BlueCross BlueShield of New Mexico

Genetic Testing
Medical Policy – Genetics

Please complete all appropriate questions fully.

Suggested medical record documentation:

- Current History & Physical
- Progress Notes
- Family Genetic History
- Genetic Counseling Evaluation

*Failure to include suggested medical record documentation may result in delay or possible denial of request.

PATIENT INFORMATION

Name:	
Member ID:	
Group ID:	

PROCEDURE INFORMATION

Genetic Counseling performed:	☐ Yes ☐ No

**Please check the requested analyte(s), identify number of units requested, and provide indication/rationale for testing.

81400 Molecular Pathology Level 1

Units

_____ ☐ ACADM (acyl-CoA dehydrogenase, C-4 to C-12 straight chain, MCAD) (e.g., medium chain acyl dehydrogenase deficiency), K304E variant

_____ ☐ ACE (angiotensin converting enzyme) (e.g., hereditary blood pressure regulation), insertion/deletion variant

_____ ☐ AGTR1 (angiotensin II receptor, type 1) (e.g., essential hypertension), 1166A>C variant

_____ ☐ BCKDHA (branched chain keto acid dehydrogenase E1, alpha polypeptide) (e.g., maple syrup urine disease, type 1A), Y438N variant

_____ ☐ CCR5 (chemokine C-C motif receptor 5) (e.g., HIV resistance), 32-bp deletion mutation/794 825del32 deletion

_____ ☐ CLRN1 (clarin 1) (e.g., Usher syndrome, type 3), N48K variant

_____ ☐ DPYD (dihydropyrimidine dehydrogenase) (e.g., 5-fluorouracil/5-FU and capecitabine drug metabolism), IVS14+1G>A variant

_____ ☐ F13B (coagulation factor XIII, B polypeptide) (e.g., hereditary hypercoagulability), V34L variant

_____ ☐ F2 (coagulation factor 2) (e.g., hereditary hypercoagulability), 1199G>A variant

_____ ☐ F5 (coagulation factor V) (e.g., hereditary hypercoagulability), HR2 variant

_____ ☐ F7 (coagulation factor VII [serum prothrombin conversion accelerator]) (e.g., hereditary hypercoagulability), R353Q variant

_____ ☐ FGB (fibrinogen beta chain) (e.g., hereditary ischemic heart disease), -455G>A variant

_____ ☐ FGFR1 (fibroblast growth factor receptor 1) (e.g., Pfeiffer syndrome type 1, craniosynostosis), P252R variant

_____ ☐ FGFR3 (fibroblast growth factor receptor 3) (e.g., Muenke syndrome), P250R variant

_____ ☐ FKTN (Fukutin) (e.g., Fukuyama congenital muscular dystrophy), retrotransposon insertion variant

_____ ☐ GNE (glucosamine [UDP-N-acetyl]-2-epimerase/N-acetylmannosamine kinase) (e.g., 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]) (e.g., neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-1a/b (L33P)

_____ ☐ Human platelet antigen 15 genotyping (HPA-15), CD109 (CD109 molecule) (e.g., neonatal thrombocytopenia [NAIT], post-transfusion purpura), HPA-15a/b(S682Y)

_____ ☐ Human platelet antigen 2 genotyping (HPA-2), GP1BA (glycoprotein Ib [platelet], alpha polypeptide [GPIba]) (e.g., 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 IIb/IIIa complex], antigen CD41 [GPIIb]) (e.g., 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]) (e.g., neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-4a/b (R143Q)

_____ ☐ Human platelet antigen 5 genotyping (HPA-5), ITGA2 (integrin, alpha 2 [CD49B, alpha 2 subunit of VLA-2 receptor] [GPIa]) (e.g., 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]) (e.g., 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 IIb/complex, antigen CD41] [GPIIb]) (e.g., neonatal alloimmune thrombocytopenia [NAIT], post-transfusion purpura), HPA-9a/b (V837M)

_____ ☐ IL28B (interleukin 28B [interferon, lambda 3]) (e.g., drug response), rs12979860 variant

_____ ☐ IVD (isovaleryl-CoA dehydrogenase) (e.g., isovaleric acidemia), A282V variant

_____ ☐ LCT (lactase-phlorizin hydrolase) (e.g., lactose intolerance), 13910 C>T variant

_____ ☐ NEB (nebulin) (e.g., nemaline myopathy 2), exon 55 deletion variant

_____ ☐ PCDH15 (protocadherin-related 15) (e.g., Usher syndrome type 1F), R245X variant

_____ ☐ SLCO1B1 (solute carrier organic anion transporter family, member 1B1) (e.g., adverse drug reaction), V174A variant

_____ ☐ SERPINE1 (serpine peptidase inhibitor clade E, member 1, plasminogen activator inhibitor -1, PAI-1) (e.g., thrombophilia), 4G variant

_____ ☐ SHOC2 (soc-2 suppressor of clear homolog) (e.g., Noonan-like syndrome with loose anagen hair), S2G variant

_____ ☐ SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy), exon 7 deletion

_____ ☐ SRY (sex determining region Y) (e.g., 46,XX testicular disorder of sex development, gonadal dysgenesis), gene analysis

_____ ☐ TOR1A (torsin family 1, member A [torsin A]) (e.g., early-onset primary dystonia [DYT1]), 907_909delGAG (904_906delGAG) variant

Indication/Rationale for Testing:

81401 Molecular Pathology Level 2

Units

_____ ☐ ABCC8 (ATP-binding cassette, sub-family C [CFTR/MRP], member 8) (e.g., familial hyperinsulinism), common variants (e.g., c.3898-9G>A [c.3992-9G>A], F1388del)

_____ ☐ ABL (c-abl oncogene 1, receptor tyrosine kinase) (e.g., acquired imatinib resistance), T315I variant

_____ ☐ ACADM (acyl-CoA dehydrogenase, C-4 to C-12 straight chain, MCAD) (e.g., medium chain acyl dehydrogenase deficiency), common variants (e.g., K304E, Y42H)

_____ ☐ ADRB2 (adrenergic beta-2 receptor surface) (e.g., drug metabolism), common variants (e.g., G16R, Q27E)

_____ ☐ AFF2 (AF4/FMR2 family, member 2 [FMR2]) (e.g., fragile X mental retardation 2 [FRAXE]), evaluation to detect abnormal (e.g., expanded) alleles

_____ ☐ APOB (apolipoprotein B) (e.g., familial hypercholesterolemia type B), common variants (e.g., R3500Q, R3500W)

_____ ☐ APOE (apolipoprotein E) (e.g., hyperlipoproteinemia type III, cardiovascular disease, Alzheimer disease), common variants (e.g., *2, *3, *4)

_____ ☐ AR (androgen receptor) (e.g., spinal and bulbar muscular atrophy, Kennedy disease, X chromosome inactivation), characterization of alleles (e.g., expanded size or methylation status)

_____ ☐ ATN1 (atrophin 1) (e.g., dentatorubral-pallidoluysian atrophy), evaluation to detect abnormal (e.g., expanded) alleles

_____ ☐ ATXN1 (ataxin 1) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles

_____ ☐ ATXN10 (ataxin 10) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles

_____ ☐ ATXN2 (ataxin 2) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles

_____ ☐ ATXN3 (ataxin 3) (e.g., spinocerebellar ataxia, Machado-Joseph disease), evaluation to detect abnormal (e.g., expanded) alleles

