Local Coverage Determination (LCD):
Molecular Pathology Procedures (L34506)

Contractor Information

Contractor Name
National Government Services, Inc.

Contract Number
14311

Contract Type
MAC - Part A

LCD Information

Document Information

LCD ID
L34506

LCD Title
Molecular Pathology Procedures

Jurisdiction
New Hampshire

Original Effective Date
For services performed on or after 03/01/2014

Revision Effective Date
For services performed on or after 03/01/2014

Revision Ending Date
N/A

Retirement Date
N/A

Notice Period Start Date
01/14/2014

Notice Period End Date
02/28/2014

CMS National Coverage Policy Language quoted from Centers for Medicare and Medicaid Services (CMS), National Coverage Determinations (NCDs) and coverage provisions in interpretive manuals is italicized throughout the policy. NCDs and coverage provisions in interpretive manuals are not subject to the Local Coverage Determination (LCD) Review Process (42 CFR 405.860[b] and 42 CFR 426 [Subpart D]). In addition, an administrative law judge may not review an NCD. See Section 1869(f)(1)(A)(i) of the Social Security Act.

Unless otherwise specified, italicized text represents quotation from one or more of the following CMS sources:

Title XVIII of the Social Security Act (SSA):
Section 1862(a)(1)(A) excludes expenses incurred for items or services which are not reasonable and necessary for the diagnosis or treatment of illness or injury or to improve the functioning of a malformed body member.

Section 1833(e) prohibits Medicare payment for any claim which lacks the necessary information to process the claim.

Section 1862(a)(7) excludes routine physical examinations, unless otherwise covered by statute.

CMS Publications:
CMS Publication 100-02, Medicare Benefit Policy Manual, Chapter 15, Section 80.1 – Laboratory services must meet applicable requirements of CLIA

Printed on 5/7/2014. Page 1 of 20
**Abstract:**
The American Medical Association (AMA) Current Procedural Terminology (CPT) manual states molecular pathology procedures are medical laboratory procedures involving the analyses of nucleic acid to detect variants in genes that may be indicative of germline (e.g., constitutional disorders) or somatic (e.g., neoplasia) conditions, or to test for histocompatibility antigens (e.g., HLA). Given the elimination of the stacking procedure codes (83890-83914) and the array based evaluation codes (88384-88386), molecular pathology codes now include all analytical services performed in the test (e.g., cell lysis, nucleic acid stabilization, extraction, digestion, amplification, and detection). (Note: molecular pathology procedure techniques may be described in other sections of the Pathology and Laboratory section of CPT. For microbial identification using molecular pathology techniques CPT codes 87149-87153, 87470-87801, and 87900-87904 apply. For in situ hybridization analyses, CPT codes 88271-88275 and 88365-88368 apply.)

Code selection is typically based on the specific gene(s) that is being analyzed. Codes that describe tests to assess for the presence of gene variants use common gene variant names. Typically, all of the listed variants would be tested. However, these lists are not exclusive. If other variants are also tested in the analysis, they would be included in the procedure and not reported separately. Full gene sequencing should not be reported using codes that assess for the presence of gene variants unless the CPT code specifically states full gene sequence in the code descriptor. In other words, you may only assign the CPT code that is described as “full gene sequence” if the test assay performed was a full gene sequence.

Tier 2 molecular pathology procedures represent medically useful procedures that are generally performed in lower volumes than Tier 1 molecular pathology procedures (e.g., the incidence of the disease being tested is rare). They are arranged by level of technical resources and interpretive work by the physician or other qualified healthcare professional. If the analyte tested is not listed under one of the Tier 2 codes or is not represented by a Tier 1 code in CPT, use of the unlisted CPT code 81479 is required.

Molecular pathology procedures have broad clinical and research applications. The following examples of applications may not be relevant to a Medicare beneficiary or may not meet a Medicare benefit category and/or reasonable and necessary threshold for coverage. Such examples include Genetic Testing and Genetic Counseling (when applicable) for:

- Disease Risk,
- Carrier Screening,
- Hereditary Cancer Syndromes,
- Gene Expression Profiling for certain cancers,
- Prenatal Diagnostic testing,
- Diagnosis and Monitoring Non-Cancer Indications, and
- Several Pharmacogenomics applications.
Based on the Centers for Medicare & Medicaid Services (CMS) Program Integrity Manual (100-08), this Local Coverage Determination (LCD) addresses the circumstances under which the item or service is reasonable and necessary under the Social Security Act, §1862(a)(1)(A). For laboratory services, a service can be reasonable and necessary if the service is safe and effective; and appropriate, including the duration and frequency that is considered appropriate for the item or service, in terms of whether it is furnished in accordance with accepted standards of medical practice for the diagnosis of the patient's condition; furnished in a setting appropriate to the patient's medical needs and condition; ordered and furnished by qualified personnel; one that meets, but does not exceed, the patient's medical need; and is at least as beneficial as an existing and available medically appropriate alternative. Preventive Services (such as secondary prevention (population based screening test) are a specific Medicare benefit category and not under the Medicare Administrative Contractor’s (MAC) jurisdiction.

Per 42 Code of Federal Regulations (CFR) section 410.32 (a) states the following requirements: All diagnostic x-rays tests, diagnostic laboratory tests, and other diagnostic tests must be ordered by the physician who is treating the beneficiary, that is, the physician who furnishes a consultation or treats a beneficiary for a specific medical problem and who uses the results in the management of the beneficiary’s specific medical problem. Tests not ordered by the physician who is treating the beneficiary are not reasonable and necessary (see §411.15(k)(1)). Also, see Medicare Benefit Policy Manual (100-02), Chapter 15, Section 80.6 for related physician order instructions.

Laboratory services must meet all applicable requirements of the Clinical Laboratory Improvement Amendments of 1988 (CLIA), as set forth at 42 CFR part 493. Section 1862(a)(1)(A) of the Act provides that Medicare payment may not be made for services that are not reasonable and necessary. Clinical laboratory services must be ordered and used promptly by the physician who is treating the beneficiary as described in 42 CFR 410.32(a), or by a qualified nonphysician practitioner, as described in 42 CFR 410.32(a)(3).

Many applications of the molecular pathology procedures are not covered services given lack of benefit category (preventive service) and/or failure to reach the reasonable and necessary threshold for coverage (based on quality of clinical evidence and strength of recommendation). Furthermore, payment of claims in the past (based on stacking codes) or in the future (based on the new code series) is not a statement of coverage since the service was not audited for compliance with program requirements and documentation supporting the reasonable and necessary testing for the beneficiary. Certain molecular pathology procedures may be subject to prepayment medical review (records requested) and paid claims must be supportable, if selected, for post payment audit by the MAC or other contractors. Molecular pathology tests for diseases or conditions that manifest severe signs or symptoms in newborns and in early childhood or that result in early death (e.g., Canavan disease) could be subject to automatic denials since these tests are not usually relevant to a Medicare beneficiary.

This LCD gives general guidance to the medically reasonable and necessary applications of the Molecular Pathology Procedures described in CPT range 81200 – 81408 (with the exception of HLA testing 81370 - 81383).

**Indications:**

Molecular pathology procedures (Tier1 and Tier 2) may be eligible for coverage when ALL of the following criteria are met:

- Alternative laboratory or clinical tests to definitively diagnose the disorder/identify the condition are unavailable or results are clearly equivocal; AND

- Availability of a clinically valid test, based on published peer reviewed medical literature; AND

- Testing assay(s) are Food and Drug Administration (FDA) approved/cleared or if LDT (lab developed test) or LDT protocol or FDA modified test(s) the laboratory documentation should support assay(s) of analytical validity and clinical utility; AND

- Results of the testing must directly impact treatment or management of the Medicare beneficiary; AND

- For testing panels, including but not limited to, multiple genes or multiple conditions, and in cases where a tiered approach/method is clinically available, testing would be covered ONLY for the number of genes or test that are reasonable and necessary to obtain necessary information for therapeutic decision making; AND

- Individual has not previously received genetic testing for the disease/condition. (In general, diagnostic genetic testing for a disease should be performed once in a lifetime.)

**Limitations:**

- Any procedures required prior to cell lysis (e.g., microdissection [CPT codes 88380 and 88381]) should be reported separately and utilization must be clearly supported based on the application and clinical utility. Such
claims may be subject to prepayment medical review.

- HCPCS code G0452 with modifier 26 should be used by pathologists when an interpretation of a molecular pathology test is performed. Non physician practitioners (e.g., PhD, scientists etc.) are not eligible to report this code, only physicians may use/bill this code. This code should not be billed without modifier 26 since it is an interpretation code only.

- Testing for quality assurance (component of the service is not separately billable per CMS National Correct Coding Initiative (NCCI).

Other Comments:
For claims submitted to the Part A MAC: this coverage determination also applies within states outside the primary geographic jurisdiction with facilities that have nominated National Government Services to process their claims.

Bill type codes only apply to providers who bill these services to the Part A MAC. Bill type codes do not apply to physicians, other professionals and suppliers who bill these services to the Part B MAC.

Limitation of liability and refund requirements apply when denials are anticipated, whether based on medical necessity or other coverage reasons. The provider/supplier must notify the beneficiary in writing, prior to rendering the service, if the provider/supplier is aware that the test, item or procedure may not be covered by Medicare. The limitation of liability and refund requirements do not apply when the test, item or procedure is statutorily excluded, has no Medicare benefit category or is rendered for screening purposes.

