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Genetic Counseling

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[Last Review](#) 03/26/2024

Effective: 11/19/1997

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Policy

Scope of Policy

This Clinical Policy Bulletin addresses genetic counseling.

Additional Information

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I. Medical Necessity

- A. Aetna considers genetic counseling in connection with pregnancy management medically necessary for evaluation of any of the following:
1. Couples who are closely related genetically (consanguinity, incest); or
 2. Familial cancer disorders; or
 3. Individuals from ethnic groups recognized to be at increased risk for specific genetic disorders (e.g., African Americans for sickle cell anemia, Ashkenazi [eastern European] Jews for Tay-Sachs disease); or
 4. Infertility cases where either parent is known to have a chromosomal abnormality; or
 5. Individuals with primary amenorrhea, azospermia, abnormal sexual development or failure in developing secondary sexual characteristics; or
 6. Mother, known, or presumed carrier of an X-linked recessive disorder; or
 7. One or both parents are known carriers of an autosomal recessive disorder; or
 8. Parents of a child born with a genetic disorder, birth defect, inborn error of metabolism or chromosome abnormality; or
 9. Parents of a child with autism, developmental delays, intellectual or learning disabilities; or
 10. Pregnant women who, based on prenatal ultrasound tests or an abnormal multiple marker screening test, maternal serum alpha-fetoprotein (AFP) test, test for sickle cell anemia, or tests for other genetic abnormalities have been told their pregnancy may be at increased risk for complications or birth defects; or
 11. Pregnant women with maternal age 35 years or greater at delivery; or
 12. Pregnant women, or women planning pregnancy, exposed to potentially teratogenic, mutagenic or carcinogenic agents (i.e., chemicals, drugs, infections, radiation); or

13. Previous unexplained stillbirth or repeated (3 or more; 2 or more among infertile couples) first trimester miscarriages, where there is suspicion of parental or fetal chromosome abnormalities; or
14. When contemplating pregnancy, either parent affected with an autosomal dominant disorder.

B. Aetna considers appropriate genetic counseling unrelated to pregnancy medically necessary for consideration of, or provided in conjunction with, medically necessary genetic testing, and in accordance with the guidelines of the American College of Medical Genetics (ACMG).

II. Experimental, Investigational, or Unproven

A. Aetna considers genetic counseling experimental, investigational or unproven for all other indications because its effectiveness for indications other than the ones listed above has not been established.

III. Policy Limitations and Exclusions

Note: Genetic counseling for pregnancy management may not be covered under plans that exclude family planning benefits. Please check benefit plan descriptions for details.

IV. Related Policies

- [CPB 0140 - Genetic Testing \(0140.html\)](#)

CPT Codes / HCPCS Codes / ICD-10 Codes

Information in the [brackets] below has been added for clarification purposes. Codes requiring a 7th character are represented by "+":

Code	Code Description
CPT codes covered if selection criteria are met:	
96040	Medical genetics and genetic counseling services, each 30 minutes face-to-face with patient/family
Other CPT codes related to the CPB:	
82106	Alpha-fetoprotein (AFP); amniotic fluid
HCPCS codes covered if selection criteria are met:	
S0265	Genetic counseling, under physician supervision, each 15 minutes
ICD-10 codes covered if selection criteria are met:	
C18.0 - C18.9	Malignant neoplasm of colon [hereditary nonpolyposis colorectal cancer (HNPCC)] [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
D45	Polycythemia vera [Osler's disease] [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
D57.00 - D57.819	Sickle-cell disorders [infertility cases where either parent is known to have a chromosomal abnormality]
E28.39	Other primary ovarian failure
E28.8	Other ovarian dysfunction [amenorrhea due to ovarian dysfunction]
E30.0 - E30.1	Delay in sexual development and puberty and precocious sexual development and puberty, not elsewhere classified [abnormal sexual development or failure in developing secondary sexual characteristics]
E75.02	Tay-Sachs disease [infertility cases where either parent is known to