_____ ☐ ATXN7 (ataxin 7) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ ATXN8OS (ATXN8 opposite strand [non-protein coding]) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ CACNA1A (calcium channel, voltage-dependent, P/Q type, alpha 1A subunit) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ CBFB/MYH11 (inv(16)) (e.g., acute myeloid leukemia), qualitative, and quantitative, if performed
_____ ☐ CBS (cystathionine-beta-synthase) (e.g., homocystinuria, cystathionine beta-synthase deficiency), common variants (e.g., I278T, G307S)
_____ ☐ CCND1/IGH (BCL1/IgH, t(11;14)) (e.g., mantle cell lymphoma) translocation analysis, major breakpoint, qualitative and quantitative, if performed
_____ ☐ CFH/ARMS2 (complement factor H/age-related maculopathy susceptibility 2) (e.g., macular degeneration), common variants (e.g., Y402H [CFH], A69S [ARMS2])
_____ ☐ CNBP (CCHC-type zinc finger, nucleic acid binding protein) (e.g., myotonic dystrophy type 2), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ CSTB (cystatin B [stefin B]) (e.g., Unverricht-Lundborg disease), evaluation to detect abnormal (e.g., expanded) alleles CYP3A4 (cytochrome P450, family 3, subfamily A, polypeptide 4) (e.g., drug metabolism), common variants (e.g., *2, *3, *4, *5, *6)
_____ ☐ CYP3A5 (cytochrome P450, family 3, subfamily A, polypeptide 5) (e.g., drug metabolism), common variants (e.g., *2, *3, *4, *5, *6)
_____ ☐ DMPK (dystrophia myotonica-protein kinase) (e.g., myotonic dystrophy, type 1), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ E2A/PBX1 (t(1;19)) (e.g., acute lymphocytic leukemia), translocation analysis, qualitative, and quantitative, if performed
_____ ☐ EML4/ALK (inv(2)) (e.g., non-small cell lung cancer), translocation or inversion analysis
_____ ☐ ETV6/NTRK3 (t(12;15)) (e.g., congenital/infantile fibrosarcoma), translocation analysis, qualitative, and quantitative, if performed
_____ ☐ ETV6/RUNX1 (t(12;21)) (e.g., acute lymphocytic leukemia), translocation analysis, qualitative and quantitative, if performed
_____ ☐ EWSR1/ATF1 (t(12;22)) (e.g., clear cell sarcoma), translocation analysis, qualitative, and quantitative, if performed
_____ ☐ EWSR1/ERG (t(21;22)) (e.g., Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative and quantitative, if performed
_____ ☐ EWSR1/FLI1 (t(11;22)) (e.g., Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative and quantitative, if performed
_____ ☐ EWSR1/WT1 (t(11;22)) (e.g., desmoplastic small round cell tumor), translocation analysis, qualitative and quantitative, if performed
_____ ☐ F11 (coagulation factor XI) (e.g., coagulation disorder), common variants (e.g., E117X [Type II], F283L [Type III], IVS14del14, and IVS14+1G>A [Type I])
_____ ☐ FGFR3 (fibroblast growth factor receptor 3) (e.g., achondroplasia, hypochondroplasia), common variants (e.g., 1138G>A, 1138G>C, 1620C>A, 1620C>G)
_____ ☐ FIP1L1/PDGFRA (del[4q12]) (e.g., imatinib-sensitive chronic eosinophilic leukemia), qualitative and quantitative, if performed
_____ ☐ FLG (filaggrin) (e.g., ichthyosis vulgaris), common variants (e.g., R501X, 2282del4, R2447X, S3247X, 3702delG)
_____ ☐ FOXO1/PAX3 (t(2;13)) (e.g., alveolar rhabdomyosarcoma), translocation analysis, qualitative and quantitative, if performed
_____ ☐ FOXO1/PAX7 (t(1;13)) (e.g., alveolar rhabdomyosarcoma), translocation analysis, qualitative and quantitative, if performed
_____ ☐ FUS/DDIT3 (t(12;16)) (e.g., myxoid liposarcoma), translocation analysis, qualitative, and quantitative, if performed FOXO1/PAX3 (t(1;13)) (e.g., Ewing sarcoma/peripheral neuroectodermal tumor), translocation analysis, qualitative and quantitative, if performed
_____ ☐ FXN (frataxin) (e.g., Friedreich ataxia), evaluation to detect abnormal (expanded) alleles
_____ ☐ GALC (galactosylceramidase) (e.g., Krabbe disease), common variants (e.g., c.857G>A, 30-kb deletion)
_____ ☐ GALT (galactose-1-phosphate uridylyltransferase) (e.g., galactosemia), common variants (e.g., Q188R, S135L, K285N, T138M, L195P, Y209C, IVS2-2A>G, P171S, del5kb, N314D, L218L/N314D)
_____ ☐ H19 (imprinted maternally expressed transcript [non-protein coding]) (e.g., Beckwith-Wiedemann syndrome), methylation analysis
_____ ☐ HBB (hemoglobin, beta) (e.g., sickle cell anemia, hemoglobin C, hemoglobin E), common variants (e.g., HbS, HbC, HbE)
_____ ☐ HTT (huntingtin) (e.g., Huntington disease), evaluation to detect abnormal expanded alleles) expanded
_____ ☐ KCNQ1OT1 (KCNQ1 overlapping transcript 1 [non-protein coding]) (e.g., Beckwith-Wiedemann syndrome), methylation analysis
_____ ☐ LRRK2 (leucine-rich repeat kinase 2) (e.g., Parkinson disease), common variants (e.g., R1441G, G2019S, I2020T)
_____ ☐ MED12 (mediator complex subunit 12) (e.g., FG syndrome type 1, Lujan syndrome), common variants (e.g., R961W, N1007S)
_____ ☐ MEG3/DLK1 (maternally expressed 3 [non-protein coding]/delta-like 1 homolog [Drosophila]) (e.g., intrauterine growth retardation), methylation analysis
_____ ☐ MLL/AFF1 (t[4:11]) (e.g. acute lymphoblastic leukemia), translocation analysis, qualitative and quantitative, if performed
_____ ☐ MLL/MLLT3 (T(9:11)) (e.g., acute myeloid leukemia) translocation analysis, qualitative and quantitative, if performed
_____ ☐ MT-ATP6 (mitochondrially encoded ATP synthase 6) (e.g., neuropathy with ataxia and retinitis pigmentosa [NARP], Leigh syndrome), common variants (e.g., m.8993T>G, m.8993T>C)
_____ ☐ MT-ND4, MT-ND6 (mitochondrially encoded NADH dehydrogenase 4, mitochondrially encoded NADH dehydrogenase 6) (e.g. Leber hereditary optic neuropathy [LHON], common variants (e.g. m.11778G>A, m34606>A, m14484T>C)
_____ ☐ MT-ND5 (mitochondrially encoded tRNA leucine 1 [UUA/G] mitochondrially encoded NADH dehydrogenase 5) (e.g., mitochondrial encephalopathy with lactic acidosis and stroke-like episodes [MELAS]), common variants (e.g., m.3243A>G, m.3271T>C, m.3252A>G, m.13513G>A)

_____ ☐ MT-RNR1 (mitochondrially encoded 12S RNA) (e.g., nonsyndromic hearing loss), common variants (e.g., m.1555>G, m1494C>T)
_____ ☐ MT-TK (mitochondrially encoded tRNA lysine) (e.g., myclonic epilepsy with ragged-red fibers [MERRF]), common variants (e.g., m8344A>G, m.8356T>C)
_____ ☐ MT-TL1 (mirochondrially encoded tRNA leucine 1 [UUA/G] (e.g., diabetes and hearing loss), common variants (e.g., m.3243A>G, m.14709 T>C) MT-TL1
_____ ☐ MT-TS1, MT-RNR1 (mitochondrially encoded tRNA serine 1 [UCN], mitochondrially encoded 12S RNA) (e.g., nonsyndromic sensorineural deafness [including aminoglycoside-induced nonsyndromic deafness]) common variants (e.g., m.7445A>G, m.1555A>G)
_____ ☐ MUTYH (mutY homolog [E.coli]) (e.g., MYH-associated polyposis), common variants (e.g., Y165C, G382D)
_____ ☐ NOD2 (nucleotide-binding oligomerization domain containing 2) (e.g., Crohn's disease, Blau syndrome), common variants (e.g., SNP 8, SNP 12, SNP 13)
_____ ☐ NPM/ALK (t(2;5)) (e.g., anaplastic large cell lymphoma), translocation analysis
_____ ☐ PABPN1 (poly[A] binding protein, nuclear 1) (e.g., oculopharyngeal muscular dystrophy), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ PAX8/PPARG (t(2;3) (q13;p25)) (e.g., follicular thyroid carcinoma), translocation analysis
_____ ☐ PPP2R2B (protein phosphatase 2, regulatory subunit B, beta) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ PRSS1 (protease, serine, 1 [trypsin 1]) (e.g. hereditary pancreatitis), common variants (e.g., N29I, A16V, R122H)
_____ ☐ PYGM (phosphorylase, glycogen, muscle) (e.g. glycogen storage disease type V, McArdle disease), common variants (e.g., R50X, G205S)
_____ ☐ RUNX1/RUNX1T1 (t(8;21)) (e.g., acute myeloid leukemia) translocation analysis, qualitative and quantitative, if performed
_____ ☐ SEPT9 (Septin 9) (e.g., colon cancer), methylation analysis
_____ ☐ SMN1/SMN2 (survival of motor neuron 1, telomeric/survival of motor neuron 2, centromeric) (e.g., spinal muscular atrophy), dosage analysis (e.g., carrier testing)
_____ ☐ SS18/SSX1 (t(X;18)) (e.g., synovial sarcoma), translocation analysis, qualitative and quantitative, if performed
_____ ☐ SS18/SSX2 (t(X;18)) (e.g., synovial sarcoma), translocation analysis, qualitative and quantitative, if performed
_____ ☐ TBP (TATA box binding protein) (e.g., spinocerebellar ataxia), evaluation to detect abnormal (e.g., expanded) alleles
_____ ☐ TPMT (thiopurine S-methyltransferase) (e.g., drug metabolism), common variants (e.g., *2, *3)
_____ ☐ TYMS (thymidylate synthetase) (e.g., 5-fluorouracil/5-FU drug metabolism), tandem repeat variant
_____ ☐ VWF (von Willebrand factor) (e.g., von Willebrand disease type 2N), common variants (e.g., T791M, R816W, R854Q)

Indication/Rationale for Testing:

81402 Molecular Pathology Level 3

Units

_____ ☐ COL1A1/PDGFB (t(17;22)) (e.g., dermatofibrosarcoma protuberans), translocation analysis, multiple breakpoints, qualitative, and quantitative, if performed
_____ ☐ CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide 2) (e.g., congenital adrenal hyperplasia, 21-hydroxylase deficiency), common variants (e.g., IVS2-13G, P30L, I172N, exon 6 mutation cluster [I235N, V236E, M238K], V281L, L307FfsX6, Q318X, R356W, P453S, G110VfsX21, 30-kb deletion variant)
_____ ☐ Chromosome 18q- (e.g., D18S55, D18S58, D18S61, D18S64, and D18S69) (e.g., colon cancer), allelic imbalance assessment (i.e., loss of heterozygosity)
_____ ☐ ESR1/PGR (receptor 1/progesterone receptor) ratio (e.g., breast cancer)
_____ ☐ KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (e.g., mastocytosis), common variants (e.g., D816V, D816Y, D816F)
_____ ☐ MEFV (Mediterranean fever) (e.g., familial Mediterranean fever), common variants (e.g., E148Q, P369S, F479L, M680I, I692del, M694V, M694I, K695R, V726A, A744S, R761H)
_____ ☐ MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (e.g., myeloproliferative disorder), common variants (e.g., W515A, W515K, W515L, W515R)

_____ ☐ TRD@ (T cell antigen receptor, delta) (e.g., leukemia and lymphoma), gene rearrangement analysis, evaluation to detect abnormal clonal population
_____ ☐ Uniparental disomy (UPD) (e.g., Russell-Silver syndrome, Prader-Willi/Angelman syndrome), short tandem repeat (STR) analysis
_____ ☐ IGH@/BCL2 (t(14;18)) (e.g., follicular lymphoma) translocation analysis; major breakpoint region (MBR) and minor cluster region (mcr) breakpoints, qualitative or quantitative

Indication/Rationale for Testing:

