UnitedHealthcare Prior Authorization Test List - Tests Previously Ordered

Highlighted tests: Frequently ordered by Pathologists

Please Note: This list is based on order history. It is not meant to be fully inclusive of all tests that may require prior authorization.

Soft code ARUP Test Number	Molecular and Genetic Pre-authorization Test Description
2007228	**5-FU Toxicity, Chemotherapeutic Response, 5 Mutations** Predict risk of dose-related toxicity and responsiveness to 5-FU therapy
2011622	**Alpha Globin (HBA1 and HBA2) Deletion/Duplication** Preferred first-tier genetic test for confirmation of suspected alpha thalassemia or alpha thalassemia trait. Detect common, rare, and novel deletions or duplications of the alpha globin gene cluster H.
0051495	**Alpha Thalassemia (HBA1 and HBA2) 7 Deletions** Acceptable first-tier genetic test for confirmation of suspected alpha thalassemia or alpha thalassemia trait. Assesses for seven common alpha globin gene deletions.
0051256	**Alpha-1-Antitrypsin (SERPINA1) Enzyme Concentration and 2 Mutations with Reflex to Alpha-1-Antitrypsin Phenotype** Preferred test to identifiy alpha-1-antitrypsin deficiency and causative DNA and protein variants.
2005077	**Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR** Preferred initial diagnostic test for Angelman syndrome or Prader-Willi syndrome.
2013341	**Apolipoprotein E (APOE) Genotyping, Alzheimer Disease Risk** Supports a clinical diagnosis of Alzheimer disease (AD) in symptomatic individuals. Use for AD risk assessment only. Genetic counseling and informed consent are strongly recommended prior to ordering and post test to discuss results.
2013337	**Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk** Provides supporting evidence for a diagnosis of type III hyperlipoproteinemia for evaluation of premature coronary heart disease.
BCPCR 2006193	**B-Cell Clonality Screening (IgH and IgK) by PCR** Aid in diagnosis and monitoring of B-cell lymphoproliferative disorders and in differentiating malignant from reactive lymphoid proliferations.
2008420	**BCR-ABL1 Mutation Analysis for Tyrosine Kinase Inhibitor Resistance by Next Generation Sequencing** Order only for patients with an established diagnosis of a BCR-ABL1 positive leukemia. This test is used to determine if a mutation is present that would interfere with response to TKI therapy in Philadelphia chromosome positive (Ph+) lymphoblastic leukemia or chronic myelogenous leukemia (CML). The test detects all common mutations, including T315I. For initial BCR-ABL1 testing, refer to BCR-ABL1, Qualitative with Reflex to BCR-ABL1 Quantitative (2005010)
BCAMJ 2005017	**BCR-ABL1, Major (p210), Quantitative** This quantitative test is appropriate for diagnosis and therapeutic monitoring for CML or ALL. The BCR-ABL1 major (p210) fusion forms are present in almost all cases of CML and in a small subset of cases of ALL.
BCAMN 2005016	**BCR-ABL1, Minor (p190), Quantitative** Order in cases of Philadelphia chromosome positive (Ph+) lymphoblastic leukemia to quantify the BCR-ABL1 p190 fusion form. For CML, use BCR-ABL1, Major (p210), Quantitative, (2005017).
BCRRX 2005010	**BCR-ABL1, Qualitative with Quantitative Reflex** For use in detecting the presence of a BCR-ABL1 fusion, for determining breakpoint status, and for baseline quantitation.
0051730	**Biotinidase Deficiency (BTD) Sequencing** Molecular DNA test to confirm a diagnosis of biotinidase (BTD) deficiency when biotinidase enzymatic activity is low. To diagnose or rule out BTD deficiency, refer to Biotinidase, Serum (with Paired Normal Control)(0093362)
0093362	**Biotinidase, Serum (with Paired Normal Control)** Initial biotinidase enzyme test to diagnose or rule out biotinidase deficiency.
