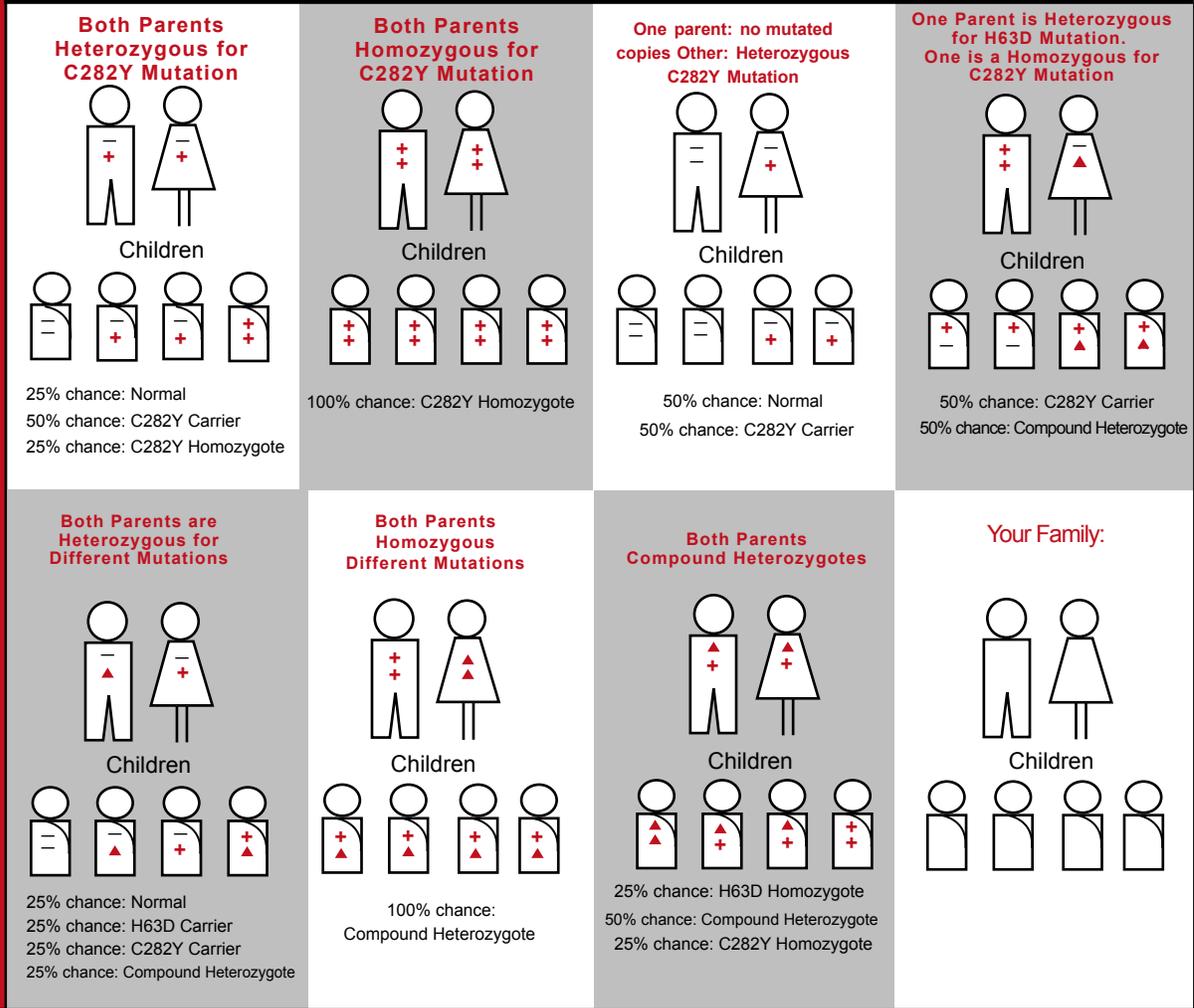


Examples of *HFe* Genotypes in Families with Hemochromatosis

Homozygote: inherits two mutated copies of the *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. Use of the term "normal" or unknown are sometimes called "wild type."
- Everyone inherits two copies of the *HFe* gene (one copy from the father, one copy from the mother.)
- Mutated copies of *HFe* are found primarily in people of Northern European descent.
- Only the mutated copies C282Y and H63D are represented in this chart because these are the most studied mutations to date. Other mutations such as the S65C can be identified with today's genetic testing availability.
- When one birth parent has two mutated copies of *HFe*, all offspring are at least obligate carriers.
- HFe* mutations are present in about 85% of Americans diagnosed with iron overload. Other inherited causes of iron overload include, but not limited to: ferroportin disease, sickle cell disease, and thalassemia.

- 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 the 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 (TS%) is one way to estimate tissue iron status
- For more information about prevalence and consequences of *HFe*, visit our websites:

www.irondisorders.org www.hemochromatosis.org

