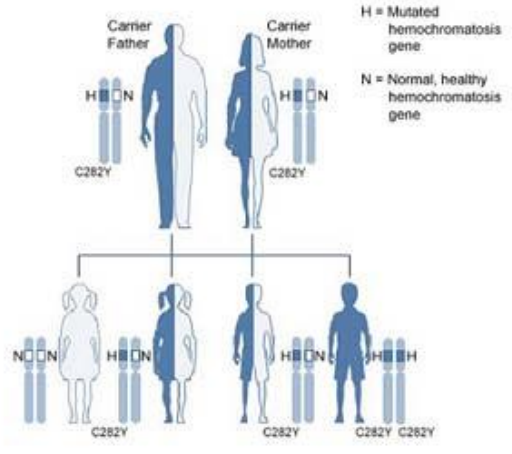
Canadian HEMOCHROMATOSIS SOCIETY
Société canadienne de l'hémochromatose

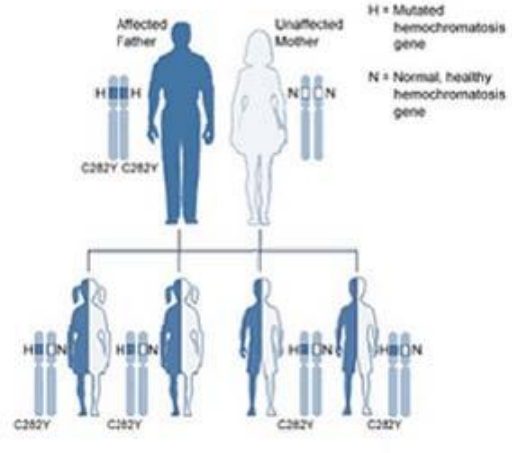
Hemochromatosis - Genetic Inheritance

Inheritance Combinations for HFE Hemochromatosis (Autosomal Recessive Inheritance)

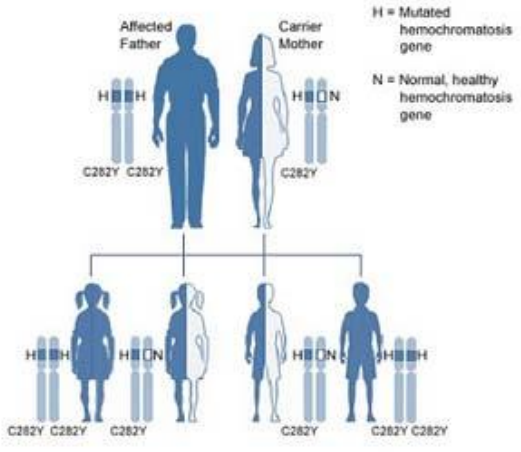
If **both parents are carriers of one C282Y mutation** for the HFE-hemochromatosis gene, for each pregnancy there is a 25% chance of inheriting two normal copies of the gene and being unaffected, a 50% chance of inheriting one mutated copy and one normal copy and being a carrier, and a 25% chance of inheriting two mutated copies and being affected.



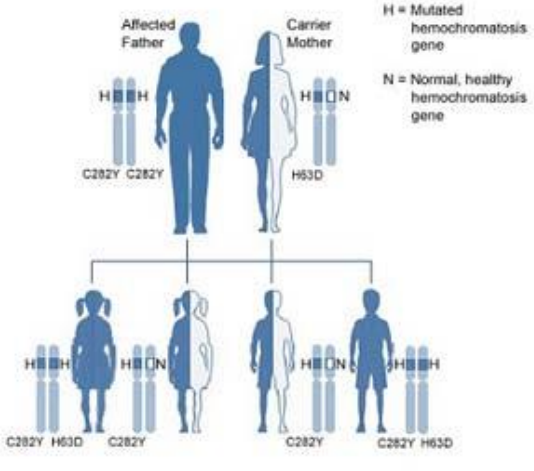
If **one parent is affected with two copies of the C282Y mutation** for the HFE-hemochromatosis gene, **and the other parent is unaffected**, there is a 100% chance that each pregnancy will inherit one mutated copy and one normal copy, which means that all offspring will be obligate carriers.



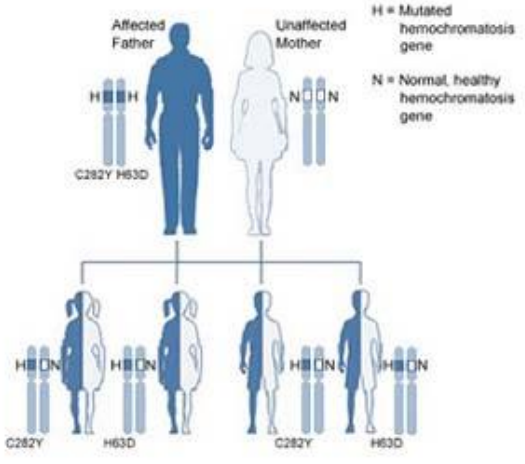
If **one parent is affected with two copies of the C282Y mutation** for the HFE-hemochromatosis gene, **and the other parent is a carrier for one C282Y mutation**, for each pregnancy there is a 50% chance of inheriting one mutated copy and one normal copy and being a carrier, and a 50% chance of inheriting two mutated copies and being affected.



