Laboratory Developed Test Coverage Criteria Guide

Genetic Test Name	Coverage Criteria
Afirma® Thyroid FNA Analysis	To aid in thyroid nodule diagnosis by reducing unnecessary surgeries in patients with indeterminate thyroid nodules
ALK	To determine response to tyrosine kinase inhibitor (TKI) therapy in patients with adenocarcinoma of the lung or mixed lung cancer with adenocarcinoma component of the lung
APC	• Testing for APC variants in individuals with clinical symptoms consistent with familial adenomatous polyposis (FAP) • Testing for APC variants in individuals with clinical symptoms consistent with attenuated familial adenomatous polyposis (AFAP) • Testing for APC variants in individuals with clinical symptoms consistent with Turcot's or Gardner's syndromes • Testing individuals with an APC-associated polyposis syndrome for the purpose of identifying a variant that may be used to screen at-risk relatives • For the presymptomatic testing of at-risk relatives for a known familial variant
ATXN1	• Diagnosis of spinocerebellar ataxia type 1 (SCA1) in patients with cerebellar ataxia of unknown etiology, along with extracerebellar symptoms associated with SCA1 and/or a family history consistent with autosomal dominant inheritance • Diagnosis of SCA1 in symptomatic family members of known SCA1 patients
ATXN2	• Diagnosis of spinocerebellar ataxia type 2 (SCA2) in patients with cerebellar ataxia of unknown etiology, along with extracerebellar symptoms associated with SCA2 and/or a family history consistent with autosomal dominant inheritance • Diagnosis of SCA2 in symptomatic family members of known SCA2 patients
ATXN3	• Diagnosis of spinocerebellar ataxia type 3 (SCA3) in patients with cerebellar ataxia of unknown etiology, along with extracerebellar symptoms associated with SCA3 and/or a family history consistent with autosomal dominant inheritance • Diagnosis of SCA3 in symptomatic family members of known SCA3 patients
ATXN7	• Diagnosis of spinocerebellar ataxia type 7 (SCA7) in patients with cerebellar ataxia and visual disturbance • Diagnosis of SCA7 in symptomatic family members of known SCA7 patients
ATXN10	• Diagnosis of spinocerebellar ataxia type 10 (SCA10) in ataxia patients whose ancestry is of American Indian origin, and whose family history is consistent with autosomal dominant inheritance • Diagnosis of SCA10 in symptomatic family members of known SCA10 patients
BCR/ABL1	• Diagnostic assessment of individuals with suspected chronic myelogenous leukemia (CML) by quantitative RT-PCR (RQ-PCR) • Diagnostic assessment of individuals with suspected CML by qualitative RT-PCR • Monitoring response to tyrosine kinase inhibitor (TKI) therapy, such as imatinib, in individuals with CML by RQ-PCR • Testing for the presence of the BCR/ABL1 p.Thr315Ile variant in CML patients to guide treatment selection following resistance to first-line imatinib therapy • Testing for the presence of BCR/ABL1 variants other than p.Thr315Ile in CML patients to guide treatment selection following resistance to first-line imatinib therapy
Biotheranostics Breast Cancer Index®	• Women with diagnosed early stage hormone-receptor positive (HR+), lymph node-negative (LN-) breast cancer being treated with adjuvant endocrine therapy • Women with diagnosed early stage hormone-receptor positive (HR+), lymph node-positive (LN+) (1-3 nodes) breast cancer being treated with adjuvant endocrine therapy

Genetic Test Name	Coverage Criteria
BMPR1A	• To clarify the diagnosis of individuals with juvenile polyposis syndrome (JPS) • If a known SMAD4 mutation is in the family, genetic testing should be performed in the first six months of life due to hereditary hemorrhagic telangiectasia risk
BRAF	• To predict response to vemurafenib therapy in patients with a positive cobas® 4800 BRAF mutation test result • To predict response to trametinib monotherapy in advanced melanoma patients with a positive BRAF p.Val600Glu and/or p.Val600Lys test result • To predict response to dabrafenib monotherapy in advanced melanoma patients with a positive BRAF p.Val600Glu test result • To predict response to trametinib and dabrafenib combination therapy in advanced melanoma patients with a positive BRAF p.Val600Glu and/or p.Val600Lys test result • For individuals with indeterminate thyroid fine-needle aspiration (FNA) biopsy cytology for diagnosis of papillary thyroid carcinoma
BRCA1/BRCA2 or BRACAnalysis CDx®	BRCA1/BRCA2 gene testing must be in accordance with the most current National Comprehensive Cancer Network (NCCN) guidelines for breast cancer
CACNA1A	• Diagnosis of spinocerebellar ataxia type 6 (SCA6) in patients with cerebellar ataxia with dysarthria and/or nystagmus • Diagnosis of SCA6 in symptomatic family members of known SCA6 patients