81403 Molecular Pathology Level 4

Units

_____ ☐ ABL1 (c-abl oncogene 1, receptor tyrosine kinase) (e.g., acquired imatinib tyrosine kinase inhibitor resistance), variants in the kinase domain
_____ ☐ ANG (angiogenin, ribonuclease, RNase A family, 5) (e.g., amyotrophic lateral sclerosis), full gene sequence
_____ ☐ ARX (aristaless-related homeobox) (e.g., X-linked lissencephaly with ambiguous genitalia, X-linked mental retardation), duplication/deletion analysis
_____ ☐ CEBPA (CCAAT/enhancer binding protein [C/EBP], alpha) (e.g., acute myeloid leukemia), full gene sequence
_____ ☐ CEL (carboxyl ester lipase [bile salt-stimulated lipase]) (e.g., maturity-onset diabetes of the young [MODY]), targeted sequence analysis of exon 11 (e.g., c.1785delC, c.1686delT)
_____ ☐ CTNNB1 (catenin [cadherin-associated protein], beta 1, 88kDa) (e.g., desmoid tumors), targeted sequence analysis (e.g., exon 3)
_____ ☐ DAZ/SRY (deleted in azoospermia and sex determining region Y) (e.g., male infertility), common deletions (e.g., AZFa, AZFb, AZFc, AZFd)
_____ ☐ DNMT3A (DNA [cytosine-5-]-methyltransferase 3 alpha) (e.g., acute myeloid leukemia), targeted sequence analysis (e.g., exon 23)
_____ ☐ EPCAM (epithelial cell adhesion molecule) (e.g., Lynch syndrome), duplication/deletion analysis
_____ ☐ F12 (coagulation factor XII [Hageman factor]) (e.g., angioedema, hereditary, type III; factor XII deficiency), targeted sequence analysis of exon 9
_____ ☐ F8 (coagulation factor VIII) (e.g., hemophilia A), inversion analysis, intron 1 and intron 22A
_____ ☐ FGFR3 (fibroblast growth factor receptor 3) (e.g., isolated craniosynostosis), targeted sequence analysis (e.g., exon 7)
_____ ☐ GJB1 (gap junction protein, beta 1) (e.g., Charcot-Marie-Tooth X-linked), full gene sequence
_____ ☐ GNAQ (guanine nucleotide-binding protein G[q] subunit alpha) (e.g., uveal melanoma), common variants (e.g., R183, Q209)
_____ ☐ HBB (hemoglobin, beta, beta-globin) (e.g., beta thalassemia), duplication/deletion analysis
_____ ☐ HRAS (v-Ha-ras Harvey rat sarcoma viral oncogene homolog) (e.g., Costello syndrome), exon 2 sequence
_____ ☐ IDH1 (isocitrate dehydrogenase 1 [NADP+], soluble) (e.g., glioma), common exon 4 variants (e.g., R132H, R132C)
_____ ☐ IDH2 (isocitrate dehydrogenase 2 [NADP+], mitochondrial) (e.g., glioma), common exon 4 variants (e.g., R140W, R172M)
_____ ☐ JAK2 (Janus kinase 2) (e.g., myeloproliferative disorder), exon 12 sequence and exon 13 sequence, if performed
_____ ☐ Killer cell immunoglobulin-like receptor (KIR) gene family (e.g., hematopoietic stem cell transplantation), genotyping of KIR family genes
_____ ☐ KCNC3 (potassium voltage-gated channel, Shaw-related subfamily, member 3) (e.g., spinocerebellar ataxia), targeted sequence analysis (e.g., exon 2)
_____ ☐ KCNJ11 (potassium inwardly-rectifying channel, subfamily J, member 11) (e.g., familial hyperinsulinism), full gene sequence
_____ ☐ KCNJ2 (potassium inwardly-rectifying channel, subfamily J, member 2) (e.g., Andersen-Tawil syndrome), full gene sequence
_____ ☐ KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene) (e.g., carcinoma), gene analysis, variant(s) in exon 3 (e.g., codon 61)
_____ ☐ MC4R (melanocortin 4 receptor) (e.g., obesity), full gene sequence
_____ ☐ MICA (MHC class I polypeptide-related sequence A) (e.g., solid organ transplantation), common variants (e.g., *001, *002)

_____ ☐ MPL (myeloproliferative leukemia virus oncogene, thrombopoietin receptor, TPOR) (e.g., myeloproliferative disorder), exon 10 sequence
_____ ☐ MT-RNR1 (mitochondrially encoded 12S RNA) (e.g., nonsyndromic hearing loss), full gene sequence
_____ ☐ MT-TS1 (mitochondrially encoded tRNA serine 1) (e.g., nonsyndromic hearing loss), full gene sequence
_____ ☐ NDP (Norrie disease [pseudoglioma]) (e.g., Norrie disease), duplication/deletion analysis
_____ ☐ NHLRC1 (NHL repeat containing 1) (e.g., progressive myoclonus epilepsy), full gene sequence
_____ ☐ PHOX2B (paired-like homeobox 2b) (e.g., congenital central hypoventilation syndrome), duplication/deletion analysis
_____ ☐ PLN (phospholamban) (e.g., dilated cardiomyopathy, hypertrophic cardiomyopathy), full gene sequence
_____ ☐ SH2D1A (SH2 domain containing 1A) (e.g., X-linked lymphoproliferative syndrome), duplication/deletion analysis
_____ ☐ SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy), known familial sequence variant(s)
_____ ☐ TWIST1 (twist homolog 1 [Drosophila]) (e.g., Saethre-Chotzen syndrome), duplication/deletion analysis
_____ ☐ UBA1 (ubiquitin-like modifier activating enzyme 1) (e.g., spinal muscular atrophy, X-linked), targeted sequence analysis (e.g., exon 15)
_____ ☐ VHL (von Hippel-Lindau tumor suppressor) (e.g., von Hippel-Lindau familial cancer syndrome), deletion/duplication analysis
_____ ☐ VWF (von Willebrand factor) (e.g., von Willebrand disease types 2A, 2B, 2M), targeted sequence analysis (e.g., exon 28)
_____ ☐ Known familial variant, not otherwise specified, for gene listed in Tier 1 or Tier 2, DNA sequence analysis, each variant exon (If known variant is common, use specific Tier 1 or Tier 2 code)

Indication/Rationale for Testing:

81404 Molecular Pathology Level 5

Units

_____ ☐ ACADS (acyl-CoA dehydrogenase, C-2 to C-3 short chain) (e.g., short chain acyl-CoA dehydrogenase deficiency), targeted sequence analysis (e.g., exons 5 and 6)
_____ ☐ AQP2 (aquaporin 2 [collecting duct]) (e.g., nephrogenic diabetes insipidus), full gene sequence
_____ ☐ ARX (aristaless related homeobox) (e.g., X-linked lissencephaly with ambiguous genitalia, X-linked mental retardation), full gene sequence
_____ ☐ BTD (biotinidase) (e.g., biotinidase deficiency), full gene sequence
_____ ☐ C10orf2 (chromosome 10 open reading frame 2) (e.g., mitochondrial DNA depletion syndrome), full gene sequence
_____ ☐ CAV3 (caveolin 3) (e.g., CAV3-related distal myopathy, limb-girdle muscular dystrophy type 1C), full gene sequence
_____ ☐ CD40LG (CD40 ligand) (e.g., X-linked hyper IgM syndrome), full gene sequence
_____ ☐ CDKN2A (cyclin-dependent kinase inhibitor 2A) (e.g., CDKN2A-related cutaneous malignant melanoma, familial atypical mole-malignant melanoma syndrome), full gene sequence
_____ ☐ CLRN1 (clarin 1) (e.g., Usher syndrome, type 3), full gene sequence
_____ ☐ COX6B1 (cytochrome c oxidase subunit VIb polypeptide 1) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ CPT2 (carnitine palmitoyltransferase 2) (e.g., carnitine palmitoyltransferase II deficiency), full gene sequence
_____ ☐ CRX (cone-rod homeobox) (e.g., cone-rod dystrophy 2, Leber congenital amaurosis), full gene sequence
_____ ☐ CSTB (cystatin B [stefin B]) (e.g., Unverricht-Lundborg disease), full gene sequence
_____ ☐ CYP1B1 (cytochrome P450, family 1, subfamily B, polypeptide 1) (e.g., primary congenital glaucoma), full gene sequence
_____ ☐ DMPK (dystrophia myotonica-protein kinase) (e.g., myotonic dystrophy type 1), characterization of abnormal (e.g., expanded) alleles
_____ ☐ EGR2 (early growth response 2) (e.g., Charcot-Marie-Tooth), full gene sequence
_____ ☐ EMD (emerin) (e.g., Emery-Dreifuss muscular dystrophy), duplication/deletion analysis

