**Glossary of Jewish Genetic Terms**

**Autosomal Recessive Inheritance:**

Pattern of inheritance in which disease results only when an individual inherits two gene mutations for a particular disease, one from each parent. If both members of a couple are carriers for the same disease gene, there is a 25% chance in each pregnancy for the child to be affected.

**Ashkenazi Jew:**

Jewish individual of Eastern/Central European ancestry.

**Carrier:**

An individual who has one altered (mutated) copy of a gene and one normal copy of the same gene. Carriers are healthy and have no features of the illness for which they are carriers. The only consequence of the carrier state is the possibility of transmitting the mutated gene to children of the carrier.

**Chromosome:**

A structure found in the cell nucleus and containing genetic information. Human cells contain 46 chromosomes (23 pairs). One of each pair of chromosomes derives from the father, and the other from the mother. Each chromosome contains thousands of individual genes. The genes determine each person's characteristics.

**Diagnostic Testing**\*:

This is performed on a person who is suspected of having a specific genetic disorder. Each disorder has different testing.

**Direct to consumer genetic testing**\*:

This is testing sold or marketed directly to the consumer. This type pf testing provides limited access to a person’s genetic information without necessarily involving a medical professional, genetic counselor or insurance company during the process.

**DNA:**

Deoxyribonucleic Acid. The molecule that encodes genetic information responsible for the structure and function of living organisms. DNA allows for the transmission of inherited information from generation to generation.

**Enzyme:**

A protein that acts as a catalyst to accelerate specific biochemical reactions. Enzymes are required for the normal metabolism (breakdown) of substances in the body.

**Gene:**

The fundamental unit of heredity. Each gene is composed of a sequence of DNA needed to produce a functional protein. Information in the genes is passed from parent to child.

**Genetic Disease:**

Disease state resulting from genetic mutations.

**Mutation:**

An alteration in the sequence of DNA in the gene. Many mutations are "silent" and do not cause disease. Other mutations disrupt the production of a functional protein leading to a genetic disease.

**Predictive testing**\*:

This is performed on an individual if a specific familial mutated gene has been responsible for a disease occurring later in life, and can be conducted before symptoms appear. It can predict likelihood of such disorders as breast, ovarian and colon cancer, Huntington disease and heart disease.

**Prenatal testing**\*:

These tests are performed during a pregnancy to determine the health status of the fetus. Since this testing is not available for every disorder, carrier screening is recommended before pregnancy.

**Pre-Implantation Genetic Diagnosis (PGD**)\*:

This is a procedure performed in conjunction with In Vitro Fertilization (IVF). In IVF, an egg is fertilized in the laboratory and allowed to grow for a few days. After fertilization, one or more cells can be removed from the embryo before implantation for testing. The genetic material (DNA), which is contained within each cell and provides the instruction for the development of an individual, can then be amplified and analyzed. After PDG is performed, only the embryos without the specific genetic changes/mutations can be re-implanted in the uterus.

**Sephardic Jew:**

Jewish individual originating from North Africa and Mediterranean countries

\*These definitions were shared with us by the Center for Jewish Genetic Disorders