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Hereditary Hemochromatosis-Special Reference to Indian Scenario

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ABSTRACT Hereditary haemochromatosis (HH) manifested as iron overload in different organs due to homozygocity of a single autosomal mutation. If untreated it leads to conditions such as liver cirrhosis, type 1 diabetes mellitus, hypogonadotropic hypogonadism, cardiomyopathy, arthritis, and bronze coloring of the skin. Two different mutations C282Y and H63D in the HFE gene have been shown to be associated with over 93 per cent of HH cases. The disease is seen in Northern European population but in India the reports of genetic study is rare. Large population based studies are required to investigate the prevalence of this disease and association with other diseases in Indian subcontinent.