LABHORIZONS
A NEWSLETTER FOR CLIENTS
Volume XIX, No. 4 — April 2019

New Procedures

11-Hydroxyandrostenedione 504677

CPT 82542

Use 11-oxo-androgens are emerging biomarkers for androgen production of adrenal origin. These biomarkers will be useful in disease in men and women and children. Although the presence of androgens with oxygen at the 11 position of the steroid backbone have been known for some time, the clinical utility and prevalence of these androgens have only recently come to light. As has been the case historically, the research has been enhanced with availability of good assay techniques, in this case HPLC MS/MS. 11-ketotestosterone and 11-ketodihydrotestosterone bind the androgen receptor as well as testosterone and DHT.[1] The 11-oxo-androgens also follow the same metabolic pathways as androgens without oxygen at 11. Interestingly, the origin of 11-oxo-androgens is entirely adrenal. All of this is important because 11-oxo-androgens appear to play a significant role in some endocrine diseases.

PCOS: Polycystic ovary syndrome (PCOS) is a disease characterized by amenorrhea or oligomenorrhea and excess androgens. Although this syndrome is found in 5-10% of women, the disease is not well understood. 11-ketotestosterone has been shown to be in excess in PCOS patients, and levels in those patients are in fact higher than levels of testosterone.[3] This finding significantly points to the adrenal as a source of the excess androgens. This idea is supported by the similarity of the levels found in adult men and women. 11-ketotestosterone may be a better biomarker than testosterone or androstenedione for androgen excess in women with PCOS.

CAH: Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency is a genetic disease affecting about 1/10,000 people.[4] The enzyme defect that causes the disease causes excess adrenal androgen production driven by ACTH, and the major androgens are 11-oxygenated.[2] Therefore, 11-oxoandrogens are important to monitor for control of CAH, especially in children and women. In men, 11-oxo-androgens are proposed to be biomarkers for disease activity because they are of adrenal origin. Although excess androgens are a lesser problem for adult men with CAH, these patients are subject to other sequelae of CAH, especially TARTS (testicular adrenal-rest tumors). Following 11-oxo-androgens is expected to be a uniquely useful biomarker since testosterone is not useful as an adrenal androgen for adult men.[6]

Puberty: Adrenarche is a stage of development that precedes puberty; clinically it is defined by axillary hair and body odor; biochemically it is defined by a rise in adrenal androgens, such as DHEA-sulfate. Testosterone levels do not rise significantly during adrenarche. Recent academic research shows that during adrenarche both DHEA-sulfate and 11-ketotestosterone rise.[7] However, DHEA-sulfate is not an active androgen, while 11-ketotestosterone is fully active. Therefore, 11-ketotestosterone may be measured along with DHEA-sulfate when investigating premature adrenarche and premature puberty.

Prostate Cancer: The goal of GnRH agonist treatment, antiandrogens and 17-hydroxylase blockade is to minimize androgens in prostate cancer. Eliminating adrenal androgens has been helpful in castration-resistant prostate cancer, and it now appears that 11-ketotestosterone is an important adrenal androgen to control in castration-resistant prostate cancer.[1,5] 11-ketotestosterone may be an effective adrenal androgen biomarker to monitor in castration-resistant prostate cancer.

Limitations This assay is limited to plasma and serum samples. Serum separator gel barrier samples also cannot be analyzed by this method.

Methodology LC-MS/MS Analysis

Specimen Serum (EDTA or heparin plasma may be used.)

Volume 2.0 mL

Minimum Volume 0.6 mL

Container Serum from red-top tube **or** EDTA plasma tube **or** heparin plasma tube

Collection Collect into vacutainer and separate within 2 hours. Send serum in a plastic transport tube. To avoid delays in turnaround time when requesting multiple tests on frozen samples, **please submit separate frozen specimens for each test requested.**

Storage Instructions **Freeze.**

Stability

Temperature	Period
Room temperature	14 days
Refrigerated	14 days
Frozen	3 months
Freeze/thaw cycles	Stable x6

Causes for Rejection Gross hemolysis, gross lipemia, incorrect specimen type, SST specimen type

Special Instructions Testing is performed at Esoterix Endocrinology Laboratory (ES No. 804677)

Footnotes

1. Storbeck KH, Bloem LM, Africander D, Schloms L, Swart P, Swart AC. 11β-Hydroxydihydrotestosterone and 11-ketodihydrotestosterone, novel C19 steroids with androgenic activity: a putative role in castration resistant prostate cancer? *Mol Cell Endocrinol.* 2013 Sep 5;377(1-2):135-146. PubMed 23856005

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3. Rodin A, Thakkar H, Taylor N, Clayton R. Hyperandrogenism in polycystic ovary syndrome. Evidence of dysregulation of 11 beta-hydroxysteroid dehydrogenase. *N Engl J Med.* 1994 Feb 17;330(7):460-465. PubMed 8289851

4. Speiser PW, White PC. Congenital adrenal hyperplasia. *N Engl J Med.* 2003 Aug 21;349(8):776-788. PubMed 12930931

5. Pretorius E, Africander DJ, Vlok M, Perkins MS, Quanson J, Storbeck KH. 11-Ketotestosterone and 11-Ketodihydrotestosterone in Castration Resistant Prostate Cancer: Potent Androgens Which Can No Longer Be Ignored. *PLoS One.* 2016 Jul 21;11(7):e0159867. PubMed 27442248

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- **The Endocrine Experience Newsletter: Endocrine Sciences Announces the Availability of 11-Oxoandrogen Testing (L20348)**
- **Thyroid Testing Services Brochure (L11755)**
- **Esoterix Endocrinology Syllabus (L5169)**

Please ask your LabCorp service representative for these titles.

7. Rege J, Turcu AF, Kasa-Vubu JZ, et al. 11-Ketotestosterone Is the Dominant Circulating Bioactive Androgen During Normal and Premature Adrenarche. J Clin Endocrinol Metab. 2018 Dec 1;103(12):4589-4598. PubMed 30137510 Eckfeldt JH, Levitt MD. Diagnostic enzymes for pancreatic disease. *Clin Lab Med.* 1989 Dec; 9(4):731-743. PubMed 2480201

11-Hydroxytestosterone 504680

CPT 82542

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Stability

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Freeze/thaw cycles	Stable x6

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7. Rege J, Turcu AF, Kasa-Vubu JZ, et al. 11-Ketotestosterone Is the Dominant Circulating Bioactive Androgen During Normal and Premature Adrenarche. J Clin Endocrinol Metab. 2018 Dec 1;103(12):4589-4598. PubMed 30137510 Eckfeldt JH, Levitt MD. Diagnostic enzymes for pancreatic disease. *Clin Lab Med.* 1989 Dec; 9(4):731-743. PubMed 2480201

11-Ketotestosterone 504674

CPT 82542

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11-oxo-Androgens Panel 504683

CPT 82542

Synonyms 11-Hydroxyandrostenedione; 11-Hydroxytestosterone; 11-Ketotestosterone

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Limitations This assay is limited to plasma and serum samples. Serum separator gel barrier samples also cannot be analyzed by this method.