Coding Information

Bill Type Codes:

Contractors may specify Bill Types to help providers identify those Bill Types typically used to report this service. Absence of a Bill Type does not guarantee that the policy does not apply to that Bill Type. Complete absence of all Bill Types indicates that coverage is not influenced by Bill Type and the policy should be assumed to apply equally to all claims.

012x Hospital Inpatient (Medicare Part B only)
013x Hospital Outpatient
014x Hospital - Laboratory Services Provided to Non-patients
022x Skilled Nursing - Inpatient (Medicare Part B only)
023x Skilled Nursing - Outpatient
071x Clinic - Rural Health
072x Clinic - Hospital Based or Independent Renal Dialysis Center
073x Clinic - Freestanding
075x Clinic - Comprehensive Outpatient Rehabilitation Facility (CORF)
077x Clinic - Federally Qualified Health Center (FQHC)
083x Ambulatory Surgery Center
085x Critical Access Hospital

Revenue Codes:

Contractors may specify Revenue Codes to help providers identify those Revenue Codes typically used to report this service. In most instances Revenue Codes are purely advisory; unless specified in the policy services reported under other Revenue Codes are equally subject to this coverage determination. Complete absence of all Revenue Codes indicates that coverage is not influenced by Revenue Code and the policy should be assumed to apply equally to all Revenue Codes.

Contractors may specify Revenue Codes to help providers identify those Revenue Codes typically used to report this service. In most instances Revenue Codes are purely advisory; unless specified in the policy services reported under other Revenue Codes are equally subject to this coverage determination. Complete absence of all Revenue Codes indicates that coverage is not influenced by Revenue Code and the policy should be assumed to apply equally to all Revenue Codes.

Printed on 5/7/2014. Page 4 of 20
Revenue codes only apply to providers who bill these services to the Part A MAC. Revenue codes do not apply to physicians, other professionals and suppliers who bill these services to the Part B MAC.

Please note that not all revenue codes apply to every type of bill code. Providers are encouraged to refer to the FISS revenue code file for allowable bill types. Similarly, not all revenue codes apply to each CPT/HCPCS code. Providers are encouraged to refer to the FISS HCPCS file for allowable revenue codes.

030X Laboratory - General Classification
031X Laboratory Pathology - General Classification

CPT/HCPCS Codes

**Group 1 Paragraph: N/A**

**Group 1 Codes:**

81200 ASPA (ASPARTOACYLASE) (EG, CANAVAN DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, E285A, Y231X)

81201 APC (ADENOMATOUS POLYPOSIS COLI) (EG, FAMILIAL ADENOMATOSIS POLYPOSIS [FAP], ATTENUATED FAP) GENE ANALYSIS; FULL GENE SEQUENCE

81202 APC (ADENOMATOUS POLYPOSIS COLI) (EG, FAMILIAL ADENOMATOSIS POLYPOSIS [FAP], ATTENUATED FAP) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS

81203 APC (ADENOMATOUS POLYPOSIS COLI) (EG, FAMILIAL ADENOMATOSIS POLYPOSIS [FAP], ATTENUATED FAP) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS

81205 BCKDHB (BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE) (EG, MAPLE SYRUP URINE DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, R183P, G278S, E422X)

81206 BCR/ABL1 (T(9;22)) (EG, CHRONIC MYELOGENOUS LEUKEMIA) TRANSLOCATION ANALYSIS; MAJOR BREAKPOINT, QUALITATIVE OR QUANTITATIVE

81207 BCR/ABL1 (T(9;22)) (EG, CHRONIC MYELOGENOUS LEUKEMIA) TRANSLOCATION ANALYSIS; MINOR BREAKPOINT, QUALITATIVE OR QUANTITATIVE

81208 BCR/ABL1 (T(9;22)) (EG, CHRONIC MYELOGENOUS LEUKEMIA) TRANSLOCATION ANALYSIS; OTHER BREAKPOINT, QUALITATIVE OR QUANTITATIVE

81209 BLM (BLOOM SYNDROME, RECQ HELICASE-LIKE) (EG, BLOOM SYNDROME) GENE ANALYSIS, 2281DEL6INS7 VARIANT

81210 BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1) (EG, COLON CANCER), GENE ANALYSIS, V600E VARIANT

81211 BRCA1, BRCA2 (BREAST CANCER 1 AND 2) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL SEQUENCE ANALYSIS AND COMMON DUPLICATION/DELETION VARIANTS IN BRCA1 (IE, EXON 13 DEL 3.835KB, EXON 13 DUP 6KB, EXON 14-20 DEL 26KB, EXON 22 DEL 510BP, EXON 8-9 DEL 7.1KB)

81212 BRCA1, BRCA2 (BREAST CANCER 1 AND 2) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; 185DELAG, 5385INSC, 6174DELT VARIANTS

81213 BRCA1, BRCA2 (BREAST CANCER 1 AND 2) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; UNCOMMON DUPLICATION/DELETION VARIANTS

81214 BRCA1 (BREAST CANCER 1) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL SEQUENCE ANALYSIS AND COMMON DUPLICATION/DELETION VARIANTS (IE, EXON 13 DEL 3.835KB, EXON 13 DUP 6KB, EXON 14-20 DEL 26KB, EXON 22 DEL 510BP, EXON 8-9 DEL 7.1KB)

81215 BRCA1 (BREAST CANCER 1) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS

81216 BRCA2 (BREAST CANCER 2) (EG, HEREDITARY BREAST AND OVARIAN CANCER) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

81217 CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; COMMON VARIANTS (EG, ACMG/ACOG GUIDELINES)

81218 CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS

81219 CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS

81220 CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; FULL GENE SEQUENCE

81221 CFTR (CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR) (EG, CYSTIC FIBROSIS) GENE ANALYSIS; INTRON 8 POLY-T ANALYSIS (EG, MALE INFERTILITY)


CYTOGENOMIC CONSTITUTIONAL (GENOME-WIDE) MICROARRAY ANALYSIS; INTERROGATION OF GENOMIC REGIONS FOR COPY NUMBER VARIANTS (EG, BACTERIAL ARTIFICIAL CHROMOSOME [BAC] OR OLIGO-BASED COMPARATIVE GENOMIC HYBRIDIZATION [CGH] MICROARRAY ANALYSIS)

CYTOGENOMIC CONSTITUTIONAL (GENOME-WIDE) MICROARRAY ANALYSIS; INTERROGATION OF GENOMIC REGIONS FOR COPY NUMBER AND SINGLE NUCLEOTIDE POLYMORPHISM (SNP) VARIANTS FOR CHROMOSOMAL ABNORMALITIES

EGFR (EPIDERMAL GROWTH FACTOR RECEPTOR) (EG, NON-SMALL CELL LUNG CANCER) GENE ANALYSIS, COMMON VARIANTS (EG, EXON 19 LREA DELETION, L858R, T790M, G719A, G719S, L861Q)

F2 (PROTHROMBIN, COAGULATION FACTOR II) (EG, HEREDITARY HYPERCOAGULABILITY) GENE ANALYSIS, 20210G>A VARIANT

F5 (COAGULATION FACTOR V) (EG, HEREDITARY HYPERCOAGULABILITY) GENE ANALYSIS, LEIDEN VARIANT

FANCC (FANCONI ANEMIA, COMPLEMENTATION GROUP C) (EG, FANCONI ANEMIA, TYPE C) GENE ANALYSIS, COMMON VARIANTS (EG, IVS4+4A>T)

FMRI1 (FRAGILE X MENTAL RETARDATION 1) (EG, FRAGILE X MENTAL RETARDATION) GENE ANALYSIS; EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES

FMRI1 (FRAGILE X MENTAL RETARDATION 1) (EG, FRAGILE X MENTAL RETARDATION) GENE ANALYSIS; CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE AND METHYLATION STATUS)

FLT3 (FMS-RELATED TYROSINE KINASE 3) (EG, ACUTE MYELOID LEUKEMIA), GENE ANALYSIS, INTERNAL TANDEM DUPLICATION (ITD) VARIANTS (IE, EXONS 14, 15)

G6PC (GLUCOSE-6-PHOSPHATASE, CATALYTIC SUBUNIT) (EG, GLYCOGEN STORAGE DISEASE, TYPE 1A, VON GIERKE DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, R83C, Q347X)

GBA (GLUCOSIDASE, BETA, ACID) (EG, GAUCHER DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, N370S, 84GG, L444P, IVS2+1G>A)

GJB2 (GAP JUNCTION PROTEIN, BETA 2, 26KDA, CONNEXIN 26) (EG, NONSYNDROMIC HEARING LOSS) GENE ANALYSIS; FULL GENE SEQUENCE

GJB2 (GAP JUNCTION PROTEIN, BETA 2, 26KDA, CONNEXIN 26) (EG, NONSYNDROMIC HEARING LOSS) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS

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)])

HEXA (HEXOSAMINIDASE A [ALPHA POLYPEPTIDE]) (EG, TAY-SACHS DISEASE) GENE ANALYSIS, COMMON VARIANTS (EG, 1278INSTATC, 1421+1G>C, G269S)

HFE (HEMOCHROMATOSIS) (EG, HEREDITARY HEMOCHROMATOSIS) GENE ANALYSIS, COMMON VARIANTS (EG, C282Y, H63D)

