Sentara® Health Plans

Medicare

Legacy Policy - Retired 12.31.2024. Use NCD/LCDS and Dex Registry.

Pre-conceptional, Prenatal, Preimplantation Genetic Testing

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Effective Date	8/2011
Next Review Date	N/A
Coverage Policy	Medical 34D
Version	1

Member-specific benefits take precedence over medical policy and benefits may vary across plans. Refer to the individual's benefit plan for details*.

Purpose:

This policy addresses genetic testing for pre-conception, prenatal and pre-implantation.

Description & Definitions:

Prenatal diagnosis or prenatal screening is testing for diseases or conditions in a fetus or embryo before it is born.

Preconceptual diagnosis for pregnancy planning and care in the form of genetic testing for members of reproductive age may be initiated.

Preimplantation genetic diagnosis (PGD or PIGD) (also known as embryo screening) refers to procedures that are performed on embryos prior to implantation, sometimes even on oocytes prior to fertilization.

Inheritest Carrier Screen: The Inheritest Carrier Screen offers a broad genetic screening option, providing genetic information regarding greater than 90 autosomal recessive inherited diseases found throughout the pan-ethnic US population, all in one simple test.

Cell-free fetal DNA-based prenatal screening for fetal aneuploidies, including but not limited to Trisomy 13 (Patau Syndrome), Trisomy 18 (Edwards Syndrome) and Trisomy 21 (Down syndrome) uses sequence analysis of cell-free fetal DNA in maternal plasma.

Autosomal recessive: A genetic condition that appears only in individuals who have received two copies of an autosomal gene, one copy from each parent. The gene is on an autosome, a nonsex chromosome. The parents are carriers who have only one copy of the gene and do not exhibit the trait because the gene is recessive to its normal counterpart gene

X-linked recessive inheritance - hereditary pattern in which a recessive gene on the X chromosome results in the manifestation of characteristics in male offspring and a carrier state in female offspring.

Criteria:

Preconceptional carrier status genetic testing for pregnancy planning for members of reproductive age OR **preimplantation genetic testing**, OR **prenatal genetic testing** to determine carrier status of a fetus, are medically necessary for **1 or more** of the following if criteria are met:

- **Ashkenazi Jewish** genetic panel testing may be indicated when **ALL** of the following are present:
  - Individual to be tested is of Ashkenazi Jewish ancestry and of reproductive age.
  - Panel testing is being ordered to assess for mutations associated with **3 or more** of the following diseases:
    - Bloom syndrome
    - Canavan disease
    - Cystic fibrosis
    - Dihydrolipoamide dehydrogenase deficiency
    - Familial dysautonomia (Riley-Day syndrome)
    - Familial hyperinsulinism
    - Fanconi anemia group C
    - Gaucher disease
    - Glycogen storage disease type 1A
    - Joubert syndrome 2
    - Maple syrup urine disease
    - Mucolipidosis IV
    - Nemaline myopathy
    - Niemann-Pick disease type A
    - Spinal muscle atrophy
    - Tay-Sachs disease
    - Usher syndrome type 1F
    - Usher syndrome type 3
- **Bloom syndrome** (BLM Gene) for **1 or more** of the following:
  - Preimplantation or prenatal genetic diagnosis when disease-causing mutation in BLM gene has been identified in both parents
  - Carrier testing for individual of Ashkenazi Jewish ancestry who is of reproductive age
  - Patient with family history of Bloom syndrome
  - Reproductive partner of BLM gene mutation carrier
  - Need to establish disease-causing mutation in patient with confirmed diagnosis
- **Canavan disease** (ASPA Gene) with **1 or more** of the following indications:
  - Preimplantation genetic diagnosis or Prenatal diagnosis, when disease-causing mutation in ASPA gene has been identified in both parents
  - Preconceptional or prenatal carrier testing when reproductive partner is an ASPA gene mutation carrier
  - Preconceptional, prenatal, or preimplantation testing if a member is of Ashkenazi Jewish ancestry and of reproductive age
  - The member has a family history of Canavan disease in first or second degree relative
  - Preconceptional or prenatal testing for a member with levels of urinary N-acetyl aspartic acid that are equivocal or indeterminate
- **Cystic Fibrosis** is medically necessary for **1 or more** of the following:
  - The Plan covers requests for common mutations with included in CPT codes 81221 (single mutation) or 81220 (common variants) endorsed by the American College of Medical Genetics (ACMG) for Cystic Fibrosis testing of members in **1 or more** of the following groups:
    - Couples seeking prenatal care;
    - Couples who are planning a pregnancy.
  - Extended CFTR mutation panels (Code 81222 and 81224) are approved for patients meeting **ANY** of the following 3 criteria (but not full sequencing, see exclusions):
    - Individuals with reproductive partners with cystic fibrosis or congenital absence of the vas deferens and no identified mutation with standard gene sequencing
    - Individuals with a family history of cystic fibrosis with no identified mutation on basic/standard gene sequencing,

