

Medical Necessity Guidelines: Genetic Testing: Prenatal, Preconception

Effective: September 16, 2020

Prior Authorization Required	Yes <input checked="" type="checkbox"/> No <input type="checkbox"/>
If <u>REQUIRED</u> , submit supporting clinical documentation pertinent to service request.	
<p>Applies to: COMMERCIAL Products <input checked="" type="checkbox"/> Tufts Health Plan Commercial products; Fax: 617.972.9409 <input checked="" type="checkbox"/> Tufts Health Freedom Plan products; Fax: 617.972.9409 • CareLinkSM – Refer to CareLink Procedures, Services and Items Requiring Prior Authorization</p> <p>TUFTS HEALTH PUBLIC PLANS Products <input checked="" type="checkbox"/> Tufts Health Direct – A Massachusetts Qualified Health Plan (QHP) (a commercial product); Fax: 888.415.9055 <input checked="" type="checkbox"/> Tufts Health Together – MassHealth MCO Plan and Accountable Care Partnership Plans; Fax: 888.415.9055 <input checked="" type="checkbox"/> Tufts Health RITogether – A Rhode Island Medicaid Plan; Fax: 857.304.6404 <input checked="" type="checkbox"/> Tufts Health Unify* – OneCare Plan (a dual-eligible product); Fax: 857.304.6304 *The MNG applies to Tufts Health Unify members unless a less restrictive LCD or NCD exists.</p> <p>SENIOR Products • Tufts Health Plan Senior Care Options (SCO), (a dual-eligible product) – Refer to the Tufts Health Plan SCO Prior Authorization List • Tufts Medicare Preferred HMO, (a Medicare Advantage product) – Refer to the Tufts Medicare Preferred HMO Prior Authorization and Inpatient Notification List</p>	

Note: While you may not be the provider responsible for obtaining prior authorization, as a condition of payment you will need to make sure that prior authorization has been obtained.

OVERVIEW

There are two types of genetic testing that are performed in the preconception and/or prenatal period:

Carrier testing is used to identify individuals who carry a gene mutation that can increase the risk of having a child with an inherited genetic disorder. Carrier testing is offered prior to or during pregnancy. Carrier screening for some genetic disorders (e.g. spinal muscular atrophy, cystic fibrosis) may be offered to woman regardless of family history or ethnicity. More targeted carrier screening tests may be offered to individuals with an increased risk of specific genetic disorders based on family history or ethnicity. For autosomal recessive genetic disorders to be present in a child, two copies of the abnormal gene are needed, therefore each partner to the pregnancy must be a carrier for the child to inherit the disorder. If an individual is confirmed to be a carrier for an autosomal recessive inherited disorder, the individual's reproductive partner should then be offered testing for this disorder.

Prenatal testing is used to detect changes in a fetus's genes or chromosomes before birth. This type of testing is offered during pregnancy if there is an increased risk that the baby will have a genetic or chromosomal disorder. In some cases, prenatal testing can lessen the uncertainty or guide decisions regarding a pregnancy. It cannot identify all possible inherited disorders and birth defects³.

CLINICAL COVERAGE CRITERIA

1. Tufts Health Plan will cover without prior authorization the following screening for a **female** member who is currently pregnant or planning a pregnancy:
 - a. Cystic fibrosis
 - b. Spinal muscular atrophy

- c. Ashkenazi panel (which includes Tay-Sachs Disease, Gaucher Disease, cystic fibrosis, Canavan Disease, Bloom Syndrome, Familial Dysautonomia, Mucopolidosis Type IV, Fanconi Anemia Type C, Nieman-Pick Disease Type A, and Glycogen Storage Disease type 1a)
 - d. First trimester prenatal screening combination of ultrasound for nuchal cord translucency and maternal blood testing for free Beta chorionic gonadotropin (free Beta), and pregnancy associated plasma protein-A (PAPP-A). This screening can be known as Early Risk Assessment, First Screen, or Ultrascreen
 - e. Second trimester screening combination second-trimester screening using four serum markers (alpha-fetoprotein, β -hCG, unconjugated estriol, inhibin-A)
2. For genetic testing not listed above, including genetic testing of male reproductive partner, Tufts Health Plan may authorize coverage for carrier testing and prenatal genetic testing if the Member meets ALL the following criteria (NOTE: Documentation, including a letter of medical necessity is required):
- a. The request is for a Member who is currently pregnant, is partner to a pregnancy or is planning a pregnancy and who falls within a high-risk group for a disease(s) based on personal history, family history, documentation of a genetic mutation and/or ethnic background.
 - b. The results of the test will significantly alter the medical management of the pregnancy and/or the reproductive choices of the Member.
 - c. Documentation is provided by an MD geneticist, a licensed genetic counselor or a physician with expertise in genetic counseling and supports the recommendation for testing based on a review of risk factors, clinical scenario and family history.
 - d. Testing of reproductive partner has not been done or reproductive partner's test results alone are insufficient to inform an appropriate treatment decision.
 - e. Requested test/testing method is considered a proven method for the identification of a genetically linked inheritable disease (i.e., the genotypes to be detected by a genetic test must be shown by scientifically valid methods to be associated with the occurrence of a disease, and the observations must be independently replicated and subject to peer review).