HBA1/HBA2 (ALPHA GLOBIN 1 AND ALPHA GLOBIN 2) (EG, ALPHA THALASSEMIA, HB BART HYDROPS FETALIS SYNDROME, HBH DISEASE), GENE ANALYSIS, FOR COMMON DELETIONS OR VARIANT (EG, SOUTHEAST ASIAN, THAI, FILIPINO, MEDITERRANEAN, ALPHA3.7, ALPHA4.2, ALPHA20.5, AND CONSTANT SPRING)

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)

IGH@ (IMMUNOGLOBULIN HEAVY CHAIN LOCUS) (EG, LEUKEMIAS AND LYMPHOMAS, B-CELL), GENE REARRANGEMENT ANALYSIS TO DETECT ABNORMAL CLONAL POPULATION(S); AMPLIFIED METHODOLOGY (EG, POLYMERASE CHAIN REACTION)

IGH@ (IMMUNOGLOBULIN HEAVY CHAIN LOCUS) (EG, LEUKEMIAS AND LYMPHOMAS, B-CELL), GENE REARRANGEMENT ANALYSIS TO DETECT ABNORMAL CLONAL POPULATION(S); DIRECT PROBE METHODOLOGY (EG, SOUTHERN BLOT)

IGH@ (IMMUNOGLOBULIN HEAVY CHAIN LOCUS) (EG, LEUKEMIA AND LYMPHOMA, B-CELL), VARIABLE REGION SOMATIC MUTATION ANALYSIS

IGH@ (IMMUNOGLOBULIN HEAVY CHAIN LOCUS) (EG, LEUKEMIA AND LYMPHOMA, B-CELL), VARIABLE REGION SOMATIC MUTATION ANALYSIS

IGK@ (IMMUNOGLOBULIN KAPPA LIGHT CHAIN LOCUS) (EG, LEUKEMIA AND LYMPHOMA, B-CELL), GENE REARRANGEMENT ANALYSIS, EVALUATION TO DETECT ABNORMAL CLONAL POPULATION(S) COMPARATIVE ANALYSIS USING SHORT TANDEM REPEAT (STR) MARKERS; PATIENT AND COMPARATIVE SPECIMEN (EG, PRE-TRANSPLANT RECIPIENT AND DONOR GERMLINE TESTING, POST-TRANSPLANT NON-HEMATOPOIETIC RECIPIENT GERMLINE [EG, BUCCAL SWAB OR OTHER GERMLINE TISSUE SAMPLE] AND DONOR TESTING, TWIN ZYGOSITY TESTING, OR MATERNAL CELL CONTAMINATION OF FETAL CELLS)

Printed on 5/7/2014. Page 6 of 20
COMPARATIVE ANALYSIS USING SHORT TANDEM REPEAT (STR) MARKERS; EACH ADDITIONAL SPECIMEN (EG, ADDITIONAL CORD BLOOD DONOR, ADDITIONAL FETAL SAMPLES FROM DIFFERENT CULTURES, OR ADDITIONAL ZYGOSITY IN MULTIPLE BIRTH PREGNANCIES) (LIST SEPARATELY IN ADDITION TO CODE FOR PRIMARY PROCEDURE)

CHIMERISM (ENGRAFTMENT) ANALYSIS, POST TRANSPLANTATION SPECIMEN (EG, HEMATOPOIETIC STEM CELL), INCLUDES COMPARISON TO PREVIOUSLY PERFORMED BASELINE ANALYSES; WITHOUT CELL SELECTION

CHIMERISM (ENGRAFTMENT) ANALYSIS, POST TRANSPLANTATION SPECIMEN (EG, HEMATOPOIETIC STEM CELL), INCLUDES COMPARISON TO PREVIOUSLY PERFORMED BASELINE ANALYSES; WITH CELL SELECTION (EG, CD3, CD33), EACH CELL TYPE

JAK2 (JANUS KINASE 2) (EG, MYELOPROLIFERATIVE DISORDER) GENE ANALYSIS, P.VAL617PHE (V617F) VARIANT

KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA VIRAL ONCOGENE) (EG, CARCINOMA) GENE ANALYSIS, VARIANTS IN CODONS 12 AND 13

LONG QT SYNDROME GENE ANALYSES (EG, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, AND ANK2); FULL SEQUENCE ANALYSIS

LONG QT SYNDROME GENE ANALYSES (EG, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, AND ANK2); KNOWN FAMILIAL SEQUENCE VARIANT

LONG QT SYNDROME GENE ANALYSES (EG, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, AND ANK2); DUPLICATION/DELETION VARIANTS

MGMT (O-6-METHYLGUANINE-DNA METHYLTRANSFERASE) (EG, GLIOBLASTOMA MULTIFORME), METHYLATION ANALYSIS

MCOLN1 (MUCOLIPIDOSIS, TYPE IV) GENE ANALYSIS, COMMON VARIANTS (EG, IVS3 -2A>G, DEL6.4KB)

MTHFR (5,10-METHYLENETETRAHYDROFOLATE REDUCTASE) (EG, HEREDITARY HYPERCOAGULABILITY) GENE ANALYSIS, COMMON VARIANTS (EG, 677T, 1298C)

MLH1 (MUTL HOMOLOG 1, COLON CANCER, NONPOLYPOSIS TYPE 2) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

MLH1 (MUTL HOMOLOG 1, COLON CANCER, NONPOLYPOSIS TYPE 2) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS

MLH1 (MUTL HOMOLOG 1, COLON CANCER, NONPOLYPOSIS TYPE 2) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS

MSH2 (MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

MSH2 (MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS

MSH2 (MUTS HOMOLOG 2, COLON CANCER, NONPOLYPOSIS TYPE 1) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS

MSH6 (MUTS HOMOLOG 6 [E. COLI]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

MSH6 (MUTS HOMOLOG 6 [E. COLI]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS

MSH6 (MUTS HOMOLOG 6 [E. COLI]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS

MICROSATELLITE INSTABILITY ANALYSIS (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) OF MARKERS FOR MISMATCH REPAIR DEFICIENCY (EG, BAT25, BAT26), INCLUDES COMPARISON OF NEOPLASTIC AND NORMAL TISSUE, IF PERFORMED

MECP2 (METHYL CPG BINDING PROTEIN 2) (EG, RETT SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

MECP2 (METHYL CPG BINDING PROTEIN 2) (EG, RETT SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANT

MECP2 (METHYL CPG BINDING PROTEIN 2) (EG, RETT SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS

NPM1 (NUCLEOPHOSMIN) (EG, ACUTE MYELOID LEUKEMIA) GENE ANALYSIS, EXON 12 VARIANTS

PML/RARALPHA, (T(15;17)), (PROMYELOCYTIC LEUKEMIA/RETINOIC ACID RECEPTOR ALPHA) (EG, PROMYELOCYTIC LEUKEMIA) TRANSLOCATION ANALYSIS; COMMON BREAKPOINTS (EG, INTRON 3 AND INTRON 6), QUALITATIVE OR QUANTITATIVE

PML/RARALPHA, (T(15;17)), (PROMYELOCYTIC LEUKEMIA/RETINOIC ACID RECEPTOR ALPHA) (EG, PROMYELOCYTIC LEUKEMIA) TRANSLOCATION ANALYSIS; SINGLE BREAKPOINT (EG, INTRON 3, INTRON 6 OR EXON 6), QUALITATIVE OR QUANTITATIVE

PMS2 (POSTMEIOTIC SEGREGATION INCREASED 2 [S. CEREVISIAE]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

PMS2 (POSTMEIOTIC SEGREGATION INCREASED 2 [S. CEREVISIAE]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANTS
PMS2 (POSTMEIOTIC SEGREGATION INCREASED 2 [S. CEREVISIAE]) (EG, HEREDITARY NON-POLYPOSIS COLORECTAL CANCER, LYNCH SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANTS

PTEN (PHOSPHATASE AND TENSIN HOMOLOG) (EG, COWDEN SYNDROME, PTEN HAMARTOMA TUMOR SYNDROME) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

PTEN (PHOSPHATASE AND TENSIN HOMOLOG) (EG, COWDEN SYNDROME, PTEN HAMARTOMA TUMOR SYNDROME) GENE ANALYSIS; KNOWN FAMILIAL VARIANT

PTEN (PHOSPHATASE AND TENSIN HOMOLOG) (EG, COWDEN SYNDROME, PTEN HAMARTOMA TUMOR SYNDROME) GENE ANALYSIS; DUPLICATION/DELETION VARIANT

PMP22 (PERIPHERAL MYELIN PROTEIN 22) (EG, CHARCOT-MARIE-TOOTH, HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES) GENE ANALYSIS; DUPLICATION/DELETION ANALYSIS

PMP22 (PERIPHERAL MYELIN PROTEIN 22) (EG, CHARCOT-MARIE-TOOTH, HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES) GENE ANALYSIS; FULL SEQUENCE ANALYSIS

PMP22 (PERIPHERAL MYELIN PROTEIN 22) (EG, CHARCOT-MARIE-TOOTH, HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES) GENE ANALYSIS; KNOWN FAMILIAL VARIANT

SMPD1 (SPHINGOMYELIN PHOSPHODIESTERASE 1, ACID LYOSOMAL) (EG, NIEMANN-PICK DISEASE, TYPE A) GENE ANALYSIS, COMMON VARIANTS (EG, R496L, L302P, FSP330)

SNRPN/UBE3A (SMALL NUCLEAR RIBONUCLEOPROTEIN POLYPEPTIDE N AND UBIQUITIN PROTEIN LIGASE E3A) (EG, PRADER-WILLI SYNDROME AND/OR ANGELMAN SYNDROME), METHYLATION ANALYSIS