        - Individuals with elevated or indeterminate sweat chloride levels where from zero to up to 2 mutations have been identified by basic/standard gene sequencing.
- **Duchene Muscular Dystrophy** for the **1 or more** of the following indications:
    - Preimplantation genetic diagnosis, when the DMD gene mutation in has been identified in mother
    - Prenatal diagnosis in fetus with 46, XY karyotype, when DMD gene mutation in has been identified in carrier mother or if linkage has been established suggesting mother is carrier
    - Carrier testing for asymptomatic female with family history of Duchenne muscular dystrophy, Becker muscular dystrophy, or DMD-associated dilated cardiomyopathy
    - Member with confirmed diagnosis of muscular dystrophy with need establish disease-causing mutation
- **Familial Dysautonomia (Riley-Day syndrome)** for **1 or more** of the following indication:
    - Preimplantation or prenatal genetic diagnosis when the IKBKAP gene mutation in has been identified in both parents
    - The member is of Ashkenazi Jewish ancestry and of reproductive age
    - The member has a family history (in a 1st or 2nd degree relative) of familial dysautonomia
    - The member is a reproductive partner of an individual that has been confirmed to be a IKBKAP gene mutation carrier
- **Familial Hemophagocytic Lymphohistiocytosis:** Covered if requested by name without criteria (there is no specific code)
- **Familial Mediterranean Fever (FMF)** for **1 or more** of the following:
    - Carrier testing for MEFV gene may be indicated when one of the following are present: Member is from an ethnic group at increased risk (eg, Armenian, Turkish, Arab, North African Jewish) and of reproductive age.
    - Prior to gamete donation if gamete recipient is known carrier
    - Reproductive partner of MEFV gene mutation carrier
- **Fanconi anemia group (FANC Gene)** for **1 or more** of the following indication:
    - Preimplantation or prenatal genetic test when the disease causing mutation has been found in both parents
    - Carrier testing for individual of Ashkenazi Jewish ancestry who is of reproductive age
    - The member has a family history of Fanconi anemia, and prior identification of disease-causing mutations in a first or second degree relative
    - Reproductive partner of FANC gene mutation carrier
    - Identification of disease-causing mutation in patient with confirmed diagnosis
    - Equivocal or indeterminate cytogenetic testing for chromosomal breakage or rearrangement in presence of DNA interstrand cross-linking agent (eg, diepoxybutane or mitomycin C);
    - Prior to gamete donation if gamete recipient is carrier;
- **Gaucher's disease** for **1 or more** of the following indication:
    - Preimplantation or prenatal genetic diagnosis for families in which disease-causing mutations have been identified in both parents or in previously affected child,
    - Carrier testing for individual of Ashkenazi Jewish ancestry
    - Carrier testing for preconception testing of partner of known carrier or affected individual
- **Glycogen storage disease (Maple syrup urine disease)** for **1 or more** of the following indication:
    - Preimplantation or prenatal genetic diagnosis when the G6PC or SLC37A4 gene mutation in has been identified in both parents.
    - The member is of Ashkenazi Jewish ancestry and of reproductive age
    - The member has a family history of glycogen storage disease type I
    - The member is a reproductive partner of an individual that has been confirmed to be a G6PC or SLC37A4 gene mutation carrier
- **Huntington's Disease** for 1 or more of the following indication:
    - Preimplantation or prenatal genetic diagnosis when the HTT gene disease-causing has been confirmed in one parent
    - Preimplantation or prenatal diagnostic testing for couples in at-risk family who do not wish to undergo presymptomatic mutation testing themselves
- **Karyotyping codes** along with 88230 and 88289 codes are covered without criteria or preauthorization.
- **Marfan Syndrome** for **1 or more** of the following indications:

- The use of Marfan syndrome gene testing in patients fulfilling the Ghent diagnostic criteria for the purpose of obtaining information for reproductive decision making or facilitating the diagnosis of Marfan syndrome in at-risk relatives;
- The prenatal diagnosis or preimplantation genetic testing for Marfan syndrome in the offspring of patients with known disease-causing

