Information Session: Discussion on New Genetic Testing Codes

Place: Centers for Medicare & Medicaid Services (CMS) Auditorium
7500 Security Boulevard
Baltimore, Maryland 21244-1850
Date: Monday, July 18, 2011
Time: 11:15a.m.—5:00 p.m. Eastern Standard Time (EST)
Check-in will begin at 8:30 a.m. EST

Immediately following the Laboratory Public Meeting on Payment for New Clinical Laboratory Tests for 2012, we will host a separate information session aimed at discussing new genetic testing codes. While we are not accepting payment recommendations at this time for genetic testing, we would like to open discussions to the public on recommendations going forward on how these codes should be addressed. It would be most helpful if we received supporting information such as –

1. Clinical Laboratory Fee Schedule (CLFS) vs. Physician Fee Schedule (PFS) assignment.
2. Current CPT codes utilized to reflect test steps.
3. How various genetic tests are similar to/different from existing laboratory tests.

Registration
Registration will begin June 15, 2011. To register for the meeting, individuals should complete the internet registration form by July 8, 2011, at www.cms.hhs.gov/ClinicalLabFeeSched/. A confirmation will be sent upon receipt of the registration by CMS. Individuals who wish to make a presentation to give recommendations on how genetic testing codes should be addressed should complete this information on the registration form, prepare a brief presentation (not to exceed 15 minutes), and provide three written copies at the time of presentation. Presenters may also make copies available for up to 50 meeting participants.

An agenda will be published at www.cms.hhs.gov/ClinicalLabFeeSched/ showing a list of presenters.

Security
The meeting will be held in a Federal government building; therefore, Federal security measures are applicable. Photo identification and registration confirmation will be required to enter the building. Vehicles are inspected, at entrance to the grounds, and persons must pass through a metal detector when entering the building. Directions and other information for visitors to the building are available at www.cms.hhs.gov/CMSHeadquarters.

Audio Listening
In addition, individuals may listen to the public meeting by dialing 877-267-1577. Registration is not required for audio listening. Registration, in-person attendance, and three written copies of the presentation are necessary to make a presentation on the assignment of payment levels at this meeting. After the on-site presentations, a question and answer period will be opened to both the participants in the room and audio listeners. The moderator of the meeting will monitor the question and answer period.

New Genetic Test Codes for 2012 Medicare
The following is a list of the newly created genetic codes that will be discussed. The coding changes have been developed by the American Medical Association's (AMA) Current Procedural Terminology (CPT) Editorial Panel and will not be further discussed at the CMS public meeting. Numbering of all new codes has not yet been finalized. The identifying information should be sufficient for those knowledgeable in coding.

As stated above, this listing represents the major actions of the CPT Editorial Panel for CPT 2012 taken at its June, October, February, and Final meetings. Please note that these codes are the most recent listing provided by the AMA, and are subject to change. Any changes will be updated as they occur.

Molecular Pathology Procedures – Tier 1

1. **812XX** ASPA (aspartoacylase (eg, Canavan disease) gene analysis, common variants (eg, E285A, Y231X)

2. **812XX** BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (eg, Maple syrup urine disease) gene analysis, common variants (eg, R183P, G278S, E422X)

3. **812XX** BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; major breakpoint, qualitative or quantitative

4. **812XX** BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; minor breakpoint, qualitative or quantitative

5. **812XX** BCR/ABL1 (t(9;22)) (eg, chronic myelogenous leukemia) translocation analysis; other breakpoint, qualitative or quantitative

6. **812XX** BLM (Bloom syndrome, RecQ helicase-like) (eg, Bloom syndrome) gene analysis, 2281del6ins7 variant

7. **812XX** BRAF (v-raf murine sarcoma viral oncogene humolog B1) (eg, colon cancer), gene analysis, V600E variant

8. **812XX** 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 26 kb, exon 22 del 510bp, exon 8-9 del 7.1kb)

9. **812XX** BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; 185delAG, 5385insC, 6174delT variants

10. **812XX** BRCA1, BRCA2 (breast cancer 1 and 2) (eg, hereditary breast and ovarian cancer) gene analysis; uncommon duplication/deletion variants

11. **812XX** 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)

12. **812XX** BRCA1 (breast cancer 1) (eg, hereditary breast and ovarian cancer) gene analysis; known familial variant

13. **812XX** BRCA2 (breast cancer 2) (eg, hereditary breast and ovarian cancer) gene analysis; full sequence analysis