Note: For maternal laboratory genetic testing for trisomy please refer to: [Genetic Testing: Maternal Tests for Fetal Trisomy Medical Necessity Guidelines](#)

LIMITATIONS

1. Tufts Health Plan will initially cover testing of only the female parent or potential female parent for autosomal recessive or X-linked recessive conditions.
2. Testing of the male partner to a pregnancy will be covered for an autosomal recessive condition only if the female partner has been found to be a carrier for the disorder, **OR** if the couple is felt to be at high risk and the pregnancy is beyond 15 weeks gestation.
3. Tufts Health Plan will not cover carrier screening for a specific genetic condition more than once in a member's lifetime.

CODES

The following CPT codes **DO NOT** require prior authorization for testing of female partner:

Codes	Description
81223	CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; full gene sequence
81224	CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; intron 8 poly-T analysis (e.g., male infertility)
81242	FANCC (Fanconi anemia, complementation group C) (e.g., Fanconi anemia, type C) gene analysis, common variant (e.g., IVS4+4A>T)
81250	G6PC (glucose-6-phosphatase, catalytic subunit) (e.g., Glycogen storage disease, type 1a, von Gierke disease) gene analysis, common variants (e.g., R83C, Q347X)
81251	GBA (glucosidase, beta, acid) (e.g., Gaucher disease) gene analysis, common variants (e.g., N370S, 84GG, L444P, IVS2+1G>A)
81255	HEXA (hexosaminidase A [alpha polypeptide]) (e.g., Tay-Sachs disease) gene analysis, common variants (e.g., 1278insTATC, 1421+1G>C, G269S)

Codes	Description
81260	IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (e.g., familial dysautonomia) gene analysis, common variants (e.g., 2507+6T>C, R696P)
81290	MCOLN1 (mucolipin 1) (e.g., Mucopolipidosis, type IV) gene analysis, common variants (e.g., IVS3-2A>G, del6.4kb)
81330	SMPD1(sphingomyelin phosphodiesterase 1, acid lysosomal) (e.g., Niemann-Pick disease, Type A) gene analysis, common variants (e.g., R496L, L302P, fsP330)
81329	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; dosage/deletion analysis (eg, carrier testing), includes SMN2 (survival of motor neuron 2, centromeric) analysis, if performed
81336	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; full gene sequence
81337	SMN1 (survival of motor neuron 1, telomeric) (eg, spinal muscular atrophy) gene analysis; known familial sequence variant(s)
81412	Ashkenazi Jewish associated disorders (e.g., Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia group C, Gaucher disease, Tay-Sachs disease), genomic sequence analysis panel, must include sequencing of at least 9 genes, including ASPA, BLM, CFTR, FANCC, GBA, HEXA, IKBKAP, MCOLN1, and SMPD1
81420	Fetal chromosomal aneuploidy (e.g., trisomy <u>21</u> , monosomy X) genomic sequence analysis panel, circulating cell-free fetal DNA in maternal blood, must include analysis of chromosomes <u>13</u> , <u>18</u> , and <u>21</u>
81507	Fetal aneuploidy (trisomy 21, 18, and 13) DNA sequence analysis of selected regions using maternal plasma, algorithm reported as a risk score for each trisomy
81511	Fetal congenital abnormalities, biochemical assays of four analytes (AFP, uE3, hCG [any form], DIA) utilizing maternal serum, algorithm reported as a risk score (may include additional results from previous biochemical testing)

REFERENCES

1. American College of Obstetricians and Gynecologists. Screening for fetal chromosomal abnormalities. Practice Bulletin #77. Obstetrics & Gynecology. January 2007.
2. American College of Obstetricians and Gynecologists. Screening for fetal chromosomal abnormalities. Practice Bulletin #163. May 2016. (Replaces Practice Bulletin #77). Last accessed July 13, 2017. U.S. National Library of Medicine. What are the types of genetic tests? Genetics Home Reference. Retrieved on January 12, 2007 from: ghr.nlm.nih.gov/handbook/testing/uses. Published July 11, 2017. Last accessed July 13, 2017.
3. Raby BA. Tools for genetics and genomics. Slavotinek A, ed. Up ToDate. Waltham, MA: Up ToDate Inc. <http://www.uptodate.com> (Accessed on August 13, 2018).
4. Schattman G, Xu K. Preimplantation genetic testing. Wilkins-Haug L, ed. Up ToDate. Waltham, MA: Up ToDate Inc. <http://www.uptodate.com> (Accessed on August 13, 2018).
5. Sanders K, Griffin D. Chromosomal Preimplantation Genetic Diagnosis: 25 Years and Counting. J. Fetal Med. (June 2017) 4:51–56 DOI 10.1007/s40556-017-0123-5.
6. American College of Obstetricians and Gynecologists. Carrier Screening in the Age of Genomic Medicine. Committee Opinion Number 690, March 2017, reaffirmed 2019. Accessed January 7, 2020.
7. American College of Obstetrics and Gynecologists. Prenatal Genetic Screening Tests. FAQ 195. September 2019. Accessed January 7, 2020.
8. American College of Obstetrics and Gynecologists. Carrier Screening for Genetic Conditions. Committee Opinion Number 691. March 2017, reaffirmed 2019. Accessed January 7, 2020.