CALM1, CASQ2, RYR2, and/or TRDN	To confirm a diagnosis of catecholaminergic polymorphic ventricular tachycardia (CPVT) in patients with clinically diagnosed or suspected CPVT
CDH1	For large rearrangements in the CDH1 gene for the treatment of hereditary diffuse gastric cancer (HDGC)
CEBPA	To guide the treatment decisions for individuals with acute myeloid leukemia (AML)
CFTR/Cystic Fibrosis Testing	• Confirmation of diagnosis in individuals showing clinical symptoms of cystic fibrosis (CF) or having a high sweat chloride level • Identification of newborns who are affected with CF • Identification of individuals with the p.Gly551Asp variant who will respond to treatment with ivacaftor • Male infertility testing and treatment **Note:** Effective Dec. 27, 2021, TRICARE covers CFTR gene testing as a preconception and prenatal carrier screening under the TRICARE basic benefit. Preconception and prenatal carrier screening for CFTR is no longer covered under the LDT Demonstration Project. Refer to TPM, Chapter 6, Section 3.2 for details.
Chimerism Analysis	For the management and treatment of stem cell transplant patients
Chromosome 22q11.2	Confirmation of diagnosis in an individual suspected of chromosome 22q11.2 deletion syndrome based on clinical findings
COL1A1/COL1A2	For sequence variants in the COL1A1/COL1A2 genes for the diagnosis of osteogenesis imperfecta (OI) when clinical and radiological examination and family history provide inadequate information for diagnosis of OI
COL3A1	To confirm or establish a diagnosis of Ehlers-Danlos syndrome type 4 (EDS IV), also known as vascular EDS, in patients with clinical symptoms or features of EDS IV
CYP2C9	For the initiation and management of warfarin treatment
CYP2C19	To manage dosing of clopidogrel
Cytogenomic Constitutional Microarray Analysis	• Diagnostic evaluation of patients suspected of having a genetic syndrome (in other words, have congenital anomalies, dysmorphic features, developmental delay and/or intellectual disability) • Diagnostic evaluation of individuals with autism spectrum disorder (ASD), including autism, Asperger's syndrome and pervasive developmental disorder

Genetic Test Name	Coverage Criteria
DAZ/SRY	To detect submicroscopic deletions involving the Y chromosome in the evaluation of men with infertility secondary to azoospermia, oligozoospermia or teratozoospermia
DermTech Pigmented Lesion Assay (PLA)	Neoplasms of uncertain behavior of skin
DMD	For diagnostic DMD testing (deletion and duplication analysis with reflex to complete gene sequencing) in males or females exhibiting symptoms of Duchenne muscular dystrophy or Becker muscular dystrophy
DMPK	• Confirmation of a diagnosis of myotonic dystrophy type 1 (DM1) or type 2 (DM2) in symptomatic patients • Diagnosis of DM1 or DM2 in asymptomatic adults who are at an increased risk of DM1 or DM2 through a positive family history
DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, and/or TMEM43	• For sequence variants in the DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, and TMEM43 genes to confirm a diagnosis of arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) in probands • For a known familial sequence variant in the DSC2, DSG2, DSP, PKP2, or TMEM43 gene for at-risk relatives of probands with International Task Force (ITF)-confirmed ARVD/C to confirm a diagnosis of ARVD/C in those whose symptoms meet the ITF diagnostic criteria
DYT1/TOR1A	• For genetic testing for sequence variants of DYT1 for patients with primary dystonia with onset < 30 years of age • For genetic testing for sequence variants of DYT1 for patients with primary dystonia with onset ≥ 30 years of age who have a relative who developed dystonia aged < 30 years
EGFR	To help guide administration of epidermal growth factor receptor (EGFR) tyrosine kinase inhibitors (TKI) in the first-line treatment of non-small cell lung cancer
F2	**Prothrombin (factor II) related thrombophilia gene testing:** • Diagnostic evaluation of individuals with a prior venous thromboembolism (VTE) during pregnancy or puerperium • For patients with VTE with a personal or family history of recurrent VTE (more than two in the same person) • For patients with their first VTE before age 50 with no precipitating factors • For venous thrombosis at unusual sites such as the cerebral, mesenteric, portal, or hepatic veins • For VTE associated with the use of estrogen-containing oral contraceptives, selective estrogen receptor modulators (SERMs), or hormone replacement therapy (HRT) • To diagnose an inherited thrombophilia in female family members of individuals with an inherited thrombophilia if the female family member is pregnant or considering pregnancy or oral contraceptive use