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Volume 2.0 mL

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Storage Instructions Freeze.

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Causes for Rejection Gross hemolysis, gross lipemia, incorrect specimen type, SST specimen type

Footnotes

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Carbamazepine Sensitivity HLA Associations 167443

CPT Call client services.

Synonyms Carbamazepine Sensitivity; HLA A*3101; HLA A*31:01; HLA A3101; HLA B*1502; HLA B*15:02; HLA B1502; Oxcarbazepine Sensitivity

Use To avoid possible adverse reactions, this test has been recommended for patients who may be prescribed Carbamazepine and oxcarbazepine. Two HLA alleles are associated with adverse reactions to these drugs. The alleles are HLA B*15:02, which is more common in individuals of Asian descent, and HLA A*31:01, which is more common in individuals of European descent. This test evaluates the presence or absence of these alleles.

Limitations A negative test result does not rule out an adverse reaction with the drug Carbamazepine and oxcarbazepine. Even with approproate precautions, an occasional specimen may not be satisfactory for testing. In such cases, an additional specimen should be collected for retesting.

This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug Administration.

Methodology Next Generation Sequencing (NGS), sequence-based typing (SBT), sequence-specific oligonucleotide probes (SSOP) and/or sequence-specific primers (SSP) as needed to obtain the required resolution

Specimen Whole blood **or** buccal swabs

Volume 7 mL whole blood **or** 4 buccal swabs

Minimum Volume 3 mL whole blood

Container Lavender-top (EDTA) tube **or** four buccal swabs in a sealed envelope (buccal swab kit). When submitting buccal swabs, please use a buccal swab kit provided by LabCorp. To obtain the buccal kit, please telephone 800-533-1037.

Collection For collection instructions or to obtain the buccal swab kit, please telephone 800-533-1037.

Storage Instructions Maintain whole blood at room temperature or refrigerate. Keep buccal swabs dry and at room temperature.

Causes for Rejection Insufficient volume of DNA

Special Instructions If you have questions, please call 800-533-1037 (HLA customer service) for assistance in selecting the proper HLA test for the patient.

References

Alfirevic A, Jorgensen AL, Williamson PR, Chadwick DW, Park BK, Pirmohamed M. HLA-B locus in Caucasian patients with carbamazepine hypersensitivity. *Pharmacogenomics.* 2006 Sep;7(6):813-818. PubMed 16981842

Chung WH, Hung SI, Hong HS, et al. Medical genetics: A marker for Stevens-Johnson syndrome. *Nature.* 2004 Apr 1;428(6982):486. PubMed 15057820

Hung SI, Chung WH, Jee SH, et al. Genetic susceptibility to carbamazepine-induced cutaneous adverse drug reactions. *Pharmacogenet Genomics.* 2006 Apr;16(4):297-306. PubMed 16538176

Lonjou C, Thomas L, Borot N, et al. Medical genetics: A marker for Stevens-Johnson syndrome ...: ethnicity matters. *Pharmacogenomics J.* 2006 Jul-Aug;6(4):265-268. PubMed 16415921

Phillips EJ, Sukasem C, Whirl-Carillo M, et al. Clinical Pharmacogenetics Implementation Consortium Guideline for HLA Genotype and Use of Carbamazepine and Oxcarbazepine: 2017 Update. *Clin Pharmacol Ther. 2018* Apr;103(4):574-581. PubMed 29392710

Gene Sequencing, ADAMTS-13 824840

CPT 81479

Use The two main forms of thrombotic microangiopathies are thrombotic thrombocytopenic purpura (TTP) and atypical hemolytic uremic syndrome, and these can have similar clinical presentations. Sequencing the ADAMTS-13 gene can aid in confirming the diagnosis.

Limitations This genetic panel is designed to detect single nucleotide variants, multiple nucleotide variants, small indels (<50bps), and large copy number variations (deletions or duplications spanning multiple exons). The panel may miss some structural variants such as inversions, translocations, medium-sized deletions/duplications, and medium/large insertions; these types of structural variants are considered to be rare and are challenging to detect for any clinical method without prior knowledge of the nature and location of the specific structural variant. Copy number assays determine the presence of zero, one, two (normal), or three plus copies (i.e., it cannot distinguish four from five copies, though either would be abnormal) of the gene or gene region. This test does not detect acquired TTP, a non-hereditary disorder.

Methodology Next Generation Sequencing (NGS)

Specimen Whole blood; acceptable alternate: cheek swab (buccal swab)

Volume 3 mL

Minimum Volume 1 mL

Container Lavender-top (EDTA) tube

Collection Invert tube 4 times to ensure adequate mixing.

Storage Instructions Room temperature

Stability

Temperature	Period
Room temperature	1 month

Causes for Rejection Sample contamination

Special Instructions Full gene sequencing of the ADAMTS13 gene uses Next Generation Sequencing (NGS). ADAMTS13 is associated with thrombotic thrombocytopenic purpura.

References

Lotta LA, Garagiola I, Palla R, Cairo A, Peyvandi F. ADAMTS13 mutations and polymorphisms in congenital thrombotic thrombocytopenic purpura. *Hum Mutat.* 2010 Jan;31(1):11-19. PubMed 19847791

Gene Sequencing, aHUS 825178

CPT Call client services.

Use The exonic regions of 12 genes are sequenced and analyzed as part of this panel, including *CFH, MCP(CD46), CFI, C3, CFB, CFHR1, CFHR3, CFHR4, CFHR5,* Thrombomodulin (*THBD*), Plasminogen (*PLG*) and *DGKE*. The sequences have been compared to the reference Human genome (Hg18) sequence.

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Bu F, Borsa N, Gianluigi A, Smith RJ. Familial atypical hemolytic uremic syndrome: a review of its genetic and clinical aspects. *Clin Dev Immunol.* 2012; 2012:370426. PubMed 23251215

Kavanagh D, Goodship TH. Atypical hemolytic syndrome, genetic basis, and clinical manifestations. Hematology *Am Soc Hematol Educ Program.* 2011;2011:15-20. PubMed 22160007

Gene Sequencing, Dysfibrinogenemia 824839

CPT Call client services.

Synonyms afibrinogenemia; FGA sequencing; FGB sequencing; FGG sequencing; hereditary fibrinogen Aa-Chain amyloidosis; hypodysfibrinogenemia

Use Inherited disorders of fibrinogen can affect either its activity (dysfibrinogenemia) or quantity (hypofibrinogenemia) or both (hypodysfibrinogenemia). Heterozygous, homozygous and compound heterozygous mutations can be found in any of the three genes (*FGA, FGB*, and *FGG*) encoding the subunits of fibrinogen.