	have a chromosomal abnormality]
E84.0 - E84.9	Cystic fibrosis [infertility cases where either parent is known to have a chromosomal abnormality]
F70 - F79	Mental retardation [infertility cases where either parent is known to have a chromosomal abnormality]
F84.0	Autistic disorder [infertility cases where either parent is known to have a chromosomal abnormality]
F95.2	Tourette's disorder [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
G10	Huntington's disease [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
G11.3, G11.8	Cerebellar and other hereditary ataxias [Machado-Joseph disease] [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
G12.1	Other inherited spinal muscular atrophy [Kugelberg-Welander] [familial spinal muscular atrophy] [infertility cases where either parent is known to have a chromosomal abnormality]
G90.3	Multi-system degeneration of the autonomic nervous system [Shy-Drager]
H80.00 - H80.93	Otosclerosis [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
M30.3	Mucocutaneous lymph node syndrome [Kawasaki] [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
N46.0 - N46.9	Male infertility [infertility cases where either parent is known to have a chromosomal abnormality]
N91.0 - N91.2	Amenorrhea
N97.0 - N97.9	Female infertility [infertility cases where either parent is known to have a chromosomal abnormality]
009.511 - 009.529	Elderly primagravida or elderly multigravida [pregnant women with maternal age 35 years or greater at delivery]
O28.0 - O28.9	Abnormal findings on antenatal screening of mother [pregnant women who, based on prenatal ultrasound tests or an abnormal multiple marker screening test, maternal serum alpha-fetoprotein (AFP) test, test for sickle cell anemia, or tests for other genetic abnormalities have been told their pregnancy may be at increased risk for complications or birth defects]
035.0xx+ - 035.9xx+, 035.AXX0 - 035.HXX9	Maternal care for known or suspected fetal abnormality and damage [pregnant women who, based on prenatal ultrasound tests or an abnormal multiple marker screening test, maternal serum alpha-fetoprotein (AFP) test, test for sickle cell anemia, or tests for other genetic abnormalities have been told their pregnancy may be at increased risk for complications or birth defects] [pregnant women exposed to potentially teratogenic, mutagenic or carcinogenic agents]
Q61.11 - Q61.19	Polycystic kidney, infantile type [infertility cases where either parent is known to have a chromosomal abnormality] [one or both parents are known carriers of an autosomal recessive disorder]
Q61.2	Polycystic kidney, adult type [infertility cases where either parent is known to have a chromosomal abnormality] [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
Q77.4	Achondroplasia [when contemplating pregnancy, either parent affected with an autosomal dominant disorder]
Q85.01	Neurofibromatosis, Type 1 [von Recklinghausen's disease] [when

	contemplating pregnancy, either parent affected with an autosomal dominant disorder]
Q90.0 - Q99.9	Chromosomal abnormalities, not elsewhere classified [infertility cases where either parent is known to have a chromosomal abnormality]
T50.0X1+ - T50.996+	Poisoning by, adverse effect of and underdosing of diuretics and other and unspecified drugs, medicaments and biological substances [pregnant women or women planning pregnancy exposed to potentially teratogenic, mutagenic or carcinogenic agents]
T65.0X1+ - T65.94X+	Toxic effect of other and unspecified substances [pregnant women or women planning pregnancy exposed to potentially teratogenic, mutagenic or carcinogenic agents]
T66.XXX+	Radiation sickness, unspecified [pregnant women, or women planning pregnancy, exposed to potentially teratogenic, mutagenic or carcinogenic agents]
Z14.1	Cystic fibrosis carrier [one or both parents are known carriers of an autosomal recessive disorder]
Z14.8	Genetic carrier of other disease [one or both parents are known carriers of an autosomal recessive disorder]
Z15.01 - Z15.09	Genetic susceptibility to malignant neoplasm [familial cancer disorders] [individuals from ethnic groups recognized to be at increased risk for specific genetic disorders (e.g., African-Americans for sickle cell anemia, Ashkenazi (eastern European) Jews for Tay-Sachs disease)]
Z15.81 - Z15.89	Genetic susceptibility to other disease [individuals from ethnic groups recognized to be at increased risk for specific genetic disorders (e.g., African-Americans for sickle cell anemia, Ashkenazi (eastern European) Jews for Tay-Sachs disease)]
Z80.0 - Z80.9	Family history of primary malignant neoplasm [familial cancer disorders]
Z81.8	Family history of other mental and behavioral disorders [parents of a child with mental retardation]
Z82.71	Family history of polycystic kidney [when contemplating pregnancy, either parent affected with an autosomal dominant disorder] [one or both parents are known carriers of an autosomal recessive disorder]
Z82.79	Family history of other congenital malformations, deformations and chromosomal abnormalities [parents of a child born with a genetic disorder, birth defect, inborn error of metabolism or chromosome abnormality]
Z84.3	Family history of consanguinity [couples who are closely related genetically (consanguinity, incest)]
Z84.81	Family history of carrier of genetic disease [mother, known, or presumed carrier of an X-linked recessive disorder] [one or both parents are known carriers of an autosomal recessive disorder]
Z85.00 - Z85.9	Personal history of malignant neoplasm [familial cancer disorders]
Z87.42	Personal history of other diseases of the female genital tract [previous unexplained stillbirth or repeated (three or more; two or more among infertile couples) first trimester miscarriages, where there is suspicion of parental or fetal chromosome abnormalities]
Z87.59	Personal history of other complications of pregnancy, childbirth and the puerperium [previous unexplained stillbirth or repeated (three or more; two or more among infertile couples) first trimester miscarriages, where there is suspicion of parental or fetal chromosome abnormalities]
Z87.790 - Z87.798	Personal history of other (corrected) congenital malformations [infertility cases where either parent is known to have a chromosomal abnormality]

