Kallmann's syndrome: a visual vignette

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DESCRIPTION

Kallmann's syndrome is a rare form of idiopathic hypogonadotropic hypogonadism; impaired sense of smell and absent olfactory bulb are the hallmarks of this disease.[1 2] A 20-year-old man presented to us for evaluation of symptoms of a small-sized penis and testis, and poorly developed secondary sexual characteristics. His birth history revealed that he had been born with a cleft lip and cleft palate. The cleft lip was surgically corrected when he was 4 years of age. His current clinical examination revealed the presence of a surgically corrected cleft lip, and a cleft palate that had not received surgical correction (figure 1), with evidence of hyposmia. He had eunuchoidal body proportions with an arm span and height difference of 15 cm and an upper segment to lower segment ratio of 74/88 cm (figure 2). He had a stretched penile length of 5 cm and testicular volume was 2 mL bilaterally, with a Tanner stage 3 pubic hair level (figure 3). Laboratory tests showed serum luteinising hormone <0.10 mIU/mL and follicle-stimulating hormone 0.325 mIU/mL with early morning serum testosterone of <20.0 ng/dL. The other hormonal axes were normal. MRI of the brain revealed a normal-sized pituitary gland with an absent olfactory bulb bilaterally (figure 4). The patient's clinical and biochemical features of idiopathic hyposmic hypogonadotropic hypogonadism was suggestive of Kallmann's syndrome. Defective migration of

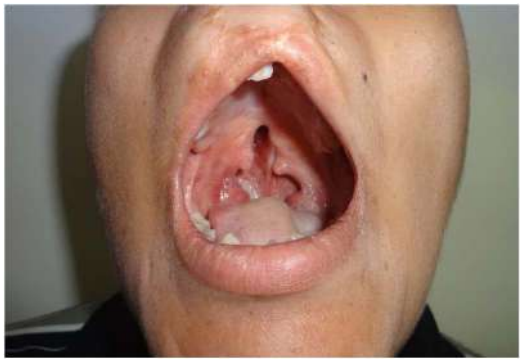

Figure 1 Picture of the patient showing the presence of a cleft palate.

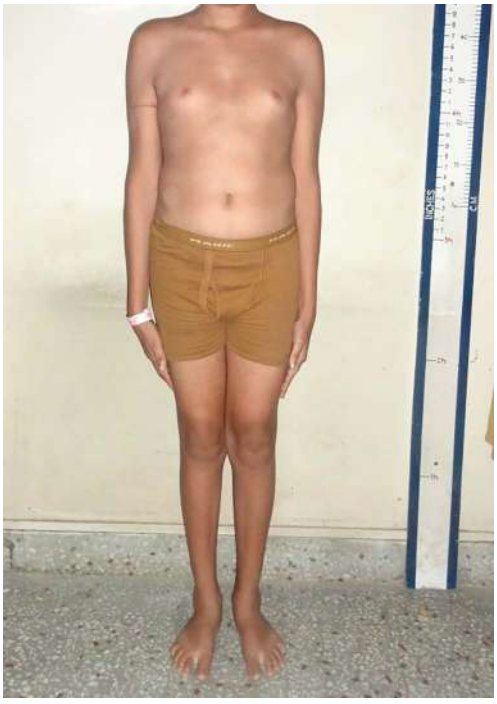

Figure 2 Picture of the patient showing a tall eunuchoid appearance without other skeletal abnormalities.

Figure 3 Picture of the external genitalia showing poorly developed pubic hair (Tanner's stage 3), small penis (5 cm) and bilaterally palpable testis (2 mL measured by Prader's orchidometer).

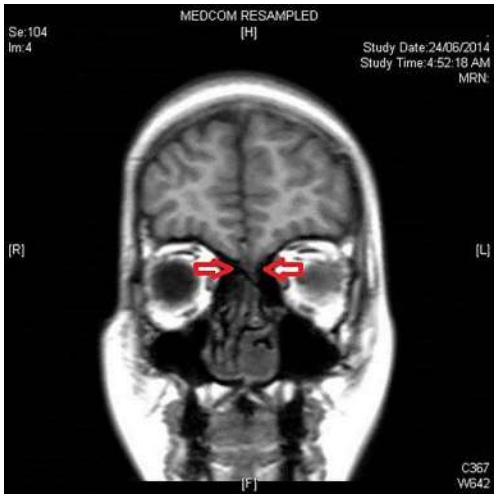

Figure 4 T1-weighted MRI (contrast enhanced) of the brain showing evidence of bilateral absence of olfactory bulbs (shown with arrows).

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gonadotropin-releasing hormone (GnRH) neurons along with olfactory neurons results in an agenesis of olfactory bulb and gonadotropin deficiency.[3] The patient was initially started on 100 mg testosterone injections once every 3 weeks, with a plan to gradually escalate the dose to 250 mg.

Learning points

- A cleft lip and cleft palate at birth are definite signs of Kallmann's syndrome.
- Impaired sense of smell distinguishes Kallmann's syndrome from most other forms of hypogonadotropic hypogonadism.

Contributors FKJ saw the patient and wrote the manuscript. RD saw the patient and helped write the manuscript. NT helped correct the manuscript.

Competing interests None declared.

Patient consent Obtained.

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