International Journal of Innovative Research in Medical Science (IJIRMS)
Volume 09, Issue 05, May 2024,
https://doi.org/10.23958/ijirms/vol09-i05/1894

Images in Clinical Medicine

Kallmann Syndrome: A Late Diagnosis

Ricardo Antunes Pereira [*1], Hugo Jorge Casimiro [1,2], Joana Carreira [1], Pedro L. De Freitas [3], Pedro Carreira [3], Susana Marques [3]

[1]Internal Medicine Department, Unidade Local de Saúde da Arrábida, Setúbal, Portugal.
[2]Hospital Palliative Care Team, Unidade Local de Saúde da Arrábida, Setúbal, Portugal.
[3]Internal Medicine Intermediate Care Unit, Unidade Local de Saúde da Arrábida, Setúbal, Portugal.

*Corresponding author: Ricardo Antunes Pereira; *ricardoapereira4@gmail.com*

Received 28 April 2024; Accepted 20 May 2024; Published 22 May 2024

Case description

We present the case of a 44-year-old man with no relevant known prior clinical history, who was initially admitted to an Internal Medicine ward with the diagnosis of intermediate-risk pulmonary embolism. On physical examination, several abnormal features were noted, such as the lack of facial and body hair, severe gynecomastia, obesity, decreased muscle mass, a penile length of 4 cm, right-sided cryptorchidism, and an upper to lower body ratio < 1 (**Figure 1**). Upon further inquiry, the patient reported having decreased libido and erectile dysfunction since puberty and anosmia since childhood. To better clarify those clinical findings, serum hormone levels were quantified, showing low levels of follicle-stimulating hormone (<0.3 mIU/ml; reference range 1.3 mIU/ml – 11.8 mIU/ml), luteinizing hormone (0.3 mIU/ml; reference range 2.8 mIU/ml – 6.8 mIU/ml), and testosterone (0.09 ng/ml; reference range 1.61 ng/ml – 11.33 ng/ml). No other hormone abnormalities were identified. A karyotype was then obtained, which was considered normal (46, XY). Since the clinical and laboratory findings were highly suggestive of Kallmann syndrome, a brain MRI was performed. The exam showed olfactory bulb hypoplasia, confirming the diagnosis (Figure 2). The patient was later referred to an Endocrinology appointment for follow-up.

Kallmann syndrome is a rare genetic disease characterized by congenital hypogonadotropic hypogonadism, along with anosmia or severe hyposmia, due to hypoplasia or aplasia of olfactory bulbs [1,2]. It has an estimated incidence of 1 in 30,000 male individuals [3]. Given the clinical features of hypogonadism, it is usually diagnosed during puberty, when a lack of sexual maturation occurs [4]. In this case, however, the patient did not seek medical care during that period, delaying the diagnosis to adulthood during a hospital admission for an unrelated disease. Since early diagnosis is of utmost importance, so the patient can start appropriate treatment (such as hormone replacement therapy) in order to live an otherwise normal life, a high clinical suspicion is vital for the correct identification of these patients.

