Primary Hemochromatosis. Early detection of Myocardial Involvement Through Cardiac Magnetic Resonance Imaging

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SUMMARY
The primary hemochromatosis is the most common genetic disease in Western regions (1 out of 300/400 individuals). The cardiac involvement during its early stages is not detected with the diagnostic imaging techniques, and it is during these stages when potentially life-threatening arrhythmias occur. Through the CMRI, with its T2 star sequence, it is possible to early detect the cardiac involvement and to stratify the risk and monitor the evolution of treatment in affected patients.

CASE REPORT
On April 2005, a 53-year old male patient asked for medical advice due to a syncopal attack lasting a few seconds. He had no previous history of cardiovascular disease.

Personal history
Bilateral congenital blepharoptosis, osteochondroma of the head of the fibula, left elbow bursitis, left meniscectomy, tonsillectomy and appendectomy. Cardiovascular examination was normal before each surgery.

Family history

History of present illness


09/02: Intense headache, vertigo and dizziness. Neurological examination: UN. Brain NMRI and CT: UN. 11/02: Ferritin level...
1680 (35 to 280 mg/ml). Iron serum level 215 (60 to 170 μg/100 ml). Transferrin serum level 538 (250 to 300 μg/100 ml). Transferrin saturation 75% (Vn B&:50% @&:30%). SGPT and SGOT: slightly increased. 12/02: Genetic testing was positive for hemochromatosis with homozygote gene mutation C 282 T. Phlebotomies of 500 cm cm were performed each week and hematological parameters were monitored. 08/03: Normal hematological parameters. Phlebotomies are performed monthly. Absence of symptoms (abdominal pain, myalgias and diarrhea). 03/05: Hypothyroidism (subacute thyroiditis confirmed by thyroid panel, ultrasound and I-131 scintigraphy). T4 50 μg/day. 04/05: The patient presented a car accident due to a syncopal attack lasting few seconds.

DISCUSSION

During early stages of hemochromatosis, iron is stored inside the myocyte; however, there is no myocardial

Figure 1 shows patient’s electrocardiogram and two tracings of the 34-hour Holter monitoring.

Fig. 1. A. ECG: sinus rhythm. Left QRS axis deviation (45°) consistent with left anterior hemiblock. B. 24-hour Holter monitoring record showing a 6-beat run self-limited monomorphic ventricular tachycardia. C. 24-hour Holter monitoring record showing isolated monomorphic ventricular premature beats.

Fig. 2. A. Normal M-mode and 2-D echocardiogram. B. Normal tissue Doppler. C. Normal color tissue Doppler.
damage leading to fibrosis, wall motion or systolic thickness anomalies. For this reason, at early stages of the disease, M mode echocardiogram, tissue Doppler imaging, color tissue Doppler and transmitral Doppler, as well as CNMRI (with its functional and anatomic sequences) fail to detect any anomaly. Biventricular systolic function, mass, thickness, volumes and diastolic function assessed by Doppler echocardiography and CNMRI, were normal (Figures 2 and 3).

Scintigraphy and delayed enhancement in CNMRI were also normal due to the lack of fibrosis.

Unlike other imaging methods, cardiac NMRI with measurement of T2 star sequence at different and progressively longer echo times (TEs) from 5 ms to 20 ms is useful to determine iron overload. The presence of «black» areas in the myocardium below 20 ms puts in evidence abnormal iron stores which may produce malignant ventricular arrhythmias (Figure 4). Through this technique it is possible to monitor the evolution of treatment and to stratify the risk of patients during early stages of this disease. (5, 6) The presence of liver iron overload was ruled out due to high levels SGOT and SGPT; the field of view (FOV) was modified in order to visualize the entire liver parenchyma.

**RESUMEN**

**Hemocromatosis primaria. Detección precoz de compromiso miocárdico a través de la resonancia magnética cardíaca**

La hemocromatosis primaria es la enfermedad genética más común de Occidente (1 de cada 300 a 400 personas). El compromiso cardíaco durante sus estadios iniciales no se detecta por las técnicas de diagnóstico por imágenes y es durante este período cuando sobrevienen arritmias potencialmente letales. A través de la resonancia magnética cardíaca (RMC) con su secuencia de T2 estrella es posible detectar precozmente la afectación cardíaca y permite estratificar el riesgo y monitorizar la evolución del tratamiento de los pacientes afectados.

**Palabras clave** > Hemocromatosis - Arritmias malignas - Resonancia magnética
BIBLIOGRAPHY