0051750	**BRAF Codon 600 Mutation Detection with Reflex to MLH1 Promoter Methylation** Recommended reflex test to differentiate between Lynch syndrome and sporadic colorectal cancer in tumors showing loss of MLH1
BRAV6 2002498	**BRAF V600 Mutation (non-melanoma/colorectal)** Detect activating BRAF mutations at codon 600, which can indicate responsiveness to BRAF inhibitors in melanomas or resistance to anti-EGFR therapy in colorectal cancer. Useful in assessing prognosis of certain thyroid cancers. Can also be used within the Lynch syndrome reflex testing pathway (for colorectal cancer specimens only).
BRAF	**BRAF V600 Mutation by PCR (melanoma/colorectal)** BRAF is a human gene that makes a protein called B-Raf. The BRAF gene belongs to a class of genes known as oncogenes. When mutated, oncogenes have the potential to cause normal cells to become cancerous. BRAF by PCR is indicated for identifying melanoma cancers that may respond to treatment with Vemurafenib.
BRCAS 2011949	**BRCA1 and BRCA2 Sequencing and Del/Dup (HBOC)** Detect activating BRAF mutations at codon 600, which can indicate responsiveness to BRAF inhibitors in melanomas or resistance to anti-EGFR therapy in colorectal cancer. Useful in assessing prognosis of certain thyroid cancers. Can also be used within the Lynch syndrome reflex testing pathway (for colorectal cancer specimens only).
2010183	**Cardiomyopathy/Arrhythmia Sequencing and Deletion/Duplication** Preferred test to assess for hereditary form of cardiomyopathy or arrhythmia.
CS137	**Carrier Scr-137 Conditions** This panel is suitable for patients of all ethnicities **except** those with Jewish ancestry
CS27	**Carrier Scr-27 Conditions** This panel is suitable for patients with any ethnic background
CS274	**Carrier Scr-274 Conditions** This panel is suitable for patients of any ethnic background OR Jewish patients who prefer more coverage than the Carrier Screen, 106 Conditions
CS4	**Carrier Scr-4 Conditions** This panel is suitable for patients of any ethnic background
2004247	**CEBPA Mutation Detection** Initial test for prognostication of CN-AML.
CELGP 2005018	**Celiac (HLA-DQ2 and HLA-DQ8) Disease Genotyping** Do not use in the initial evaluation for celiac disease. Useful in ruling out celiac disease in selective clinical situations, such as equivocal small bowel histologic finding or individuals on a gluten-free diet not tested for celiac disease prior to starting the diet

2012155	**Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, PMP22 Deletion/Duplication with Reflex to Sequencing Panel** Recommended initial test to confirm a suspected diagnosis of CMT1 or CMT1A. PMP22 gene deletion/duplication analysis is performed first. If negative or inconclusive, testing reflexes to sequencing of 78 genes related to CMT and hereditary neuropathies
2002066	**Chimerism Post-Transplant** Monitor engraftment of donor cells post allogenic stem cell transplantation.
2002065	**Chimerism Recipient Pre-Transplant** Assess recipient genotype.
CRCGP 2011616	**Colon Cancer Gene Panel** Indicated for individuals with metastatic colorectal cancer to guide treatment with anti-EGFR monoclonal antibodies (ie, cetuximab and panitumumab). Detects mutations in BRAF, KRAS, NRAS, extended KRAS, and PIK3CA.
0051374	**Connexin 26 (GJB2), Full Gene Sequencing** Diagnostic testing or carrier screening for GJB2-related nonsyndromic hearing loss (NHSL). May be used as first-tier genetic test for individuals with NSHL.
2013661	**Cystic Fibrosis (CFTR) 165 Pathogenic Variants** Carrier screening for expectant individuals and those planning a pregnancy AND diagnostic testing for individuals with symptoms of classic CF.
CFBR3	**Cystic Fibrosis Carrier Screen** This is a qualitative genotyping test to be used for CF carrier screening as recommended by the American College of Medical Genetics (ACMG). The panel includes testing and interpretation of 23 mutations and variants plus a reflex test for the 5/7/9T polymorphism as recommended by the ACMG
C2C19 2012769	**Cytochrome P450 2C19, CYP2C19 - 9 Variants** Assess genetic risk of abnormal drug metabolism for drugs metabolized by CYP2C19. May aid in drug selection and dose planning for drugs metabolized by CYP2C19.
