Arthropathy in hereditary hemochromatosis

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Arthropathy is one of the leading clinical manifestations of hereditary hemochromatosis (HH). Although cirrhosis of the liver is crucial for mortality in patients with HH, arthropathy has the greatest impact on the quality of life. Several mutations in the HFE and other genes have recently been identified, and the prevalence of some of these mutations has already been investigated in population studies in greater detail. Even though cofactors other than genetic predisposition may play a role in the establishment of the disease, the new understanding of the genetic background of this iron storage disorder may help in identifying patients before the onset of clinical symptoms. Early initiation of iron depletion therapy, not effective in established arthropathy of HH, might prevent the manifestation of arthropathy or reduce its severity.