- **Mucolipidosis Type IV (MCOLN1)** for **1 or more** of the following indications
  - Preimplantation or Prenatal diagnosis genetic diagnosis, when disease-causing mutation in MCOLN1 gene has been identified in both parents
  - Carrier testing for an individual of Ashkenazi Jewish ancestry
  - The member has a family history of mucolipidosis IV in first or second degree relative
  - Reproductive partner of MCOLN1 gene mutation carrier
  - Need to establish disease-causing mutation in patient with confirmed diagnosis
- **Myotonic Dystrophy, DMPK and CNBP gene** testing for **1 or more** of the following indications:
  - Prenatal diagnosis when CNBP expansion has been identified in affected parent
  - Preimplantation, prenatal, or preconceptional genetic testing when a CNBP mutation has been identified in an affected first-degree relative
- **Nieman Pick Disease** for **1 or more** of the following indication:
  - Preimplantation or Prenatal genetic diagnosis, when disease-causing mutations, SMPD1 NPC1 or NPC2 gene have been identified in both parents or in a couple with previously affected child
  - Carrier testing for individual of Ashkenazi Jewish ancestry who is of reproductive age
  - Carrier testing for individual with family history of Niemann-Pick disease type A or type B
  - Establishment of disease-causing mutation in patient with confirmed diagnosis of Niemann-Pick disease type A or type B
- **Nonsyndromic Deafness:** Genes GJB2, GJB6, POU3F4, PRPS1, and SMPX. See Milliman guideline A-0823 Deafness and Hearing Loss, Nonsyndromic - Gene and Gene Panel Testing for criteria.
- **Neurofibromatosis Type1 and 2 (NF1/ NF2) gene** testing may be indicated for **1 or more** of the following Indications:
  - Preimplantation genetic diagnosis when NF1 or NF2 gene mutation has been identified in parent
  - Prenatal testing, when parent has NF1 or NF2 gene mutation, or linkage is established in family.
  - Carrier testing, when there is a first-degree relative with either NF1 or NF2.
- **Paraganglioma-Pheochromocytoma Syndromes**, Hereditary - SDHB, SDHC, SDHD, and TMEM127 Genes: See Milliman guideline Paraganglioma-Pheochromocytoma (Hereditary) - Gene Testing and Gene Panel A-0798 for criteria.
- **Retinoblastoma RB1 gene** testing may be indicated for **1 or more** of the following indication:
  - Preimplantation or prenatal genetic diagnosis for families RB1 mutation has been identified in either parent.
  - The member has a first degree relative with a known RB1 mutation
  - Prenatal diagnosis for pregnancies at increased risk when disease-causing allele of affected family member has been identified or linkage has been established in family
- **Spinal Muscular Atrophy (SMA)** testing of the SMN1 and SM2 genes is approved for carrier screening in prospective parents who wish to reproduce
- **Tay-Sach's disease** (HEXA gene) is approved for **1 or more** of the following:
  - Carrier testing for **1 or more** of the following:
    - Individual of Ashkenazi Jewish ancestry and of reproductive age
    - Individual with deficiency of beta-hexosaminidase A enzyme activity on carrier screening assay
    - Individual with family history of Tay-Sachs disease and of reproductive age, when both disease-causing mutations in HEXA gene have been identified in affected relative
    - Prior to gamete donation if gamete recipient is carrier
  - Confirmation of diagnosis of Tay-Sachs disease in symptomatic patient with inconclusive leukocyte or serum activity of beta-hexosaminidase A
  - Establishment of disease-causing mutation in patient with confirmed diagnosis of Tay-Sachs disease

- Preimplantation genetic diagnosis when disease-causing mutation in HEXA gene has been identified in both parents
- Prenatal diagnosis when disease-causing mutation in HEXA gene has been identified in both parents