CP2D6 2014547	**Cytochrome P450 2D6 (CYP2D6)** Assess genetic risk of abnormal drug metabolism for drugs metabolized by CYP2D6. May aid in drug selection and dose planning for drugs metabolized by CYP2D6.
2013098	**Cytochrome P450 Genotype Panel** Assess genetic risk of abnormal drug metabolism for drugs metabolized by CYP2D6, CYP2C9, CYP2C19, and CYP3A5. May aid in drug selection and dose planning for many drugs.
2010795	**Cytogenomic Molecular Inversion Probe Array, FFPE Tissue – Products of Conception** For detection of copy number alterations and loss of heterozygosity in FFPE specimens from products of conception.
SNP5C 2009353	**Cytogenomic SNP Array w/Five-Cell Study, Peripheral Blood** Useful when chromosome and array tests would otherwise have been ordered concurrently. Assay may identify cytogenetically visible rearrangements and provide information regarding mechanism of gains and losses.
2002366	**Cytogenomic SNP Microarray - Fetal** • Diagnostic test designed to identify genomic abnormalities (eg, aneuploidy and microdeletions). • Performed on direct or cultured amniotic fluid and chorionic villus sampling (CVS) specimens.
CSNPM 2003414	**Cytogenomic SNP Microarray** Preferred first-tier test for developmental delay, multiple anomalies, and autism-spectrum disorders. Testing is performed on peripheral blood.
2012166	**Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants** Predict risk of dose-related toxicity to 5-FU therapy.
0051463	**Dysautonomia, Familial (IKBKAP), 2 Variants** Carrier screening or diagnostic testing for familial dysautonomia for individuals of Ashkenazi Jewish descent.
EGFRM	**EGFR Mutation by PCR** EGFR by PCR is indicated for identifying non-small cell lung cancers that may respond to epidermal growth factor receptor tyrosine kinase inhibitor therapy
2002440	**EGFR Mutation Detection by Pyrosequencing** Predicts response to tyrosine kinase inhibitor (TKI) therapy.
EGFRR	**EGFR Mutation with Reflex to ALK** Indicated for use in identifying non-small cell lung cancers that may benefit from treatment with epidermal growth factor receptor tyrosine kinase or anaplastic lumphoma kinase inhibitors
2012868	**EGFR T790M Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR** Monitor for development of EGFR T790M drug-resistant mutation in patients administered tyrosine kinase inhibitor (TKI) therapy for EGFR-mutant non-small cell lung cancer. Monitor response to therapy and disease progression in patients treated with EGFR T790M-specific TKIs.
EPPRC 2013906	**Epi proColon** The Epi proColon test is indicated to screen adults of either sex, 50 years or older, defined as average risk for CRC, who have been offered and have a history of not completing CRC screening. Tests that are available and recommended in the USPSTF 2008 CRC screening guidelines should be offered and declined prior to offering the Epi proColon test. Patients with a positive Epi proColon test result should be referred for diagnostic colonoscopy. The Epi proColon test results should be used in combination with physician's assessment and individual risk factors in guiding patient management
FACTV	**Factor V Leiden (DNA)** This test aids in the evaluation of the risk for venous thrombosis. The Xpert® Factor II & Factor V Assay is a qualitative in vitro diagnostic genotyping test for the detection of Factor II and Factor V alleles from sodium citrate or EDTA anticoagulated whole blood. The test is performed on the Cepheid GeneXpert® Dx System. This test is intended to provide results for Factor II (G20210A)) and Factor V (G1691A) Leiden mutations as an aid in the diagnosis in individuals with suspected thrombophilia. The association of Factor II and Factor V Leiden mutations with an increased risk for venous thrombosis has been well documented. The Factor II or Prothrombin mutation refers to the G to A transition at nucleotide in the untranslated region of the gene and is associated with increased plasma levels of prothrombin. Factor V Leiden (G1691A) refers to the G to A transition at nucleotide position 1691 of the Factor V gene, resulting in the substitution of the amino acid arginine by glutamine in the Factor V protein, causing resistance to cleavage by Activated Protein C (APC). Factor II (G20210A) and Factor V Leiden (G1691A) mutations are present in 2% and 5% of the general population, respectively.
0097720	**Factor V Leiden (F5) R506Q Mutation** Order to detect factor V Leiden variant.