_____ ☐ EPM2A (epilepsy, progressive myoclonus type 2A, Lafora disease [laforin]) (e.g., progressive myoclonus epilepsy), full gene sequence
_____ ☐ FGF23 (fibroblast growth factor 23) (e.g., hypophosphatemic rickets), full gene sequence
_____ ☐ FGFR2 (fibroblast growth factor receptor 2) (e.g., craniosynostosis, Apert syndrome, Crouzon syndrome), targeted sequence analysis (e.g., exons 8, 10)
_____ ☐ FGFR3 (fibroblast growth factor receptor 3) (e.g., achondroplasia, hypochondroplasia), targeted sequence analysis (e.g., exons 8, 11, 12, 13)
_____ ☐ FHL1 (four and a half LIM domains 1) (e.g., Emery-Dreifuss muscular dystrophy), full gene sequence
_____ ☐ FKRP (Fukutin related protein) (e.g., congenital muscular dystrophy type 1C [MDC1C], limb-girdle muscular dystrophy [LGMD] type 2I), full gene sequence
_____ ☐ FOXG1 (forkhead box G1) (e.g., Rett syndrome), full gene sequence
_____ ☐ FSHMD1A (facioscapulohumeral muscular dystrophy 1A) (e.g., facioscapulohumeral muscular dystrophy), characterization of haplotype(s) (i.e., chromosome 4A and 4B haplotypes)
_____ ☐ FSHMD1A (facioscapulohumeral muscular dystrophy 1A) (e.g., facioscapulohumeral muscular dystrophy), evaluation to detect abnormal (e.g., deleted) alleles
_____ ☐ GH1 (growth hormone 1) (e.g., growth hormone deficiency), full gene sequence
_____ ☐ GP1BB (glycoprotein Ib [platelet], beta polypeptide) (e.g., Bernard-Soulier syndrome type B), full gene sequence
_____ ☐ FXN (frataxin) (e.g., Friedreich ataxia), full gene sequence
_____ ☐ HBA1/HBA2 (alpha globin 1 and alpha globin 2) (e.g., alpha thalassemia), duplication/deletion analysis (For common deletion variants of alpha globin 1 and alpha globin 2 genes, use 81257)
_____ ☐ HBB (hemoglobin, beta, beta-globin) (e.g., thalassemia), full gene sequence
_____ ☐ HNF1B (HNF1 homeobox B) (e.g., maturity-onset diabetes of the young [MODY]), duplication/deletion analysis
_____ ☐ HRAS (v-Ha-ras Harvey rat sarcoma viral oncogene homolog) (e.g., Costello syndrome), full gene sequence
_____ ☐ HSD11B2 (hydroxysteroid [11-beta] dehydrogenase 2) (e.g., mineralocorticoid excess syndrome), full gene sequence
_____ ☐ HSD3B2 (hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 2) (e.g., 3-beta-hydroxysteroid dehydrogenase type II deficiency), full gene sequence
_____ ☐ HSPB1 (heat shock 27kDa protein 1) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ INS (insulin) (e.g., diabetes mellitus), full gene sequence
_____ ☐ KCNJ1 (potassium inwardly-rectifying channel, subfamily J, member 1) (e.g., Bartter syndrome), full gene sequence
_____ ☐ KCNJ10 (potassium inwardly-rectifying channel, subfamily J, member 10) (e.g., SeSAME syndrome, EAST syndrome, sensorineural hearing loss), full gene sequence
_____ ☐ KIT (C-kit) (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (e.g., GIST, acute myeloid leukemia, melanoma), targeted gene analysis (e.g., exons 8, 11, 13, 17, 18)
_____ ☐ LITAF (lipopolysaccharide-induced TNF factor) (e.g., Charcot-Marie-Tooth), full gene sequence
_____ ☐ MEFV (Mediterranean fever) (e.g., familial Mediterranean fever), full gene sequence
_____ ☐ MEN1 (multiple endocrine neoplasia I) (e.g., multiple endocrine neoplasia type 1, Wermer syndrome), duplication/deletion analysis
_____ ☐ MMACHC (methylmalonic aciduria [cobalamin deficiency] cblC type, with homocystinuria) (e.g., methylmalonic acidemia and homocystinuria), full gene sequence
_____ ☐ NDP (Norrie disease [pseudoglioma]) (e.g., Norrie disease), full gene sequence
_____ ☐ NDUFA1 (NADH dehydrogenase [ubiquinone] 1 alpha subcomplex, 1, 7.5kDa) (e.g., Leigh syndrome, mitochondrial complex I deficiency), full gene sequence
_____ ☐ NDUFAF2 (NADH dehydrogenase [ubiquinone] 1 alpha subcomplex, assembly factor 2) (e.g., Leigh syndrome, mitochondrial complex I deficiency), full gene sequence
_____ ☐ NDUFS4 (NADH dehydrogenase [ubiquinone] Fe-S protein 4, 18kDa [NADH-coenzyme Q reductase]) (e.g., Leigh syndrome, mitochondrial complex I deficiency), full gene sequence
_____ ☐ NIPA1 (non-imprinted in Prader-Willi/Angelman syndrome 1) (e.g., spastic paraplegia), full gene sequence
_____ ☐ NLGN4X (neuroligin 4, X-linked) (e.g., autism spectrum disorders), duplication/deletion analysis
_____ ☐ NPC2 (Niemann-Pick disease, type C2 [epididymal secretory protein E1]) (e.g., Niemann-Pick disease type C2), full gene sequence
_____ ☐ NROB1 (nuclear receptor subfamily 0, group B, member 1) (e.g., congenital adrenal hypoplasia), full gene sequence
_____ ☐ NRAS (neuroblastoma RAS viral oncogene homolog) (e.g., colorectal carcinoma), exon 1 and exon 2 sequences
_____ ☐ PDGFRA (platelet-derived growth factor receptor alpha polypeptide) (e.g., gastrointestinal stromal tumor), targeted sequence analysis (e.g., exons 12, 18)
_____ ☐ PDX1 (pancreatic and duodenal homeobox 1) (e.g., maturity-onset diabetes of the young [MODY]), full gene sequence
_____ ☐ PHOX2B (paired-like homeobox 2b) (e.g., congenital central hypoventilation syndrome), full gene sequence
_____ ☐ PLP1 (proteolipid protein 1) (e.g., Pelizaeus-Merzbacher disease, spastic paraplegia), duplication/deletion analysis
_____ ☐ PQBP1 (polyglutamine binding protein 1) (e.g., Renpenning syndrome), duplication/deletion analysis
_____ ☐ PRNP (prion protein) (e.g., genetic prion disease), full gene sequence
_____ ☐ PROP1 (PROP paired-like homeobox 1) (e.g., combined pituitary hormone deficiency), full gene sequence
_____ ☐ PRSS1 (protease, serine, 1 [trypsin 1]) (e.g., hereditary pancreatitis), full gene sequence RAF1 (v-raf-1)
_____ ☐ RAF1 (v-raf-murine leukemia viral oncogene homolog 1) (e.g., LEOPARD syndrome), targeted sequence analysis (e.g., exons 7, 12, 14, 17)

_____ ☐ RHO (rhodopsin) (e.g., retinitis pigmentosa), full gene sequence
_____ ☐ RP1 (retinitis pigmentosa 1) (e.g., retinitis pigmentosa), full gene sequence
_____ ☐ SCN1B (sodium channel, voltage-gated, type I, beta) (e.g., Brugada syndrome), full gene sequence
_____ ☐ SCO2 (SCO cytochrome oxidase deficient homolog 2 [SCO1L]) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ SDHC (succinate dehydrogenase complex, subunit C, integral membrane protein, 15kDa) (e.g., hereditary paraganglioma-pheochromocytoma syndrome), duplication/deletion analysis
_____ ☐ SDHD (succinate dehydrogenase complex, subunit D, integral membrane protein) (e.g., hereditary paraganglioma), full gene sequence
_____ ☐ SGCG (sarcoglycan, gamma [35kDa dystrophin-associated glycoprotein]) (e.g., limb-girdle muscular dystrophy), duplication/deletion analysis
_____ ☐ SH2D1A (SH2 domain containing 1A) (e.g., X-linked lymphoproliferative syndrome), full gene sequence
_____ ☐ SLC16A2 (solute carrier family 16, member 2 [thyroid hormone transporter]) (e.g., specific thyroid hormone cell transporter deficiency, Allan-Herndon-Dudley syndrome), duplication/deletion analysis
_____ ☐ SLC25A20 (solute carrier family 25 [carnitine/acylcarnitine translocase], member 20) (e.g., carnitine-acylcarnitine translocase deficiency), duplication/deletion analysis
_____ ☐ SLC25A4 (solute carrier family 25 [mitochondrial carrier; adenine nucleotide translocation], member 4) (e.g., progressive external ophthalmoplegia), full gene sequence
_____ ☐ SOD1 (superoxide dismutase 1, soluble) (e.g., amyotrophic lateral sclerosis), full gene sequence
_____ ☐ SPINK1 (serine peptidase inhibitor, Kazal type 1) (e.g., hereditary pancreatitis), full gene sequence
_____ ☐ STK11 (serine/threonine kinase 11) (e.g., Peutz-Jeghers syndrome), duplication/deletion analysis
_____ ☐ TACO1 (translational activator of mitochondrial encoded cytochrome c oxidase I) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ THAP1 (THAP domain containing, apoptosis associated protein 1) (e.g., torsion dystonia), full gene sequence
_____ ☐ TOR1A (torsin family 1, member A [torsin A]) (e.g., torsion dystonia), full gene sequence
_____ ☐ TP53 (tumor protein 53) (e.g., tumor samples), targeted sequence analysis of 2-5 exons
_____ ☐ TTPA (tocopherol [alpha] transfer protein) (e.g., ataxia), full gene sequence
_____ ☐ TTR (transthyretin) (e.g., familial transthyretin amyloidosis), full gene sequence
_____ ☐ TWIST1 (twist homolog 1 [Drosophila]) (e.g., Saethre-Chotzen syndrome), full gene sequence
_____ ☐ TYR (tyrosinase [oculocutaneous albinism IA]) (e.g., oculocutaneous albinism IA), full gene sequence
_____ ☐ USH1G (Usher syndrome 1G [autosomal recessive]) (e.g., Usher syndrome, type 1), full gene sequence
_____ ☐ VHL (von Hippel-Lindau tumor suppressor) (e.g., von Hippel-Lindau familial cancer syndrome), full gene sequence
_____ ☐ VWF (von Willebrand factor) (e.g., von Willebrand disease type 1C), targeted sequence analysis (e.g., exons 26, 27, 37)
_____ ☐ ZEB2 (zinc finger E-box binding homeobox 2) (e.g., Mowat-Wilson syndrome), duplication/deletion analysis
_____ ☐ ZNF41 (zinc finger protein 41) (e.g., X-linked mental retardation 89), full gene sequence

Indication/Rationale for Testing:

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Units

_____ ☐ ABCD1 (ATP-binding cassette, sub-family D [ALD], member 1) (e.g., adrenoleukodystrophy), full gene sequence
_____ ☐ ACADS (acyl-CoA dehydrogenase, C-2 to C-3 short chain) (e.g., short chain acyl-CoA dehydrogenase deficiency), full gene sequence
_____ ☐ ACTA2 (actin, alpha 2, smooth muscle, aorta) (e.g., thoracic aortic aneurysms and aortic dissections), full gene sequence