14. **812XX** BRCA2 (breast cancer 2) (eg, hereditary breast and ovarian cancer) gene analysis; known familial variant

15. **812XX** CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; common variants (eg, ACMG/ACOG guidelines)

16. **812XX** CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; known familial variants

17. **812XX** CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; duplication/deletion variants

18. **812XX** CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; full gene sequence

19. **812XX** CFTR (cystic fibrosis transmembrane conductance regulator) (eg, cystic fibrosis) gene analysis; intron 8 poly-T analysis (eg, male infertility)

20. **812XX** CYP2C19 (cytochrome P450, family 2, subfamily C, polypeptide 19) (eg, drug metabolism), gene analysis, common variants (eg, *2, *3, *4, *8, *17)

21. **812XX** CYP2D6 (cytochrome P450, family 2, subfamily D, polypeptide 6) (eg, drug metabolism), gene analysis, common variants (eg, *2, *3, *4, *5, *6, *9, *10, *17, *19, *29, *35, *41, *1XN, *2XN, *4XN)

22. **812XX** CYP2C9 (cytochrome P450, family 2, subfamily C, polypeptide 9) (eg, drug metabolism), gene analysis, common variants (eg, *2, *3, *5, *6)

23. **812XX** 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)

24. **812XX** Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants for chromosomal abnormalities

25. **812XX** F2 (prothrombin, coagulation factor II) (eg, hereditary hypercoagulability) gene analysis, 20210G>A variant

26. **812XX** F5 (coagulation Factor V) (eg, hereditary hypercoagulability) gene analysis, Leiden variant

27. **812XX** FANCC (Fanconi anemia, complementation group C) (eg, Fanconi anemia, type C) gene analysis, common variant (eg, IVS4+4A>T)

28. **812XX** FMR1 (Fragile 1 mental retardation 1) (eg, fragile X mental retardation) gene analysis; evaluation to detect abnormal (eg, expanded) alleles

29. **812XX** FMR1 (Fragile 1 mental retardation 1) (eg, fragile X mental retardation) gene analysis; characterization of alleles (eg, expanded size and methylation status)

30. **812XX** FLT3 (fms-related tyrosine kinase 3) (eg, acute myeloid leukemia), gene analysis, internal tandem duplication (ITD) variants (ie, exons 14, 15)

31. **812XX** G6PC (glucose-6-phosphatase, catalytic subunit) (eg, Glycogen storage disease, Type 1a, von Gierke disease) gene analysis, common variants (eg, R83C, Q347X)

32. **812XX** GBA (glucosidase, beta, acid) (eg, Gaucher disease) gene analysis, common variants (eg, N370S, 84GG, L444P, IVS2+1G>A)

33. **812XX** HEXA (hexosaminidase A (alpha polypeptide) (eg, Tay-Sachs disease) gene analysis, common variants (eg, 1278insTATC, 1421+1G>C, G269S)

34. **812XX** HFE (hemochromatosis) (eg, hereditary hemochromatosis) gene analysis, common variants (eg, C282Y, H63D)

35. **812XX** 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)

36. **812XX** 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)

37. **812XX** 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)

38. **812XX** 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)

39. **812XX** IGH@ (Immunoglobulin heavy chain locus) (eg, leukemia and lymphoma, B-cell), variable region somatic mutation analysis

40. **812XX** IGK@ (Immunoglobulin kappa light chain locus) (eg, leukemia and lymphoma, B-cell), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)

41. **812XX** 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

42. **812XX** 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)

43. **812XX** Chimerism (engraftment) analysis, post hematopoietic stem cell transplantation specimen, includes comparison to previously performed baseline analyses; without cell selection

44. **812XX** Chimerism (engraftment) analysis, post hematopoietic stem cell transplantation specimen, includes comparison to previously performed baseline analyses; with cell selection (eg, CD3, CD33), each cell type

45. **812XX** JAK2 (Janus kinase 2) (eg, myeloproliferative disorder) gene analysis, p.Val617Phe (V617F) variant

46. **812XX** KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene) (eg, carcinoma) gene analysis, variants in codons 12 and 13

47. **812XX** Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); full sequence analysis

48. **812XX** Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); known familial sequence variant

49. **812XX** Long QT syndrome gene analyses (eg, KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP, SNTA1, and ANK2); duplication/deletion variants

50. **812XX** MCOLN1 (mucolipin 1) (eg, Mucolipidosis, type IV) gene alalysis, common variants (eg, IVS3-2A>G, del6.4kb)