2004863	**Familial Adenomatous Polyposis (APC) Sequencing** Acceptable diagnostic or predictive test for familial adenomatous polyposis. For classic FAP, consider Familial Adenomatous Polyposis Panel: (APC) Sequencing and Deletion/Duplication, (MUTYH) 2 Mutations (2004915). Whole Blood.
2004915	**Familial Adenomatous Polyposis Panel: (APC) Sequending and Deletion/Duplicaiton, (MUTYH)2 Mutations.** Preferred diagnostic or predictive test for familial adenomatous polyposis and MUTYH-associated polyposis. Whole Blood

2002658	**Familial Mediterranean Fever (MEFV) Sequencing** Preferred test for suspected familial Mediterranean fever.
2005400	**FLT3 Mutation Detection by PCR - INACTVE AFTER 11/13/2017 Replacement: See LeukoStrat CDx FLT3 (2014683)** First-line test for prognostication of CN-AML.
FMR1 2009033	**Fragile X (FMR1) w/Reflex to Methylation** Preferred test to diagnose fragile X syndrome and carrier screening in individuals with a positive family history.
0051175	**Galactosemia (GALT) Enzyme Activity and 9 Mutations** Initial test to diagnose or rule out classic galactosemia. Recommended carrier testing for galactosemia.
2013449	**Gastrointestinal Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 16 Genes** Confirm a diagnosis of hereditary gastrointestinal (GI) cancer in individuals with a personal or family history of GI cancer and/or polyposis.
2002674	**Gastrointestinal Stromal Tumor Mutation** Detect activating mutations in KIT and PDGFRA. Predict response to tyrosine kinase inhibitor (TKI) therapy.
0051438	**Gaucher Disease (GBA), 8 Variants** Carrier screening or diagnostic testing for Gaucher disease for individuals of Ashkenazi Jewish descent.
2005633	**Genomic SNP Microarray, Products of Conception** Use for intrauterine fetal demise or stillbirth when further cytogenetic analysis is desired, pregnancy loss or termination in the presence of fetal anomalies, further characterization of fetal chromosomal abnormalities seen by conventional cytogenetic methods, multiple fetal losses of unknown etiology, or POC samples that fail to grow in culture.
HMCH3 0055656	**Hemochromatosis (HFE) DNA Mutations** Confirm clinical diagnosis of hereditary hemochromatosis (HH) in an individual with biochemical findings of iron overload. Screen adult family members of individuals with known HH. Test reproductive partner of an individual with HH for carrier status. Not recommended for initial hemochromatosis testing
2001614	**Hemophilia A (F8) 2 Inversions with Reflex to Sequencing and Reflex to Deletion/Duplication** Detect causal F8 variant in individuals with established severe hemophilia A and determine carrier status in at-risk females with severely affected male relatives. For mild to moderate hemophilia A, Hemophilia A (F8) sequencing (2001747) is preferred.
2001747	**Hemophilia A (F8) Sequencing** Identify causal F8 variant in individuals with established mild to moderate hemophilia A and determine carrier status for those with a family history of mild to moderate hemophilia A. For severe hemophilia A, Hemophilia A (F8) 2 Inversions with Reflex to Sequencing and Reflex to Deletion/Duplication (2001614) is recommended.
0051381	**Hereditary Hemorrhagic Telangiectasia (ACVRL1 and ENG) Sequencing** Alternate test when clinical/family history is classic for HHT, but this test does not detect large duplications/deletions.
2007167	**Hereditary Paraganglioma-Pheochromocytoma,HPCG-PCC (SDHB, SDHC, and SDHD) Sequencing and Deletion/Duplication Panel** Preferred initial test when hereditary paraganglioma-pheochromocytoma is suspected.
2011264	**HLA Class I Panel (ABC) by NGS** Test is intended for pre-transplant allele matching. Do not use for specific disease screening or diagnosis (eg, celiac disease, rheumatologic diseases).
0051067	**HLA DRB3, 4, 5** May be useful in immunization/vaccination trials or may aid in the clinical diagnosis of diseases strongly associated with the HLA-DRB 3*, 4*, 5* loci.