- **Wiskott-Aldrich syndrome (WAS)** gene mutation testing **1 or more** of the following:
  - Individual is male with **All** of the following:
    - Initial testing points to a WAS related disorder (Wiskott Aldrich Syndrome, X linked thrombytopenia, X-linked congenital neutropenia)
  - Individual is female with **All** of the following:
    - There is a known family history of WAS gene mutation (testing is to identify female carriers)
  - Testing is prenatal with **All** of the following indications:
    - Fetus is male. Testing is being done with chorionic villi sampling or cultured amniocytes
    - There is known risk of WAS gene mutation (positive family history of WAS gene mutation and/or of known positive carrier females)
- **Preconceptional carrier status genetic testing** for pregnancy planning for members of reproductive age or prenatal genetic testing to determine **carrier status** of a fetus for **other** inherited disorders when **ALL** of the following criteria are met:
  - **Family history** genetic testing are considered **medically necessary** when **1 or more** of the following criteria is met:
    - An affected child is identified with either an autosomal recessive disorder, an x-linked disorder, or an inherited disorder with variable penetrance;
    - One or both parents or prospective parent(s) have another first or a second degree relative who is affected, or the first degree relative has an affected child, with either an autosomal recessive disorder, an x-linked disorder, or an inherited disorder with variable penetrance;
    - The parent or prospective parent is at high risk for a genetic disorder with a late onset presentation;
    - The parents or prospective parents are members of an ethnic group with a high risk of a specific genetic disorder with an autosomal recessive pattern of inheritance and testing is performed to determine carrier status and to guide subsequent reproductive decisions.
  - **Specific genetic testing** is considered medically necessary when **ALL** of the following criteria are met:
    - Testing is accompanied by genetic counseling.
    - Testing is for **1 or more** of the following:
      - A biochemical or other test is identified and the results are indeterminate
      - The genetic disorder cannot be identified through biochemical or other testing
  - **Approved Disorders** for targeted disease processes, Testing is for **1 or more** of the following:
    - 22q11.2 deletion syndromes (Velocardiofacial / DiGeorge syndrome, Catch22, Familial third and fourth pharyngeal pouch syndrome, Hypoplasia of thymus and parathyroid, Pharyngeal pouch syndrome, Sedlackova syndrome, Third and fourth pharyngeal pouch syndrome, Thymic aplasia syndrome, and Velofacial hypoplasia)
    - Adrenoleukodystrophy DNA Sequencing (ABCD1 gene) 8/23/16
    - Alpha Thalassemia, Beta Thalassemia and Sickle Cell
    - Autosomal recessive or autosomal dominant centronuclear myopathy (DNM2 and/or BIN1)
    - Congenital muscular dystrophy
    - Deficiency, Familial hyperinsulinism,
    - Dihydrolipoamide dehydrogenase
    - Ellis-van Creveld syndrome EVC/EVC2 gene
    - Emery-Dreifuss muscular dystrophy (EDMD1, 2, and 3) (FGFR2, Facioscapulohumeral muscular dystrophy (FSHMD1A)FGFR3)
    - Familial HEMOPHAGOCYTIC,
    - Familial hyperinsulinism
    - Familial Myotonic Dystrophy, (FMD)
    - Fascioscapulohumeral Muscular Dystrophy (FSHD)
    - Hemophilia A or B