SERPINA1 (SERPIN PEPTIDASE INHIBITOR, CLADE A, ALPHA-1 ANTIPROTEINASE, ANTITRYPSIN, MEMBER 1) (EG, ALPHA-1-ANTITRYPSIN DEFICIENCY), GENE ANALYSIS, COMMON VARIANTS (EG, *S AND *Z)

TRB@ (T CELL ANTIGEN RECEPTOR, BETA) (EG, LEUKEMIA AND LYMPHOMA), GENE REARRANGEMENT ANALYSIS TO DETECT ABNORMAL CLONAL POPULATION(S); USING AMPLIFICATION METHODOLOGY (EG, POLYMERASE CHAIN REACTION)

TRB@ (T CELL ANTIGEN RECEPTOR, BETA) (EG, LEUKEMIA AND LYMPHOMA), GENE REARRANGEMENT ANALYSIS TO DETECT ABNORMAL CLONAL POPULATION(S); USING DIRECT PROBE METHODOLOGY (EG, SOUTHERN BLOT)

TRG@ (T CELL ANTIGEN RECEPTOR, GAMMA) (EG, LEUKEMIA AND LYMPHOMA), GENE REARRANGEMENT ANALYSIS, EVALUATION TO DETECT ABNORMAL CLONAL POPULATION(S)