HLB57 2002429	**HLA-B*57:01 for Abacavir Sensitivity** Standard of care prior to abacavir therapy per FDA. Predict risk of abacavir hypersensitivity syndrome. Relevant to most populations
2014079	**HLA-DQ Genotyping** May be useful in immunization/vaccination trials or may aid in the clinical diagnosis of diseases strongly associated with the HLA-DQ locus. To rule out celiac disease (when other testing is inconclusive), refer to Celiac Disease (HLA-DQ2 and HLA-DQ8) Genotyping (2005018). When narcolepsy is suspected in symptomatic individuals, refer to Narcolepsy (HLA-DQB1*06:02) Genotyping (2005023).
0040018	**Huntington Disease Mutation by PCR** Diagnostic confirmation for Huntington disease (HD) in a symptomatic individual. Presymptomatic testing for adults with a family history of HD.
2006444	**IDH1 and IDH2 Mutation Analysis, exon 4** Detect IDH1 and IDH2 mutations in whole blood or bone marrow. May have prognostic significance in patients with hematologic malignancies, depending on the clinical and genetic context. For FFPE tissues, use IDH1 and IDH2 Mutation Analysis, Exon 4, Formalin-Fixed, Paraffin-Embedded (FFPE) Tissue (2014188).
2014188	**IDH1 and IDH2 Mutation Analysis, Exon 4, Formalin-Fixed, Paraffin-Embedded (FFPE) Tissue** IDH1/IDH2 mutational status is a prognostic marker in individuals with low- and high-grade gliomas. Aid in distinguishing a primary from a secondary glioblastoma
IGHVS 40227	**IGHV Mutation Analysis by Sequencing** Determine risk group in newly diagnosed CLL.
JACAR 2012084	**JAK2 , (V617F) QL w/reflex to CALR Exon 9 by PCR w/reflex to MPL codon 515 by Pyrosequencing, Quant** Use when diagnosis of essential thrombocytemia (ET) or primary myelofibrosis (PMF) is suspected.
JAE12 2002357	**JAK2 Exon 12 Mutation Analysis by PCR** Most appropriate in cases of high suspicion of polycythemia vera with negative JAK2 V617F mutation status
NJAK2 0051245	**JAK2 Gene, V617F Mutation, Qualitative** Aids in the workup of suspected myeloproliferative neoplasms. Detects the JAK2 V617F mutation in peripheral blood or bone marrow.
JAK2M 0040168	**JAK2 Gene, V617F Mutation, Quantitative** Quantitates JAK2 V617F allele frequency in enriched granulocytes from peripheral whole blood. Aids in risk stratification and therapeutic monitoring of JAK2 V617F mutation positive myeloproliferative neoplasms.
JAK2R 2012085	**JAK2 V617F with reflex to JAK2 Exon 12** Use when diagnosis of polycythemia vera (PV) is suspected
0040137	**KIT (D816V) Mutation by PCR** Aid in the diagnosis of mastocytosis. Provide prognostic and predictive information for tyrosine kinase inhibitor (TKI) therapy planning.
KRASM 0040248	**KRAS Mutation Analysis** Predicts response to anti-EGFR and MAPK pathway therapies in a variety of malignancies (eg, colorectal and lung cancer).
2001932	**KRAS Mutation Detection with Reflex to BRAF Codon 600 Mutation Detection** Determine eligibility for anti-EGFR (cetuximab and panitumumab) therapy in patients with metastatic colorectal cancer.
2014683	**LeukoStrat CDx FLT3 Mutation Detection by PCR** Aid in the assessment of acute myeloid leukemia patients for whom midostaurin (RYDAPT) treatment is being considered.
NAFLD 2012521	**Liver Fibrosis, FibroMeter NAFLD** Only intended for use in patients with non-alcoholic liver disease (NAFLD); results may be inaccurate in patients with other etiologies of liver disease.

LUNGK 2008895	**Lung Cancer Limited Panel with KRAS** Screening panel detects EGFR and KRAS mutations (pyrosequencing), ALK and ROS1 fusion proteins (IHC).
2008894	**Lung Cancer Limited Panel** Screening panel detects EGFR mutations (pyrosequencing), ALK and ROS1 fusion proteins (IHC).