APPROVAL HISTORY

February 1, 2007: Reviewed by the Clinical Coverage Criteria Committee

Subsequent endorsement date(s) and changes made:

- January 30, 2008: Preimplantation Genetic Determination limitation removed from guideline
- March 16, 2009: Reviewed and renewed without changes
- December 16, 2009: Reviewed and no changes made

- February 1, 2010: Reviewed by Medical Policy Advisory Group Committee (MSPAC), no changes
- March 2011: Reviewed by MSPAC. Preconception added to title. Cystic fibrosis and spinal muscular atrophy added to covered without prior authorization section
- March 7, 2012: Reviewed by Integrated Medical Policy Advisory Committee (IMPAC), no changes
- November 28, 2012: Reviewed at IMPAC. A link was added for access to the Medical Necessity Guideline 'Genetic Testing: Maternal Tests for Fetal Trisomy'.
- October 9, 2013: Reviewed by IMPAC, renewed without changes.
- September 30, 2014: Adopted by Tufts Health Plan – Network Health Commercial Plans and Tufts Health Plan – Network Health Medicaid Plans.
- October 8, 2014: Reviewed by IMPAC, renewed without changes.
- November 19, 2014: Reviewed by IMPAC, renewed without changes.
- September 2015: Branding and template change to distinguish Tufts Health Plan products in "Applies to" section. Added Tufts Health Freedom Plan products, effective January 1, 2016.
- November 16, 2015: Reviewed by IMPAC, renewed without changes
- November 9, 2016: Reviewed by IMPAC, renewed without changes
- December 14, 2016: Reviewed by IMPAC, renewed without changes
- July 2017: Added RITogether Plan product to template. For MNGs applicable to RITogether, effective date is August 1, 2017
- October 11, 2017: Reviewed by IMPAC, renewed without changes
- December 13, 2017: Reviewed by IMPAC, renewed without changes
- July 25, 2018: Reviewed by IMPAC, renewed without changes
- September 12, 2018: Reviewed by IMPAC, renewed without changes
- October 2018: Template and disclaimer updated
- July 17, 2019: Reviewed by IMPAC, renewed with no changes
- September 18, 2019: Reviewed by IMPAC, renewed without changes
- February 19, 2020: Reviewed at IMPAC. For effective date July 1, 2020, criteria clarified that testing covered without prior authorization applies to female partner, genetic testing for male reproductive partner requires prior authorization. Added MD geneticist to criteria regarding required documentation of counseling. Added to limitations section carrier screening for a specific genetic condition more than once in a member's lifetime. CPT code table of covered testing added.
- April 6, 2020: Fax number for Unify updated.
- June 17, 2020: Reviewed by IMPAC, renewed without changes.
- September 16, 2020: Reviewed by IMPAC, renewed without changes.

BACKGROUND, PRODUCT AND DISCLAIMER INFORMATION

Medical Necessity Guidelines are developed to determine coverage for benefits, and are published to provide a better understanding of the basis upon which coverage decisions are made. We make coverage decisions using these guidelines, along with the Member's benefit document, and in coordination with the Member's physician(s) on a case-by-case basis considering the individual Member's health care needs.

Medical Necessity Guidelines are developed for selected therapeutic or diagnostic services found to be safe and proven effective in a limited, defined population of patients or clinical circumstances. They include concise clinical coverage criteria based on current literature review, consultation with practicing physicians in our service area who are medical experts in the particular field, FDA and other government agency policies, and standards adopted by national accreditation organizations. We revise and update Medical Necessity Guidelines annually, or more frequently if new evidence becomes available that suggests needed revisions.

For self-insured plans, coverage may vary depending on the terms of the benefit document. If a discrepancy exists between a Medical Necessity Guideline and a self-insured Member's benefit document, the provisions of the benefit document will govern.

Treating providers are solely responsible for the medical advice and treatment of Members. The use of this guideline is not a guarantee of payment or a final prediction of how specific claim(s) will be adjudicated. Claims payment is subject to eligibility and benefits on the date of service, coordination of benefits, referral/authorization, utilization management guidelines when applicable, and adherence to plan policies, plan procedures, and claims editing logic.

[Provider Services](#)