

GLOSSARY OF GENETIC TERMS

Allele: One version of a **gene** at a given location (**locus**) along a chromosome

ATP7B gene – The WD gene, encodes a copper transporting ATPase mainly expressed in the liver that is mutated and rendered absent or dysfunctional in Wilson disease.

Autosomal recessive: Describes a trait or disorder that requires the presence of two copies of a **gene mutation** at a particular **locus** in order to express observable phenotype; specifically refers to genes on one of the 22 pairs of autosomes (non-sex chromosomes).

Carrier: (Heterozygote) A person who carries one normal and one abnormal copy of a gene and therefore does not have the disease. [assuming autosomal recessive]

Chromosome: A circular strand of **DNA** that contains the **genes** and carries hereditary information.

DNA: Genetic material of all living organisms.

First-degree relative: Any relative who is one meiosis away from a particular individual in a **pedigree**; a relative with whom one-half of an individual's genes is shared (i.e., parent, sibling, offspring).

Gene: The basic unit of heredity, consisting of a segment of **DNA** arranged in a linear manner along a chromosome. A gene codes for a specific protein or segment of protein, leading to a particular characteristic or function.

Genotype: The genetic constitution of an organism or cell; also refers to the specific set of alleles inherited at a locus.

Gene sequencing (mutation screening of the entire ATP7B gene: Analysis of the entire ATP7B gene to detect and identify disease-causing mutations. An individual with confirmed Wilson disease needs to be tested first. If both mutations are identified, other family members can then be offered testing. Gene sequencing will identify both mutations in most but not all cases of Wilson disease. Useful for family members to learn if they could be affected but do not yet have symptoms, to learn they are carriers, or to allow for prenatal testing for confirmed carriers.

Haplotype analysis: (Linkage analysis) - Molecular genetic testing to identify a set of closely linked segments of **DNA** (a **marker** or set of markers), comparing the markers of family members to those of an affected patient. Useful for screening siblings of an identified patient.

Heterozygote: An individual who has two different alleles at a particular **locus**, one on each chromosome of a pair; one **allele** is usually normal and the other abnormal.

Homozygote: An individual who has two identical alleles at a particular **locus** one on each **chromosome** of a pair; a disease-affected individual.

Locus: The physical site or location of a specific **gene** on a chromosome.

Marker: An identifiable segment of **DNA**.

Molecular genetic testing: (synonyms: DNA testing, DNA-based testing, molecular testing) Testing that involves the analysis of **DNA** either through **linkage analysis** or sequencing, or one of several methods of detecting a **mutation**.

Mutation: A **gene** alteration that causes or predisposes an individual to a specific disease.

Phenotype: The observable physical and/or biochemical characteristics of the expression of a gene; the clinical presentation of an individual with a particular genotype.

Proband: The family member who is affected with a genetic disease (**homozygote**) whose **markers** are used to determine if other family members have the disease (**haplotype analysis**) or **same mutation (mutation analysis by sequencing)**.

Pedigree: A diagram of the genetic relationships and medical history of a family using standard symbols and terminology.

Second-degree relative: Any relative who is two meioses away from a particular individual in a pedigree; a relative with whom one-quarter of an individual's genes is shared (i.e., grandparent, grandchild, uncle, aunt, nephew, niece, half-sibling)

Targeted mutation analysis: Analysis of a specific location in the **ATP7B** gene for a known particular mutation. Useful for specific populations