51. **812XX** MTHFR (5, 10-methylenetetrahydrofolate reductase) (eg, hereditary hypercoagulability) gene alalysis, common variants (eg, 677T, 1298C)

52. **812XX** MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

53. **812XX** MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

54. **812XX** MLH1 (mutL homolog 1, colon cancer, nonpolyposis type 2) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

55. **812XX** MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

56. **812XX** MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

57. **812XX** MSH2 (mutS homolog 2, colon cancer, nonpolyposis type 1) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

58. **812XX** MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

59. **812XX** MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

60. **813XX** MSH6 (mutS homolog 6 [E. coli]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

61. **813XX** 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

62. **813XX** MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; full sequence analysis

63. **813XX** MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; known familial variant

64. **813XX** MECP2 (methyl CpG binding protein 2) (eg, Rett syndrome) gene analysis; duplication/deletion variants

65. **813XX** NPM1 (nucleophosmin) (eg, acute myeloid leukemia) gene analysis; exon 12 variants

66. **813XX** PML/RARalpha, (t(15;17)), (PML-RARA regulated adaptor molecule 1) (eg, promyelocytic leukemia) translocation analysis; common breakpoints (eg, intron 3 and intron 6), qualitative or quantitative

67. **813XX** PML/RARalpha, (t(15;17)), (PML-RARA regulated adaptor molecule 1) (eg, promyelocytic leukemia) translocation analysis; single breakpoint (eg, intron 3, intron 6, or exon 6), qualitative or quantitative

68. **813XX** PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis

69. **813XX** PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants

70. **813XX** PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

71. **813XX** SMPD1 (sphingomyelin phosphodiesterase 1, acid sysosomal) (eg, Niemann-Pick disease, Type A) gene analysis, common variants (eg, R496L, L302P, fsP330)

72. **813XX** SNRPN/UBE3A, (small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A) (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis

73. **813XX** 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)

74. **813XX** TCB@ (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)

75. **813XX** TCB@ (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)

76. **813XX** TCG@ (T cell receptor, gamma) (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)

77. **813XX** UGT1A1 (UDP glucuronosyltransferase 1 family, polypeptide A1) (eg, irinotecan metabolism), gene analysis, common variants (eg, *28, *36, *37)

78. **813XX** VKORC1 (vitamin K epoxide reductase complex, subunit 1) (eg, warfarin metabolism), gene analysis, common variants (eg, -1639/3673)

79. **813XX** HLA Class I and II typing, low resolution (eg, antigen equivalents); HLA-A, -B, -C, -DRB1/3/4/5, and -DQB1

80. **813XX** HLA Class I and II typing, low resolution (eg, antigen equivalents); HLA-A, -B, and -DRB1/3/4/5 (eg, verification typing)

81. **813XX** HLA Class I typing, low resolution (eg, antigen equivalents); complete (ie, HLA-A, -B, and -C)

82. **813XX** HLA Class I typing, low resolution (eg, antigen equivalents); one locus (eg, HLA-A, -B, or -C), each

83. **813XX** HLA Class I typing, low resolution (eg, antigen equivalents); one antigen equivalent (eg, B*27), each

84. **813XX** HLA Class II typing, low resolution (eg, antigen equivalents); HLA-DRB1/3/4/5 and -DQB1

85. **813XX** HLA Class II typing, low resolution (eg, antigen equivalents); one locus (eg, HLA-DRB1/3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1), each

86. **813XX** HLA Class II typing, low resolution (eg, antigen equivalents); one antigen equivalent, each

87. **813XX** HLA Class I and II typing, high resolution (ie, alleles or allele groups), HLA-A, -B, -C, and -DRB1

88. **813XX** HLA Class I typing, high resolution (ie, alleles or allele groups); complete (ie, HLA-A, -B, and -C)

89. **813XX** HLA Class I typing, high resolution (ie, alleles or allele groups); one locus (eg, HLA-A, -B, or -C), each

90. **813XX** HLA Class I typing, high resolution (ie, alleles or allele groups); one allele or allele group (eg, B*57:01P), each

91. **813XX** HLA Class II typing, high resolution (ie, alleles or allele groups); one locus (eg, HLA-DRB1, -DRB3, -DRB4, -DRB5, -DQB1, -DQA1, -DPB1, or -DPA1), each

92. **813XX** HLA Class II typing, high resolution (ie, alleles or allele groups); one allele or allele group (eg, HLA-DQB1*06:02P), each

