DEFINITION

1. **Haemochromatosis** is a condition in which the total body iron is increased. The excess iron is stored in several organs, including the liver, causing damage to them.

CLINICAL MANIFESTATIONS

2. The condition does not become clinically manifest until it is either discovered as an incidental finding on blood testing or when tissue damage becomes evident.

3. The presenting features depend on the organs mainly damaged by iron deposition. The skin becomes pigmented, initially causing browning of the exposed areas, later producing a more generalised slate-grey appearance. The commonest clinical finding is hepatomegaly, which may present before there is any disturbance or liver metabolism or any symptoms, although there may be abdominal pain or discomfort. Eventually hepatic cirrhosis develops. Splenomegaly may also occur.

4. Iron deposits in the pancreas may cause insulin dependent diabetes mellitus. Deposits in the synovial tissues may cause arthralgia and an arthropathy distinct from rheumatoid arthritis or osteoarthritis. Hypogonadism may result in loss of libido, impotence and scanty body hair. Deposits in the heart may result in cardiac failure and arrhythmias.

AETIOLOGY

5. **Haemochromatosis** may be **Primary** or **Secondary**.

    5.1. **Primary (hereditary) haemochromatosis** is caused by an increased absorption of dietary iron. The inability to limit iron absorption is inherited as an autosomal recessive gene associated with the HLA-B3, B7 and B14 histocompatibility antigens. The disease only develops in homozygotes.

    5.2. **Secondary haemochromatosis** may be produced by repeated blood transfusions. It may also arise from excess iron absorption (in idiopathic haemochromatosis or in conditions resulting in ineffective erythropoiesis) or from excess iron intake, orally or parenterally. It may arise secondary to liver disease. In all the secondary forms of haemochromatosis the aetiology is that of the underlying condition.

CONCLUSION

6. Haemochromatosis is a condition in which the total body iron becomes excessive. The primary or hereditary form is the result of a genetic disorder and is unrelated to external factors. Secondary forms occur in association with other conditions which have been listed above, the aetiology in these cases being that of the underlying condition.

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