brain showed diminished character of olfactory bulbs and olfactory sulcus. Gyrus rectus and medial orbital gyrus formed a single gyrus (Figure 3). All these findings were consistent with the diagnosis of Kallmann syndrome.¹² Work-up for splenomegaly led to the diagnosis of Philadelphia chromosome [t(9;22) i.e. BCR and ABL fusion] positive Chronic myeloid leukemia in chronic phase.

There is no case report of Kallmann syndrome with CML till date and no association has been established. Philadelphia negative CML patients have other tranlocations [t(8;22) i.e. BCR and FGFR1 fusion].³⁴ Loss-of-function mutation of the gene encoding FGFR1 has been described in autosomal dominant KS.³ However, our patient had X-linked Kallmann syndrome not related to CML.

References


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