VKORC1 (VITAMIN K EPOXIDE REDUCTASE COMPLEX, SUBUNIT 1) (EG, WARFARIN METABOLISM), GENE ANALYSIS, COMMON VARIANTS (EG, -1639/3673)
MOLECULAR PATHOLOGY PROCEDURE, LEVEL 1 (EG, IDENTIFICATION OF SINGLE GERMLINE VARIANT [EG, SNP] BY TECHNIQUES SUCH AS RESTRICTION ENZYME DIGESTION OR MELT CURVE ANALYSIS) ACADM (ACYL-COA DEHYDROGENASE, C-4 TO C-12 STRAIGHT CHAIN, MCAD) (EG, MEDIUM CHAIN ACYL DEHYDROGENASE DEFICIENCY), K304E VARIANT ACE (ANGIOTENSIN CONVERTING ENZYME) (EG, HEREDITARY BLOOD PRESSURE REGULATION), INSERTION/DELETION VARIANT AGTR1 (ANGIOTENSIN II RECEPTOR, TYPE 1) (EG, ESSENTIAL HYPERTENSION), 1166A>C VARIANT BCKDHA (BRANCHED CHAIN KETO ACID DEHYDROGENASE E1, ALPHA POLYPEPTIDE) (EG, MAPLE SYRUP URINE DISEASE, TYPE 1A), Y438N VARIANT CCR5 (CHEMOKINE C-C MOTIF RECEPTOR 5) (EG, HIV RESISTANCE), 32-BP DELETION MUTATION/794 825DEL32 DELETION CLRN1 (CLARIN 1) (EG, USHER SYNDROME, TYPE 3), N48K VARIANT DPYD (DIHYDROPYRIMIDINE DEHYDROGENASE) (EG, 5-FLUOROURACIL/5-FU AND CAPECITABINE DRUG METABOLISM), IVS14+1G>A VARIANT F2 (COAGULATION FACTOR II) (EG, HEREDITARY HYPERCOAGULABILITY), 1199G>A VARIANT F5 (COAGULATION FACTOR V) (EG, HEREDITARY HYPERCOAGULABILITY), HR2 VARIANT F7 (COAGULATION FACTOR VII [SERUM PROTHROMBIN CONVERSION ACCELERATOR]) (EG, HEREDITARY HYPERCOAGULABILITY), R353Q VARIANT F13B (COAGULATION FACTOR XIII, B POLYPEPTIDE) (EG, HEREDITARY HYPERCOAGULABILITY), V34L VARIANT FGB (FIBRINOGEN BETA CHAIN) (EG, HEREDITARY ISCHEMIC HEART DISEASE), -455G>A VARIANT FGFR1 (FIBROBLAST GROWTH FACTOR RECEPTOR 1) (EG, PFIEFFER SYNDROME TYPE 1, CRANIOSYNOSTOSIS), P252R VARIANT FGFR3 (FIBROBLAST GROWTH FACTOR RECEPTOR 3) (EG, MUEKENE SYNDROME), P250R VARIANT FKN (FUKUTIN) (EG, FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY), RETROTRANSPOSON INSERTION VARIANT GNE (GLUCOSAMINE [UDP-N-ACETYL]-2-EPIMERASE/N-ACETYLMANNOSAMINE KINASE) (EG, INCLUSION BODY MYOPATHY 2 [IBM2], NONAKA MYOPATHY), M712T VARIANT HUMAN PLATELET ANTIGEN 1 GENOTYPING (HPA-1), ITGB3 (INTEGRIN, BETA 3 [PLATELET GLYCOPROTEIN IIIA], ANTIGEN CD61 [GPIIIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-1A/B (L33P) HUMAN PLATELET ANTIGEN 2 GENOTYPING (HPA-2), GP1BA (GLYCOPROTEIN IB [PLATELET], ALPHA POLYPEPTIDE [GPIBA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-2A/B (T145M) HUMAN PLATELET ANTIGEN 3 GENOTYPING (HPA-3), ITGA2B (INTEGRIN, ALPHA 2B [PLATELET GLYCOPROTEIN IIB OF II/III COMPLEX], ANTIGEN CD41 [GPIIIB]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-3A/B (I843S) HUMAN PLATELET ANTIGEN 4 GENOTYPING (HPA-4), ITGB3 (INTEGRIN, BETA 3 [PLATELET GLYCOPROTEIN IIIA], ANTIGEN CD61 [GPIIIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-4A/B (R143Q) HUMAN PLATELET ANTIGEN 5 GENOTYPING (HPA-5), ITGA2 (INTEGRIN, ALPHAS 2 [CD49B, ALPHA 2 SUBUNIT OF VLA-2 RECEPTOR] [GPIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-5A/B (K505E) HUMAN PLATELET ANTIGEN 6 GENOTYPING (HPA-6W), ITGB3 (INTEGRIN, BETA 3 [PLATELET GLYCOPROTEIN IIIA], ANTIGEN CD61 [GPIIIA]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-6A/B (R489Q) HUMAN PLATELET ANTIGEN 9 GENOTYPING (HPA-9W), ITGA2B (INTEGRIN, ALPHA 2B [PLATELET GLYCOPROTEIN IIB OF II/III COMPLEX], ANTIGEN CD41 [GPIIIB]) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-9A/B (V837M) HUMAN PLATELET ANTIGEN 15 GENOTYPING (HPA-15), CD109 (CD109 MOLECULE) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-15A/B (CD109) HUMAN PLATELET ANTIGEN 15 GENOTYPING (HPA-15), CD109 (CD109 MOLECULE) (EG, NEONATAL ALLOIMMUNE THROMBOCYTOPENIA [NAIT], POST-TRANSFUSION PURPURA), HPA-15A/B (S682Y) IL28B (INTERLEUKIN 28B [INTERFERON, LAMBDA 3]) (EG, DRUG RESPONSE), RS12979860 VARIANT IVD (ISOVALERYL-COA DEHYDROGENASE) (EG, ISOVALERIC ACIDEMIA), A282V VARIANT LCT (LACTASE-PHLOРИZIN HYDROLASE) (EG, LACTOSE INTOLERANCE), 13910 C>T VARIANT NEB (NEBULIN) (EG, NEMALINE MYOPATHY 2), EXON 55 DELETION VARIANT PCDH15 (PROTOCADHERIN-RELATED 15) (EG, USHER SYNDROME TYPE 1F), R245X VARIANT SERPINE1 (SERPINE PEPTIDASE INHIBITOR CLADE E, MEMBER 1, PLASMINOGEN ACTIVATOR INHIBITOR -1, PAI-1) (EG, THROMBOPHILIA), 4G
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) ABC28 (ATP-BINDING CASSETTE, SUB-FAMILY C [CFTR/MRP], MEMBER 8) (EG, FAMILIAL HYPERINSULINISM), COMMON VARIANTS (EG, C.3898-9G>A [C.3992-9G>A], F1388DEL) ABL (C-ABL ONCOGENE 1, RECEPTOR TYROSINE KINASE) (EG, ACQUIRED IMATINIB RESISTANCE), T315I VARIANT ACADM (ACYL-COA DEHYDROGENASE, C-4 TO C-12 STRAIGHT CHAIN, MCAD) (EG, MEDIUM CHAIN ACYL DEHYDROGENASE DEFICIENCY), COMMONS VARIANTS (EG, K304E, Y42H) ADRB2 (ADRENERGIC BETA-2 RECEPTOR SURFACE) (EG, DRUG METABOLISM), COMMON VARIANTS (EG, G16R, Q27E) AFF2 (AF4/FMR2 FAMILY, MEMBER 2 [FMR2]) (EG, FRAGILE X MENTAL RETARDATION 2 [FRAXE]), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES APOB (APOLIPOPROTEIN B) (EG, FAMILIAL HYPERCHOLESTEROLEMIA TYPE B), COMMON VARIANTS (EG, R3500Q, R3500W) APOE (APOLIPOPROTEIN E) (EG, HYPERLIPOPROTEINEMIA TYPE III, CARDIOVASCULAR DISEASE, ALZHEIMER DISEASE), COMMON VARIANTS (EG, *2, *3, *4) AR (ANDROGEN RECEPTOR) (EG, SPINAL AND BULBAR MUSCULAR ATROPHY, KENNEDY DISEASE, X CHROMOSOME INACTIVATION), CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE OR METHYLATION STATUS) ATN1 (ATROPHIN 1) (EG, DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES ATXN1 (ATAXIN 1) (EG, SPINOCEREBELLAR ATAXIA), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES ATXN2 (ATAXIN 2) (EG, SPINOCEREBELLAR ATAXIA), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES ATXN3 (ATAXIN 3) (EG, SPINOCEREBELLAR ATAXIA, MACHADO-JOSEPH DISEASE), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES ATXN7 (ATAXIN 7) (EG, SPINOCEREBELLAR ATAXIA), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES ATXN8OS (ATXN8 OPPOSITE STRAND [NON-PROTEIN CODING]) (EG, SPINOCEREBELLAR ATAXIA), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES ATXN10 (ATAXIN 10) (EG, SPINOCEREBELLAR ATAXIA), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES CACNA1A (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA 1A SUBUNIT) (EG, SPINOCEREBELLAR ATAXIA), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES CBS (CYSTATHIONINE-BETA-SYNTHASE) (EG, HOMOCYSTINURIA, CYSTATHIONINE BETA-SYNTHASE DEFICIENCY), COMMON VARIANTS (EG, I278T, G307S) CCND1/IGH (BCL1/IGH, T(11;14)) (EG, MANTLE CELL LYMPHOMA) TRANSLOCATION ANALYSIS, MAJOR BREAKPOINT, QUALITATIVE, AND QUANTITATIVE, IF PERFORMED CBS (CYSTATHIONINE-BETA-SYNTHASE) (EG, HOMOCYSTINURIA, CYSTATHIONINE BETA-SYNTHASE DEFICIENCY), COMMON VARIANTS (EG, I278T, G307S) CCND1/IGH (BCL1/IGH, T(11;14)) (EG, MANTLE CELL LYMPHOMA) TRANSLOCATION ANALYSIS, MAJOR BREAKPOINT, QUALITATIVE, AND QUANTITATIVE, IF PERFORMED CFH/ARMS2 (COMPLEMENT FACTOR H/AGE-RELATED MACULOPATHY SUSCEPTIBILITY 2) (EG, MACULAR DEGENERATION), COMMON