Limitations This genetic panel is designed to detect single nucleotide variants, multiple nucleotide variants, small indels (<50bps), and large copy number variations (deletions or duplications spanning multiple exons). The panel may miss some structural variants such as inversions, translocations, medium-sized deletions/duplications, and medium/large insertions; these types of structural variants are considered to be rare and are challenging to detect for any clinical method without prior knowledge of the nature and location of the specific structural variant. Copy number assays determine the presence of zero, one, two (normal), or three plus copies (i.e., it cannot distinguish four from five copies, though either would be abnormal) of the gene or gene region. This test does not detect acquired dysfibrinogenemia, a nonhereditary disorder.

Methodology Next Generation Sequencing (NGS)

Specimen Whole blood; acceptable alternate: cheek swab (buccal swab)

Volume 3 mL

Minimum Volume 1 mL

Container Lavender-top (EDTA) tube

Collection Invert tube 4 times to ensure adequate mixing.

Storage Instructions Room temperature

Stability

Temperature	Period
Room temperature	1 month

Causes for Rejection Sample contamination

Special Instructions The following genes associated with dysfibrinogenemia are sequenced using Next Generation Sequencing (NGS): *FGA, FGB, FGG*. This panel is designed to detect single nucleotide variants, multiple nucleotide variants, small indels (<50bps), and large copy number variations (deletions or duplications spanning multiple exons).

References

Everse SJ, Spraggon G, Doolittle RF. A three-dimensional consideration of of variant human fibrinogens. *Thromb Haemost.* 1998 Jul;80(1):1-9. PubMed 9684777

Mosesson MW. Dysfibrinogenemia and thrombosis. *Semin Thromb Hemost.* 1999;25(3):311-319. PubMed 10443961

Roberts HR, Stinchcombe TE, Gabriel DA. The dysfibrinogenaemias. *Br J Haematol.* 2001 Aug;114(2):249-257. PubMed 11529842

Gene Sequencing, Hemophilia-Complete 824833

CPT Call client services.

Synonyms Factor IX Genetic Sequencing; Factor VIII Genetic Sequencing; Inversion testing; Von Willbrand Genetic Sequencing

Use Hemophilia A (HA) and hemophilia B (HB) are both X-liked recessive bleeding disorders caused by genetic defects found in the human coagulation factor VIII (F8) and IX (F9) genes, respectively. While hemophilia is the most well-known bleeding disorder, von Willebrand disease (VWD), caused by a lack or dysfunction of the von Willebrand factor (VWF), is thought to be the most common inherited bleeding disorder. VWD can exhibit either an autosomal recessive (Type 2N and 3) or an autosomal dominant (Type 1, 2A, 2B and 2M) inheritance pattern. Type 2N patients can present clinically as HA and are often misdiagnosed. Genetic analysis can help with this differential diagnosis. Our targeted next generation sequencing (NGS) plus inversion detection panel can confirm the diagnosis of HA, HB and VWD. Identification of the causative mutations can facilitate proper diagnosis and treatment of inherited bleeding disorders.

Limitations This test targets all exons and untranslated regions of the selected genes, 25-bp of intronic DNA flanking the exon-intron boundary, plus several additional known variants of interest elsewhere in the genome for sequencing. This test would not detect a causative mutation within promoter regions or elsewhere in the genome that were not specifically targeted. A rare variant that disrupts primer binding during PCR could potentially lead to a false negative. All our reports are based on the current understanding of the genes and disease. This understanding changes with time as new papers are published. We recommend annual follow-up for more current interpretations. This test will not detect inhibitors to F8, F9 or VWF proteins, which are non-hereditary. This test does not detect pseudo-VWD (also called platelet VWD), caused by mutations in GP1B.

Methodology Next Generation Sequencing (NGS)

Specimen Whole blood; acceptable alternate: cheek swab (buccal swab)

Volume 3 mL

Minimum Volume 1 mL

Container Lavender-top (EDTA) tube

Collection Invert tube 4 times to ensure adequate mixing.

Storage Instructions Room temperature

Stability

Temperature	Period
Room temperature	1 month

Causes for Rejection Sample contamination

Special Instructions The following genes associated with hemophillia are sequenced using Next Generation Sequencing (NGS) and PCR: *F8, F9,* and *VWF.*

References

Aledort LM, Coates J. Can Health Care Plans Afford Hemophilia Costs? Yes. *Blood.* 2005;106(11):5551.

Federici AB. Current and emerging approaches for assessing von Willebrand disease in 2016. *Int J Lab Hematol.* 2016 May;38 Suppl 1:41-49. PubMed 27426859

James PD, Lillicrap D. The molecular characterization of von Willebrand disease: good in parts. *Br J Haematol.* 2013 Apr;161(2):166-176. PubMed 23406206

Kirtava A, Crudder S, Dilley A, Lally C, Evatt B. Trends in clinical management of women with von Willebrand disease: a survey of 75 women enrolled in haemophilia treatment centres in the United States. *Haemophilia* 2004; 10(2): 158-161. Haemophilia. 2004 Mar;10(2):158-161. PubMed 14962204

Leebeek FW, Eikenboom JC. Von Willebrand's Disease. *N Engl J Med.* 2016 Nov 24;375(21):2067-2080. PubMed 27959741

Lillicrap D, James P. Von Willebrand Disease: An Introduction for the Primary Care Physician. Treatment of Hemophilia. World Federation of Hemophilia (WFH): 2009; No. 47.

Santagostino E, Fasulo MR. Hemophilia a and hemophilia B: different types of diseases? *Semin Thromb Hemost.* 2013 Oct;39(7):697-701. PubMed 24014073

Gene Sequencing, VWD-Complete 824841

CPT 81408; 81479

Use The exonic regions of the *VWF* and *GP1BA* genes are sequenced and analyzed as part of this panel. The sequences have been compared to the reference Human genome (Hg19) sequence. This assay may also discover novel, deleterious variants.

Limitations The clinical sensitivity of this test is approximately 85% for subtypes 2A, 2B, 2N, 2M and 3 vWD.

Methodology Next Generation Sequencing (NGS)

Specimen Whole blood; acceptable alternate: cheek swab (buccal swab), extracted DNA, dried blood

Volume 3 mL

Minimum Volume 2 mL

Container Lavender-top (EDTA) tube

Collection Invert tube 4 times to ensure adequate mixing.

Storage Instructions Room temperature

Stability

Temperature	Period
Room temperature	1 month

Causes for Rejection Sample contamination

Special Instructions The following genes associated with von Willebrand Disease are sequenced using Next Generation Sequencing (NGS): *VWF* and *GP1BA*.

