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29 C.F.R. § 2590.702-1

Additional requirements prohibiting discrimination based on genetic information.

(a) *Definitions.* Unless otherwise provided, the definitions in this paragraph (a) govern in applying the provisions of this section.

(1) *Collect* means, with respect to information, to request, require, or purchase such information.

(2) *Family member* means, with respect to an individual—

(i) A dependent (as defined for purposes of § 2590.701-2 of this Part) of the individual; or

(ii) Any other person who is a first-degree, second-degree, third-degree, or fourth-degree relative of the individual or of a dependent of the individual. Relatives by affinity (such as by marriage or adoption) are treated the same as relatives by consanguinity (that is, relatives who share a common biological ancestor). In determining the degree of the relationship, relatives by less than full consanguinity (such as half-siblings, who share only one parent) are treated the same as relatives by full consanguinity (such as siblings who share both parents).

(A) First-degree relatives include parents, spouses, siblings, and children.

(B) Second-degree relatives include grandparents, grandchildren, aunts, uncles, nephews, and nieces.

(C) Third-degree relatives include great-grandparents, great-grandchildren, great aunts, great uncles, and first cousins.

(D) Fourth-degree relatives include great-great grandparents, great-great grandchildren, and children of first cousins.

(3) *Genetic information* means—

(i) Subject to paragraphs (a)(3)(ii) and (a)(3)(iii) of this section, with respect to an individual, information about —

(A) The individual's genetic tests (as defined in paragraph (a)(5) of this section);

(B) The genetic tests of family members of the individual;

(C) The manifestation (as defined in paragraph (a)(6) of this section) of a disease or disorder in family members of the individual; or

(D) Any request for, or receipt of, genetic services (as defined in paragraph (a)(4) of this section), or participation in clinical research which includes genetic services, by the individual or any family member of the individual.

(ii) The term *genetic information* does not include information about the sex or age of any individual.

(iii) The term *genetic information* includes—

(A) With respect to a pregnant woman (or a family member of the pregnant woman), genetic information of any fetus carried by the pregnant woman; and

(B) With respect to an individual (or a family member of the individual) who is utilizing an assisted reproductive technology, genetic information of any embryo legally held by the individual or family member.

(4) *Genetic services* means—

(i) A genetic test, as defined in paragraph (a)(5) of this section;

(ii) Genetic counseling (including obtaining, interpreting, or assessing genetic information); or

(iii) Genetic education.

(5)

(i) *Genetic test* means an analysis of human DNA, RNA, chromosomes, proteins, or metabolites, if the analysis detects genotypes, mutations, or chromosomal changes. However, a genetic test does not include an analysis of proteins or metabolites that is directly related to a manifested disease, disorder, or pathological condition. Accordingly, a test to determine whether an individual has a BRCA1 or BRCA2 variant is a genetic test. Similarly, a test to determine whether an individual has a genetic variant associated with hereditary nonpolyposis colorectal cancer is a genetic test. However, an HIV test, complete blood count, cholesterol test, liver function test, or test for the presence of alcohol or drugs is not a genetic test.

(ii) The rules of this paragraph (a)(5) are illustrated by the following example:

Example.

(i) *Facts.* Individual *A* is a newborn covered under a group health plan. *A* undergoes a phenylketonuria (PKU) screening, which measures the concentration of a metabolite, phenylalanine, in *A*'s blood. In PKU, a mutation occurs in the phenylalanine hydroxylase (PAH) gene which contains instructions for making the enzyme needed to break down the amino acid phenylalanine. Individuals with the mutation, who have a deficiency in the enzyme to break down phenylalanine, have high concentrations of phenylalanine.

(ii) *Conclusion.* In this *Example,* the PKU screening is a genetic test with respect to *A* because the screening is an analysis of metabolites that detects a genetic mutation.

(6)

(i) *Manifestation* or *manifested* means, with respect to a disease, disorder, or pathological condition, that an individual has been or could reasonably be diagnosed with the disease, disorder, or pathological condition by a health care professional with appropriate training and expertise in the field of medicine involved. For purposes of this section, a disease, disorder, or pathological condition is not manifested if a diagnosis is based principally on genetic information.

(ii) The rules of this paragraph (a)(6) are illustrated by the following examples:

Example 1.

(i) *Facts.* Individual *A* has a family medical history of diabetes. *A* begins to experience excessive sweating, thirst, and fatigue. *A*'s physician examines *A* and orders blood glucose testing (which is not a genetic test). Based on the physician's examination, *A*'s symptoms, and test results that show elevated levels of blood glucose, *A*'s physician diagnoses *A* as having adult onset diabetes mellitus (Type 2 diabetes).

(ii) *Conclusion.* In this *Example 1, A* has been diagnosed by a health care professional with appropriate training and expertise in the field of medicine involved. The diagnosis is not based principally on genetic information. Thus, Type 2 diabetes is manifested with respect to *A.*

Example 2.

(i) *Facts.* Individual *B* has several family members with colon cancer. One of them underwent genetic testing which detected a mutation in the MSH2 gene associated with hereditary nonpolyposis colorectal cancer (HNPCC). *B*'s physician, a health care professional with appropriate training and expertise in the field of medicine involved, recommends that *B* undergo a targeted genetic test to look for the specific mutation found in *B*'s relative to determine if *B* has an elevated risk for cancer. The genetic test with respect to *B* showed that *B* also carries the mutation and is at increased risk to develop colorectal and other cancers associated with HNPCC. *B* has a colonoscopy which indicates no signs of disease, and *B* has no symptoms.

(ii) *Conclusion.* In this *Example 2,* because *B* has no signs or symptoms of colorectal cancer, *B* has not been and could not reasonably be diagnosed with HNPCC. Thus, HNPCC is not manifested with respect to *B.*

Example 3.

(i) *Facts.* Same facts as *Example 2,* except that *B*'s colonoscopy and subsequent tests indicate the presence of HNPCC. Based on the colonoscopy and subsequent test results, *B*'s physician makes a diagnosis of HNPCC.

(ii) *Conclusion.* In this *Example 3,* HNPCC is manifested with respect to *B* because a health care professional with appropriate training and expertise in the field of medicine involved has made a diagnosis that is not based principally on genetic information.

Example 4.

(i) *Facts.* Individual *C* has a family member that has been diagnosed with Huntington's Disease. A genetic test indicates that *C* has the Huntington's Disease gene variant. At age 42, *C* begins suffering from occasional moodiness and disorientation, symptoms which are associated with Huntington's Disease. *C* is examined by a neurologist (a physician with appropriate training and expertise for diagnosing Huntington's Disease). The examination includes a clinical neurological exam. The results of the examination do not support a diagnosis of Huntington's Disease.

(ii) *Conclusion.* In this *Example 4, C* is not and could not reasonably be diagnosed with Huntington's Disease by a health care professional with appropriate training and expertise. Therefore, Huntington's Disease is not manifested with respect to *C.*