VARIANTS (EG, Y402H [CFH], A69S [ARMS2]) CNBP (CCHC-TYPE ZINC FINGER, NUCLEIC ACID BINDING PROTEIN) (EG, MYOTONIC DYSTROPHY TYPE 2), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES CSTB (CYSTATIN B [STEFIN B]) (EG, UNVERRICHT-LUNDBORG DISEASE), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES CYP3A4 (CYTOCHROME P450, FAMILY 3, SUBFAMILY A, POLYPEPTIDE 4) (EG, DRUG METABOLISM), COMMON VARIANTS (EG, *2, *3, *4, *5, *6) CYP3A5 (CYTOCHROME P450, FAMILY 3, SUBFAMILY A, POLYPEPTIDE 5) (EG, DRUG METABOLISM), COMMON VARIANTS (EG, *2, *3, *4, *5, *6) DMPK (DYSTROPHIA MYOTONICA-PROTEIN KINASE) (EG, MYOTONIC DYSTROPHY, TYPE 1), EVALUATION TO DETECT ABNORMAL (EG, EXPANDED) ALLELES E2A/PBX1 (T(1;19)) (EG, ACUTE LYMPHOCYTIC LEUKEMIA), TRANSLOCATION ANALYSIS, QUALITATIVE, AND QUANTITATIVE, IF PERFORMED EML4/ALK (INV(2)) (EG, NON-SMALL CELL LUNG CANCER), TRANSLOCATION OR INVERSION ANALYSIS ETV6/NTRK3 (T(12;15)) (EG, CONGENITAL/INFANTILE FIBROSARCOMA), TRANSLOCATION ANALYSIS, QUALITATIVE, AND QUANTITATIVE, IF PERFORMED ETV6/RUNX1 (T(12;21)) (EG, ACUTE LYMPHOCYTIC LEUKEMIA), TRANSLOCATION ANALYSIS, QUALITATIVE, AND QUANTITATIVE, IF PERFORMED EWSR1/ATF1 (T(12;22)) (EG, CLEAR CELL SARCOMA), TRANSLOCATION ANALYSIS, QUALITATIVE, AND QUANTITATIVE, IF PERFORMED EWSR1/ERG (T(21;22)) (EG, EWING SARCOMA/PERIP
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]) CHROMOSOME 18Q- (EG, D18S55, D18S58, D18S61, D18S64, AND D18S69) (EG, COLON CANCER), Allelic imbalance assessment (IE, Loss of heterozygosity) COL1A1/PGDFB (T(17;22)) (EG, DERMATOFIBROSARCOMA PROTUBERANS), Translocation analysis, Multiple breakpoints, Qualitative, and quantitative, IF performed CYP21A2 (Cytochrome P450, FAMILY 21, Subfamily A, Polypeptide 2) (EG, Congenital adrenal hyperplasia, 21-hydroxylase deficiency), Common variants (EG, IVS2-13G, P30L, I172N, EXON 6 mutation cluster [I235N, V236E, M238K], V281L, L307FSX6, Q318X, R356W, P453S, G110VFSX21, 30-KB deletion variant) ESR1/PGR (Receptor 1/Progesterone receptor) Ratio (EG, Breast cancer) IGH@/BCL2 (T(14;18)) (EG, Follicular lymphoma), Translocation analysis; Major breakpoint region (MBR) and minor cluster region (MCR) Breakpoints, Qualitative or Quantitative KIT (V-KIT HARDY-ZUCKERMAN 4 FELINE SARCOMA VIRAL ONCOGENE HOMOLOG) (EG, Mastocytosis), Common variants (EG, D816V, D816Y, D816F) MEFV (MEDITERRANEAN FEVER) (EG, FAMILIAL MEDITERRANEAN FEVER), Common variants (EG, E148Q, P369S, F479L, M680I, I692DEL, M694V, M694I, K695R, V726A, A744S, R761H) MPL (MYELOPROLIFERATIVE LEUKEMIA VIRUS ONCOGENE, THROMBOPOIETIN RECEPTOR, TPOR) (EG, MYELOPROLIFERATIVE DISORDER), Common variants (EG, W515A, W515K, W515L, W515R) TRD@ (T CELL ANTIGEN RECEPTOR, DELTA) (EG, LEUKEMIA AND LYMPHOMA), GENE REARRANGEMENT ANALYSIS, EVALUATION TO DETECT ABNORMAL CLONAL POPULATION UNIPARENTAL DISOMY (UPD) (EG, RUSSELL-SILVER SYNDROME, PRADER-WILLI/ANGELMAN SYNDROME), SHORT TANDEM REPEAT (STR) ANALYSIS
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) ABL1 (C-ABL ONCOPROTEIN 1, RECEPTOR TYROSINE KINASE) (EG, ACQUIRED IMATINIB TYROSINE KINASE INHIBITOR RESISTANCE), VARIANTS IN THE KINASE DOMAIN ANG (ANGIOGENIN, RIBONUCLEASE, RNASE A FAMILY), 5 (EG, AMYOTROPHIC LATERAL SCLEROSIS), FULL GENE SEQUENCE ARX (ARISTALESS-RELATED HOMEBOX) (EG, X-LINKED LISSENCEPHALY WITH AMBIGUOUS GENITALIA, X-LINKED MENTAL RETARDATION), DUPLICATION/DELETION ANALYSIS CEBPA (CCAAT/ENHANCER BINDING PROTEIN [C/EBP], ALPHA) (EG, ACUTE MYELOID LEUKEMIA), FULL GENE SEQUENCE CEL (CARBOXYL ESTER LIPOASE [BILE SALT-STIMULATED LIPOASE]) (EG, MATURITY-ONSET DIABETES OF THE YOUNG [MODY]), TARGETED SEQUENCE ANALYSIS OF EXON 11 (EG, C.1785DELC, C.1686DELT) CTNNB1 (CATENIN [CADHERIN-ASSOCIATED PROTEIN], BETA 1, 88KDA) (EG, DESMOID TUMORS), TARGETED SEQUENCE ANALYSIS (EG, EXON 3) DAZ/SRY (DELETED IN AZOOSPERMIA AND SEX DETERMINING REGION Y) (EG, MALE INFERTILITY), COMMON DELETIONS (EG, AZFA, AZFB, AZFC, AZFD) DNMT3A (DNA [CYTOSINE-5-] Methyltransferase 3 Alpha) (EG, ACUTE MYELOID LEUKEMIA), TARGETED SEQUENCE ANALYSIS (EG, EXON 23) EPCAM (EPITHELIAL CELL ADHESION MOLECULE) (EG, LYMPH SYNDROME), DUPLICATION/DELETION ANALYSIS F8 (COAGULATION FACTOR VIII) (EG, HEMOPHILIA A), INVERSION ANALYSIS, INTRON 1 AND INTRON 22A F12 (COAGULATION FACTOR XII [HAGEMAN FACTOR]) (EG, ANGIOEDEMA, HEREDITARY, TYPE III; FACTOR XII DEFICIENCY), TARGETED SEQUENCE ANALYSIS OF EXON 9 FGFR3 (FIBROBLAST GROWTH FACTOR RECEPTOR 3) (EG, ISOLATED CRANIOSYNOSTOSIS), TARGETED SEQUENCE ANALYSIS (EG, EXON 7) (FOR TARGETED SEQUENCE ANALYSIS OF MULTIPLE FGFR3 EXONS, USE 81404) GJB1 (GAP JUNCTION PROTEIN, BETA 1) (EG, CHARCOT-MARIE-TOOTH X-LINKED), FULL GENE SEQUENCE GNAQ (GUANINE NUCLEOTIDE-BINDING PROTEIN G[Q] SUBUNIT ALPHA) (EG, UVEAL MELANOMA), COMMON VARIANTS (EG, R183, Q209) HBB (HMSOGLLOBIN, BETA, BETA-GLOBIN) (EG, BETA THALASSEMIA), DUPLICATION/DELETION ANALYSIS HRAS (V-HA-RAS HARVEY RAS SARCOMA VIRAL ONCOPROTEIN HOMOLOG) (EG, COSTELLO SYNDROME), EXON 2 SEQUENCE IDH1 (ISOCITRATE DEHYDROGENASE 1 [NADP+], SOLUBLE) (EG, GLIOMA), COMMON EXON 4 VARIANTS (EG, R132H, R132C) IDH2 (ISOCITRATE DEHYDROGENASE 2 [NADP+], MITOCHONDRIAL) (EG, GLIOMA), COMMON EXON 4 VARIANTS (EG, R140W, R172M) JAK2 (JANUS KINASE 2) (EG, MYELOPROLIFERATIVE DISORDER), EXON 12 SEQUENCE AND EXON 13 SEQUENCE, IF PERFORMED KILLER CELL IMMUNOGLOBULIN-LIKE RECEPTOR (KIR) GENE FAMILY (EG, HEMATOPOIETIC STEM CELL TRANSPLANTATION), GENOTYPING OF KIR FAMILY GENES KNOWN FAMILIAL VARIANT NOT OTHERWISE SPECIFIED, FOR GENE LISTED IN TIER 1 OR TIER 2, DNA SEQUENCE ANALYSIS, EACH VARIANT EXON (FOR A KNOWN FAMILIAL VARIANT THAT IS CONSIDERED A COMMON VARIANT, USE SPECIFIC COMMON VARIANT TIER 1 OR TIER 2 CODE) KCN3 (POTASSIUM VOLTAGE-GATED CHANNEL, SHAW-RELATED SUBFAMILY, MEMBER 3) (EG, SPINOCEREBELLAR ATAXIA), TARGETED SEQUENCE ANALYSIS (EG, EXON 2) KCNJ2 (POTASSIUM INWARDLY-RECTIFYING CHANNEL, SUBFAMILY J, MEMBER 2) (EG, ANDERSEN-TAWIL SYNDROME), FULL GENE SEQUENCE KCNJ11 (POTASSIUM INWARDLY-RECTIFYING CHANNEL, SUBFAMILY J, MEMBER 11) (EG, FAMILIAL HYPERINSULINISM), FULL GENE SEQUENCE KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA VIRAL ONCOPROTEIN) (EG, CARCINOMA), GENE ANALYSIS, VARIANT(S) IN EXON 3 (EG, CODON 61) MC4R (MELANOCORTIN 4 RECEPTOR) (EG, OBESITY), FULL GENE SEQUENCE MICA (MH CLASS I POLYPEPTIDE-RELATED SEQUENCE A) (EG, SOLID ORGAN TRANSPLANTATION), COMMON VARIANTS (EG, *001, *002) MPL (MYELOPROLIFERATIVE LEUKEMIA VIRUS ONCOGENE, THROMBOPOIETIN RECEPTOR, TPOR) (EG, MYELOPROLIFERATIVE DISORDER), EXON 10 SEQUENCE MT-RNR1 (MITOCHONDRIALLY ENCODED 12S RNA) (EG, NONSYNDROMIC HEARING LOSS), FULL GENE SEQUENCE MT-TS1 (MITOCHONDRIALLY ENCODED TRNA SERINE 1) (EG, NONSYNDROMIC HEARING LOSS), FULL GENE SEQUENCE NDP (NORRIE DISEASE [PSEUDOGLIOMA]) (EG, NORRIE DISEASE), DUPLICATI