_____ ☐ ACTC1 (actin, alpha, cardiac muscle 1) (e.g., familial hypertrophic cardiomyopathy), full gene sequence
_____ ☐ ANKRD1 (ankyrin repeat domain 1) (e.g., dilated cardiomyopathy), full gene sequence
_____ ☐ APTX (aprataxin) (e.g., ataxia with oculomotor apraxia 1), full gene sequence
_____ ☐ AR (androgen receptor) (e.g., androgen insensitivity syndrome), full gene sequence
_____ ☐ ARSA (arylsulfatase A) (e.g., arylsulfatase A deficiency), full gene sequence
_____ ☐ BCKDHA (branched chain keto acid dehydrogenase E1, alpha polypeptide) (e.g., maple syrup urine disease, type 1A), full gene sequence
_____ ☐ BCS1L (BCS1-like [S. cerevisiae]) (e.g., Leigh syndrome, mitochondrial complex III deficiency, GRACILE syndrome), full gene sequence
_____ ☐ BMPR2 (bone morphogenetic protein receptor, type II [serine/threonine kinase]) (e.g., heritable pulmonary arterial hypertension), duplication/deletion analysis
_____ ☐ CASQ2 (calsequestrin 2 [cardiac muscle]) (e.g., catecholaminergic polymorphic ventricular tachycardia), full gene sequence
_____ ☐ CASR (calcium-sensing receptor) (e.g., hypocalcemia), full gene sequence
_____ ☐ CDKL5 (cyclin-dependent kinase-like 5) (e.g., early infantile epileptic encephalopathy), duplication/deletion analysis
_____ ☐ CHRNA4 (cholinergic receptor, nicotinic, alpha 4) (e.g., nocturnal frontal lobe epilepsy), full gene sequence
_____ ☐ CHRNB2 (cholinergic receptor, nicotinic, beta 2 [neuronal]) (e.g., nocturnal frontal lobe epilepsy), full gene sequence
_____ ☐ COX10 (COX10 homolog, cytochrome c oxidase assembly protein) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ COX15 (COX15 homolog, cytochrome c oxidase assembly protein) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ CYP11B1 (cytochrome P450, family 11, subfamily B, polypeptide 1) (e.g., congenital adrenal hyperplasia), full gene sequence
_____ ☐ CYP17A1 (cytochrome P450, family 17, subfamily A, polypeptide 1) (e.g., congenital adrenal hyperplasia), full gene sequence
_____ ☐ CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide2) (e.g., steroid 21-hydroxylase isoform, congenital adrenal hyperplasia), full gene sequence
_____ ☐ DBT (dihydrolipoamide branched chain transacylase E2) (e.g., maple syrup urine disease, type 2), duplication/deletion analysis
_____ ☐ DCX (doublecortin) (e.g., X-linked lissencephaly), full gene sequence
_____ ☐ DES (desmin) (e.g., myofibrillar myopathy), full gene sequence
_____ ☐ DFNB59 (deafness, autosomal recessive 59) (e.g., autosomal recessive nonsyndromic hearing impairment), full gene sequence
_____ ☐ DGUOK (deoxyguanosine kinase) (e.g., hepatocerebral mitochondrial DNA depletion syndrome), full gene sequence
_____ ☐ DHCR7 (7-dehydrocholesterol reductase) (e.g., Smith-Lemli-Opitz syndrome), full gene sequence
_____ ☐ EIF2B2 (eukaryotic translation initiation factor 2B, subunit 2 beta, 39kDa) (e.g., leukoencephalopathy with vanishing white matter), full gene sequence
_____ ☐ EMD (emerin) (e.g., Emery-Dreifuss muscular dystrophy), full gene sequence
_____ ☐ ENG (endoglin) (e.g., hereditary hemorrhagic telangiectasia, type 1), duplication/deletion analysis
_____ ☐ EYA1 (eyes absent homolog 1 [Drosophila]) (e.g., branchio-oto-renal [BOR] spectrum disorders), duplication/deletion analysis
_____ ☐ F9 (coagulation factor IX) (e.g., hemophilia B), full gene sequence
_____ ☐ FGFR1 (fibroblast growth factor receptor 1) (e.g., Kallmann syndrome 2), full gene sequence
_____ ☐ FH (fumarate hydratase) (e.g., fumarate hydratase deficiency, hereditary leiomyomatosis with renal cell cancer), full gene sequence
_____ ☐ FKTN (fukutin) (e.g., limb-girdle muscular dystrophy [LGMD] type 2M or 2L), full gene sequence
_____ ☐ FTSJ1 (FtsJ RNA methyltransferase homolog 1 [E. coli]) (e.g., X-linked mental retardation 9), duplication/deletion analysis
_____ ☐ GABRG2 (gamma-aminobutyric acid [GABA] A receptor, gamma 2) (e.g., generalized epilepsy with febrile seizures), full gene sequence
_____ ☐ GCH1 (GTP cyclohydrolase 1) (e.g., autosomal dominant dopa-responsive dystonia), full gene sequence
_____ ☐ GDAP1 (ganglioside-induced differentiation-associated protein 1) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ GFAP (glial fibrillary acidic protein) (e.g., Alexander disease), full gene sequence
_____ ☐ GHR (growth hormone receptor) (e.g., Laron syndrome), full gene sequence
_____ ☐ GHRHR (growth hormone releasing hormone receptor) (e.g., growth hormone deficiency), full gene sequence
_____ ☐ GLA (galactosidase, alpha) (e.g., Fabry disease), full gene sequence
_____ ☐ HBA1/HBA2 (alpha globin 1 and alpha globin 2) (e.g., thalassemia), full gene sequence
_____ ☐ HNF1A (HNF1 homeobox A) (e.g., maturity-onset diabetes of the young [MODY]), full gene sequence
_____ ☐ HNF1B (HNF1 homeobox B) (e.g., maturity-onset diabetes of the young [MODY]), full gene sequence
_____ ☐ HTRA1 (HtrA serine peptidase 1) (e.g., macular degeneration), full gene sequence
_____ ☐ IDS (iduronate 2-sulfatase) (e.g., mucopolysaccharidosis, type II), full gene sequence
_____ ☐ IL2RG (interleukin 2 receptor, gamma) (e.g., X-linked severe combined immunodeficiency), full gene sequence
_____ ☐ ISPD (isoprenoid synthase domain containing) (e.g., muscle-eye-brain disease, Walker-Warburg syndrome), full gene sequence

_____ ☐ KRAS (v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog) (e.g., Noonan syndrome), full gene sequence
_____ ☐ LAMP2 (lysosomal-associated membrane protein 2) (e.g., Danon disease), full gene sequence
_____ ☐ LDLR (low density lipoprotein receptor) (e.g., familial hypercholesterolemia), duplication/deletion analysis
_____ ☐ MEN1 (multiple endocrine neoplasia I) (e.g., multiple endocrine neoplasia type 1, Wermer syndrome), full gene sequence
_____ ☐ MMAA (methylmalonic aciduria [cobalamine deficiency] type A) (e.g., MMAA-related methylmalonic acidemia), full gene sequence
_____ ☐ MMAB (methylmalonic aciduria [cobalamine deficiency] type B) (e.g., MMAA-related methylmalonic acidemia), full gene sequence
_____ ☐ MPI (mannose phosphate isomerase) (e.g., congenital disorder of glycosylation 1b), full gene sequence
_____ ☐ MPV17 (MpV17 mitochondrial inner membrane protein) (e.g., mitochondrial DNA depletion syndrome), full gene sequence
_____ ☐ MPZ (myelin protein zero) (e.g., Charcot-Marie-Tooth), full gene sequence
_____ ☐ MTM1 (myotubularin 1) (e.g., X-linked centronuclear myopathy), duplication/deletion analysis
_____ ☐ MYL2 (myosin, light chain 2, regulatory, cardiac, slow) (e.g., familial hypertrophic cardiomyopathy), full gene sequence
_____ ☐ MYL3 (myosin, light chain 3, alkali, ventricular, skeletal, slow) (e.g., familial hypertrophic cardiomyopathy), full gene sequence
_____ ☐ MYOT (myotilin) (e.g., limb-girdle muscular dystrophy), full gene sequence
_____ ☐ NDUFS7 (NADH dehydrogenase [ubiquinone] Fe-S protein 7, 20kDa [NADH-coenzyme Q reductase]) (e.g., Leigh syndrome, mitochondrial complex I deficiency), full gene sequence
_____ ☐ NDUFS8 (NADH dehydrogenase [ubiquinone] Fe-S protein 8, 23kDa [NADH-coenzyme Q reductase]) (e.g., Leigh syndrome, mitochondrial complex I deficiency), full gene sequence
_____ ☐ NDUFV1 (NADH dehydrogenase [ubiquinone] flavoprotein 1, 51kDa) (e.g., Leigh syndrome, mitochondrial complex I deficiency), full gene sequence
_____ ☐ NEFL (neurofilament, light polypeptide) (e.g., Charcot-Marie-Tooth), full gene sequence
_____ ☐ NF2 (neurofibromin 2 [merlin]) (e.g., neurofibromatosis, type 2), duplication/deletion analysis
_____ ☐ NLGN3 (neuroligin 3) (e.g., autism spectrum disorders), full gene sequence
_____ ☐ NLGN4X (neuroligin 4, X-linked) (e.g., autism spectrum disorders), full gene sequence
_____ ☐ NPHP1 (nephronophthisis 1 [juvenile]) (e.g., Joubert syndrome), deletion analysis, and duplication analysis, if performed
_____ ☐ NPHS2 (nephrosis 2, idiopathic, steroid-resistant [podocin]) (e.g., steroid-resistant nephrotic syndrome), full gene sequence
_____ ☐ NSD1 (nuclear receptor binding SET domain protein 1) (e.g., Sotos syndrome), duplication/deletion analysis
_____ ☐ OTC (ornithine carbamoyltransferase) (e.g., ornithine transcarbamylase deficiency), full gene sequence
_____ ☐ PAFAH1B1 (platelet-activating factor acetylhydrolase 1b, regulatory subunit 1 [45kDa]) (e.g., lissencephaly, Miller-Dieker syndrome), duplication/deletion analysis
_____ ☐ PARK2 (Parkinson protein 2, E3 ubiquitin protein ligase [parkin]) (e.g., Parkinson disease), duplication/deletion analysis
_____ ☐ PCCA (propionyl CoA carboxylase, alpha polypeptide) (e.g., propionic acidemia, type 1), duplication/deletion analysis
_____ ☐ PCDH19 (protocadherin 19) (e.g., epileptic encephalopathy), full gene sequence
_____ ☐ PDHA1 (pyruvate dehydrogenase [lipoamide] alpha 1) (e.g., lactic acidosis), duplication/deletion analysis
_____ ☐ PDHB (pyruvate dehydrogenase [lipoamide] beta) (e.g., lactic acidosis), full gene sequence
_____ ☐ PINK1 (PTEN induced putative kinase 1) (e.g., Parkinson disease), full gene sequence
_____ ☐ PLP1 (proteolipid protein 1) (e.g., Pelizaeus-Merzbacher disease, spastic paraplegia), full gene sequence
_____ ☐ POU1F1 (POU class 1 homeobox 1) (e.g., combined pituitary hormone deficiency), full gene sequence
_____ ☐ PQBP1 (polyglutamine binding protein 1) (e.g., Renpenning syndrome), full gene sequence
_____ ☐ PRX (periaxin) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ PSEN1 (presenilin 1) (e.g., Alzheimer's disease), full gene sequence
_____ ☐ RAB7A (RAB7A, member RAS oncogene family) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ RAI1 (retinoic acid induced 1) (e.g., Smith-Magenis syndrome), full gene sequence
_____ ☐ REEP1 (receptor accessory protein 1) (e.g., spastic paraplegia), full gene sequence
_____ ☐ RET (ret proto-oncogene) (e.g., multiple endocrine neoplasia, type 2A and familial medullary thyroid carcinoma), targeted sequence analysis (e.g., exons 10, 11, 13-16)
_____ ☐ RPS19 (ribosomal protein S19) (e.g., Diamond-Blackfan anemia), full gene sequence
_____ ☐ RRM2B (ribonucleotide reductase M2 B [TP53 inducible]) (e.g., mitochondrial DNA depletion), full gene sequence
_____ ☐ SCO1 (SCO cytochrome oxidase deficient homolog 1) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ SDHB (succinate dehydrogenase complex, subunit B, iron sulfur) (e.g., hereditary paraganglioma), full gene sequence
_____ ☐ SDHC (succinate dehydrogenase complex, subunit C, integral membrane protein, 15kDa) (e.g., hereditary paraganglioma-pheochromocytoma syndrome), full gene sequence