References

Lillicrap D. von Willebrand disease: advances in pathogenetic understanding, diagnosis, and therapy. *Blood.* 2013 Nov 28;122(23):3735-3740. PubMed 24065240

Nichols WL, Hultin MB, James AH, et al. von Willebrand disease (VWD): evidence-based diagnosis and management guidelines, the National Heart, Lung, and Blood Institute (NHLBI) Expert Panel report (USA). *Haemophilia.* 2008 Mar;14(2):171-232. PubMed 18315614

Von Willebrand disease (VWD). Von Willebrand Factor Variant Database (VWFdb). University of Sheffield. http://www.vwf.group.shef.ac.uk/vwd.html. Accessed March 31, 2015.

Naloxone, UR, MS Confirm 701195

CPT Call client services.
Use Detect the presence of Naloxone
Limitations This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug Administration.
Methodology Liquid chromatography tandem mass spectrometry (LC/MS-MS)
Specimen Urine (random)
Volume 10 mL
Minimum Volume 3 mL
Container Plastic urine container without preservative
Storage Instructions Room temperature
Causes for Rejection Urine in preservative tube

Plasminogen Gene Sequencing 824837

CPT 81479
Use PLG is the human gene coding for the plasminogen protein. Plasminogen deficiency is the major disorder associated with mutations in PLG.
Limitations This genetic panel is designed to detect single nucleotide variants, multiple nucleotide variants, smallindels (<50bps), and large copy number variations (deletions or duplications spanning multiple exons). The panel may miss some structural variants such as inversions, translocations, medium sized deletions/duplications, and medium/large insertions; these types of structural variants are considered to be rare, and are challenging to detect for any clinical method without prior knowledge of the nature and location of the specific structural variant. Copy number assays determine the presence of zero, one, two (normal), or three plus copies (i.e., it cannot distinguish four from five copies, though either would be abnormal) of the gene or gene region. This test does not detect acquired or non-hereditary disorder.
Methodology Next Generation Sequencing (NGS)
Specimen Whole blood; acceptable alternate: cheek swab (buccal swab)
Volume 3 mL
Minimum Volume 1 mL
Container Lavender-top (EDTA) tube
Collection Invert tube 4 times to ensure adequate mixing.
Storage Instructions Room temperature
Stability

Temperature	Period
Room temperature	1 month

Causes for Rejection Sample contamination
Special Instructions The PLG gene is sequenced using Next Generation Sequencing (NGS).
References

Mehta R, Shapiro AD. Plasminogen deficiency. *Haemophilia.* 2008 Nov;14(6):1261-1268. PubMed 19141167

Okamoto A, Sakata T, Mannami T, et al. Population-based distribution of plasminogen activity and estimated prevalence and relevance to thrombotic diseases of plasminogen deficiency in the Japanese: the Suita Study. *J Thromb Haemost.* 2003 Nov;1(11):2397-2403. PubMed 14629475

Tefs K, Gueorguieva M, Klammt J, et al. Molecular and clinical spectrum of type I plasminogen deficiency: A series of 50 patients. *Blood.* 2006 Nov 1;108(9):3021-3026. PubMed 16849641

PlateletGenex Functional Defect Panel 824842

CPT 81404; 81406; 81408; 81479
Use This panel sequences 31 genes associated with clinically relevant defects in platelet function, activation, secretio and binding. The clinical importance of bleeding symptoms is difficult to assess and concurrent bleeding disorders are commonly seen in the same patient. Therefore, gene sequencing of the platelet, von Willebrand and fibrinogen genes, all of which are contained in this panel. can provide valuable understanding of the physiology of bleeding diathesis.
Methodology Next Generation Sequencing (NGS)
Specimen Whole blood; acceptable alternate: cheek swab (buccal swab)
Volume 3 mL
Minimum Volume 1 mL
Container Lavender-top (EDTA) tube
Collection Invert tube 4 times to ensure adequate mixing.
Storage Instructions Room temperature
Stability

Temperature	Period
Room temperature	1 month

Causes for Rejection Sample contamination
Special Instructions The following genes associated with the clinically significant defects in platelet function, activation, secretion and binding are sequenced using Next Generation Sequencing (NGS): *ANO6, AP3B1, BLOC1S3, BLOC1S6, DTNBP1, FGA, FGB, FGG, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGAB3, LYST, MYH9, P2RY12, PLA2GA, PLAU, RASGRP2, TBXA2R, TBXAS1, VIPAS39, VPS33B, VWF* and *WAS*. This panel can confirm the diagnosis of more common platelet aggregation, platelet secretion (ATP release) and platelet signaling defects; it can also diagnose (and subtype) von Willebrand disease, dysfibrinogenemias and rare platelet defects (Glanzmann thrombasthenia, Bernard-Soulier, WiskottAldrich, Hermansky-Pudlak, Scott, MayHegglin syndromes, others).
This is an NGS panel and is not a platelet count assay.
References

Buinmov N, Fuller N, Hayward CP. Genetic loci associated with platelet traits and platelet disorders. *Semin Thromb Hemost.* 2013 Apr;39(3):291-305. PubMed 23468379

Kunishima S, Saito H. Advances in the understanding of MYH9 disorders. *Curr Opin Hematol.* 2010 Sep;17(5):405-410. PubMed 20601875

Mikkelsson J et al. Genetics of the platelet glycoprotein receptors: risk of thrombotic events and pharmacogenetic implications. *Clin Appl Thromb Hemost.* 2005 Apr;11(2):113-125. PubMed 15821818

PlateletGenex Thrombocytopenia Panel 824834

CPT 81406(x2); 81408; 81479
Synonyms Platelet Quantitative Defect Genetic Panel
Use The PlateletGenex Thrombocytopenia Panel targets the exons exons and splice sites of 26 genes that have been associated with low platelet counts. Thrombocytopenia can be an acquired or inherited condition and genetic results can help differentiate between the two.
Limitations This test targets all exons of the selected 26 genes, plus 5bp of intronic DNA flanking the exon-intron boundary. This test would not detect a causative mutation within the promoter or deep intronic regions or elsewhere in the genome unless it was part of the subset we specifically targeted based on the published literature. A rare mutation that disrupts primer binding during PCR could potentially lead to a false negative.
Methodology Next Generation Sequencing (NGS)
Specimen Whole blood; acceptable alternate: cheek swab (buccal swab)
Volume 3 mL
Minimum Volume 1 mL
Container Lavender-top (EDTA) tube
Collection Invert tube 4 times to ensure adequate mixing.
Storage Instructions Room temperature
Stability

Temperature	Period
Room temperature	1 month

Causes for Rejection Sample contamination
Special Instructions The following genes associated with thrombocytopenia are sequenced using Next Generation Sequencing (NGS): *ACTN1, ADAMTS13, ANKRD26, CD36, CYCS, ETV6, FERMT3, FLI1, FLNA, GATA1, GFI1B, GNE, HOXA11, HRG, MPL, NBEA, NBEAL2, ORAI1, RBM8A, RUNX1, STIM1,*

STXBP2, THPO, TUBB1, VWF and *WAS*. This is an NGS panel and is not a platelet count assay.