LUNGP	**Lung Cancer Panel** This panel evaluates formalin-fixed paraffin-embedded tumor from patients with lung cancer. Mutational status in non-small-cell cancer is critically important in determining the most effective therapy. Since these mutations are not simultaneously presented, EGFR positive tumors are reported with no further testing. EGFR negative tumors are reflexed to ALK and ROS1 rearrangements. This panel is useful for identifying lung tumors that may respond to targeted therapies such as the epidermal growth factor receptor (EGFR) tyrosine kinase inhibitor, erlotinib, or the anaplastic lymphoma kinase (ALK) inhibitor, crizotinib, for NSCLC with ALK and ROS1 rearrangements.
2005584	**Marfan Syndrome (FBN1) Sequencing and Deletion/Duplication** Preferred test to confirm diagnosis when Marfan syndrome is strongly suspected by consensus criteria.
2005589	**Marfan Syndrome (FBN1) Sequencing** Acceptable test to confirm diagnosis for individuals with clinical phenotype of Marfan syndrome.
2009310	**MGMT Methylation Detection by PCR** Aids in therapeutic decisions in individuals with gliomas.
0051740	**Microsatellite Instability (MSI), HNPCC/Lynch Syndrome, by PCR** First-line screening test for Lynch syndrome. Directs subsequent genetic diagnostic testing for Lynch syndrome.
2006054	**Mitochondrial Disorders Panel (mtDNA and Nuclear) by Sequencing and Delection/Duplication** Comprehensive test to confirm a suspected diagnosis of a mitochondrial disorder.
0051650	**MLH1 HNPCC/Lynch Syndrome Sequencing and Deletion andDuplication** Detect germline MLH1 variants. Use in MMR-deficient carcinoma with suggestive IHC results (loss of MLH1 and PMS2 proteins), negative for the BRAF codon 600 pathogenic variant, and normal MLH1 methylation studies.
2002499	**MLH1 Methylation by PCR, Paraffin** Recommended test to distinguish between Lynch syndrome and sporadic noncolorectal tumors with loss of MLH1.
0051654	**MSH2 HNPCC/Lynch Syndrome Sequencing and Deletion andDuplication** Detect germline MSH2 variants. Use in MMR-deficient carcinoma with suggestive IHC results (loss of MSH2 and MSH6 proteins). Includes evaluation of EPCAM exon 9 deletions.
0051656	**MSH6 HNPCC/Lynch Syndrome Sequencing and Deletion andDuplication** Detect germline MSH6 variants. Use in MMR-deficient carcinoma with suggestive IHC results (isolated loss of MSH6 protein).
MTHFM 0055655	**MTHFR Mutations** Determine genetic cause for hyperhomocysteinemia and potential sensitivity to antifolate drugs. Test is not recommended for women who have recurrent pregnancy loss, thrombophilia screening, or neural tube defect risk assessment
0051390	**Multiple Endocrine Neoplasia Type 2 (MEN2), RET Gene Mutations by Sequencing** Diagnostic and predictive testing for multiple endocrine neoplasia type 2.
2009318	**MYD88 L265P Mutation Detection by PCR, Quantitative** Useful in distinguishing lymphoplasmacytic lymphoma (LPL) from other low-grade B-cell lymphoproliferative disorders which may be in the differential diagnosis. Use when monitoring patients with LPL diagnosis and previously identified MYD88 L265P mutation.
2011117	**Myeloid Malignancies Mutation Panel by NGS** Assess for single gene mutations, including substitutions and insertions and deletions that may have diagnostic, prognostic, and/or therapeutic significance in acute myeloid leukemia, myelodysplastic syndromes, myeloproliferative neoplasms, or MDS/MPN overlap disorders such as chronic myelomonocytic leukemia.
2012182	**Myeloid Malignancies Somatic Mutation and Copy Number Analysis Panel** Detects important genomic abnormalities in acute myeloid leukemia (AML), myeloproliferative neoplasms (MPN), myelodysplastic syndromes (MDS), and MDS/MPN neoplasms that may have diagnostic, prognostic, and/or therapeutic significance: • Loss/gain of DNA • Loss of heterozygosity (LOH) • Single gene mutations (substitutions and small insertions and deletions)
HLANC 2005023	**Narcolepsy (HLA-DQB1 06:02) Genotyping** May help rule out narcolepsy when clinical history and sleep studies are inconclusive.