F5	**Factor V Leiden thrombophilia gene testing:** • Diagnostic evaluation of individuals with a prior venous thromboembolism (VTE) during pregnancy or puerperium • For patients with VTE with a personal or family history of recurrent VTE (more than two in the same person) • For patients with their first VTE before age 50 with no precipitating factors • For venous thrombosis at unusual sites such as the cerebral, mesenteric, portal, or hepatic veins • For VTE associated with the use of estrogen-containing oral contraceptives, selective estrogen receptor modulators (SERMs), or hormone replacement therapy (HRT) • To diagnose an inherited thrombophilia in female family members of individuals with an inherited thrombophilia if the female family member is pregnant or considering pregnancy or oral contraceptive use

Genetic Test Name	Coverage Criteria
FBN1	• To facilitate the diagnosis of Marfan syndrome in patients who do not fulfill the Ghent diagnostic criteria, but have at least one major feature of the condition • To facilitate the diagnosis of Marfan syndrome in the at-risk relatives of patients carrying known disease-causing variants
FLCN	To confirm a diagnosis of Birt-Hogg-Dubé syndrome (BHD) in patients with suspected BHD
FLT3	For diagnosis and prognosis in acute myeloid leukemia (AML)
FMR1	**FMR1 gene testing:** For CGG repeat length for diagnosis of patients of either sex with mental retardation, intellectual disability, developmental delay, or autism **FMR1 testing for fragile X-associated tremor/ataxia syndrome:** – Males and females older than age 50 years who have progressive cerebellar ataxia and intention tremor with or without a positive family history of FMR1-related disorders in whom other common causes of ataxia have been excluded – Women with unexplained premature ovarian insufficiency (POI)
FoundationOne® Heme	• Assessment of gene alterations in hematologic malignancies • Assessment of gene alterations in sarcomas
GCK	Diagnosis of maturity-onset diabetes of the young type 2 (MODY2) in patients with hyperglycemia or non-insulin–dependent diabetes who have a family history of abnormal glucose metabolism in at least two consecutive generations, with the patient or ≥ 1 family member(s) diagnosed before age 25
GJB2	Diagnosis of DFNB1 or DFNA3 in individuals with nonsyndromic hearing loss to aid in treatment
GJB6	Diagnosis of DFNB1 or DFNA3 in individuals with nonsyndromic hearing loss to aid in treatment
HBA1/HBA2	• To confirm the diagnosis of alpha-thalassemia in a symptomatic individual • To confirm the diagnosis in a pregnant woman with low hemoglobin when alpha-thalassemia is suspected
HEXA	As an adjunct to biochemical testing in patients with low hexosaminidase A levels in blood; When individuals are identified with apparent deficiency of hexosaminidase A enzymatic activity, targeted mutation analysis can then be used to distinguish pseudodeficiency alleles from disease-causing alleles
HFE	Diagnosis of patients with or without symptoms of iron overload with a serum transferrin saturation > 45% and/or elevated serum ferritin
HLA	• To determine histocompatibility of tissue between organ and bone marrow donors and recipients prior to transplant • For platelet transfusion for patients refractory to treatment due to alloimmunization • Diagnosis of celiac disease in symptomatic patients with equivocal results on small bowel biopsy and serology, or in previously symptomatic patients who are asymptomatic while on a gluten-free diet • Testing for the HLA-B*1502 allele prior to initiating treatment with carbamazepine in patients from high-risk ethnic groups • Testing for the HLA-B*5701 allele for hypersensitivity reactions in patients prior to initiation or re-initiation with treatments containing abacavir • Testing for the HLA-B*58:01 allele in patients prior to initiating treatment with allopurinol
HNF1A	Diagnosis of maturity-onset diabetes of the young type 3 (MODY3) in patients with hyperglycemia or non-insulin–dependent diabetes who have a family history of abnormal glucose metabolism in at least two consecutive generations, with the patient or ≥ 1 family member(s) diagnosed before age 25
HNF1B	Diagnosis of maturity-onset diabetes of the young type 5 (MODY5) in patients with hyperglycemia or non-insulin–dependent diabetes who have a family history of abnormal glucose metabolism in at least two consecutive generations, with the patient or ≥ 1 family member(s) diagnosed before age 25, and who have structural or functional abnormalities of the kidneys

Genetic Test Name	Coverage Criteria
