Pituitary hemochromatosis
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Section: Neuroradiology
Area of Interest: Education Head and neck Abdomen
Procedure: Imaging sequences
Technique: MR
Special Focus: Epidemiology Endocrine disorders Metabolic disorders Case Type: Clinical Cases
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Patient: 27 years, female

Clinical History:

27 years old female who initially presented amenorrhea, hypogonadism and syncopal episodes, with an EKG with atrial fibrillation, a laboratory with alteration of the hepatogram and serum ferritin of 5000 ng/mL.

Imaging Findings:

MRI shows an hypointense in T1 and T2 WS pituitary gland with no intravenous contrast enhancement, findings compatible with hemosiderin deposits.

Hepatic MRI was performed, which reported a severe hepatic iron overload.

Discussion:

Hemochromatosis is a disorder characterized by excessive iron deposition in reticular tissue affecting mainly the liver and heart [1, 2]. Etiologically, it can be classified as a genetic (or primary) cause, or acquired (or secondary) [1]. Primary hemochromatosis usually manifests in the fifth decade of life, but when it manifests in people younger than 30 years it is called juvenile hemochromatosis (JH) like in our case, a more severe variant due to the involvement of two genes (autosomal recessive).

JH presents multisystemic affection, producing cirrhosis, diabetes mellitus, myocardiopathy, cutaneous pigmentation and hypogonadism (affecting CNS organs that are not incorporated in the blood-brain barrier such as pineal gland, choroid plexus and pituitary gland). [2]

When the iron capacity is saturated it can accumulate, among others, in the hypophysis, most commonly manifesting as hypogonadotrophic hypogonadism [1, 3, 4]. The iron overload in the pituitary gland can be demonstrated with MRI given the superparamagnetic conditions. In pituitary hemochromatosis, a signal drop in the T2 WS of the neurohypophysis is observed pathognomonically and, in more advanced cases, signal drop in T1 WS can be observed, which can be accompanied by a decrease in pituitary volume [1, 4]. The most sensitive sequence for the detection of iron deposition is the T2 * gradient echo sequence (GRE) demonstrating a marked decrease in the signal intensity. [1, 4, 5]

Among the entities that could present a shortening in T2 WS are pituitary hemorrhage, melanoma metastasis, an aneurysm that generates flow void, a Rathke pouch cyst or other tumors that have calcifications (chordoma,
Treatment is based on bleeding and chelation therapy

TEACHING POINT
1) Juvenile Hemochromatosis with pituitary affection manifests clinically as hypogonadotropic hypogonadism
2) MRI detects iron as a low intensity signal in T2 WS and, in advance cases, also in T1 WS
3) The most sensitive sequence for the detection of iron deposition is T2* GRE, showing a low signal intensity

Written informed patient consent for publication has been obtained.

Differential Diagnosis List: Juvenile Hemochromatosis, Pituitary hemorrhage, Melanoma metastasis, Rathke pouch cyst, Aneurism

Final Diagnosis: Juvenile Hemochromatosis

References:
Bonneville, J.-F., Bonneville, F., Cattin, F., Nagi, S (2016) MRI of the Pituitary Gland. Pages 327-328
Description: Hypointense pituitary gland (arrow). Origin: Italian Hospital of Buenos Aires
Description: Hypointense pituitary gland (arrow) Origin: Italian Hospital of Buenos Aires
Description: Hypointense pituitary gland (arrow) without IV. contrast enhancement (arrowhead)
Origin: Italian Hospital of Buenos Aires
Description: Low signal of hepatic parenchyma due to iron deposit
Origin: Hospital Italiano de Buenos Aires