[REDACTED]	
287.898	Personal history of other specified conditions [previous unexplained stillbirth or repeated (three or more; two or more among infertile couples) first trimester miscarriages, where there is suspicion of parental or fetal chromosome abnormalities]

Background

Genetic counseling is a process of communication between patients and trained professionals intended to provide patients who have a genetic disease, or risk of such a disease, with information about their condition and its effect on their family. This allows patients and their families to make informed reproductive and other medical decisions. The counselor will evaluate medical problems or risks present in a family, analyze and explain inheritance patterns of any disorders found, provide information about management and treatment of these disorders, and discuss available options with the family or individual.

According to the American College of Medical Genetics, an important issue in genetic testing is defining the scope of informed consent. The obligation to counsel and obtain consent is inherent in the clinician-patient and investigator-subject relationships. In the case of most genetic tests, the patient or subject should be informed that the test might yield information regarding a carrier or disease state that requires difficult choices regarding their current or future health, insurance coverage, career, marriage, or reproductive options. The objective of informed consent is to preserve the individual's right to decide whether to have a genetic test. This right includes the right of refusal should the individual decide the potential harm outweighs the potential benefits.

Moyer (2014) noted that the U.S. Preventive Services Task Force (USPSTF) reviewed the evidence on risk assessment, genetic counseling, and genetic testing for potentially harmful BRCA mutations in asymptomatic women with a family history of breast or ovarian cancer but no personal history of cancer or known potentially harmful BRCA mutations in their family. The USPSTF also reviewed interventions aimed at reducing the risk for BRCA-related cancer in women with potentially harmful BRCA mutations, including intensive cancer screening, medications, and risk-reducing surgery. This recommendation applies to asymptomatic women who have not been diagnosed with BRCA-related cancer. The USPSTF recommends that primary care providers screen women who have family members with breast, ovarian, tubal, or peritoneal cancer with 1 of several screening tools designed to identify a family history that may be associated with an increased risk for potentially harmful mutations in breast cancer susceptibility genes (BRCA1 or BRCA2). Women with positive screening results should receive genetic counseling and, if indicated after counseling, BRCA testing (B recommendation). The USPSTF recommends against routine genetic counseling or BRCA testing for women whose family history is not associated with an increased risk for potentially harmful mutations in the BRCA1 or BRCA2 genes (D recommendation).

Telephone Versus In-Person Genetic Counseling in BRCA1/BRCA2 Genetic Testing

Bracke and colleagues (2021) noted that pathogenic variants in the BRCA1 and BRCA2 genes increase the risk of breast and ovarian cancer. Individuals with identified pathogenic variants in the BRCA1 or BRCA2 gene could benefit from cancer risk-reducing strategies. In the recent years, there has been an increase in the demand of genetic services. In light of the ongoing COVID19 pandemic, alternatives to face-to-face consultations have had to be considered and adopted, including telemedicine. Informed consent is needed for genetic testing. Studies