        - Hereditary sensory and autonomic neuropathies
        - Inheritest Universal screening
        - Leopard syndrome
        - Limb girdle muscular dystrophy (LGMD1, LGMD2) (FKRP (Fukutin related protein))
        - Lissencephaly
        - LYMPHOHISTIOCYTOSIS (FHL),
        - Mucopolysaccharidosis (MPS)
        - Nemaline myopathy,
        - Noonan syndrome
        - Pontocerebellar Hypoplasia (TSEN54, EXOSC8)
        - Primary Ciliary Dyskinesia (PCD)
        - type 1C (MDC1C) (FKRP (Fukutin related protein))
        - Ullrich Muscular Dystrophy COL6A2
        - Usher syndrome type 1F or Usher syndrome type 3,
        - von Willebrand factor
        - Walker-Warburg syndrome (POMGNT1)
        - X-linked centronuclear myopathy (MTM1)
        - X-linked Lymphoproliferative Syndromes for transplant patients
- **Prenatal diagnosis testing** is covered for **1 or more** of the following:
    - Echogenic bowel is detected on ultrasound examination of fetus during pregnancy
    - VLDLR Associated Cerebellar Hypoplasia.
    - Disease-causing mutation in RYR1 (Malignant Hyperthermia) gene has been identified in one parent
- **Cell-free fetal DNA-based prenatal screening for common fetal aneuploidies:** Covered without precertification. (i.e., Trisomy 13 (Patau Syndrome), Trisomy 18 (Edwards Syndrome), and Trisomy 21 (Down syndrome)) (e.g., MaterniT21, Informaseq, Verifi)
- **Comparative Genomic Hybridization Microarray testing or Single Nucleotide Polymorphism (SNP) Chromosomal Microarray Analysis** for the evaluation of a fetus for **1 or more** of the following:
    - Evaluating abnormal fetal anatomic findings detected on fetal ultrasound or fetal magnetic resonance imaging which are characteristic of a genetic abnormality;
    - Women undergoing invasive prenatal diagnostic testing (i.e. amniocentesis, chorionic villus sampling or fetal tissue sampling).
    - Evaluation of recurrent pregnancy loss, after the second consecutive loss
    - Evaluation of intrauterine fetal demise (IUFD) or stillbirth after 20 weeks of gestational age
    - Evaluation of a pregnancy loss with one or more major structural anomalies
- **Preimplantation genetic diagnosis** when **1 or more** of the following:
    - used to determine the sex of an embryo only when there is a documented history of an X-linked disorder, such that deselection of an affected embryo can be made on the basis of sex alone.
    - used to evaluate human leukocyte antigen (HLA) status alone is in families with a child with a bone marrow disorder requiring a stem cell transplant, and in whom there is no other source of a compatible bone marrow donor other than an HLA matched sibling.
    - used as a technique to improve the implantation rate of in vitro fertilization (IVF) procedures in otherwise infertile couples, when **1 or more** of the following below are met:
        - Three prior failed attempts at IVF;
        - One of the partners is known to harbor a balanced translocation
    - used to deselect embryos with genetic mutations in partners who meet any criteria in **All** of the following:
        - Must meet at **1 or more** of the following:
            - Both partners are known carriers of the same autosomal recessive disorder;
            - One partner is a known carrier of an autosomal recessive disorder, and the couple has previously produced offspring affected by that disorder;
            - One partner is a known carrier of a single gene autosomal dominant disorder;
            - One partner is a known carrier of a single X-linked disorder;
        - Must meet **ALL** of the following:
            - A specific mutation, or set of mutations, has been identified, that specifically identifies the genetic disorder with a high degree of reliability;

- The genetic disorder is associated with severe disability or has a lethal natural history
- Testing is accompanied by genetic counseling.

Pre-conception, prenatal and pre-implantation genetic testing is considered **not medically necessary** for any use other than those indicated in clinical criteria, to include but not limited to:

- Ambry's Cystic Fibrosis 508 First and reflex testing if negative to Cystic Fibrosis Full Gene Sequencing Analysis or Cystic Fibrosis Amplified genetic testing
- Any other test not listed above as covered is considered among those that are not medically necessary.
- APOE 4 or other Genetic Testing for Alzheimer's Disease (S3852)
- CFTR Full Gene Sequencing (81223)
- Comparative genomic hybridization microarray testing and single nucleotide polymorphism (SNP) chromosomal microarray analysis are unproven and not medically necessary for preimplantation genetic diagnosis or screening in embryos.
- EPG5 Gene-VICI Syndrome testing
- EpiSign Complete
- Exome Sequence Analysis (CPT 81415, 81416, 81417)
- FGFR3 mutation Achondroplasia (Dwarfism)
- Genetic disease carrier panel testing for multiple heritable disorders in the general population is considered not medically necessary if the genetic testing includes genes not recommended for routine preconceptional or prenatal screening by the American College of Medical Genetics and the American College of Obstetricians and Gynecologists (e.g. HerediT, Inheritest, NxGen MDx Super Panel/Universal Panels)
- Genetic testing related to seizure disorders
- Genome Sequence Analysis (CPT 81425, 81426, 81427)
- GPR56 gene for polymicrogyria
- Hereditary Retinal Disorders Genetic Panel Lab Test
- Holoprosencephaly, schizencephaly & craniosynostosis genetic testing including, but not limited to genes SHH, ZIC2, SIX3, and TGIF1
- Human leukocyte antigen (HLA) typing of an embryo to identify a future suitable stem cell, tissue or organ transplantation donor
- Hyperimmunoglobulin D syndrome (HIDS)
- JAG1 and/or NOTCH2 testing for Alagille Syndrome
- KCNC2 or KCNC3
- Mevalonate kinase deficiency (MKD)
- MTHFR
- MYH8 Gene(Trismus-pseudocamptodactyly syndrome)
- Repeat/Duplicative genetic testing
- Routine requests for cell-free prenatal genetic testing beyond fetal trisomies 21, 18, and 13 are considered not medically necessary (e.g., microdeletion testing, MaterniT21 Plus, InformaSeq with Y analysis, InformaSeq with XY analysis).
- Signature Precision Panel™ | Prenatal
- SPRED1 (sprout-related, EVH1 domain containing 1) (eg, Legius syndrome)
- Tumor necrosis factor receptor-1 associated periodic syndrome (TRAPS) genetic testing
- Uniparental Disomy
- Whole Exome Sequencing (WES)
- Whole Genome Sequencing (WGS)
- Y Chromosome Microdeletion Analysis