_____ ☐ SGCA (sarcoglycan, alpha [50kDa dystrophin-associated glycoprotein]) (e.g., limb-girdle muscular dystrophy), full gene sequence
_____ ☐ SGCB (sarcoglycan, beta [43kDa dystrophin-associated glycoprotein]) (e.g., limb-girdle muscular dystrophy), full gene sequence
_____ ☐ SGCD (sarcoglycan, delta [35kDa dystrophin-associated glycoprotein]) (e.g., limb-girdle muscular dystrophy), full gene sequence
_____ ☐ SGCE (sarcoglycan, epsilon) (e.g., myoclonic dystonia), duplication/deletion analysis
_____ ☐ SGCG (sarcoglycan, gamma [35kDa dystrophin-associated glycoprotein]) (e.g., limb-girdle muscular dystrophy), full gene sequence
_____ ☐ SHOC2 (soc-2 suppressor of clear homolog) (e.g., Noonan-like syndrome with loose anagen hair), full gene sequence
_____ ☐ SHOX (short stature homeobox) (e.g., Langer mesomelic dysplasia), full gene sequence
_____ ☐ SIL1 (SIL1 homolog, endoplasmic reticulum chaperone [S. cerevisiae]) (e.g., ataxia), full gene sequence
_____ ☐ SLC16A2 (solute carrier family 16, member 2 [thyroid hormone transporter]) (e.g., specific thyroid hormone cell transporter deficiency, Allan-Herndon-Dudley syndrome), full gene sequence
_____ ☐ SLC22A5 (solute carrier family 22 [organic cation/carnitine transporter], member 5) (e.g., systemic primary carnitine deficiency), full gene sequence
_____ ☐ SLC25A20 (solute carrier family 25 [carnitine/acylcarnitine translocase], member 20) (e.g., carnitine-acylcarnitine translocase deficiency), full gene sequence
_____ ☐ SLC2A1 (solute carrier family 2 [facilitated glucose transporter], member 1) (e.g., glucose transporter type 1 [GLUT 1] deficiency syndrome), full gene sequence
_____ ☐ SMAD4 (SMAD family member 4) (e.g., hemorrhagic telangiectasia syndrome, juvenile polyposis), duplication/deletion analysis
_____ ☐ SMN1 (survival of motor neuron 1, telomeric) (e.g., spinal muscular atrophy), full gene sequence
_____ ☐ SPAST (spastin) (e.g., spastic paraplegia), duplication/deletion analysis
_____ ☐ SPG7 (spastic paraplegia 7 [pure and complicated autosomal recessive]) (e.g., spastic paraplegia), duplication/deletion analysis
_____ ☐ SPRED1 (sprouty-related, EVH1 domain containing 1) (e.g., Legius syndrome), full gene sequence
_____ ☐ STAT3 (signal transducer and activator of transcription 3 [acute-phase response factor]) (e.g., autosomal dominant hyper-IgE syndrome), targeted sequence analysis (e.g., exons 12, 13, 14, 16, 17, 20, 21)
_____ ☐ STK11 (serine/threonine kinase 11) (e.g., Peutz-Jeghers syndrome), full gene sequence
_____ ☐ SURF1 (surfeit 1) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ TARDBP (TAR DNA binding protein) (e.g., amyotrophic lateral sclerosis), full gene sequence
_____ ☐ TBX5 (T-box 5) (e.g., Holt-Oram syndrome), full gene sequence
_____ ☐ TCF4 (transcription factor 4) (e.g., Pitt-Hopkins syndrome), duplication/deletion analysis
_____ ☐ TGFBR1 (transforming growth factor, beta receptor 1) (e.g., Marfan syndrome), full gene sequence
_____ ☐ TGFBR2 (transforming growth factor, beta receptor 2) (e.g., Marfan syndrome), full gene sequence
_____ ☐ THRB (thyroid hormone receptor, beta) (e.g., thyroid hormone resistance, thyroid hormone beta receptor deficiency), full gene sequence or targeted sequence analysis of >5 exons
_____ ☐ TK2 (thymidine kinase 2, mitochondrial) (e.g., mitochondrial DNA depletion syndrome), full gene sequence
_____ ☐ TNNC1 (troponin C type 1 [slow]) (e.g., hypertrophic cardiomyopathy or dilated cardiomyopathy), full gene sequence
_____ ☐ TNNI3 (troponin 1, type 3 [cardiac]) (e.g., familial hypertrophic cardiomyopathy), full gene sequence
_____ ☐ TP53 (tumor protein 53) (e.g., Li-Fraumeni syndrome, tumor samples), full gene sequence or targeted sequence analysis of >5 exons
_____ ☐ TPM1 (tropomyosin 1 [alpha]) (e.g., familial hypertrophic cardiomyopathy), full gene sequence
_____ ☐ TSC1 (tuberous sclerosis 1) (e.g., tuberous sclerosis), duplication/deletion analysis
_____ ☐ TYMP (thymidine phosphorylase) (e.g., mitochondrial DNA depletion syndrome), full gene sequence
_____ ☐ VWF (von Willebrand factor) (e.g., von Willebrand disease type 2N), targeted sequence analysis (e.g., exons 18-20, 23-25)
_____ ☐ WT1 (Wilms tumor 1) (e.g., Denys-Drash syndrome, familial Wilms tumor), full gene sequence
_____ ☐ ZEB2 (zinc finger E-box binding homeobox 2) (e.g., Mowat-Wilson syndrome), full gene sequence
_____ ☐ 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
_____ ☐ 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
_____ ☐ Do not report analyte-specific molecular pathology services separately when the analytes are part of the microarray analysis of chromosome 22q13
_____ ☐ Do not report analyte-specific molecular pathology services separately when the analytes are part of the microarray analysis of the X chromosome
_____ ☐ Do not report with (88271)
_____ ☐ Do not report with (88271)

_____ ☐ Mitochondrial genome deletions (e.g., Kearns-Sayre syndrome [KSS], chronic progressive external ophthalmoplegia [CPEO], Pearson syndrome), deletion analysis, and duplication analysis, if performed

Indication/Rationale for Testing:

81406 Molecular Pathology Level 7

Units

_____ ☐ NOTCH3 (notch 3) (e.g., cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy [CADASIL]), targeted sequence analysis (e.g., exons 1-23)
_____ ☐ RAF1 (v-raf-1 murine leukemia viral oncogene homolog 1) (e.g., LEOPARD syndrome), full gene sequence
_____ ☐ ACADVL (acyl-CoA dehydrogenase, very long chain) (e.g., very long chain acyl-coenzyme A dehydrogenase deficiency), full gene sequence
_____ ☐ ACTN4 (actinin, alpha 4) (e.g., focal segmental glomerulosclerosis), full gene sequence
_____ ☐ AFG3L2 (AFG3 ATPase family gene 3-like 2 [S. cerevisiae]) (e.g., spinocerebellar ataxia), full gene sequence
_____ ☐ AIRE (autoimmune regulator) (e.g., autoimmune polyendocrinopathy syndrome type 1), full gene sequence
_____ ☐ ALDH7A1 (aldehyde dehydrogenase 7 family, member A1) (e.g., pyridoxine-dependent epilepsy), full gene sequence
_____ ☐ ANO5 (anoctamin 5) (e.g., limb-girdle muscular dystrophy), full gene sequence
_____ ☐ APP (amyloid beta [A4] precursor protein) (e.g., Alzheimer's disease), full gene sequence
_____ ☐ ASS1 (argininosuccinate synthase 1) (e.g., citrullinemia type I), full gene sequence
_____ ☐ ATL1 (atlastin GTPase 1) (e.g., spastic paraplegia), full gene sequence
_____ ☐ ATP1A2 (ATPase, Na+/K+ transporting, alpha 2 polypeptide) (e.g., familial hemiplegic migraine), full gene sequence
_____ ☐ ATP7B (ATPase, Cu++ transporting, beta polypeptide) (e.g., Wilson disease), full gene sequence
_____ ☐ BBS1 (Bardet-Biedl syndrome 1) (e.g., Bardet-Biedl syndrome), full gene sequence
_____ ☐ BBS2 (Bardet-Biedl syndrome 2) (e.g., Bardet-Biedl syndrome), full gene sequence
_____ ☐ BCKDHB (branched-chain keto acid dehydrogenase E1, beta polypeptide) (e.g., maple syrup urine disease, type 1B), full gene sequence
_____ ☐ BEST1 (bestrophin 1) (e.g., vitelliform macular dystrophy), full gene sequence
_____ ☐ BMPR2 (bone morphogenetic protein receptor, type II [serine/threonine kinase]) (e.g., heritable pulmonary arterial hypertension), full gene sequence
_____ ☐ BRAF (v-raf murine sarcoma viral oncogene homolog B1) (e.g., Noonan syndrome), full gene sequence
_____ ☐ BSCL2 (Berardinelli-Seip congenital lipodystrophy 2 [seipin]) (e.g., Berardinelli-Seip congenital lipodystrophy), full gene sequence
_____ ☐ BTK (Bruton agammaglobulinemia tyrosine kinase) (e.g., X-linked agammaglobulinemia), full gene sequence
_____ ☐ CACNB2 (calcium channel, voltage-dependent, beta 2 subunit) (e.g., Brugada syndrome), full gene sequence
_____ ☐ CAPN3 (calpain 3) (e.g., limb-girdle muscular dystrophy [LGMD] type 2A, calpainopathy), full gene sequence
_____ ☐ CBS (cystathionine-beta-synthase) (e.g., homocystinuria, cystathionine beta-synthase deficiency), full gene sequence
_____ ☐ CDH1 (cadherin 1, type 1, E-cadherin [epithelial]) (e.g., hereditary diffuse gastric cancer), full gene sequence
_____ ☐ CDKL5 (cyclin-dependent kinase-like 5) (e.g., early infantile epileptic encephalopathy), full gene sequence
_____ ☐ CLCN1 (chloride channel 1, skeletal muscle) (e.g., myotonia congenita), full gene sequence
_____ ☐ CLCNKB (chloride channel, voltage-sensitive Kb) (e.g., Bartter syndrome 3 and 4b), full gene sequence
_____ ☐ CNTNAP2 (contactin-associated protein-like 2) (e.g., Pitt-Hopkins-like syndrome 1), full gene sequence
_____ ☐ COL6A2 (collagen, type VI, alpha 2) (e.g., collagen type VI-related disorders), duplication/deletion analysis