have suggested that increased levels of cancer-specific distress may impair the patient's ability to retain information; thus, providing informed consent. In a systematic review and meta-analysis, these investigators examined if telephone genetic counseling for BRCA1 and BRCA2 genetic testing is non-inferior to in-person genetic counseling for the outcomes of cancer-specific distress and genetic knowledge. Databases of Medline, Embase, PsycINFO, CINAHL, SciELO, Web of Science, CENTRAL, ProQuest Dissertation & Theses Database, Clinicaltrials.gov, EU clinical trials register were searched to identify any published or unpublished relevant literature. Random-effects models were used for the meta-analysis. A total of 4 studies were included in the qualitative synthesis of the results; 3 studies were included in the quantitative synthesis of the results. Telephone genetic counseling was non-inferior compared to in-person genetic counseling for the outcomes of cancer-specific distress and genetic knowledge. Sensitivity analysis corroborated the main results. The authors concluded that telephone genetic counseling for BRCA1/BRCA2 genetic testing may be an alternative model of delivering genetic services in front of the increased demand/or when required by social context; however, the paucity of the evidence prevents from drawing strong conclusions regarding the generalizability of these results. These researchers stated that further research is needed to strengthen the conclusions.

Genetic Counseling for Hypertrophic Cardiomyopathy

Cirino et al (2022) stated that genetic testing and genetic counseling are routinely indicated for patients with hypertrophic cardiomyopathy (HCM); however, the uptake and utility of these services is not entirely understood. In a systematic review and meta-analysis, these investigators examined the uptake and utility of genetic counseling and genetic testing for patients with HCM and their at-risk family members, as well as the impact of genetic counseling/testing on patient-reported outcomes (PROs). They carried out a systematic search through March 12, 2021. Meta-analyses were carried out whenever possible; other findings were qualitatively summarized. A total of 48 studies met inclusion criteria (47 observational, 1 randomized). Uptake of genetic testing in probands was 57 % (95 % confidence interval [CI]: 40 to 73). Uptake of cascade screening for at-risk relatives were as follows: 61 % for cascade genetic testing (95 % CI: 45 to 75), 58 % for cardiac screening (e.g., echocardiography) (95 % CI: 40 to 73), and 69 % for either/both approaches (95 % CI: 43 to 87). Furthermore, relatives of probands with a positive genetic test result were significantly more likely to undergo cascade screening compared to relatives of probands with a negative result (odds ratio [OR] = 3.17, 95 % CI: 2.12 to 4.76). Overall, uptake of genetic counseling in both probands and relatives ranged from 37 % to 84 %. Multiple studies found little difference in PROs between individuals receiving positive versus negative genetic test results; however, other studies found that individuals with positive genetic test results experienced worse psychological outcomes. Genetic testing may also inform life choices, especially decisions related to reproduction and insurance. The authors concluded that genetic counseling was associated with high satisfaction, increased perceived personal control and empowerment, and decreased anxiety. Approximately 50 % to 75 % of patients with HCM and their relatives undergo genetic testing or cascade screening; and PROs following genetic testing varied and genetic counseling was associated with high satisfaction and improved PROs.

Genetic Counseling for Late-Onset Neurodegenerative Disease

In a systematic review, Crook et al (2022) examined genetic counseling and testing practices for late-onset neurodegenerative diseases (LONDs), and examined if practices address the internationally accepted objectives of genetic counseling: interpretation, counseling, education, and support. A total of 4 databases were systematically searched for studies published between 2009 and 2020. Peer-reviewed studies in English that reported research and clinical genetic counseling and testing practices for LONDs were included. A narrative synthesis was

performed to describe different practices and map genetic counseling activities to the objectives. Risk of bias was evaluated using the Qualsyst tool. A total of 61 studies from 68 papers were included. Most papers focused on predictive testing (58/68) and Huntington's disease (41/68). There was variation between papers in study design, study population, outcomes, interventions, and settings. Although there were commonalities, novel and inconsistent genetic counseling practices were identified. A total of 18 studies addressed all 4 objectives of genetic counseling. The authors concluded that contemporary genetic counseling and testing practices for LONDs are varied and informed by regional differences and the presence of different healthcare providers. A flexible, multi-disciplinary, client- and family-centered care continues to emerge. These investigators stated that as genetic testing becomes a routine part of care for patients (and their relatives), healthcare providers must balance their limited time and resources with ensuring clients are safely and effectively counseled, and all 4 genetic counseling objectives are addressed. Areas of further research include diagnostic and reproductive genetic counseling/testing practices, evaluations of novel approaches to care, and the role and use of different healthcare providers in practice.

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