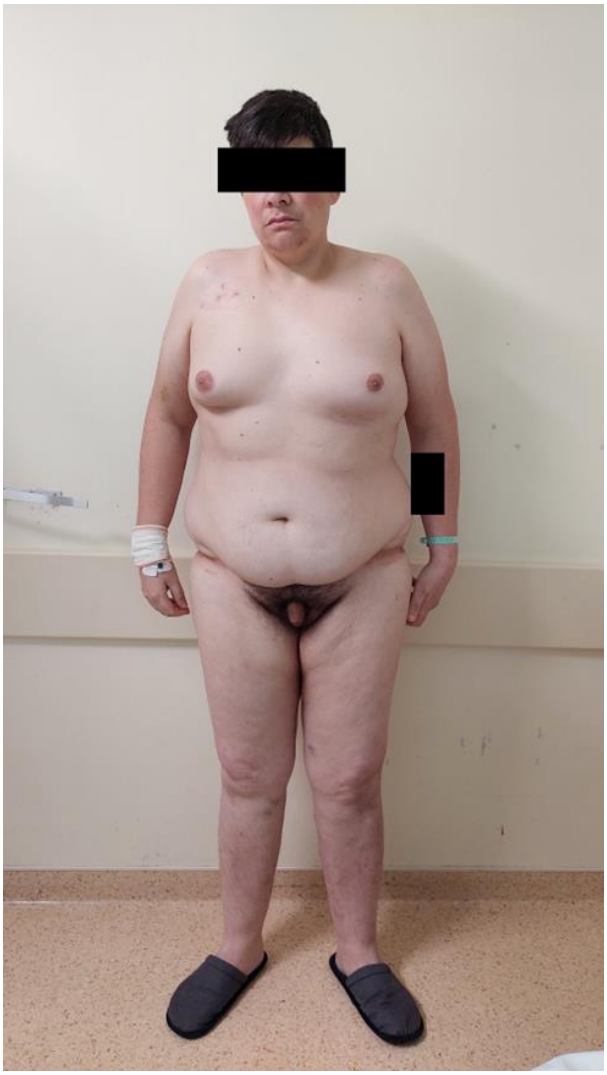

Figure 1: Picture of the patient showing clinical features of hypogonadotropic hypogonadism.

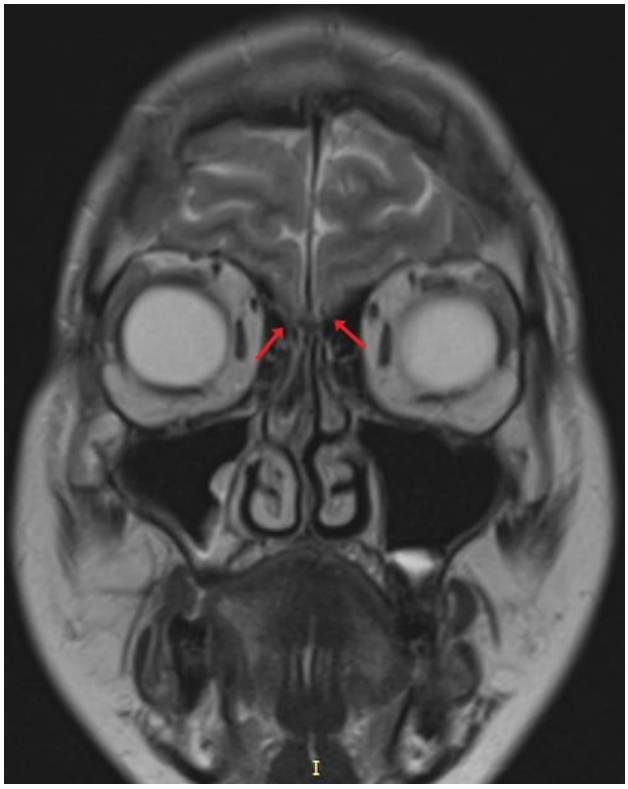

Figure 2: T2-weighted coronal MRI image showing olfactory bulbs hypoplasia (red arrows).

Ethics approval and consent to participate

Oral and written consent was obtained from the patient for publication of this case and any accompanying images.

List of abbreviations

MRI: Magnetic resonance imaging

Data Availability

Not applicable.

Conflicts of Interest

The author(s) declare(s) that there is no conflict of interest regarding the publication of this paper.

Funding Statement

The author(s) received no financial support for the research, authorship and publication of this article.

Authors' contributions

RAP was responsible for data collection, drafting of the paper and spelling revision. PLDF, PC and SM were responsible for data collection and pictures. JC and HC were responsible for revision. All authors read and approved the final manuscript.

References

[1] Zaghouani H, Slim I, Zina NB, Mallat N, Tajouri H, Kraiem C. Kallmann syndrome: MRI findings. Indian J Endocrinol Metab. 2013 Oct;17(Suppl 1): S142-5. doi: 10.4103/2230-8210.119536.

[2] Dodé C, Hardelin JP. Kallmann syndrome. Eur J Hum Genet. 2009 Feb;17(2):139-46. doi: 10.1038/ejhg.2008.206.

[3] Sonne J, Lopez-Ojeda W. Kallmann Syndrome. Treasure Island (FL): StatPearls Publishing; 2024 Jan–. PMID: 30855798.

[4] Swee DS, Quinton R, Maggi R. Recent advances in understanding and managing Kallmann syndrome. Fac Rev. 2021 Apr 13; 10:37. doi: 10.12703/r/10-37.