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Non HFE-Related *Hemochromatosis*; A Case Report from Iran

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ABSTRACT

Hemochromatosis, especially with cardiac and liver problem, is rare in Iran. We report a young female with pulmonary hypertension and abnormal liver function tests due to non HFE- related hemochromatosis. *Govaresh* 2004; **9**: 66-9

Keywords: Hemochromatosis, Pulmonary hypertension, Iran

INTRODUCTION

Hereditary hemochromatosis (HH) (also called genetic hemochromatosis) is the term used for the inherited disease of iron overload, that is *HFE*[1]-related and that is characterized by an inappropriately elevated rate of intestinal iron absorption. Trousseau was the first to describe a case of hemochromatosis in the French pathology literature in 1865[(1)]. Several population surveys have shown that the frequency of the homozygous disease ranges from 1 in 100 to 1 in 400 in white populations in several areas of the world[(2)].

1. The gene involved in the most common form of hemochromatosis is termed HFE.

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In 1996, the gene for HH, called *HFE*, was identified, thereby allowing genetic testing for the two major mutations (C282Y, H63D) that are responsible for HH[(3)]. Subsequently, numerous clinical and pathophysiologic studies have led to improved diagnosis, better family screening, and new insights into normal and abnormal iron homeostasis. Mutations in other genes, currently unidentified, are responsible for non-HFE associated hemochromatosis. But hemochromatosis seems to be a rare condition in Iran, so a few case reports about the subject exist in our country.

CASE REPORT

A 35-year-old woman presented with pallor, dyspnea and hypermenorrhea from many years ago. There was no history of jaundice, pruritus, smoking or alcohol use. The patient had been followed by a cardiologist with impression of pulmonary hypertension (Echocardiography: mild TR, mild MR, mean pulmonary artery pressure; 32 mmHg, EF; 70%, thickness of right