Medicare Pathology Procedures – Tier 2

1. **814XX** 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

CCR5 (chemokine C-C motif receptor 5) (eg, HIV resistance), 32 bp deletion mutation/794 825del32 deletion

DPYD (dihydropyrimidine dehydrogenase) (eg, 5-fluorouracil/5-FU and capecitabine drug metabolism), IVS14+1G>A variant

F2 (coagulation factor 2) (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

SERPINE1 (serpine peptidase inhibitor clade E, member 1, plasminogen activator inhibitor -1, PAI-1) (eg, thrombophilia), 4G variant

2. **814XX** 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)

ABL (c-abl oncogene 1, receptor tyrosine kinase) (eg, acquired imatinib resistance), T315I variant

ACADM (acyl-CoA deydrogenase, 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)

APOE (apolipoprotein E) (eg, hyperlipoproteinemia type III, cardiovascular disease, Alzheimer disease), common variants (eg, *2, *3, *4)

CBFB/MYH11 (inv(16)) (eg, acute myeloid leukemia), qualitative, and quantitative, if performed

CCND1/lgH (BCL1/lgH, t[(11;14]d) (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])

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

F11 (coagulation factor XI) (eg, coagulation disorder), common variants (eg, E117X [Type II], F283L [Type III], IVS14del14, and IVS14+1G>A [Type I])

FGFR3 (fibroblast growth factor receptor 3) (eg, achondroplasia), common variants (eg, 1138G>A, 1138G>C)

FIP1L1/PDGFRA (del[4q12]) (eg, imatinib-sensitive chronic eosinophilic leukemia), qualitative, and quantitative, if performed

GALT (galactose-1-phosphate uridylyltransferase (eg, galactosemia), common variants (eg, Q188R, S135L, K285N, T138M, L195P, Y209C, IVS2-2A>G, P171S, del5kb, N314D, L218L/N314D

HBB (hemoglobin, beta) (eg, sickle cell anemia, hemoglobin C, hemoglobin E), common variants (eg, HbS, HbC, HbE)

HTT (huntingtin) (eg, Huntington disease), evaluation to detect abnormal (eg, expanded) alleles

RUNX1/RUNX1T1 (t([8;21])) (eg, acute myeloid leukemia) translocation analysis, qualitative, and quantitative, if performed

TPMT (thiopurine S-methyltransferase) (eg, drug metabolism), common variants (eg, *2, *3)

VWF (von Willebrand factor) (eg, von Willebrand disease type 2N), common variants (eg, T791M, R816W, R854Q)

3. **814XX** 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 1 exon)

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, L307FfsX6, Q318X, R356W, P453S, G110VfsX21, 30-kb deletion variant

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)

TCD@ (T cell antigen receptor, delta) (eg, leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population

4. **814XX** 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 oncogene1, receptor tyrosine kinase) (eg, acquired imatinib tyrosine kinase inhibitor resistance), variants in the kinase domain

DAZ/SRY (deleted in azoospermia and sex determining region Y) (eg, male infertility), common deletions (eg, AZFa, AZFb, AZFc, AZFd)

JAK2 (Janus kinase 2) (eg, myeloproliferative disorder), exon 12 sequence and exon 13 sequence, if performed

MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (eg, myeloproliferative disorder), exon 10 sequence

VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), deletion/duplication analysis

VWF (von Willebrand factor) (eg, von Willebrand disease types 2A, 2B, 2M), targeted sequence analysis (eg, exon 28)

5. **814XX** 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)

BTD (biotinidase) (eg, biotinidase deficiency), 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

FKRP (Fukutin related protein) (eg, congenital muscular dystrophy type 1C [MDC1C], limb-girdle muscular dystrophy [LGMD] type 2I), full gene sequence

FOXG1 (forkhead box G1) (eg, Rett syndrome), full gene sequence

FSHMD1A (facioscapulohumeral muscular dystrophy 1A) (eg, facioscapulohumeral muscular dystrophy), evaluation to detect abnormal (eg, deleted) alleles

HBB (hemoglobin, beta, Beta-Globin) (eg, thalassemia), full gene sequence

KIT (C-kit) (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (eg, GIST, acute myeloid leukemia, melanoma), targeted gene analysis (eg, exons 8, 11, 13, 17, 18)

MEFV (Mediterranean fever) (eg, familial Mediterranean fever), full gene sequence

NRAS (neuroblastoma RAS viral oncogene homolog) (eg, colorectal carcinoma), exon 1 and exon 2 sequences