HNF4A	Diagnosis of maturity-onset diabetes of the young type 1 (MODY1) in patients with hyperglycemia or non-insulin–dependent diabetes diabetes who have a family history of abnormal glucose metabolism in at least two consecutive generations, with the patient or ≥ 1 family member(s) diagnosed before age 25
HTT	To test for CAG repeat length for diagnosis of Huntington's/chorea disease (HD) in patients suspected of having HD in the absence of a family history of HD
IGH	• For medical management of patients with acute lymphoblastic leukemia (ALL) through analysis of rearrangements in the IGH gene to estimate minimal residual disease (MRD) levels • For diagnostic evaluation of rearrangements in the IGH gene in patients with suspected B-cell non-Hodgkin lymphoma (NHL), but in whom clinical, immunophenotypic, and histologic evaluation have provided inconclusive results
IGK	• For medical management of patients with acute lymphoblastic leukemia (ALL) through analysis of rearrangements in the IGK gene to estimate minimal residual disease (MRD) levels • For diagnostic evaluation of rearrangements in the IGK gene in patients with suspected B-cell non-Hodgkin lymphoma (NHL), but in whom clinical, immunophenotypic, and histologic evaluation have provided inconclusive results
IL28B	For IL28B single nucleotide polymorphism (SNP) testing in patients with chronic hepatitis C virus (HCV) genotype 1 being considered for treatment with PegIFN/RBV dual therapy
JAK2	• Diagnostic evaluation of individuals presenting with clinical, laboratory, or pathological findings suggesting classic forms of myeloproliferative neoplasms (MPN), that is, polycythemia vera (PV), essential thrombocythemia (ET), or primary myelofibrosis (PMF) • Diagnostic evaluation of PV through JAK2 exon 12 variant detection in JAK2 p.Val617Phe-negative individuals
KCNQ1, KCNH2, SCN5A, KCNE1, and/or KCNE2	For patients with suspected familial long QT syndrome for confirmation of diagnosis and treatment
KIT	• To confirm a diagnosis of a gastrointestinal stromal tumor (GIST) in patients who are negative by immunostaining • To determine primary resistance to treatment with tyrosine kinase inhibitors (TKI) in patients with an advanced metastatic or unresectable GIST • To determine primary resistance to preoperative or postoperative treatment of a GIST with TKIs
KMT2D and/or KDM6A	Diagnosis of Kabuki syndrome (KS) in patients with symptoms compatible with KS
KRAS	To help guide administration of anti-epidermal growth factor receptor (EGFR) monoclonal antibodies
MDxHealth Confirm MDx	• Men with a previous diagnosis of prostate cancer that have undergone a previous prostate biopsy (within prior 24 months) and are being considered for a repeat prostate biopsy due to persistent cancer-risk factors • Men with a previous diagnosis of prostate cancer that have undergone a previous prostate biopsy (within prior 24 months) and are being considered for a repeat prostate biopsy due to elevated cancer risk factors
MDxHealth Select MDx	Men with previous diagnosis of prostate cancer that are suspected of harboring prostate cancer
MECP2	• Testing for MECP2 sequence variants in patients who meet established clinical diagnostic criteria for classic or variant Rett syndrome (RS) • Testing for MECP2 sequence variants in patients who have symptoms of RS, but do not meet established clinical diagnostic criteria
MEFV	• In patients exhibiting symptoms of familial Mediterranean fever (FMF), including periodic episodes of fever in combination with peritonitis, pleuritic, arthritis, and erysipelas-like erythema • In patients from ethnic groups considered at high risk for FMF who present with nephrotic syndrome or amyloidosis, but do not meet the diagnostic criteria for FMF

Genetic Test Name	Coverage Criteria
MLH1, MSH2, MSH6, MSI, PMS2, and/or EPCAM	Genetic testing for Lynch syndrome (LS) must be in accordance with the most current National Comprehensive Cancer Network (NCCN) guidelines for colon cancer
MPL	Diagnostic evaluation of myeloproliferative leukemia (MPL) variants to include Trp515Leu and Trp515Lys in JAK2 p.Val617Phe-negative individuals showing symptoms
MUTYH	• Diagnosis of MUTYH (MYH)-associated polyposis (MAP) in APC-negative colorectal polyposis patients, or in polyposis patients who have a family history consistent with autosomal recessive inheritance • Diagnosis of MAP in asymptomatic siblings of patients with known MYH variants
Noninvasive Prenatal Screening for Trisomies 13, 18, 21, X & Y	Screening is covered for the following: • In singleton pregnancies with a high risk of fetal aneuploidy (for dates March 5, 2015-August 16, 2020) • In accordance with the most current ACOG guidelines **Note:** Pre-authorization is not required.
