

Juvenile hemochromatosis is a rare genetic disorder characterized by the accumulation of iron in various organs of the body. Symptoms usually become apparent before the age of 30. The specific symptoms and severity of juvenile hemochromatosis vary from one person to another. Common symptoms include absent or decreased function of the testes in males or ovaries in females (hypotrophic hypogonadism), heart (cardiac) disease, scarring of the liver (cirrhosis), joint disease, diabetes, and dark discoloration of patches of skin (hyperpigmentation). These symptoms are similar to those seen in classic hereditary hemochromatosis. However, the symptoms associated with juvenile hemochromatosis occur at an early age and are usually more severe. If untreated, juvenile hemochromatosis can potentially cause life-threatening complications. Juvenile hemochromatosis is caused by mutations of one of at least two genes (the HJV and HAMP genes). These mutations are inherited as an autosomal recessive trait. Juvenile hemochromatosis affects males and females in equal numbers. The disorder is rare, but the actual incidence in the general population is unknown. Juvenile hemochromatosis has been reported worldwide. The disorder usually becomes apparent between 10-30 years of age. Mutations of the HJV gene account for the majority of cases of juvenile hemochromatosis.