FTANM 2010232	**NIPT Fetal Aneuploidy w/ Microdeletions** First- or second-tier screening test for the common fetal aneuploidy disorders: trisomy 13, trisomy 18, trisomy 21 (Down syndrome), Turner syndrome, sex chromosome aneuploidies (XXX, XXY, XYY), and triploidy; as well as microdeletions causing 22q11.2 deletion (DiGeorge or velocardiofacial [VCFS] syndrome), 1p36 deletion, Angelman, Prader-Willi, and cri-du-chat (5p-) syndromes. Testing may be offered to pregnant women from 9 weeks 0 days gestation to term. Test is not recommended for women who are carrying more than one fetus or have a known twin demise, patients who have used an egg donor, surrogates who have not used their own egg, or women who have had an allogenic bone marrow transplant
FA22Q 2013142	**NIPT Fetal Aneuploidy w/22q11.2** First- or second-tier screening test for the most common fetal aneuploidy disorders (trisomy 13, trisomy 18, trisomy 21 [Down syndrome], Turner syndrome, sex chromosome aneuploidies [XXX, XXY, XYY], triploidy) and microdeletions causing 22q11.2 deletion (DiGeorge or velocardiofacial [VCFS] syndrome). Testing may be offered to pregnant women with singleton or monozygotic twin pregnancies from 9 weeks 0 days gestation to term. Test is not recommended for women who are carrying dizygotic twins, triplets or higher-order multiples, who have a known twin demise, who have used an egg donor, who are surrogates not using their own egg, or who have had an allogenic bone marrow transplant. IMPORTANT: Monozygotic twin specimens will be run at Natera and reported through ARUP.
FETAN 2007537	**NIPT Fetal Aneuploidy (Panorama)** First- or second-tier screening test for the most common fetal aneuploidy disorders (trisomy 13, trisomy 18, trisomy 21 [Down syndrome], Turner syndrome, sex chromosome aneuploidies [XXX, XXY, XYY], triploidy). Testing may be offered to pregnant women with singleton or twin pregnancies from 9 weeks 0 days gestation to term. Test may also be ordered for women who have used an egg donor or for surrogate pregnancies. Test is not recommended for women carrying triplets or higher-order multiples, who have a known twin demise, who are carrying twins and used an egg donor/surrogate, or who have had an allogenic bone marrow transplant. IMPORTANT: Twin, egg donor/surrogate specimens will be run at Natera and reported through ARUP.
2003123	**NRAS Mutation Detection, Pyrosequencing** Predicts response to anti-EGFR and MAPK pathway therapies in a variety of malignancies (eg, melanoma and colorectal cancer).
2002016	**Pancreatitis (PRSS1) Sequencing** Preferred test for individuals with idiopathic pancreatitis who are <20 years of age OR have two affected first-degree relatives.

2002012	**Pancreatitis (SPINK1) Sequencing** For adults with idiopathic pancreatitis if other components of panel (CFTR, CTRC, PRSS1 sequencing) have been sequenced without providing a complete explanation for the pancreatitis.
2010876	**Pancreatitis, Panel (CFTR, CTRC, PRSS1, SPINK1) Sequencing** Preferred test for individuals with history of idiopathic pancreatitis.
2010102	**PCA3 - Prostate Cancer Biomarker by Transcription-Mediated Amplification** Do not use for initial prostate cancer screening. Preferred test is Prostate Specific Antigen, Total (PSA) in conjunction with digital rectal exam. The PCA3 test may be useful, in conjunction with other patient information, to aid in the decision for repeat biopsy in men 50 years of age or older who have had one or more negative prostate biopsies and for whom a repeat biopsy would be recommended by a urologist based on current standard of care.
2007370	**Periodic Fever Syndromes Panel, Sequencing (7 Genes) and Deletion/Duplication (6 Genes)** Most comprehensive test to identify causative periodic fever syndromes mutations.