NPM1	To guide treatment decisions for individuals with acute myeloid leukemia (AML)
NRAS	For patients with metastatic colorectal cancer who are being considered for treatment with anti-epidermal growth factor receptor (EGFR) monoclonal antibodies, and who have had negative KRAS gene testing
Oncotype DX® Breast Cancer Assay (Oncotype DX®)	• Estrogen receptor (ER) positive (+), lymph node (LN) negative (-), human epidermal growth factor receptor (EGFR) 2 negative (HER2-) breast cancer patients who are considering whether to use adjuvant chemotherapy in addition to standard hormone therapy • ER+, HER2- breast cancer patients with 1-3 involved ipsilateral axillary lymph nodes who are considering whether to use adjuvant chemotherapy in addition to hormonal therapy
PAX8	For individuals with indeterminate thyroid fine-needle aspiration (FNA) biopsy cytology for diagnosis of papillary thyroid carcinoma
PDGFRA	• To confirm a diagnosis of a gastrointestinal stromal tumor (GIST) in patients who are negative by immunostaining • To determine primary resistance to treatment with tyrosine kinase inhibitors (TKI) in patients with an advanced metastatic or unresectable GIST • To determine primary resistance to preoperative or postoperative treatment of a GIST with TKIs
PML/RARalpha	• Diagnostic assessment of individuals with suspected acute promyelocytic leukemia (APL) by quantitative RT-PCR (RQ-PCR) • Diagnostic assessment of individuals with suspected APL by qualitative RT-PCR • Monitoring response to treatment and disease progression in individuals with APL by RQ-PCR
PMP22	For the accurate diagnosis and classification of hereditary polyneuropathies
PPP2R2B	• Diagnosis of spinocerebellar ataxia type 12 (SCA12) in patients with action tremor of the upper extremities and signs of cerebellar and cortical dysfunction, in addition to Indian ancestry and a family history consistent with autosomal dominant inheritance • Diagnosis of SCA12 in symptomatic family members of known SCA12 patients
PRSS1	**To confirm diagnosis of hereditary pancreatitis in symptomatic patients with any of the following:** • A family history of pancreatitis in a first-degree (parent, sibling, child) or second-degree (aunt, uncle, grandparent) relative; • An unexplained episode of documented pancreatitis occurring in a child that has required hospitalization, and where there is significant concern that hereditary pancreatitis should be excluded; • Recurrent (two or more separate, documented episodes with hyperamylasemia) attacks of acute pancreatitis for which there is no explanation (anatomical anomalies, ampullary or main pancreatic strictures, trauma, viral infection, gallstones, alcohol, drugs, hyperlipidemia, etc.); or • Unexplained (idiopathic) chronic pancreatitis

Genetic Test Name	Coverage Criteria
PTEN	• For patients with autism spectrum disorders (ASD) and macrocephaly (head circumference greater than 2 standard above the mean for age) • PTEN variant testing in individuals suspected of being affected with Cowden syndrome (CS) or Bannayan-Riley-Ruvalcaba syndrome (BRRS)
RET	• Multiple endocrine neoplasia type 2 (MEN2) gene testing in patients with the clinical manifestations of MEN2A, MEN2B, or familial medullary thyroid carcinoma (FMTC), including those with apparently sporadic medullary thyroid carcinoma (MTC) or pheochromocytoma • MEN2 gene testing to confirm a diagnosis in the at-risk relatives of genetically confirmed MEN2 patients
ROS1	For patients who have wild type (negative) epidermal growth factor receptor (EGFR) or ALK gene testing, reflex testing to ROS1 should be ordered for the treatment of non-small cell lung carcinoma
RYR1	• To test clinically confirmed malignant hyperthermia susceptibility (MHS) patients for variants in the RYR1 gene to facilitate diagnostic testing in at-risk relatives • To diagnose MHS in at-risk relatives of patients with clinically confirmed MHS
SDHA, SDHB, SDHC, SDHD, SDHAF2, MAX, and/or TMEM127	To diagnose a hereditary paraganglioma (PGL) or pheochromocytoma (PCC) syndrome in patients with PGLs and/or PCCs
