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A. Subject

Genetic Testing and Counseling

B. Background

Advancements in technology have contributed to the rapid expansion of identified genetic variations. Some of these variations have been identified as disease-causing, while others are considered common variants with no clinical impact. With the ever-expanding number of genetic tests available, it can be clinically difficult to determine the most appropriate tests for a particular patient. When clinically appropriate, genetic testing may provide diagnostic and/or actionable therapeutic results which can impact a patient's outcome. Due to the complexity of genetic tests and possible results, consultation with medical genetics professionals and counselors may be required to assist members.

According to the National Society of Genetic Counselors of the United States, genetic counseling is meant to integrate the following goals: 1) interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; 2) education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources, and research; 3) counseling to promote informed choices in view of risk assessment, family goals, ethical and religious values; and 4) support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder. Genetic counselors are healthcare professionals trained to provide this care; however, access issues may require other healthcare professionals to assume this role. Genetic counseling, whether provided by a certified genetic counselor or other qualified healthcare professional, is an integral component of genetic testing that is informative and supportive to members, both before and after the completion of testing.

C. Definitions

- **Genetic Screening** – The process of testing a population for a genetic disease to identify a subgroup of people who either have the disease or the potential to pass it to offspring.
- **Genetic Testing** – A medical test that identifies changes in genes, chromosomes, or proteins to confirm or rule out a suspected genetic condition either hereditary or acquired.
- **Human Leukocyte Antigen (HLA) Typing** – A test used to match patients and donors for bone marrow or cord blood transplants.
- **Inherited Genetic Variant** – A type of DNA sequence change passed from parent to offspring (ie, germline).
- **Precision Medicine** – A field of medicine that selects pharmacotherapies based on the patient's genetics.
- **Somatic Gene Variant** – A type of DNA sequence change that is not inherited from a parent but acquired during a person's life.

The MEDICAL Policy Statement detailed above has received due consideration as defined in the
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