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_____ ☐ CPT1A (carnitine palmitoyltransferase 1A [liver]) (e.g., carnitine palmitoyltransferase 1A [CPT1A] deficiency), full gene sequence
_____ ☐ CRB1 (crumbs homolog 1 [Drosophila]) (e.g., Leber congenital amaurosis), full gene sequence
_____ ☐ CREBBP (CREB binding protein) (e.g., Rubinstein-Taybi syndrome), duplication/deletion analysis
_____ ☐ DBT (dihydrolipoamide branched chain transacylase E2) (e.g., maple syrup urine disease, type 2), full gene sequence
_____ ☐ DLAT (dihydrolipoamide S-acetyltransferase) (e.g., pyruvate dehydrogenase E2 deficiency), full gene sequence
_____ ☐ DLD (dihydrolipoamide dehydrogenase) (e.g., maple syrup urine disease, type III), full gene sequence
_____ ☐ DSC2 (desmocollin) (e.g., arrhythmogenic right ventricular dysplasia/cardiomyopathy 11), full gene sequence
_____ ☐ DSG2 (desmoglein 2) (e.g., arrhythmogenic right ventricular dysplasia/cardiomyopathy 10), full gene sequence
_____ ☐ DSP (desmoplakin) (e.g., arrhythmogenic right ventricular dysplasia/cardiomyopathy 8), full gene sequence
_____ ☐ EFHC1 (EF-hand domain [C-terminal] containing 1) (e.g., juvenile myoclonic epilepsy), full gene sequence
_____ ☐ EIF2B3 (eukaryotic translation initiation factor 2B, subunit 3 gamma, 58kDa) (e.g., leukoencephalopathy with vanishing white matter), full gene sequence
_____ ☐ EIF2B4 (eukaryotic translation initiation factor 2B, subunit 4 delta, 67kDa) (e.g., leukoencephalopathy with vanishing white matter), full gene sequence
_____ ☐ EIF2B5 (eukaryotic translation initiation factor 2B, subunit 5 epsilon, 82kDa) (e.g., childhood ataxia with central nervous system hypomyelination/vanishing white matter), full gene sequence
_____ ☐ ENG (endoglin) (e.g., hereditary hemorrhagic telangiectasia, type 1), full gene sequence
_____ ☐ EYA1 (eyes absent homolog 1 [Drosophila]) (e.g., branchio-oto-renal [BOR] spectrum disorders), full gene sequence
_____ ☐ F8 (coagulation factor VIII) (e.g., hemophilia A), duplication/deletion analysis
_____ ☐ FAH (fumarylacetoacetate hydrolase [fumarylacetoacetase]) (e.g., tyrosinemia, type 1), full gene sequence
_____ ☐ FASTKD2 (FAST kinase domains 2) (e.g., mitochondrial respiratory chain complex IV deficiency), full gene sequence
_____ ☐ FIG4 (FIG4 homolog, SAC1 lipid phosphatase domain containing [S. cerevisiae]) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ FTSJ1 (FtsJ RNA methyltransferase homolog 1 [E. coli]) (e.g., X-linked mental retardation 9), full gene sequence
_____ ☐ FUS (fused in sarcoma) (e.g., amyotrophic lateral sclerosis), full gene sequence
_____ ☐ GAA (glucosidase, alpha; acid) (e.g., glycogen storage disease type II [Pompe disease]), full gene sequence
_____ ☐ GALC (galactosylceramidase) (e.g., Krabbe disease), full gene sequence
_____ ☐ GALT (galactose-1-phosphate uridylyltransferase) (e.g., galactosemia), full gene sequence
_____ ☐ GARS (glycyl-tRNA synthetase) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ GCDH (glutaryl-CoA dehydrogenase) (e.g., glutaricacidemia type 1), full gene sequence
_____ ☐ GCK (glucokinase [hexokinase 4]) (e.g., maturity-onset diabetes of the young [MODY]), full gene sequence
_____ ☐ GLUD1 (glutamate dehydrogenase 1) (e.g., familial hyperinsulinism), full gene sequence
_____ ☐ GNE (glucosamine [UDP-N-acetyl]-2-epimerase/N-acetylmannosamine kinase) (e.g., inclusion body myopathy 2 [IBM2], Nonaka myopathy), full gene sequence
_____ ☐ GRN (granulin) (e.g., frontotemporal dementia), full gene sequence
_____ ☐ HADHA (hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase [trifunctional protein] alpha subunit) (e.g., long chain acyl-coenzyme A dehydrogenase deficiency), full gene sequence
_____ ☐ HADHB (hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase [trifunctional protein], beta subunit) (e.g., trifunctional protein deficiency), full gene sequence
_____ ☐ HEXA (hexosaminidase A, alpha polypeptide) (e.g., Tay-Sachs disease), full gene sequence
_____ ☐ HLCS (HLCS holocarboxylase synthetase) (e.g., holocarboxylase synthetase deficiency), full gene sequence
_____ ☐ HNF4A (hepatocyte nuclear factor 4, alpha) (e.g., maturity-onset diabetes of the young [MODY]), full gene sequence
_____ ☐ IDUA (iduronidase, alpha-L-) (e.g., mucopolysaccharidosis type I), full gene sequence
_____ ☐ INF2 (inverted formin, FH2 and WH2 domain containing) (e.g., focal segmental glomerulosclerosis), full gene sequence
_____ ☐ IVD (isovaleryl-CoA dehydrogenase) (e.g., isovaleric acidemia), full gene sequence
_____ ☐ JAG1 (jagged 1) (e.g., Alagille syndrome), duplication/deletion analysis
_____ ☐ JUP (junction plakoglobin) (e.g., arrhythmogenic right ventricular dysplasia/cardiomyopathy 11), full gene sequence
_____ ☐ KAL1 (Kallmann syndrome 1 sequence) (e.g., Kallmann syndrome), full gene sequence
_____ ☐ KCNH2 (potassium voltage-gated channel, subfamily H [eag-related], member 2) (e.g., short QT syndrome, long QT syndrome), full gene sequence
_____ ☐ KCNQ1 (potassium voltage-gated channel, KQT-like subfamily, member 1) (e.g., short QT syndrome, long QT syndrome), full gene sequence
_____ ☐ KCNQ2 (potassium voltage-gated channel, KQT-like subfamily, member 2) (e.g., epileptic encephalopathy), full gene sequence

_____ ☐ LDB3 (LIM domain binding 3) (e.g., familial dilated cardiomyopathy, myofibrillar myopathy), full gene sequence
_____ ☐ LDLR (low density lipoprotein receptor) (e.g., familial hypercholesterolemia), full gene sequence
_____ ☐ LEPR (leptin receptor(e.g., obesity with hypogonadism), full gene sequence
_____ ☐ LHCGR (luteinizing hormone/choriogonadotropin receptor) (e.g., precocious male puberty), full gene sequence
_____ ☐ LMNA (lamin A/C) (e.g., 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
_____ ☐ LRP5 (low density lipoprotein receptor-related protein 5) (e.g., osteopetrosis), full gene sequence
_____ ☐ MAP2K1 (mitogen-activated protein kinase 1) (e.g., cardiofaciocutaneous syndrome), full gene sequence
_____ ☐ MAP2K2 (mitogen-activated protein kinase 2) (e.g., cardiofaciocutaneous syndrome), full gene sequence
_____ ☐ MAPT (microtubule-associated protein tau) (e.g., frontotemporal dementia), full gene sequence
_____ ☐ MCCC1 (methylcrotonoyl-CoA carboxylase 1 [alpha]) (e.g., 3-methylcrotonyl-CoA carboxylase deficiency), full gene sequence
_____ ☐ MCCC2 (methylcrotonoyl-CoA carboxylase 2 [beta]) (e.g., 3-methylcrotonyl carboxylase deficiency), full gene sequence
_____ ☐ MFN2 (mitofusin 2) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ MTM1 (myotubularin 1) (e.g., X-linked centronuclear myopathy), full gene sequence
_____ ☐ MUT (methylmalonyl CoA mutase) (e.g., methylmalonic acidemia), full gene sequence
_____ ☐ MUTYH (mutY homolog [E. coli]) (e.g., MYH-associated polyposis), full gene sequence
_____ ☐ NDUFS1 (NADH dehydrogenase [ubiquinone] Fe-S protein 1, 75kDa [NADH-coenzyme Q reductase]) (e.g., Leigh syndrome, mitochondrial complex I deficiency), full gene sequence
_____ ☐ NF2 (neurofibromin 2 [merlin]) (e.g., neurofibromatosis, type 2), full gene sequence
_____ ☐ NPC1 (Niemann-Pick disease, type C1) (e.g., Niemann-Pick disease), full gene sequence
_____ ☐ NPHP1 (nephronophthisis 1 [juvenile]) (e.g., Joubert syndrome), full gene sequence
_____ ☐ NSD1 (nuclear receptor binding SET domain protein 1) (e.g., Sotos syndrome), full gene sequence
_____ ☐ OPA1 (optic atrophy 1) (e.g., optic atrophy), duplication/deletion analysis
_____ ☐ OPTN (optineurin) (e.g., amyotrophic lateral sclerosis), full gene sequence
_____ ☐ PAFAH1B1 (platelet-activating factor acetylhydrolase 1b, regulatory subunit 1 [45kDa]) (e.g., lissencephaly, Miller-Dieker syndrome), full gene sequence
_____ ☐ PAH (phenylalanine hydroxylase) (e.g., phenylketonuria), full gene sequence
_____ ☐ PALB2 (partner and localizer of BRCA2) (e.g., breast and pancreatic cancer), full gene sequence
_____ ☐ PARK2 (Parkinson protein 2, E3 ubiquitin protein ligase [parkin]) (e.g., Parkinson disease), full gene sequence
_____ ☐ PAX2 (paired box 2) (e.g., renal coloboma syndrome), full gene sequence
_____ ☐ PC (pyruvate carboxylase) (e.g., pyruvate carboxylase deficiency), full gene sequence
_____ ☐ PCCA (propionyl CoA carboxylase, alpha polypeptide) (e.g., propionic acidemia, type 1), full gene sequence
_____ ☐ PCCB (propionyl CoA carboxylase, beta polypeptide) (e.g., propionic acidemia), full gene sequence
_____ ☐ PCDH15 (protocadherin-related 15) (e.g., Usher syndrome type 1F), duplication/deletion analysis
_____ ☐ PDHA1 (pyruvate dehydrogenase [lipoamide] alpha 1) (e.g., lactic acidosis), full gene sequence
_____ ☐ PDHX (pyruvate dehydrogenase complex, component X) (e.g., lactic acidosis), full gene sequence
_____ ☐ PHEX (phosphate-regulating endopeptidase homolog, X-linked) (e.g., hypophosphatemic rickets), full gene sequence
_____ ☐ PKD2 (polycystic kidney disease 2 [autosomal dominant]) (e.g., polycystic kidney disease), full gene sequence
_____ ☐ PKP2 (plakophilin 2) (e.g., arrhythmogenic right ventricular dysplasia/cardiomyopathy 9), full gene sequence
_____ ☐ PNKD (e.g., paroxysmal nonkinesigenic dyskinesia), full gene sequence
_____ ☐ POLG (polymerase [DNA directed], gamma) (e.g., Alpers-Huttenlocher syndrome, autosomal dominant progressive external ophthalmoplegia), full gene sequence
_____ ☐ POMGNT1 (protein O-linked mannose beta1, 2-N acetylglucosaminyltransferase) (e.g., muscle-eye-brain disease, Walker-Warburg syndrome), full gene sequence
_____ ☐ POMT1 (protein-O-mannosyltransferase 1) (e.g., limb-girdle muscular dystrophy [LGMD] type 2K, Walker-Warburg syndrome), full gene sequence
_____ ☐ POMT2 (protein-O-mannosyltransferase 2) (e.g., limb-girdle muscular dystrophy [LGMD] type 2N, Walker-Warburg syndrome), full gene sequence
_____ ☐ PRKAG2 (protein kinase, AMP-activated, gamma 2 non-catalytic subunit) (e.g., familial hypertrophic cardiomyopathy with Wolff-Parkinson-White syndrome, lethal congenital glycogen storage disease of heart), full gene sequence
_____ ☐ PRKCG (protein kinase C, gamma) (e.g., spinocerebellar ataxia), full gene sequence
_____ ☐ PSEN2 (presenilin 2[Alzheimer's disease 4]) (e.g., Alzheimer's disease), full gene sequence