References

Balduini CL, Savoia A. Inherited thrombocytopenias: molecular mechanisms. *Semin Thromb Hemost.* 2004 Oct;30(5):513-523. PubMed 15497094

Balduini CL, Pecci A, Noris P. Diagnosis and management of inherited thrombocytopenias. *Semin Thromb Hemost.* 2013 Mar;39(2):161-171. PubMed 23397552

Cines DB, Bussel JB, McMillan RB, Zehnder JL. Congenital and acquired thrombocytopenia. *Hematology Am Soc Hematol Educ Program.* 2004:390-406. PubMed 15561694

Toluene, as o-Cresol, Occupational Exposure, Urine 702378

CPT 82570; 84600
Synonyms 2-Methylphenol; Toluene Metabolite
Use Evaluate exposure to o-cresol or toluene
Methodology Gas chromatography/flame ionization detection (GC/FID)
Specimen Urine (random)
Volume 10 mL
Minimum Volume 0.5 mL
Container Plastic urine container without preservative
Collection Tubes should be filled to prevent loss of volatile compound to headspace.
Stability

Temperature	Period
Refrigerated	14 days
Frozen	14 days

Causes for Rejection Urine from preservative tube

Updates to the *Directory of Services and Interpretive Guide (DoS)*

Test Name	Test No.	**Field**/Change (Only fields that change are included here.)
Abnormal Bleeding Profile	**116004**	**Volume** 5 mL EDTA whole blood, one tube citrated whole blood (unopened), **and** 2 mL citrated plasma, frozen **Minimum Volume** 5 mL EDTA whole blood, one tube citrated whole blood (unopened), **and** 1 mL citrated plasma, frozen
Activated Protein C Resistance (APCR)	**117762**	**Volume** 1 mL **Minimum Volume** (Removed field)
Adalimumab and Anti-Adalimumab Antibody, Dose*ASSURE*™ ADL	**504575**	**Stability** Temperature / Period: Room temperature 7 days; Refrigerated 7 days; Frozen 308 days; Freeze/thaw cycles Stable x6
Adalimumab and Anti-Adalimumab Antibody (Serial Monitor), *Dose*ASSURE™ ADL	**503890**	
ADAMTS13 Antibody	**117915**	**Minimum Volume** (Removed field)
α-Thalassemia, DNA Analysis	**511972**	**Special Instructions** If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.
α_2-Antiplasmin	**117739**	**Minimum Volume** (Removed field)
Antithrombin (AT) Activity	**015040**	**Volume** 1 mL **Minimum Volume** (Removed field)
Antithrombin (AT) Antigen (Immunologic)	**015057**	
Antithrombin (AT) Deficiency Profile	**015594**	
aPTT Mixing Studies	**117199**	**Volume** 2 mL **Minimum Volume** (Removed field)
β_2-Glycoprotein 1 Antibodies, IgA	**163900**	**Volume** 1 mL **Minimum Volume** 0.5 mL
β_2-Glycoprotein 1 Antibodies, IgA, IgG, IgM	**163915**	
β_2-Glycoprotein 1 Antibodies, IgG	**163882**	
β_2-Glycoprotein 1 Antibodies, IgG, IgM	**163002**	
β_2-Glycoprotein 1 Antibodies, IgM	**163908**	
Bloom Syndrome, DNA Analysis	**512145**	**Special Instructions** If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.
Calcium-Sensing Receptor (*CASR*) Gene Sequencing Analysis	**504513**	**Stability** Temperature / Period: Room temperature 28 days; Refrigerated 28 days
Canavan Disease, DNA Analysis	**511147**	**Special Instructions** If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.
Certolizumab and Anti-Certolizumab Antibody, *Dose*ASSURE™ CTZ	**504627**	**Storage Instructions** Refrigerate or **freeze**.

Note: Please consult the online Directory of Services and Interpretive Guide at https://www.labcorp.com/tests for the most current test information.

<table>
<tr><th>Test Name</th><th>Test No.</th><th>Field/Change (Only fields that change are included here.)</th></tr>
<tr><td>Congenital Adrenal Hyperplasia (CAH) 21-Hydroxylase (CYP21) Mutation (Endocrine Sciences)</td><td>500768</td><td>Container Lavender-top (EDTA) tube, yellow top (ACD) tube
Stability<table><tr><th>Temperature</th><th>Period</th></tr><tr><td>Room temperature</td><td>28 days</td></tr></table></td></tr>
<tr><td>Cortisol, Urinary Free</td><td>004432</td><td>Reference Interval<table><tr><th>Age</th><th>Male (µg/24h)</th><th>Female (µg/24h)</th></tr><tr><td>0 days to 1 year</td><td>Not established</td><td>Not established</td></tr><tr><td>2 to 5 years</td><td>2 – 16</td><td>2 – 16</td></tr><tr><td>6 to 11 years</td><td>4 – 28</td><td>4 – 24</td></tr><tr><td>12 years</td><td>4 – 36</td><td>4 – 36</td></tr><tr><td>13 to 17 years</td><td>6 – 45</td><td>6 – 42</td></tr><tr><td>18 to 80 years</td><td>5 – 64</td><td>6 – 42</td></tr><tr><td>>80 years</td><td>3 – 49</td><td>3 – 49</td></tr></table></td></tr>
<tr><td>Cryptococcus Antigen</td><td>183025</td><td>Storage Instructions Refrigerated (2°-8°C) or frozen (<–20°C)
Stability<table><tr><th>Temperature</th><th>Period</th></tr><tr><td>Refrigerated</td><td>72 hours (stability determined by manufacturer or literature reference)</td></tr><tr><td>Frozen</td><td>>72 hours (stability determined by manufacturer or literature reference)</td></tr></table></td></tr>
<tr><td>Cryptococcus Antigen, Cerebrospinal Fluid</td><td>183016</td><td>Storage Instructions Refrigerated (2°-8°C) or frozen (<–20°C)
Stability<table><tr><th>Temperature</th><th>Period</th></tr><tr><td>Refrigerated</td><td>5 days</td></tr></table></td></tr>
<tr><td>Cystic Fibrosis (CF) Profile, 32 Mutations, Fetal Analysis</td><td>480581</td><td rowspan="2">Special Instructions (added second paragraph) If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>Cystic Fibrosis (CF) Profile, 97 Mutations, CFplus®, Fetal Analysis</td><td>480819</td></tr>
<tr><td>Dihydrolipoamide Dehydrogenase (DLD)</td><td>450080</td><td>Special Instructions If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>Dilute Prothrombin Time</td><td>005200</td><td>Volume 1 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>Disseminated Intravascular Coagulation (DIC) Profile</td><td>116012</td><td>Volume 5 mL EDTA whole blood, one citrated whole blood tube, and 1 mL frozen sodium citrate plasma</td></tr>
<tr><td colspan="3">Note: Please consult the online Directory of Services and Interpretive Guide at https://www.labcorp.com/tests for the most current test information.</td></tr>
</table>