81404

Printed on 5/7/2014. Page 12 of 20
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) ACADS (ACYL-COA DEHYDROGENASE, C-2 TO C-3 SHORT CHAIN) (EG, SHORT CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY), TARGETED SEQUENCE ANALYSIS (EG, EXONS 5 AND 6) AFF2 (AF4/FMR2 FAMILY, MEMBER 2 [FMR2]) (EG, FRAGILE X MENTAL RETARDATION 2 [FRAXE]), CHARACTERIZATION OF ALLELES (EG, EXPANDED SIZE AND METHYLATION STATUS) AQP2 (AQUAPORIN 2 [COLLECTING DUCT]) (EG, NEPHROGENIC DIABETES INSIPIDUS), FULL GENE SEQUENCE ARX (ARISTALESS RELATED HOMEBOX) (EG, X-LINKED LISSENCEPHALY WITH AMBIGUOUS GENITALIA, X-LINKED MENTAL RETARDATION), FULL GENE SEQUENCE AVPR2 (ARGININE VASOPRESSIN RECEPTOR 2) (EG, NEPHROGENIC DIABETES INSIPIDUS), FULL GENE SEQUENCE BBS10 (BARDET-BIEDL SYNDROME 10) (EG, BARDET-BIEDL SYNDROME), FULL GENE SEQUENCE BTD (BIOTINIDASE) (EG, BIOTINIDASE DEFICIENCY), FULL GENE SEQUENCE C10orf2 (CHROMOSOME 10 OPEN READING FRAME 2) (EG, MITOCHONDRIAL DNA DEPLETION SYNDROME), FULL GENE SEQUENCE CAV3 (CAVEOLIN 3) (EG, CAV3-RELATED DISTAL MYOPATHY, LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 1C), FULL GENE SEQUENCE CD40lg (CD40 LIGAND) (EG, HYPER IGM SYNDROME), FULL GENE SEQUENCE CDKN2a (CYCLIN-DEPENDENT KINASE INHIBITOR 2A) (EG, CDKN2a-RELATED CUTANEOUS MALIGNANT MELANOMA, FAMILIAL ATYPICAL MOLE-MALIGNANT MELANOMA SYNDROME), FULL GENE SEQUENCE CLRN1 (CLARIN 1) (EG, USHER SYNDROME, TYPE 3), FULL GENE SEQUENCE COX6B1 (CYTOCHROME C OXIDASE SUBUNIT VIB POLYPEPTIDE 1) (EG, MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX IV DEFICIENCY), FULL GENE SEQUENCE CPT2 (CARNITINE PALMITOYLTRANSFERASE 2) (EG, CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY), FULL GENE SEQUENCE CRX (CONE-ROD DYSTROPHY 2, LEBER CONGENITAL AMAUROSIS), FULL GENE SEQUENCE CSTB (CYSTATIN B [STEFIN B]) (EG, UNVERRICHT-LUNDBORG DISEASE), FULL GENE SEQUENCE CYP1B1 (CYTOCHROME P450, FAMILY 1, SUBFAMILY B, POLYPEPTIDE 1) (EG, PRIMARY CONGENITAL GLAUCOMA), FULL GENE SEQUENCE DMPK (DYSTROPHIA MYOTONICA-PROTEIN KINASE) (EG, MYOTONIC DYSTROPHY TYPE 1), CHARACTERIZATION OF ABNORMAL (EG, EXPANDED) ALLELES EGR2 (EARLY GROWTH RESPONSE 2) (EG, CHARCOT-MARIE-TOOTH), FULL GENE SEQUENCE EMD (EMERIN) (EG, EMERY-DREIFUSS MUSCULAR DYSTROPHY), DUPLICATION/DELETION ANALYSIS EPM2A (EPILEPSY, PROGRESSIVE MYOCLONUS TYPE 2A, LAFORA DISEASE [LAFORIN]) (EG, PROGRESSIVE MYOCLONUS EPILEPSY), FULL GENE SEQUENCE FGF23 (FIBROBLAST GROWTH FACTOR 23) (EG, HYPOPHOSPHATEMIC RICKETS), FULL GENE SEQUENCE FGFR2 (FIBROBLAST GROWTH FACTOR RECEPTOR 2) (EG, CRANIOSYNOSTOSIS, APERT SYNDROME, CROUZON SYNDROME), FULL GENE SEQUENCE FOXG1 (FORKHEAD BOX G1) (EG, RETT SYNDROME), FULL GENE SEQUENCE FSHMD1A (FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY TYPE 1A) (EG, FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY), EVALUATION TO DETECT ABNORMAL (EG, DELETED) ALLELES FSHMD1A (FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A) (EG, FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY), CHARACTERIZATION OF HAPLOTYPE(S) (IE, CHROMOSOME 4A AND 4B HAPLOTYPES) FXN (FRATAXIN) (EG, FRIEDREICH ATAXIA), FULL GENE SEQUENCE GCD1 (GROWTH HORMONE 1) (EG, GROWTH HORMONE DEFICIENCY), FULL GENE SEQUENCE GP1bb (GLYCOPROTEIN 1B [PLATELET], BETA POLYPEPTIDE) (EG, BERNARD-SOULIER SYNDROME TYPE B), FULL GENE SEQUENCE HBA1/HBA2 (ALPHA GLOBIN 1 AND ALPHA GLOBIN 2) (EG, ALPHA THALASSEMIA), DUPLICATION/DELETION ANALYSIS (FOR COMMON DELETION VARIANTS OF ALPHA GLOBIN 1 AND ALPHA GLOBIN 2 GENES, USE 81257) HBB (HEMOGLOBIN, BETA, BETA-GLOB
MOLECULAR PATHOLOGY PROCEDURE, LEVEL 6 (EG, ANALYSIS OF 6-10 EXONS BY DNA SEQUENCE ANALYSIS, MUTATION SCANNING OR DUPLICATION/DELETION VARIANTS OF 11-25 EXONS), REGIONALLY TARGETED CYTOGENOMIC ARRAY ANALYSIS ABCD1 (ATP-BINDING CASSETTE, SUB-FAMILY D [ALD], MEMBER 1) (EG, ADRENOLEUKODYSTROPHY), FULL GENE SEQUENCE ACADS (ACYL-COA DEHYDROGENASE, C-2 TO C-3 SHORT CHAIN) (EG, SHORT CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY), FULL GENE SEQUENCE ACTA2 (ACTIN, ALPHA 2, SMOOTH MUSCLE, AORTA) (EG, THORACIC AORTIC ANEURYSMS AND AORTIC DISSECTIONS), FULL GENE SEQUENCE ACTC1 (ACTIN, ALPHA, CARDIAC MUSCLE 1) (EG, FAMILIAL HYPERTROPHIC CARDIOMYOPATHY), FULL GENE SEQUENCE ANKRD1 (ANKYRIN REPEAT DOMAIN 1) (EG, DILATED CARDIOMYOPATHY), FULL GENE SEQUENCE APTX (APRATAXIN) (EG, ATAXIA WITH OCULOMOTOR APRAXIA 1), FULL GENE SEQUENCE AR (ANDROGEN RECEPTOR) (EG, ANDROGEN INSensitivity SYNDROME), FULL GENE SEQUENCE ARSA (ARYLSULFATASE A) (EG, ARYLSULFATASE A DEFICIENCY), FULL GENE SEQUENCE BCKDHA (BRANCHED CHAIN KETO ACID DEHYDROGENASE E1, ALPHA POLYPEPTIDE) (EG, MAPLE SYRUP URINE DISEASE, TYPE 1A), FULL GENE SEQUENCE BCS1L (BCS1-LIKE [S. CEREVISIAE]) (EG, LEIGH SYNDROME, MITOCHONDRIAL COMPLEX III DEFICIENCY, GRACLE SYNDROME), FULL GENE SEQUENCE BMPR2 (BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE II [SERINE/THREONINE KINASE]) (EG, HERITABLE PULMONARY ARTERIAL HYPERTENSION), DUPLICATION/DELETION ANALYSIS CASQ2 (CALESEQUSTRIN 2 [CARDIAC MUSCLE]) (EG, CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA), FULL GENE SEQUENCE CASR (CALCIUM-SENSING RECEPTOR) (EG, HYPOCALCEMIA), FULL GENE SEQUENCE CDKL5 (CYCLIN-DEPENDENT KINASE-LIKE 5) (EG, EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY), DUPLICATION/DELETION ANALYSIS CHRNA4 (CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA 4) (EG, NOCTURNAL FRONTAL LOBE EPILEPSY), FULL GENE SEQUENCE CHRNBA2 (CHOLINERGIC RECEPTOR, NICOTINIC, BETA 2 [NEURONAL]) (EG, NOCTURNAL FRONTAL LOBE EPILEPSY), FULL GENE SEQUENCE COX10 (COX10 HOMOLOG, CYTOCHROME C OXIDASE ASSEMBLY PROTEIN) (EG, MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX IV DEFICIENCY), FULL GENE SEQUENCE COX15 (COX15 HOMOLOG, CYTOCHROME C OXIDASE ASSEMBLY PROTEIN) (EG, MITOCHONDRIAL RESPIRATORY CHAIN COMPLEX IV DEFICIENCY), FULL GENE SEQUENCE CYP11B1 (CYTOCHROME P450, FAMILY 11, SUBFAMILY B, POLYPEPTIDE 1) (EG, CONGENITAL ADRENAL HYPERPLASIA), FULL GENE SEQUENCE CYP17A1 (CYTOCHROME P450, FAMILY 17, SUBFAMILY A, POLYPEPTIDE 1) (EG, CONGENITAL ADRENAL HYPERPLASIA), FULL GENE SEQUENCE CYP21A2 (CYTOCHROME P450, FAMILY 21, SUBFAMILY A, POLYPEPTIDE 2) (EG, STERIOD 21-HYDROXYLASE ISOFORM, CONGENITAL ADRENAL HYPERPLASIA), FULL GENE SEQUENCE CYTOGENOMIC CONSTITUTIONAL TARGETED MICROARRAY ANALYSIS OF THE X CHROMOSOME BY INTERROGATION OF GENOMIC REGIONS FOR COPY NUMBER AND SINGLE NUCLEOTIDE POLYMORPHISM (SNP) VARIANTS FOR CHROMOSOMAL ABNORMALITIES (WHEN PERFORMING GENOME-WIDE CYTOGENOMIC CONSTITUTIONAL MICROARRAY ANALYSIS, SEE 81228, (DO NOT REPORT ANALYTE-SPECIFIC MOLECULAR PATHOLOGY PROCEDURES SEPARATELY WHEN THE SPECIFIC ANALYTES ARE INCLUDED AS PART OF THE MICROARRAY ANALYSIS OF THE X CHROMOSOME) (DO NOT REPORT 88271 WHEN PERFORMING CYTOGENOMIC MICROARRAY ANALYSIS) CYTOGENOMIC CONSTITUTIONAL TARGETED MICROARRAY ANALYSIS OF CHROMOSOME 22Q13 BY INTERROGATION OF GENOMIC REGIONS FOR COPY NUMBER AND SINGLE NUCLEOTIDE POLYMORPHISM (SNP) VARIANTS FOR CHROMOSOMAL ABNORMALITIES (WHEN PERFORMING GENOME-WIDE CYTOGENOMIC CONSTITUTIONAL MICROARRAY ANALYSIS, SEE 81228, (DO NOT REPORT ANALYTE-SPECIFIC MOLECULAR PATHOLOGY PROCEDURES SEPARATELY WHEN THE SPECIFIC ANALYTES ARE INCLUDED AS PART OF THE MICROARRAY ANALYSIS OF CHROMOSOME 22Q13) (DO NOT REPORT 88271 WHEN PERFORMING CYTOGENOMIC MICROARRAY ANALYSIS) DBT (DIHYDROLIPOAMIDE BRANCHED CHAIN TRANSACYLASE E2) (EG, MAPLE SYRUP URINE DISEASE, TYPE 2), DUPLICATION/DELETION ANALYSIS DCX (DOUBLECORTIN) (EG, X-LINKED LISSENCEPHALY), FULL GENE SEQUENCE DFNB59 (DEAFNESS, AUTOSOMAL RECESSIVE 59) (EG, AUTOSOMAL RECES
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) ABCB8 (ATP-BINDING CASSETTE, SUB-FAMILY C [CFTR/MPR], MEMBER 8) (EG, FAMILIAL HYPERINSULINISM), FULL GENE SEQUENCE AGL (AMYOLO-ALPHA-1, 6-GLUCOSIDASE, 4-ALPHA-GLUCANOTRANSFERASE) (EG, GLYCOCEN STORAGE DISEASE TYPE III), FULL GENE SEQUENCE AHI1 (ABELSON HELPER INTEGRATION SITE 1) (EG, JOUBERT SYNDROME), FULL GENE SEQUENCE ASPM (ASP [ABNORMAL SPINDLE] HOMOLOG, MICROCEPHALY ASSOCIATED [DROSOPHILA]) (EG, PRIMARY MICROCEPHALY), FULL GENE SEQUENCE CACNA1A (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA 1A SUBUNIT) (EG, FAMILIAL HEMIPLEGIC MIGRAINE), FULL GENE SEQUENCE CHD7 (CHROMODOMAIN HELICASE DNA BINDING PROTEIN 7) (EG, CHARGE SYNDROME), FULL GENE SEQUENCE COL4A4 (COLLAGEN, TYPE IV, ALPHA 4) (EG, ALPORT SYNDROME), FULL GENE SEQUENCE COL4A5 (COLLAGEN, TYPE IV, ALPHA 5) (EG, ALPORT SYNDROME), DUPLICATION/DELETION ANALYSIS COL6A1 (COLLAGEN, TYPE VI, ALPHA 1) (EG, COLLAGEN TYPE VI-RELATED DISORDERS), FULL GENE SEQUENCE COL6A2 (COLLAGEN, TYPE VI, ALPHA 2) (EG, COLLAGEN TYPE VI-RELATED DISORDERS), FULL GENE SEQUENCE COL6A3 (COLLAGEN, TYPE VI, ALPHA 3) (EG, COLLAGEN TYPE VI-RELATED DISORDERS), FULL GENE SEQUENCE CREBBP (CREB BINDING PROTEIN) (EG, RUBINSTEIN-TAYBI SYNDROME), FULL GENE SEQUENCE F8 (COAGULATION FACTOR VIII) (EG, HEMOPHILIA A), FULL GENE SEQUENCE JAG1 (JAGGED 1) (EG, ALAGILLE SYNDROME), FULL GENE SEQUENCE KDM5C (LYSINE [K]-SPECIFIC DEMETHYLASE 5C) (EG, X-LINKED MENTAL RETARDATION), FULL GENE SEQUENCE KIAA0196 (KIAA0196) (EG, SPASTIC PARAPLEGIA), FULL GENE SEQUENCE L1CAM (L1 CELL ADHESION MOLECULE) (EG, MASA SYNDROME, X-LINKED HYDROCEPHALY), FULL GENE SEQUENCE MYBPC3 (MYOSIN BINDING PROTEIN C, CARDIAC) (EG, FAMILIAL HYPTERTROPHIC CARDIOMYOPATHY), FULL GENE SEQUENCE MYH6 (MYOSIN, HEAVY CHAIN 6, CARDIAC MUSCLE, ALPHA) (EG, FAMILIAL DILATED CARDIOMYOPATHY), FULL GENE SEQUENCE MYH7 (MYOSIN, HEAVY CHAIN 7, CARDIAC MUSCLE, BETA) (EG, FAMILIAL HYPTERTROPHIC CARDIOMYOPATHY, LIANG DISTAL MYOPATHY), FULL GENE SEQUENCE MYO7A (MYOSIN VIIA) (EG, USHER SYNDROME, TYPE 1), FULL GENE SEQUENCE NOTCH1 (NOTCH 1) (EG, AORTIC VALVE DISEASE), FULL GENE SEQUENCE NPHS1 (NEPHROSIS 1, CONGENITAL, FINNISH TYPE [NEPHRIN]) (EG, CONGENITAL FINNISH NEPHROSIS), FULL GENE SEQUENCE OPAL (OPTIC ATROPHY 1) (EG, OPTIC ATROPHY), FULL GENE SEQUENCE PCDH15 (PROTOCADHERIN-RELATED 15) (EG, USHER SYNDROME, TYPE 1), FULL GENE SEQUENCE PKD1 (POLYCYSTIC KIDNEY DISEASE 1 [AUTOSOMAL DOMINANT]) (EG, POLYCYSTIC KIDNEY DISEASE), FULL GENE SEQUENCE PLCB1 (PHOSPHOLIPASE C, EPSILON 1) (EG, NEPHROTIC SYNDROME TYPE 3), FULL GENE SEQUENCE SCN1A (SODIUM CHANNEL, VOLTAGE-GATED, TYPE 1, ALPHA SUBUNIT) (EG, GENERALIZED EPILEPSY WITH FEBRILE SEIZURES), FULL GENE SEQUENCE SCN5A (SODIUM CHANNEL, VOLTAGE-GATED, TYPE V, ALPHA SUBUNIT) (EG, FAMILIAL DILATED CARDIOMYOPATHY), FULL GENE SEQUENCE SLC12A1 (SOLUTE CARRIER FAMILY 12 [SODIUM/POTASSIUM/CHLORIDE TRANSPORTERS], MEMBER 1) (EG, BARTTER SYNDROME), FULL GENE SEQUENCE SLC12A3 (SOLUTE CARRIER FAMILY 12 [SODIUM/CHLORIDE TRANSPORTERS], MEMBER 3) (EG, GITELMAN SYNDROME), FULL GENE SEQUENCE SPG11 (SPASTIC PARAPLEGIA 11 [AUTOSOMAL RECESSIVE]) (EG, SPASTIC PARAPLEGIA), FULL GENE SEQUENCE SPTBN2 (SPECTRIN, BETA, NON-ERYTHROCYTIC 2) (EG, SPINOCEREBELLAR ATAXIA), FULL GENE SEQUENCE TMEM67 (TRANSMEMBRANE PROTEIN 67) (EG, JOUBERT SYNDROME), FULL GENE SEQUENCE TSC2 (TUBEROUS SCLEROSIS 2) (EG, TUBEROUS SCLEROSIS), FULL GENE SEQUENCE USH1C (USHER SYNDROME 1C [AUTOSOMAL RECESSIVE, SEVERE]) (EG, USHER SYNDROME, TYPE 1), FULL GENE SEQUENCE VPS13B (VACUOLAR PROTEIN SORTING 13 HOMOLOG B [YEAST]) (EG, COHEN SYNDROME), DUPLICATION/DELETION ANALYSIS WDR62 (WD REPEAT DOMAIN 62) (EG, PRIMARY AUTOSOMAL RECESSIVE MICROCEPHALY), FULL GENE SEQUENCE
General Information

