Hypogonadism

Hypogonadism is a medical term which describes a diminished functional activity of the gonads – the testes and ovaries in males and females, respectively – that may result in diminished sex hormone biosynthesis. In layman's terms, it is sometimes called “interrupted stage 1 puberty”. Low androgen (e.g., testosterone) levels are referred to as hypoandrogenism and low estrogen (e.g., estradiol) as hypoestrogenism, and may occur as symptoms of hypogonadism in both sexes, but are generally only diagnosed in males and females respectively. Other hormones produced by the gonads which may be decreased by hypogonadism include progesterone, DHEA, anti-Müllerian hormone, activin, and inhibin. Spermatogenesis and ovulation in males and females may be impaired by hypogonadism, which, depending on the degree of severity, may result in partial or complete infertility.

The association of cerebellar ataxia and hypogonadism occurs in a heterogeneous group of disorders, caused by different genetic mutations often associated with recessive inheritance. In these patients, magnetic resonance imaging (MRI) plays a pivotal role in the diagnostic workflow, with variable involvement of the cerebellar cortex, alone or in combination with other brain structures. Neuroimaging involvement of the pituitary gland is also variable. Here, we provide an overview of the central clinical and conventional brain and pituitary gland MRI imaging findings of the most common genetic mutations associated with the clinical phenotype of ataxia and hypogonadism, with the aim of helping neuroradiologists in the identification of these disorders [1)].

The clinician must differentiate between primary (testicular) and secondary (pituitary-hypothalamic or central) hypogonadisms and be aware of adult-onset hypogonadism. If gonadotropins are low or inappropriately normal, the clinician must consider potential reversible causes in the hypothalamus-pituitary axis. Also, it is critical to understand the pitfalls of testosterone testing. When clinically indicated, evaluation of other pituitary hormone functions as well as pituitary magnetic resonance imaging may be necessary. Furthermore, it is essential to recognize that pituitary incidentalomas are common. Patients with microprolactinoma are more likely to present with symptoms of sexual dysfunction while those with macroprolactinoma are more likely to present with symptoms of mass effect. Some functional pituitary tumors respond to drug therapy while other nonfunctional tumors require surgical intervention. It is important for the clinician to understand the proper work-up of the hypogonadal patient with pituitary dysfunction and when necessary to refer to an endocrinologist or a neurosurgeon [2)].

1)
Scaravilli A, Tranfa M, Pontillo G, Brais B, De Michele G, La Piana R, Saccà F, Santorelli FM, Synofzik M, Brunetti A, Cocozza S. A Review of Brain and Pituitary Gland MRI Findings in Patients with Ataxia and Hypogonadism. Cerebellum. 2023 May 8. doi: 10.1007/s12311-023-01562-0. Epub ahead of print. PMID: 37155088.
2)
Levy S, Arguello M, Macki M, Rao SD. Pituitary Dysfunction Among Men Presenting with Hypogonadism. Curr Urol Rep. 2019 Nov 16;20(11):78. doi: 10.1007/s11934-019-0935-z. Review. PubMed PMID: 31734886.

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