

Examples of *HFE* Genotypes in Families with Hemochromatosis

Homozygote: inherits two copies of the same mutated *HFE* gene.
 Heterozygote: inherits one copy of a mutated *HFE* gene. Also called a carrier.
 Compound Heterozygote: inherits two different mutated copies of the *HFE* gene.

— Normal or Unknown Mutation + C282Y Mutation ▲ H63D Mutation



IMPORTANT NOTES:

- The inheritance pattern of classical (Type I) Hemochromatosis is autosomal, recessive
- Everyone inherits two copies of *HFE*
- Mutated copies of *HFE* are found primarily in Caucasians
- Only the mutated copies C282Y and H63D are represented in this chart because these are the most important known mutations to date
- When one parent has two mutated copies of *HFE*, all offspring are at least obligate carriers
- *HFE* mutations are present in about 85% of Caucasians in the USA with hereditary hemochromatosis
- *HFE* related iron overload is an adult onset disorder. Other genes that can cause iron overload in children are not included in this chart
- The risk of iron loading is presently known to be greatest in men who are C282Y homozygotes
- Heterozygotes, especially compound heterozygotes are also at increased risk of iron loading, but likelihood and severity are lower
- Informed consent: Anyone considering genetic testing should be made aware of the potential consequences, such as possible insurance and employer discrimination or paternity identification
- Genetic status provides no information about tissue iron levels. Clinical evaluation of serum ferritin and transferrin iron saturation percentage is one way to estimate tissue iron status
- For more information about prevalence and penetrance of *HFE*, contact Iron Disorders Institute: info@irondisorders.org