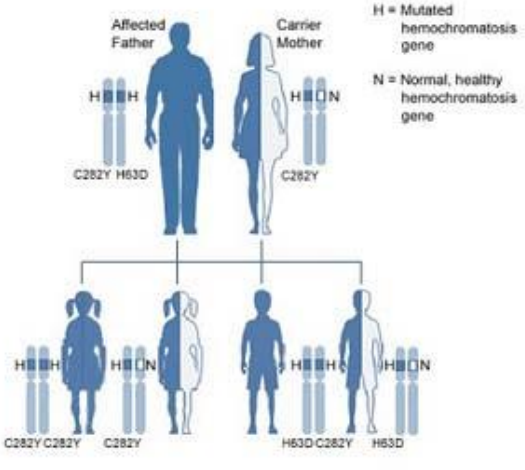
If **one parent is affected with two copies of the C282Y mutation** for the HFE-hemochromatosis gene, **and the other parent is a carrier for one H63D mutation**, for each pregnancy there is a 50% chance of inheriting one mutated C282Y copy and one normal copy and being a carrier, and a 50% chance of inheriting two mutated copies (one C282Y and one H63D) and being affected.



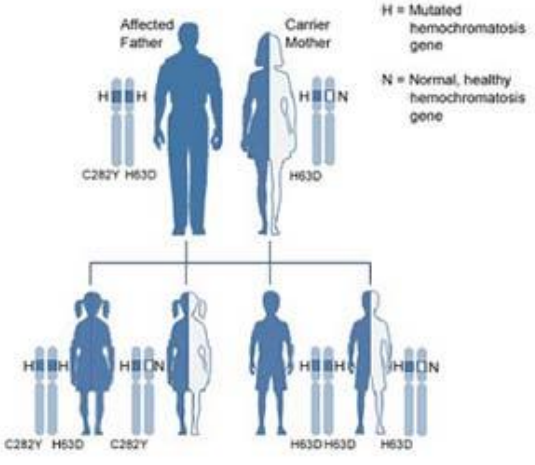
If **one parent is affected with two mutated copies (one C282Y and one H63D)** for the HFE-hemochromatosis gene, **and the other parent is unaffected**, there is a 100% chance that each pregnancy will inherit one mutated copy and one normal copy, which means that all offspring will be obligate carriers. 50% of the offspring will be C282Y carriers and 50% of the offspring will be H63D carriers.



If **one parent is affected with two mutated copies (one C282Y and one H63D)** for the HFE-hemochromatosis gene, **and the other parent is a carrier for one C282Y mutation**, for each pregnancy: there is a 50% chance of inheriting one mutated copy (either one C282Y or one H63D) and one normal copy and being a carrier, and a 50% chance of inheriting two mutated copies (either two C282Y copies, or one C282Y and one H63D) and being affected.



If **one parent is affected with two mutated copies (one C282Y and one H63D)** for the HFE-hemochromatosis gene, **and the other parent is a carrier for one H63D mutation**, for each pregnancy: there is a 50% chance of inheriting one mutated copy (either one C282Y or one H63D) and one normal copy and being a carrier, and a 50% chance of inheriting two mutated copies (either two H63D copies, or one C282Y and one H63D) and being affected.



Glossary

Autosomal

Humans have 23 pairs of chromosomes. Chromosomes 1-22 have nothing to do with sexual characteristics and are called 'autosomal' chromosomes (or autosomes).

C282Y

Refers to the most common mutation of the HFE gene. The C282Y mutation results in a change in the amino acid in the protein produced, and affects how the protein 'folds,' thereby limiting its ability to perform its task.

Carrier

A person who carries a gene that causes a disorder, but does not show symptoms.

Compound Heterozygotes

Individuals who inherited two different mutated versions of a gene, one on each of their chromosomes.

DNA

Short for deoxyribonucleic acid, DNA is the molecule that carries genetic information.

Ferritin

A protein that stores iron. High ferritin levels in the blood can be one indication of hemochromatosis.

Gene

A portion of DNA that contains instructions for making a protein.

H63D

A mutation of the HFE gene that can cause hereditary hemochromatosis.

HFE

The gene on chromosome 6 that en-codes the HFE protein. People with Type 1 hereditary hemochromatosis have abnormal HFE proteins due to mutations in the HFE gene.

HHC

Abbreviation for hereditary hemochromatosis.

Heterozygotes

Individuals who inherited one normal and one mutated version of the same gene, one on each of their chromosomes.

Homozygotes

Individuals with two identical mutations of a gene, one on each of their chromosomes.

Phlebotomy

Bloodletting, the common treatment for hemochromatosis.

Transferrin

A protein that circulates in the bloodstream and binds to iron thus "ferrying" iron to other parts of the body. Transferrin is normally 30% bound to iron.