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Variations on a gene

investigating the genetic basis of iron overload

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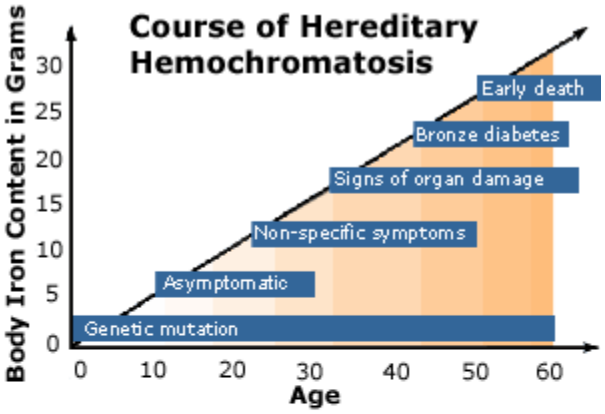

Chart reflecting the course of hereditary hemochromatosis This graph depicts the clinical course of iron overload due to hereditary hemochromatosis. The Y axis reflects increasing amounts of iron concentration in the body and the X axis reflects increasing age in years from birth to age 60. The diagonal line shows that as a person's iron concentration increases over time, the effects of iron on the body are initially seen as non-specific symptoms, followed by more serious signs of organ damage, bronze diabetes, and ultimately early death.

Slope depends on individual.

Hereditary hemochromatosis is a common disease that results in the accumulation of iron in the body. Approximately 5 per 1000 Americans inherit this disorder, but symptoms may never appear, leaving many people undiagnosed.

Mutations of the *HFE* gene are commonly at fault. In this tutorial, we examine the link between SNPs of the *HFE* gene and the development of disease. In the course of deciphering whether a SNP is significant, we look to see whether the SNP causes an amino acid change, the nature of this change (e.g., a switch of cysteine to another amino acid may disrupt a disulfide bond), and whether this change takes place in a conserved domain. We also take a look at the amino acid change on a three-dimensional representation of the HFE protein structure.

So... can a switch of a single nucleotide be responsible for a mutant HFE protein that allows iron regulation to go astray?

Search the SNPs database for HFE.

Do single nucleotide polymorphisms in the *HFE* gene have functional consequences?

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