<table>
<tr><th>Test Name</th><th>Test No.</th><th>Field/Change (Only fields that change are included here.)</th></tr>
<tr><td>Epstein-Barr Virus (EBV) Acute Infection Antibodies Profile</td><td>216655</td><td>Additional Information Epstein-Barr (EB) virus is a herpes group virus that is ubiquitous. It is the cause of classic infectious mononucleosis and is causally implicated in the pathogenesis of Burkitt lymphoma, some nasopharyngeal carcinomas, and rare hereditary lymphoproliferative disorders. The serologic response to EB virus includes antibody to early antigen, IgM and IgG antibodies to viral capsid antigen (VCA), and antibodies to Epstein-Barr nuclear antigen (EBNA).
Although most cases of infectious mononucleosis can be diagnosed on the basis of clinical findings, blood count and morphology, and a positive test for heterophile antibody, as many as 20% may be heterophile-negative, at least at presentation (Heterophile may become positive when repeated in a few days). In some of these cases, a test for Epstein-Barr virus antibodies may be useful.
The most controversial use of EBV serology is in chronic fatigue syndrome, a complaint predominantly (but not exclusively) of young to middle-aged women, characterized by long persistent debilitating fatigue and a panoply of usually mild somatic complaints. The high levels of EBV antibodies in the general population, their long persistence, and the poor correlation of antibody titers with symptoms combine to make EBV serology useless in diagnosing, following, or ruling out chronic fatigue syndrome. See table.
<table>
<tr><th>Interpretation</th><th>EBV-IgM</th><th>EA(D)-IgG</th><th>VCA-IgG</th><th>EBNA-IgG</th></tr>
<tr><td>EBV seronegative</td><td>-</td><td>-</td><td>-</td><td>-</td></tr>
<tr><td>Early phase</td><td>+</td><td>-</td><td>-</td><td>-</td></tr>
<tr><td>Acute primary infection</td><td>+</td><td>±</td><td>+</td><td>-</td></tr>
<tr><td>Convalescence/past infection</td><td>-</td><td>±</td><td>+</td><td>+</td></tr>
<tr><td>Reactivated infection</td><td>±</td><td>±</td><td>+</td><td>+</td></tr>
<tr><td colspan="5">Key — Antibody present: + Antibody absent: –</td></tr>
</table></td></tr>
<tr><td>Etanercept and Anti-Etanercept Antibody (Serial Monitor), DoseASSURE™ ETN</td><td>504245</td><td>Storage Instructions Refrigerate or freeze. Stable at room temperature or frozen for 14 days. Freeze/thaw cycles: stable x6.</td></tr>
<tr><td>Extrinsic Pathway Coagulation Factor Profile</td><td>500041</td><td>Minimum Volume 1 mL</td></tr>
<tr><td>Factor II Activity</td><td>086231</td><td rowspan="4">Volume 1 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>Factor V Activity</td><td>086249</td></tr>
<tr><td>Factor VII Activity</td><td>800599</td></tr>
<tr><td>Factor VIII Activity</td><td>086264</td></tr>
<tr><td>Factor VIII Inhibitor Profile, Comprehensive</td><td>117157</td><td>Volume 3 mL</td></tr>
<tr><td>Factor IX Activity</td><td>086298</td><td rowspan="6">Volume 1 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>Factor X Activity</td><td>086306</td></tr>
<tr><td>Factor X, Chromogenic</td><td>117904</td></tr>
<tr><td>Factor XI Activity</td><td>086314</td></tr>
<tr><td>Factor XII Activity</td><td>086322</td></tr>
<tr><td>Factor XIII</td><td>086330</td></tr>
<tr><td>Familial Dysautonomia, DNA Analysis</td><td>511352</td><td rowspan="3">Special Instructions If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>Familial Hyperinsulinism (FHI)</td><td>450070</td></tr>
<tr><td>Fanconi Anemia (Type C), DNA Analysis</td><td>511212</td></tr>
<tr><td>Fibrinogen Degradation Products (FDP), Plasma</td><td>115402</td><td>Volume 1 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>Fibrinogen Evaluation Profile</td><td>336624</td><td>Volume One 2 mL aliquot frozen citrate plasma for Fibrinogen Activity and Thrombin Mixing, and one 1 mL frozen citrate plasma aliquot for Fibrinogen Antigen.</td></tr>
<tr><td>Fragile X Syndrome, DNA Analysis, Prenatal With Southern Blot Analysis</td><td>510300</td><td>Special Instructions (added) If culture is needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>Gaucher Disease, DNA Analysis</td><td>511048</td><td rowspan="2">Special Instructions If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>Glycogen Storage Disease 1a</td><td>511290</td></tr>
<tr><td>Heart Disease and Stroke Risk Profile</td><td>500140</td><td>Volume 1 mL serum and 1 mL frozen plasma</td></tr>
<tr><td colspan="3">Note: Please consult the online Directory of Services and Interpretive Guide at https://www.labcorp.com/tests for the most current test information.</td></tr>
</table>