Coding:

Medically necessary with criteria:

Coding	Description

81130	SMPD1(sphingomyelin phosphodiesterase 1, acid lysosomal) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330)
81161	DMD (dystrophin) (eg, Duchenne/Becker muscular dystrophy) deletion analysis, and duplication analysis, if performed
81187	CNBP (CCHC-type zinc finger nucleic acid binding protein) (eg, myotonic dystrophy type 2) gene analysis, evaluation to detect abnormal (eg, expanded) alleles
81200	ASPA (aspartoacylase) (eg, Canavan disease) gene analysis, common variants (eg, E285A, Y231X)
81209	BLM (Bloom syndrome, RecQ helicase-like) (eg, Bloom syndrome) gene analysis, 2281del6ins7 variant
81220	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)
81221	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; known familial variants
81222	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; duplication/deletion variants
81224	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; intron 8 poly-T analysis (eg, male infertility)
81228	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number variants, comparative genomic hybridization [CGH] microarray analysis
81229	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants, comparative genomic hybridization (CGH) microarray analysis
81234	DMPK (DM1 protein kinase) (eg, myotonic dystrophy type 1) gene analysis; evaluation to detect abnormal (expanded) alleles
81239	DMPK (DM1 protein kinase) (eg, myotonic dystrophy type 1) gene analysis; characterization of alleles (eg, expanded size)
81242	FANCC (Fanconi anemia, complementation group C) (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A>T)
81250	G6PC (glucose-6-phosphatase, catalytic subunit) (eg, Glycogen storage disease, type 1a, von Gierke disease) gene analysis, common variants (eg, R83C, Q347X)
81251	GBA (glucosidase, beta, acid) (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+1G>A)
81252	GJB2 (gap junction protein, beta 2, 26kDa, connexin 26) (eg, nonsyndromic hearing loss) gene analysis; full gene sequence
81253	GJB2 (gap junction protein, beta 2, 26kDa, connexin 26) (eg, nonsyndromic hearing loss) gene analysis; known familial variants
81254	GJB6 (gap junction protein, beta 6, 30kDa, connexin 30) (eg, nonsyndromic hearing loss) gene analysis, common variants (eg, 309kb [del(GJB6-D13S1830)] and 232kb [del(GJB6-D13S1854)])
81255	HEXA (hexosaminidase A [alpha polypeptide]) (eg, Tay-Sachs disease) gene analysis, common variants (eg, 1278insTATC, 1421+1G>C, G269S)
81257	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; common deletions or variant (eg, Southeast Asian, Thai, Filipino, Mediterranean, alpha3.7, alpha4.2, alpha20.5, Constant Spring)

81258	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; known familial variant
81259	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; full gene sequence
81260	IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (eg, familial dysautonomia) gene analysis, common variants (eg, 2507+6T>C, R696P)
81261	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); common variant(s) (eg, HbS, HbC, HbE)
81269	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (eg, alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis; duplication/deletion variants
81271	HTT (huntingtin) (eg, Huntington disease) gene analysis; evaluation to detect abnormal (eg, expanded) alleles
81272	HTT (huntingtin) (eg, Huntington disease) gene analysis; characterization of alleles (eg, expanded size)
81290	MCOLN1 (mucolipin 1) (eg, Mucolipidosis, type IV) gene analysis, common variants (eg, IVS3-2A>G, del6.4kb)
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)
81362	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); known familial variant(s)
81363	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); duplication/deletion variant(s)
81364	HBB (hemoglobin, subunit beta) (eg, sickle cell anemia, beta thalassemia, hemoglobinopathy); full gene sequence
81401	Molecular pathology procedure, Level 2 (eg, 2-10 SNPs, 1 methylated variant, or 1 somatic variant [typically using nonsequencing target variant analysis], or detection of a dynamic mutation disorder/triplet repeat)
81402	Molecular pathology procedure, Level 3 (eg, >10 SNPs, 2-10 methylated variants, or 2-10 somatic variants [typically using non-sequencing target variant analysis], immunoglobulin and T-cell receptor gene rearrangements, duplication/deletion variants of 1 exon, loss of heterozygosity [LOH], uniparental disomy [UPD])
81403	Molecular pathology procedure, Level 4 (eg, analysis of single exon by DNA sequence analysis, analysis of >10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)
81404	Molecular pathology procedure, Level 5 (eg, analysis of 2-5 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 6-10 exons, or characterization of a dynamic mutation disorder/triplet repeat by Southern blot analysis)
81406	Molecular pathology procedure, Level 7 (eg, analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons)