SERPINA1	For guidance in diagnosis of inconclusive cases of alpha-1 antitrypsin (AAT) deficiency (AATD) in individuals with chronic obstructive pulmonary disease (COPD), unexplained liver disease, family history of AATD, or environmental exposures leading to airflow obstruction after serum AAT protein levels and protein phenotyping has been completed
SMAD4	• To clarify the diagnosis of individuals with juvenile polyposis syndrome (JPS) • If a known SMAD4 mutation is in the family, genetic testing should be performed in the first six months of life due to hereditary hemorrhagic telangiectasia risk
SMN1/SMN2	Diagnosis of patients with hypotonia and muscle weakness who are suspected of having spinal muscular atrophy (SMA)
SNRPN/UBE3A	**When a clinical diagnosis of Prader-Willi syndrome is suspected, the following findings justify genetic testing:** • From birth to age two: hypotonia with poor suck (neonatal period) • From age two to age six: hypotonia with history of poor suck, global developmental delay • From age six to age 12: hypotonia with history of poor suck, global developmental delay, excessive eating with central obesity if uncontrolled • From age 13 years to adulthood: cognitive impairment, usually mild intellectual disability; excessive eating with central obesity if uncontrolled, hypothalamic hypogonadism and/or typical behavior problems **When a clinical diagnosis of Angelman syndrome is suspected, the following findings justify genetic testing:** • As part of the evaluation of patients with developmental delay, regardless of age • As part of the evaluation of patients with a balance or movement disorder such as ataxia of gait; may not appear as frank ataxia but can be forward lurching, unsteadiness, clumsiness, or quick, jerky motions • As part of the evaluation of patients with uniqueness of behavior: any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with uplifted hand-flapping or waving movements; hypermotoric behavior • Speech impairment, none or minimal use of words; receptive and non-verbal communication skills higher than verbal ones
STK11	To confirm a diagnosis of Peutz-Jeghers syndrome (PJS) in proband patients with a presumptive or probable diagnosis of PJS

Genetic Test Name	Coverage Criteria
TBP	• Diagnosis of spinocerebellar ataxia type 17 (SCA17) in ataxia patients exhibiting variable combinations of cognitive decline, psychiatric disturbance, and movement disorders • Diagnosis of SCA17 in symptomatic family members of known SCA17 patients • Diagnosis of SCA17 in patients suspected of having Huntington's disease (HD) who have tested negative for a pathogenic variant in the HD gene
TGFBR2	To facilitate the diagnosis of Marfan syndrome in patients testing negative for FBN1 gene variants
TP53	Diagnosis of patients satisfying the criteria for classic Li-Fraumeni syndrome or Li-Fraumeni-like syndrome, or the Chompret criteria for TP53 gene testing
TPMT	TPMT genotyping or phenotyping in patients with inflammatory bowel disease (IBD) prior to administration of thiopurines (azathioprine, 6-MP, and 6-TG)
TRG	Diagnosis and treatment of T-cell neoplasms
UGT1A1	• Prior to irinotecan administration in patients with colorectal cancer (CRC) to lower the starting does of irinotecan in patients with the UGT1A1*28/UGT1A1*28 genotype • Prior to irinotecan administration in patients with CRC to increase the starting does of irinotecan in patients with the UGT1A1*1/ UGT1A1*1 or UGT1A1*1/UGT1A1*28 genotypes
UPD	For neonates, infants, children or adults symptomatic for Beckwith-Wiedermann syndrome (BWS) to diagnose uniparental disomy (UPD) for chromosome 11
VHL	• Diagnosis of Von Hippel-Lindau (VHL) syndrome in patients presenting with pheochromocytoma, paraganglioma or central nervous system hemangioblastoma • Confirmation of diagnosis in individuals with symptoms consistent with VHL syndrome
VKORC1	For the initiation and management of warfarin treatment
Y Chromosome Microdeletion Analysis	For detecting submicroscopic deletions involving the Y chromosome in men with azoospermia, oligozoospermia or teratozoospermia