Associated Information

Documentation Requirements

Documentation must be adequate to verify that coverage guidelines listed above have been met. Thus, the medical record must contain documentation that the testing is expected to influence treatment of the condition toward which the testing is directed. The laboratory or billing provider must have on file the physician requisition which sets forth the diagnosis or condition (ICD-9-CM code) that warrants the test(s).

Examples of documentation requirements of the ordering physician/nonphysician practitioner (NPP) include, but are not limited to, history and physical or exam findings that support the decision making, problems/diagnoses, relevant data (e.g., lab testing, imaging results).
Documentation requirements of the performing laboratory (when requested) include, but are not limited to, lab accreditation, test requisition, test record/procedures, reports (preliminary and final), and quality control record.

Documentation requirements for LDT(s)/protocols (when requested) include diagnostic test/assay, lab/manufacturer, names of comparable assays/services (if relevant), description of assay, analytical validity evidence, clinical validity evidence, and clinical utility.

Providers are required to code to specificity however, if CPT 81479 (unlisted molecular pathology procedure) is used the documentation must clearly identify the unique molecular pathology procedure performed. When multiple procedure codes are submitted on a claim (unique and/or unlisted) the documentation supporting each code should be easily identifiable. If on review the contractor cannot link a billed code to the documentation, these services will be denied based on Title XVIII of the Social Security Act, §1833(e).

For these tests, the ordering provider must provide to the laboratory copies of the signed informed consent documentation.

An Advance Beneficiary Notice of Noncoverage (ABN) is required before furnishing a beneficiary a test which the physician or laboratory believes to be noncovered by Medicare as not reasonable or necessary. The physician or laboratory must obtain a signed ABN from the beneficiary (or representative) that the physician or laboratory has informed him/her on the non-coverage of the test and that there will be a charge for the test.

When the documentation does not meet the criteria for the service rendered or the documentation does not establish the medical necessity for the services, such services will be denied as not reasonable and necessary under Section 1862(a)(1)(A) of the Social Security Act.

**Utilization Guidelines**

Screening services such as pre-symptomatic genetic tests and services used to detect an undiagnosed disease or disease predisposition are not a Medicare benefit and are not covered. Similarly, Medicare may not reimburse the costs of tests/examinations that assess the risk of a condition unless the risk assessment clearly and directly effects the management of the patient.

Title XVIII of the Social Security Act (SSA) §1862(a)(1)(A) states that no Medicare payment shall be made for items and services which are not reasonable and necessary for the diagnosis or treatment of illness or injury. Based on this statute, CMS states that "tests that are performed in the absence of signs, symptoms, complaints, or personal history of disease or injury are non-covered unless explicitly authorized by statute."

A specific genetic test may only be performed once in a lifetime per beneficiary for inherited conditions; however, when medically reasonable and necessary, genetic testing may be done on acquired conditions such as malignancies (including separate malignancies developing at different times) as they are treated and are being followed, in order to assess response or other relevant clinical criteria. Likewise, there are situations where medical record and literature documentation are able to demonstrate that serial testing can be reasonably predicted to provide additional clinically useful information. When the record documents that this information, such as confirmed significant response to current therapy, is likely to assist in modifying treatment, serial testing can be considered reasonable and necessary and eligible for coverage.

**Appendices**

N/A

Sources of Information and Basis for Decision


LCDs and policies from other Medicare contractors and private insurers


**Revision History Information**

Please note: Most Revision History entries effective on or before 01/24/2013 display with a Revision History Number of "R1" at the bottom of this table. However, there may be LCDs where these entries will display as a separate and distinct row.

<table>
<thead>
<tr>
<th>Revision History Date</th>
<th>Revision History Number</th>
<th>Revision History Explanation</th>
</tr>
</thead>
<tbody>
<tr>
<td>03/01/2014 R1</td>
<td></td>
<td>Added the following references to Sources of Information:</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Added the following references to CMS National Coverage Policy section:</td>
</tr>
<tr>
<td></td>
<td></td>
<td>CMS Publication 100-02, <em>Medicare Benefit Policy Manual</em>, Chapter 15, Section 80.6. 5 which describes the Surgical/Cytopathology Exception.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>CMS National Correct Coding Initiative (NCCI) <em>Policy Manual for Medicare Services</em>, Chapter 10 Pathology/Laboratory Services which addresses reflex testing.</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Under the Indications of Coverage section, revised the language in the 5th bullet from &quot;to establish a diagnosis&quot; to &quot;to obtain necessary information for therapeutic decision making&quot;</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Under CPT/HCPCS Codes section, added CPT code 81287, effective for services rendered on or after 1/1/2014</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Provider Education/Guidance</td>
</tr>
<tr>
<td></td>
<td></td>
<td>• Aberrant Local Utilization</td>
</tr>
</tbody>
</table>

**Associated Documents**

Attachments [Molecular Pathology Procedures opens in new window](#) (a comment and response document) (PDF - Printed on 5/7/2014. Page 19 of 20
Keywords

- Molecular, genes, genetics, molecular pathology, gene tests, genetic testing, genetic screening, lab tests