81407	Molecular pathology procedure, Level 8 (eg, analysis of 26-50 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of >50 exons, sequence analysis of multiple genes on one platform)
81408	Molecular pathology procedure, Level 9 (eg, analysis of >50 exons in a single gene by DNA sequence analysis)
81412	Ashkenazi Jewish associated disorders (eg, 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
81430	Hearing loss (eg, nonsyndromic hearing loss, Usher syndrome, Pendred syndrome); genomic sequence analysis panel, must include sequencing of at least 60 genes, including CDH23, CLRN1, GJB2, GPR98, MTRNR1, MYO7A, MYO15A, PCDH15, OTOF, SLC26A4, TMC1, TMPRSS3, USH1C, USH1G, USH2A, and WFS1
81431	Hearing loss (eg, nonsyndromic hearing loss, Usher syndrome, Pendred syndrome); duplication/deletion analysis panel, must include copy number analyses for STRC and DFNB1 deletions in GJB2 and GJB6 genes
81443	Genetic testing for severe inherited conditions (eg, cystic fibrosis, Ashkenazi Jewish-associated disorders [eg, Bloom syndrome, Canavan disease, Fanconi anemia type C, mucolipidosis type VI, Gaucher disease, Tay-Sachs disease], beta hemoglobinopathies, phenylketonuria, galactosemia), genomic sequence analysis panel, must include sequencing of at least 15 genes (eg, ACADM, ARSA, ASPA, ATP7B, BCKDHA, BCKDHB, BLM, CFTR, DHCR7, FANCC, G6PC, GAA, GALT, GBA, GBE1, HBB, HEXA, IKBKAP, MCOLN1, PAH)
81479	Unlisted molecular pathology procedure
83080	b-Hexosaminidase, each assay
84999	Unlisted chemistry procedure
88230	Tissue culture for non-neoplastic disorders; lymphocyte
89290	Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); less than or equal to 5 embryos
89291	Biopsy, oocyte polar body or embryo blastomere, microtechnique (for pre-implantation genetic diagnosis); greater than 5 embryos
S3841	Genetic testing for retinoblastoma
S3844	DNA analysis of the connexin 26 gene (GJB2) for susceptibility to congenital, profound deafness
S3845	Genetic testing for alpha-thalassemia
S3846	Genetic testing for hemoglobin E beta-thalassemia
S3849	Genetic testing for Niemann-Pick disease
S3850	Genetic testing for sickle cell anemia

S3853	Genetic testing for myotonic muscular dystrophy

Considered Not Medically Necessary:

Coding	Description
81223	CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; full gene sequence
81241	F5 (coagulation factor V) (eg, hereditary hypercoagulability) gene analysis, Leiden variant
81331	SNRPN/UBE3A (small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A) (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis
81415	Exome (eg, unexplained constitutional or heritable disorder or syndrome); sequence analysis
81416	Exome (eg, unexplained constitutional or heritable disorder or syndrome); sequence analysis, each comparator exome (eg, parents, siblings) (List separately in addition to code for primary procedure)
81417	Exome (eg, unexplained constitutional or heritable disorder or syndrome); re-evaluation of previously obtained exome sequence (eg, updated knowledge or unrelated condition/syndrome)
81425	Genome (eg, unexplained constitutional or heritable disorder or syndrome); sequence analysis
81426	Genome (eg, unexplained constitutional or heritable disorder or syndrome); sequence analysis, each comparator genome (eg, parents, siblings) (List separately in addition to code for primary procedure)
81427	Genome (eg, unexplained constitutional or heritable disorder or syndrome); re-evaluation of previously obtained genome sequence (eg, updated knowledge or unrelated condition/syndrome)
81434	Hereditary retinal disorders (eg, retinitis pigmentosa, Leber congenital amaurosis, cone-rod dystrophy), genomic sequence analysis panel, must include sequencing of at least 15 genes, including ABCA4, CNGA1, CRB1, EYS, PDE6A, PDE6B, PRPF31, PRPH2, RDH12, RHO, RP1, RP2, RPE65, RPGR, and USH2A
0318U	Pediatrics (congenital epigenetic disorders), whole genome methylation analysis by microarray for 50 or more genes, blood
S3842	Genetic testing for Von Hippel-Lindau disease
S3852	DNA analysis for APOE epsilon 4 allele for susceptibility to Alzheimer's disease

U.S. Food and Drug Administration (FDA) - approved only products only.