PDGFRA (platelet-derived growth factor receptor alph polypeptide) (eg, gastrointestinal stromal tumor), targeted sequence analysis (eg, exons 12, 18)

RET (ret proto-oncogene) (eg, multiple endocrine neoplasia, type 2B and familial medullary thyroid carcinoma), common variants (eg, M918T, 2647_2648delinsTT, A883F)

SDHD (succinate dehydrogenase complex, subunit D, integral membrane protein) (eg, hereditary paraganglioma), full gene sequence

VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), full gene sequence

VWF (von Willebrand factor) (eg, von Willebrand disease type 1C), targeted sequence analysis (eg, exons 26, 27, 37)

6. **814XX** 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

CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide2) (eg, steroid 21-hydroxylase isoform, congenital adrenal hyperplasia), full gene sequence

FKTN (Fukutin) (eg, limb-girdle muscular dystrophy [LGMD] type 2M or 2L), full gene sequence

RET (ret proto-oncogene) (eg, multiple endocrine neoplasia, type 2A and familial medullary thyroid carcinoma), targeted sequence analysis (eg, exons 10, 11, 13-16)

SDHB (succinate dehydrogenase complex, subunit B, iron sulfur) (eg, hereditary paraganglioma), full gene sequence

TGFBR1 (transforming growth factor, beta receptor 1) (eg, Marfan syndrome), full gene sequence

TGFBR2 (transforming growth factor, beta receptor 2) (eg, Marfan syndrome), full gene sequence

THRB (thyroid hormone receptor, beta) (eg, thyroid hormone resistance, thyroid hormone beta receptor deficiency), full gene sequence or targeted sequence analysis of >5 exons

TP53 (tumor protein 53) (eg, Li-Fraumeni syndrome, tumor samples), full gene sequence or targeted sequence analysis of >5 exons

VWF (von Willebrand factor) (eg, von Willebrand disease type 2N), targeted sequence analysis (eg, exons 18-20, 23-25)

7. **814XX** Molecular pathology procedure, Level 7 (eg, analysis of 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons, cytogenomic array analysis for neoplasia)

CAPN3 (Calpain 3) (eg, limb-girdle muscular dystrophy [LGMD] type 2A, alpainopathy), full gene sequence

GALT (galactose-1-phosphate uridylyltransferase (eg, galactosemia), full gene sequence

HEXA (hexosaminidase A, alpha polypeptide) (eg, Tay-Sachs disease), full gene sequence

LMNA (lamin A/C) (eg, Emery-Dreifuss muscular dystrophy [EDMD1, 2 and 3] limb-girdle muscular dystrophy [LGMD] type 1B, dilated cardiomyopathy [CMD1A], familial partial lipodystrophy [FPLD2]), full gene sequence

PAH (phenylalanine hydroxylase) (eg, phenylketonuria), full gene sequence

POMGNT1 (protein O-linked mannose beta1,2-N acetylglucosaminytransferase) (eg, Muscle-Eye-Brain disease, Walker-Warburg syndrome), full gene sequence

POMT1 (protein-O-mannosyltransferase 1) (eg, limb-girdle muscular dystrophy [LGMD] type 2K, Walker-Warburg syndrome), full gene sequence

POMT2 (protein-O-mannosyltransferase 2) (eg, limb-girdle muscular dystrophy [LGMD] type 2N, Walker-Warburg syndrome), full gene sequence

RYR1 (ryanodine receptor 1, skeletal) (eg, malignant hyperthermia), targeted sequence analysis of exons with functionally-confirmed mutations

VWF (von Willebrand factor) (von Willebrand disease type 2A), extended targeted sequence analysis (eg, exons 11-16, 24-26, 51, 52)

8. **814XX** 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)

   SCN1A (sodium channel, voltage-gated, type 1, alpha subunit) (eg, generalized epilepsy with febrile seizures), full gene sequence

9. **814XX** Molecular pathology procedure, Level 9 (eg, analysis of >50 exons in a single gene by DNA sequence analysis)

   FBN1 (fibrillin 1) (eg, Marfan syndrome), full gene sequence

   NF1 (neurofibromin 1) (eg, neurofibromatosis, type 1), full gene sequence

   RYR1 (ryanodine receptor 1, skeletal) (eg, malignant hyperthermia), full gene sequence

   VWF (von Willebrand factor) (eg, von Willebrand disease types 1 and 3), full gene sequence