<table>
<tr><th>Test Name</th><th>Test No.</th><th>Field/Change (Only fields that change are included here.)</th></tr>
<tr><td>Heparin Anti-Xa</td><td>117101</td><td>Reference Interval Reference intervals indicate therapeutic levels.[11]
Target Ranges for Treatment of Venous Thromboembolism (VTE)
Unfractionated Heparin (UFH): 0.3-0.7 IU/mL (UFH sample obtained 6 hours after initiation or dose adjustment)
Low Molecular Weight Heparins (LMWH) (LMWH sample obtained 4 hours following subcutaneous injection)
Enoxaparin (Lovenox): 0.6-1.0 IU/mL for twice daily dosing; >1.0 IU/mL for once daily dosing
Nadroparin (Fraxiparine): 0.6-1.0 IU/mL for twice daily dosing; 1.3 IU/mL for once daily dosing
Tinzaparin (Innohep): 0.85 IU/mL for once daily dosing
Dalteparin (Fragmin): 1.05 IU/mL for once daily dosing
Volume 1 mL
Minimum Volume (Removed field)
Footnotes (added)
11. Garcia DA, Baglin TP, Weitz JI, Samama MM. Parenteral anticoagulants: Antithrombotic Therapy and Prevention of Thrombosis, 9th ed: American College of Chest Physicians Evidence-Based Clinical Practice Guidelines. Chest. 2012 Feb;141(2 Suppl):e24S-e43S.</td></tr>
<tr><td>Heparin-induced Platelet Antibody (HIPA)</td><td>150075</td><td>Volume 1 mL
Minimum Volume 0.5 mL</td></tr>
<tr><td>Hepatitis Be Antigen</td><td>006619</td><td>Use The HBe antigen assay, in conjunction with other serological and clinical information, is intended only for the determination of chronic infection with hepatitis B virus.
Limitations (added) This assay has not been FDA-approved for the diagnosis of individuals with acute hepatitis B infection.</td></tr>
<tr><td>Hexagonal Phase Phospholipid (HPP)</td><td>117838</td><td>Volume 1 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>IA_2 Autoantibodies (Endocrine Sciences)</td><td>141531</td><td>Volume 1 mL
Minimum Volume 0.5 mL (Note: This volume does not allow for repeat testing.)</td></tr>
<tr><td>Infliximab and Anti-Infliximab Antibody, DoseASSURE™ IFX</td><td>503770</td><td>Storage Instructions Frozen (preferred) or refrigerated
Stability
<table>
<tr><th>Temperature</th><th>Period</th></tr>
<tr><td>Room temperature</td><td>7 days</td></tr>
<tr><td>Refrigerated</td><td>7 days</td></tr>
<tr><td>Frozen</td><td>359 days</td></tr>
<tr><td>Freeze/thaw cycles</td><td>Stable x6</td></tr>
</table></td></tr>
<tr><td>Infliximab and Anti-Infliximab Antibody (Serial Monitor), DoseASSURE™ IFX</td><td>503870</td><td>Use Provides infliximab drug concentration levels as well as levels of anti-infliximab antibodies.
In the absence of anti-infliximab antibodies, the infliximab drug level typically reflects the total infliximab concentration in serum. In the presence of anti-infliximab antibodies, the infliximab drug level typically reflects the antibody-unbound fraction of infliximab concentration in serum. The presence of infliximab drug, even at concentrations well above target treatment levels (>50 ug/mL), does not interfere with the anti-infliximab antibody detection. This assay may be helpful for any patients on infliximab therapy for diseases such as Crohn's disease, inflammatory bowel disease, ulcerative colitis, rhematoid arthritis, or other autoimmune conditions. This test includes long-term serial monitoring of results.
Limitations Failure of infliximab therapy may not always be due to the presence of anti-infliximab antibodies. In addition, the absence of anti-infliximab antibodies does not guarantee positive response to treatment.
Methodology Electrochemiluminescence Immunoassay (ECLIA)
Specimen Serum
Volume 2 mL
Minimum Volume 1 mL (Note: This volume does not allow for repeat testing).
Container Red-top tube or gel-barrier tube
Collection Serum must be separated from cells within 45 minutes of venipuncture. Send serum in a plastic transport tube. To avoid delays in turnaround time when requesting multiple test on frozen samples, please submit separate frozen specimens for each test requested.
Storage Instructions Frozen (preferred) or refrigerated
Stability
<table>
<tr><th>Temperature</th><th>Period</th></tr>
<tr><td>Room temperature</td><td>7 days</td></tr>
<tr><td>Refrigerated</td><td>7 days</td></tr>
<tr><td>Frozen</td><td>359 days</td></tr>
<tr><td>Freeze/thaw cycles</td><td>Stable x6</td></tr>
</table></td></tr>
<tr><td>Inherited Thrombophilias of Pregnancy Profile</td><td>365500</td><td>Volume 1 mL frozen plasma and 7 mL whole blood or LabCorp buccal swab kit
Minimum Volume (Removed field)</td></tr>
<tr><td>Intrauterine Fetal Demise/Stillborn Follow-up Profile</td><td>365200</td><td>Volume 4 mL serum; 1 mL frozen citrate plasma; whole blood and whole blood or LabCorp buccal swab kit (buccal swab collection kit contains instructions for use of a buccal swab)</td></tr>
<tr><td>Intrauterine Fetal Demise/Stillborn Profile (Extended)</td><td>365300</td><td>Volume 1 mL serum, 2 mL frozen sodium citrate plasma, full whole blood tube, and 7 mL whole blood or LabCorp buccal swab kit</td></tr>
<tr><td colspan="3">Note: Please consult the online Directory of Services and Interpretive Guide at https://www.labcorp.com/tests for the most current test information.</td></tr>
</table>

<table>
<tr><th>Test Name</th><th>Test No.</th><th>Field/Change (Only fields that change are included here.)</th></tr>
<tr><td>Intrinsic Pathway Coagulation Factor Profile</td><td>500033</td><td>Minimum Volume 1 mL</td></tr>
<tr><td>Joubert Syndrome Type II, DNA Analysis</td><td>511490</td><td>Special Instructions If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>KRAS Gene Mutation Analysis, Extended</td><td>481075</td><td>Special Instructions Provide a copy of the pathology report; the KRAS test will be delayed if the pathology report is not received. Direct any questions regarding this test to customer service at 800-345-4363.</td></tr>
<tr><td>Lupus Anticoagulant Comprehensive</td><td>117054</td><td>Volume 3 mL
Minimum Volume 2 mL</td></tr>
<tr><td>Lupus Anticoagulant With Reflex</td><td>117892</td><td>Volume 2 mL
Minimum Volume 1 mL (Note: This volume does not allow for repeat testing.)</td></tr>
<tr><td>Maple Syrup Urine Disease Carrier Test, DNA</td><td>511310</td><td rowspan="2">Special Instructions (added second paragraaph) If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>Maternal Cell Contamination</td><td>511402</td></tr>
<tr><td>MEN2: RET Gene, Sequencing Analysis (Endocrine Sciences)</td><td>504008</td><td>Container Lavender-top (EDTA) tube or yellow-top (ACD) tube
Storage Intructions Maintain specimen at room temperature.
Stability<table><tr><th>Temperature</th><th>Period</th></tr><tr><td>Room temperature</td><td>28 days</td></tr><tr><td>Refrigerated</td><td>28 days</td></tr></table></td></tr>
<tr><td>Menorrhagia Profile</td><td>336572</td><td>Volume 3 mL
Minimum Volume 2 mL</td></tr>
<tr><td>Methamphetamines D and L, Urine (LabCorp MedWatch®)</td><td>721600</td><td>Minimum Volume (Removed field)</td></tr>
<tr><td>Mucolipidosis Type IV Mutation Detection</td><td>511386</td><td rowspan="3">Special Instructions If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.</td></tr>
<tr><td>Nemaline Myopathy</td><td>450040</td></tr>
<tr><td>Niemann-Pick Disease, DNA Analysis</td><td>511329</td></tr>
<tr><td>Plasminogen Activator Inhibitor 1 (PAI-1) Activity</td><td>146787</td><td>Volume 0.5 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>Plasminogen Activator Inhibitor 1 (PAI-1) Antigen</td><td>500057</td><td>Volume 0.5 mL</td></tr>
<tr><td>Plasminogen Activity</td><td>117713</td><td>Volume 1 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>Porphyrins, Quantitative, 24-Hour Urine</td><td>003194</td><td>Additional Information (updated last paragraph) Hepatic complications are found with porphyria cutanea tarda and protoporphyria. Fluorescence is demonstrable in liver biopsies from patients with the former, as well as siderosis. Crystalline deposits may be found in protoporphyria.[2] The amount of porphobilinogen excreted in acute intermittent porphyria is usually greater than the excretion of δ-aminolevulinic acid (Δ-ALA). When there is more Δ-ALA, another diagnosis should be considered, including lead poisoning, another type of porphyria, or hereditary tyrosinemia.[2] See also Zinc Protoporphyrin (ZPP) [010170], which pertains to lead poisoning, and erythropoietic protoporphyria. The differential diagnosis of lead poisoning is relevant.[4]</td></tr>
<tr><td>Prolonged Activated Partial Thromboplastin Time (aPTT)</td><td>117796</td><td>Volume 3 mL
Minimum Volume 2 mL</td></tr>
<tr><td>Prolonged Protime Profile</td><td>117866</td><td>Volume 3 mL</td></tr>
<tr><td>Protein C Antigen</td><td>080465</td><td rowspan="6">Volume 1 mL
Minimum Volume (Removed field)</td></tr>
<tr><td>Protein C Deficiency Profile</td><td>283655</td></tr>
<tr><td>Protein C, Functional</td><td>117705</td></tr>
<tr><td>Protein S Deficiency Profile</td><td>117754</td></tr>
<tr><td>Protein S, Free</td><td>164519</td></tr>
<tr><td>Protein S, Functional</td><td>164525</td></tr>
<tr><td>Prothrombin Antibodies, IgG</td><td>117065</td><td>Volume 1 mL
Minimum Volume 0.5 mL</td></tr>
<tr><td>Prothrombin Time (PT) Mixing Study</td><td>117028</td><td>Volume 2 mL
Minimum Volume 1 mL</td></tr>
<tr><td>Reptilase Time</td><td>117180</td><td>Volume 1 mL</td></tr>
<tr><td>Rituximab and Anti-Rituximab Antibody, DoseASSURE™ RTX</td><td>504355</td><td>Specimen Serum (preferred) or plasma (EDTA or heparin)
Container Gel-barrier tube, red-top tube, lavender-top (EDTA) tube, or green-top (heparin) tube
Storage Instructions Refrigerate or freeze</td></tr>
<tr><td colspan="3">Note: Please consult the online Directory of Services and Interpretive Guide at https://www.labcorp.com/tests for the most current test information.</td></tr>
</table>

