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Short Communication

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Hereditary haemochromatosis

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Short communication

Iron is an essential element for the living body. It is stored in human body in the form of ferritin and hemosiderin in various locations like liver, spleen, marrow, duodenum, skeletal muscle and other anatomic areas. Human body lack iron excretion function, the transformation of ferritin into hemosiderin reduces iron toxicity. If there is an increase in level of non transferrin-bound iron, both in whole body iron overload and in localized iron deposition it leads to hazardous free radicals which further leads to various disorders. Hereditary hemochromatosis is generally caused by certain variants in the HFE gene. If two of these variants are inherited, one from each parent, then it leads to hereditary hemochromatosis and risk of developing high iron levels. The faulty haemochromatosis gene (HFE) is present on the short arm of 6 chromosome. The candidate gene HFE for Hereditary haemochromatosis (HH) , encodes a major histocompatability complex class 1-like molecule that is involved in iron uptake. C282Y and H63D are two common mutations.A single mutation, 845A in the gene leads to substitution of tyrosine for cysteine at 282 amino acid in C282Y, it is a missense mutation. The C282Y mutation is most strongly implicated in the development of hemochromatosis. The histidine-to-aspartic acid substitution at amino acid position 63 is a H63D HFE mutation. It is also associated with hemochromatosis, but not to much extent than C282Y.

Body builds up too much iron in the skin, heart, liver, pancreas, pituitary gland, and joints during Hemochromatosis. High levels of iron damages tissues and organs and lead to Cirrhosis (liverdamage) external icon, Hepatocellular carcinoma (liver cancer), Heart problems, Arthritis (joint pain), and Diabetes.

Symptoms: It include feeling of tiredness or weakness, weight loss, joint pain, bronze or grey skin color, abdominal pain and loss of sex drive.

Treatment: It involves regularly scheduled blood removal as it is effective way to lower the amount of iron in body, annual blood tests to check your iron levels; Liver biopsy to check for cirrhosis; Iron chelation therapy, which involves takeing medicine orally or injected to decrease the level of iron in body if one cannot get their blood removed; avoiding multivitamins, vitamin C supplements, and iron supplements, which increases iron in body; No consumption of alcohol, as it increases the risk of liver damage and not eating uncooked fish and shellfish and getting recommended vaccinations, including those against hepatitis A and B. To reduce iron toxicity, phlebotomy is performed for treating hereditary hemochromatosis.

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