Aceruloplasminemia: This is a rare recessive disorder due to mutations in the CP gene encoding ceruloplasmin, the principal copper transport protein in plasma also involved in iron release from macrophages and other cells. Deficiency of CP causes moderate anemia with iron accumulation in liver, pancreas and basal ganglia.

What causes the disease and how common is it?

This is a genetic disease linked to mutations of the CP gene, encoding ceruloplasmin, a plasma metalloprotein member of the multi-copper oxidase enzyme family. These mutations lead to a total or reduced amount of Cp or to a reduced activity.

About 60 patients with aceruloplasminemia from several countries including Japan, China, Ireland, Belgium, France, Italy and the US, have been described. Most patients are of Japanese origin. The frequency of homozygosity for deleterious Cp mutations in non-consanguineous couples in Japan was estimated to be 1 per 2000000.

What are the most frequent symptoms if I have the disease?

Laboratory and clinical expression of aceruloplasminemia includes low or absence serum ceruloplasmin, low serum copper levels, mild-moderate microcytic anemia with low serum iron and high serum ferritin, diabetes mellitus, and late-onset neurological symptoms, including retinal degeneration, ataxia, involuntary movements and dementia.

Which treatment must I follow if I have the disease?

There is no established treatment for neurological symptoms. Liver iron overload can be reduced by phlebotomy therapy, although the volume of blood removed at each session and the timing of repeat phlebotomies must be carefully controlled. In patients with very low haemoglobin levels or those intolerant to phlebotomies iron chelation therapy (such as deferoxamine, deferiprone or deferasirox chelation) should be done.

What is the risk of passing the condition on to my children?

Aceruloplasminemia is an autosomal recessive disease. An individual can be heterozygous for the disorder (healthy carrier) when only one of the CP allelel is mutated, or homozygous or compound heterozygous (affected individual) when two CP allelels are mutated.
A couple who carries each one a mutated CP allele has a 25 percent risk of having a child affected by the disorder at each pregnancy. The probability of having a child who is a healthy carrier of the disorder is 50 percent at each pregnancy, and the risk that a child will not have the disorder and will not be a carrier is 25 percent. Ask for genetic counselling to get a complete explanation.

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