Test Name	Test No.	Field/Change (Only fields that change are included here.)
***SHOX,* DHPLC (Endocrine Sciences)**	**500110**	**Storage Instructions** Maintain specimen at room temperature. **Stability** Temperature: Room temperature — Period: 28 days Temperature: Refrigerated — Period: 28 days
Tay-Sachs Disease, DNA Analysis	**510404**	**Special Instructions** (added second paragraph) If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.
Thiopurine Methyltransferase (TPMT), Enzyme Activity, Erythrocytes	**510750**	**Stability** Temperature: Refrigerated — Period: 14 days Temperature: Frozen — Period: Unstable Temperature: Freeze/thaw cycles — Period: Unstable
Thiopurine Methyltransferase (TPMT) Genotyping	**504142**	**Stability** Temperature: Room temperature — Period: 28 days Temperature: Refrigerated — Period: 28 days
Thrombin Mixing Study	**117170**	**Minimum Volume** (Removed field)
Thrombin Time	**015230**	**Volume** 1 mL **Minimum Volume** (Removed field)
Thrombotic Risk Assessment	**117720**	**Volume** 2 mL frozen sodium citrate plasma **and** 1 mL serum **and** 7 mL whole blood **or** buccal swab **Minimum Volume** (Removed field)
Thrombotic Risk, Congenital	**117706**	**Volume** 2 mL frozen sodium citrate plasma and 2 mL serum and 7 mL whole blood or buccal swab **Minimum Volume** (Removed field)
Thrombotic Risk Profile I	**117702**	**Volume** 2 mL frozen sodium citrate plasma **and** 1 mL serum **and** 7 mL whole blood **or** buccal swab **Minimum Volume** (Removed field)
Thrombotic Risk Profile II	**117090**	
Thrombotic Risk Profile, Acquired	**117024**	**Volume** 1 mL serum **and** 1 mL frozen sodium citrate plasma **Minimum Volume** (Removed field)
Thrombotic Risk Profile, Acquired, Comprehensive	**117074**	**Volume** 4 mL serum and 4 mL frozen sodium citrate plasma (2 mL of plasma into each of two tubes) **Minimum Volume** (Removed field)
Uniparental Disomy (UPD) Proband, DNA Analysis	**470074**	**Special Instructions** (added second paragraph) If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.
Usher Syndrome Type IF	**450060**	**Special Instructions** If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.
Usher Syndrome Type III	**450050**	
Ustekinumab and Anti-Ustekinumab Antibody, *Dose*ASSURE™ UST	**504594**	**Specimen** Serum (preferred) **or** plasma **Container** Red-top tube, gel-barrier tube, plasma EDTA tube, **or** plasma heparin tube
Vedolizumab and Anti-Vedolizumab Antibody, *Dose*ASSURE™ VDZ	**504567**	**Specimen** Serum (preferred) **or** plasma **Container** Serum-gel tube, red-top tube, green top (heparin) tube, **or** lavender top (EDTA) tube **Storage Instructions** Refrigerate or **freeze**
von Willebrand Factor (vWF) Activity	**164509**	**Volume** 1 mL **Minimum Volume** (Removed field)
von Willebrand Factor (vWF) Antigen	**086280**	
von Willebrand Factor (vWF) Profile	**084715**	
Walker-Warburg Syndrome	**511480**	**Special Instructions** If cultured cells are needed, an additional 7-12 days may be required. Additional culture fee may be included.

Note: Please consult the online Directory of Services and Interpretive Guide at https://www.labcorp.com/tests for the most current test information.

CPT Code Updates

Test Name	Test No.	CPT(s)
Allergen Profile With Total IgE, Respiratory–Area 12	**603719**	82785, 86003(x23)
Allergen Profile With Total IgE, Respiratory–Area 17	**602985**	82785, 86003(x24)
Allergen Profile With Total IgE, Respiratory–Area 18	**602986**	82785, 86003(x16)

Deleted Procedures

Deleted Tests	Test No.	LabCorp Offers	Test No.
Acute Myelocytic Leukemia (AML) Profile, Chromosome Analysis With Reflex to FLT3, CEBPA, and NPM1	**511972**	Please contact your LabCorp representative for testing options.	
Toluene Metabolite Profile, Urine	**723221**	Toluene, as o-Cresol, Occupational Exposure, Urine	702378

The CPT codes listed are in accordance with the current edition of Current Procedural Terminology, a publication of the American Medical Association. CPT codes are provided for the convenience of our clients; however, correct coding often varies from one carrier to another. Consequently, the codes presented here are intended as general guidelines and should not be used without confirming with the applicable payer that their use is appropriate in each case.

LOINC® Map. The Logical Observation Identifiers Names and Codes (LOINC®) corresponding to the individual LabCorp published assays is updated on a regular basis at www.labcorp.com.

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