Document History:

Revised Dates:

- 2022: July
- 2022: March
- 2021: April
- 2020: June
- 2020: January
- 2019: December
- 2019: October
- 2018: March
- 2016: August
- 2016: June
- 2016: May
- 2016: April
- 2016: March
- 2016: February
- 2016: January
- 2015: December
- 2015: November
- 2015: October
- 2015: September
- 2015: August
- 2015: July

- 2015: June
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- 2014: July
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- 2014: May
- 2014: March
- 2014: January
- 2013: December
- 2013: October
- 2013: July
- 2013: April
- 2013: February
- 2012: November
- 2012: October
- 2012: July

Reviewed Dates:

- 2021: February
- 2020: December
- 2019: October
- 2017: April
- 2016: April
- 2015: November
- 2014: January
- 2011: July

Effective Date:

- August 2011

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Special Notes: *

This medical policy expresses Sentara Health Plan's determination of medically necessity of services, and they are based upon a review of currently available clinical information. Medical policies are not a substitute for clinical judgment or for any prior authorization requirements of the health plan. These policies are not an explanation of benefits.

Medical policies can be highly technical and complex and are provided here for informational purposes. These medical policies are intended for use by health care professionals. The medical policies do not constitute medical advice or medical care. Treating health care professionals are solely responsible for diagnosis, treatment and medical advice. Sentara Health Plan members should discuss the information in the medical policies with their treating health care professionals. Medical technology is constantly evolving and these medical policies are subject to change without notice,

although Sentara Health Plan will notify providers as required in advance of changes that could have a negative impact on benefits.

Keywords:
22q11.2 deletion syndrome, Adrenoleukodystrophy DNA Sequencing, Alpha Thalassemia, Beta Thalassemia and Sickle Cell, Analysis, Ashkenazi Jewish, Autosomal recessive or autosomal dominant centronuclear myopathy, Bloom syndrome, Canavan disease, Cell-free fetal DNA-based, chromosomal microarray analysis, Congenital muscular dystrophy, Cystic Fibrosis, Deficiency, Familial hyperinsulinism, Dihydrolipoamide dehydrogenase, disease carrier, Disease-causing mutation, Duchene Muscular Dystrophy, Duplicative, Echogenic bowel, Ellis-van Creveld syndrome, Emery-Dreifuss muscular dystrophy, Facioscapulohumeral muscular dystrophy, Familial Dysautonomia, Familial HEMOPHAGOCYTIC, Familial Hemophagocytic Lymphohistiocytosis, Familial hyperinsulinism, Familial Mediterranean Fever, Familial Myotonic Dystrophy, Fanconi anemia group, Fascioscapulohumeral Muscular Dystrophy, Gaucher's disease, Gene Sequencing, Genetic testing, Glycogen storage disease, Hemophilia A or B, Hereditary sensory and autonomic neuropathies, Huntington's Disease, Hybridization, Inheritest Universal screening, Leopard syndrome, Limb girdle muscular dystrophy, Lissencephaly, LYMPHOHISTIOCYTOSIS, Maple syrup urine disease, Marfan Syndrome, MTHFR, Mucolipidosis Type IV, Mucopolysaccharidosis, Myotonic Dystrophy, Nemaline myopathy, Nieman Pick Disease, Nonsyndromic Deafness, Noonan syndrome, Paraganglioma-Pheochromocytoma Syndromes, Pontocerebellar Hypoplasia, Pre-conception, pre-implantation, Prenatal, Primary Ciliary Dyskinesia, Repeat, Retinoblastoma, Riley-Day syndrome, single nucleotide polymorphism (SNP), Spinal Muscular Atrophy, Syndrome testing, Tay-Sach's disease, Ullrich Muscular Dystrophy COL6A2, Usher syndrome, VLDLR Associated Cerebellar Hypoplasia, von Willebrand factor, Walker-Warburg syndrome, Whole Exome Sequencing (WES), Wiskott-Aldrich syndrome, X-linked centronuclear myopathy, X-linked Lymphoproliferative Syndromes