2002871	**PML-RARA Translocation, t(15;17) by RT-PCR, Quantitative** Provides genetic confirmation of APL. Predict relapse risk and monitor for minimal residual disease post-consolidation therapy.
0051737	**PMS2 HNPCC/Lynch Syndrome Sequencing and Deletion/Duplication** Detect germline PMS2 variants. Use in MMR-deficient carcinoma with suggestive IHC results (isolated loss of PMS2 protein).
FACII	**Prothrombin Mutation (Prothrombin), DNA 20210** Factor II Mutation analysis aids in evaluation of risk for patients with suspected venous thrombosis. People who have prothrombin mutation G20210A have a 2-to-3 fold increase in the risk of Deep Venous Thrombosis. This test aids in the evaluation of the risk for venous thrombosis. The Xpert® Factor II & Factor V Assay is a qualitative in vitro diagnostic genotyping test for the detection of Factor II and Factor V alleles from sodium citrate or EDTA anticoagulated whole blood. The test is performed on the Cepheid GeneXpert® Dx System. This test is intended to provide results for Factor II (G20210A)) and Factor V (G1691A) Leiden mutations as an aid in the diagnosis in individuals with suspected thrombophilia. The association of Factor II and Factor V Leiden mutations with an increased risk for venous thrombosis has been well documented. The Factor II or Prothrombin mutation refers to the G to A transition at nucleotide in the untranslated region of the gene and is associated with increased plasma levels of prothrombin. Factor V Leiden (G1691A) refers to the G to A transition at nucleotide position 1691 of the Factor V gene, resulting in the substitution of the amino acid arginine by glutamine in the Factor V protein, causing resistance to cleavage by Activated Protein C (APC). Factor II (G20210A) and Factor V Leiden (G1691A) mutations are present in 2% and 5% of the general population, respectively.
0051378	**Rett Syndrome (MECP2), Full Gene Sequencing** Acceptable initial test for clinical diagnosis of Rett syndrome or MECP2-related disorder. Consultation with a genetic counselor is recommended to plan the optimal MECP2 genetic testing sequence. ARUP's genetic counselors are available at 800-242-2787 x2141
2007991	**Solid Tumor Mutation Panel by NGS** Aid in therapeutic decisions for solid tumor cancers. Does not detect translocations.
2013436	**Spinal Muscular Atrophy(SMA) Copy Number Analysis** Diagnostic testing to confirm a suspected diagnosis of spinal muscular atrophy (SMA). Prenatal or preconception carrier screening for SMA in the general population. Carrier screening for reproductive partner of known SMA carrier. Carrier screening for parents of a child with a deletion of the SMN1 gene or other family history of SMA.
TCPCR 0055567	**T-Cell Clonality Screening by PCR** Aid in the assessment of acute myeloid leukemia patients for whom midostaurin (RYDAPT) treatment is being considered.
2012233	**Thiopurine Methyltransferase (TPMT) Genotyping, 4 Variants** Genotype test to assess risk, due to genetics, for severe myelosuppression with standard dosing of thiopurine drugs. Use for individuals being considered for thiopurine therapy or who have had an adverse reaction to thiopurine therapy. Preferred test for patients with recent heterologous blood transfusion. Can be performed irrespective of thiopurine therapy.
0030133	**Thrombosis, Common Etiologies with Reflex to Factor V Leiden** Acceptable screening panel for common inherited thrombophilias.
0051332	**UDP Glucuronosyltransferase 1A1 (UGT1A1) Genotyping** May be useful in dosage planning for individuals who will receive high-dose irinotecan, have a history of irinotecan sensitivity, or experience neutropenia while receiving irinotecan. Confirm suspected diagnosis of Gilbert syndrome.
2002965	**von Hippel-Lindau (VHL) Sequencing and Deletion/Duplication** Preferred test to confirm a suspected diagnosis of von Hippel-Lindau syndrome.
2005766	**WT1 Mutation Detection by Sequencing** WT1 testing is appropriate for detecting mutations in exons 7 and 9 as well as for the presence of SNP rs16754 in cases of cytogenetically normal AML.
2001778	**Y Chromosome Microdeletion** Aids in determining the cause of azoospermia or oligospermia and helps predict effectiveness of assisted reproductive technologies in men with Y chromosome microdeletions.