_____ ☐ PTPN11 (protein tyrosine phosphatase, non-receptor type 11) (e.g., Noonan syndrome, LEOPARD syndrome), full gene sequence
_____ ☐ PYGM (phosphorylase, glycogen, muscle) (e.g., glycogen storage disease type V, McArdle disease), full gene sequence
_____ ☐ RET (ret proto-oncogene) (e.g., Hirschsprung disease), full gene sequence
_____ ☐ RPE65 (retinal pigment epithelium-specific protein 65kDa) (e.g., retinitis pigmentosa, Leber congenital amaurosis), full gene sequence
_____ ☐ RYR1 (ryanodine receptor 1, skeletal) (e.g., malignant hyperthermia), targeted sequence analysis of exons with functionally-confirmed mutations
_____ ☐ SCN4A (sodium channel, voltage-gated, type IV, alpha subunit) (e.g., hyperkalemic periodic paralysis), full gene sequence
_____ ☐ SCNN1A (sodium channel, nonvoltage-gated 1 alpha) (e.g., pseudohypoaldosteronism), full gene sequence
_____ ☐ SCNN1B (sodium channel, nonvoltage-gated 1, beta) (e.g., Liddle syndrome, pseudohypoaldosteronism), full gene sequence
_____ ☐ SCNN1G (sodium channel, nonvoltage-gated 1, gamma) (e.g., Liddle syndrome, pseudohypoaldosteronism), full gene sequence
_____ ☐ SDHA (succinate dehydrogenase complex, subunit A, flavoprotein [Fp]) (e.g., Leigh syndrome, mitochondrial complex II deficiency), full gene sequence
_____ ☐ SETX (senataxin) (e.g., ataxia), full gene sequence
_____ ☐ SGCE (sarcoglycan, epsilon) (e.g., myoclonic dystonia), full gene sequence
_____ ☐ SH3TC2 (SH3 domain and tetratricopeptide repeats 2) (e.g., Charcot-Marie-Tooth disease), full gene sequence
_____ ☐ SLC26A4 (solute carrier family 26, member 4) (e.g., Pendred syndrome), full gene sequence
_____ ☐ SLC37A4 (solute carrier family 37 [glucose-6-phosphate transporter], member 4) (e.g., glycogen storage disease type Ib), full gene sequence
_____ ☐ SLC9A6 (solute carrier family 9 [sodium/hydrogen exchanger], member 6) (e.g., Christianson syndrome), full gene sequence
_____ ☐ SMAD4 (SMAD family member 4) (e.g., hemorrhagic telangiectasia syndrome, juvenile polyposis), full gene sequence
_____ ☐ SOS1 (son of sevenless homolog 1) (e.g., Noonan syndrome, gingival fibromatosis), full gene sequence
_____ ☐ SPAST (spastin) (e.g., spastic paraplegia), full gene sequence
_____ ☐ SPG7 (spastic paraplegia 7 [pure and complicated autosomal recessive]) (e.g., spastic paraplegia), full gene sequence
_____ ☐ STXBP1 (syntaxin-binding protein 1) (e.g., epileptic encephalopathy), full gene sequence
_____ ☐ TAZ (tafazzin) (e.g., methylglutaconic aciduria type 2, Barth syndrome), full gene sequence
_____ ☐ TCF4 (transcription factor 4) (e.g., Pitt-Hopkins syndrome), full gene sequence
_____ ☐ TH (tyrosine hydroxylase) (e.g., Segawa syndrome), full gene sequence
_____ ☐ TMEM43 (transmembrane protein 43) (e.g., arrhythmogenic right ventricular cardiomyopathy), full gene sequence
_____ ☐ TNNT2 (troponin T, type 2 [cardiac]) (e.g., familial hypertrophic cardiomyopathy), full gene sequence
_____ ☐ TRPC6 (transient receptor potential cation channel, subfamily C, member 6) (e.g., focal segmental glomerulosclerosis), full gene sequence
_____ ☐ TSC1 (tuberous sclerosis 1) (e.g., tuberous sclerosis), full gene sequence
_____ ☐ TSC2 (tuberous sclerosis 2) (e.g., tuberous sclerosis), duplication/deletion analysis
_____ ☐ UBE3A (ubiquitin protein ligase E3A) (e.g., Angelman syndrome) full gene sequence
_____ ☐ UMOD (uromodulin) (e.g., glomerulocystic kidney disease with hyperuricemia and isosthenuria), full gene sequence
_____ ☐ VWF (von Willebrand factor) (von Willebrand disease type 2A), extended targeted sequence analysis (e.g., exons 11-16, 24-26, 51, 52)
_____ ☐ WAS (Wiskott-Aldrich syndrome [eczema-thrombocytopenia]) (e.g., Wiskott-Aldrich syndrome), full gene sequence
_____ ☐ Cytogenomic microarray analysis, neoplasia (e.g., interrogation of copy number, and loss-of-heterozygosity via single nucleotide polymorphism [SNP]-based comparative genomic hybridization [CGH] microarray analysis)
_____ ☐ Do not report analyte-specific molecular pathology services separately when the analytes are part of the cytogenomic microarray analysis for neoplasia

Indication/Rationale for Testing:

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81407 Molecular Pathology Level 8

Units

_____ ☐ ABCC8 (ATP-binding cassette, sub-family C [CFTR/MRP], member 8) (e.g., familial hyperinsulinism), full gene sequence
_____ ☐ AGL (amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase) (e.g., glycogen storage disease type III), full gene sequence
_____ ☐ AHI1 (Abelson helper integration site 1) (e.g., Joubert syndrome), full gene sequence
_____ ☐ ASPM (asp [abnormal spindle] homolog, microcephaly associated [Drosophila]) (e.g., primary microcephaly), full gene sequence
_____ ☐ CACNA1A (calcium channel, voltage-dependent, P/Q type, alpha 1A subunit) (e.g., familial hemiplegic migraine), full gene sequence
_____ ☐ CHD7 (chromodomain helicase DNA binding protein 7) (e.g., CHARGE syndrome), full gene sequence
_____ ☐ COL4A4 (collagen, type IV, alpha 4) (e.g., Alport syndrome), full gene sequence
_____ ☐ COL6A1 (collagen, type VI, alpha 1) (e.g., collagen type VI-related disorders), full gene sequence
_____ ☐ COL6A2 (collagen, type VI, alpha 2) (e.g., collagen type VI-related disorders), full gene sequence
_____ ☐ COL6A3 (collagen, type VI, alpha 3) (e.g., collagen type VI-related disorders), full gene sequence
_____ ☐ CREBBP (CREB binding protein) (e.g., Rubinstein-Taybi syndrome), full gene sequence
_____ ☐ F8 (coagulation factor VIII) (e.g., hemophilia A), full gene sequence
_____ ☐ JAG1 (jagged 1) (e.g., Alagille syndrome), full gene sequence
_____ ☐ KDM5C (lysine [K]-specific demethylase 5C) (e.g., X-linked mental retardation), full gene sequence
_____ ☐ KIAA0196 (KIAA0196) (e.g., spastic paraplegia), full gene sequence
_____ ☐ L1CAM (L1 cell adhesion molecule) (e.g., MASA syndrome, X-linked hydrocephaly), full gene sequence
_____ ☐ LAMB2 (laminin, beta 2 [laminin S]) (e.g., Pierson syndrome), full gene sequence
_____ ☐ MYBPC3 (myosin binding protein C, cardiac) (e.g., familial hypertrophic cardiomyopathy), full gene sequence
_____ ☐ MYH6 (myosin, heavy chain 6, cardiac muscle, alpha) (e.g., familial dilated cardiomyopathy), full gene sequence
_____ ☐ MYH7 (myosin, heavy chain 7, cardiac muscle, beta) (e.g., familial hypertrophic cardiomyopathy, Liang distal myopathy), full gene sequence
_____ ☐ MYO7A (myosin VIIA) (e.g., Usher syndrome, type 1), full gene sequence
_____ ☐ NOTCH1 (notch 1) (e.g., aortic valve disease), full gene sequence
_____ ☐ NPHS1 (nephrosis 1, congenital, Finnish type [nephrin]) (e.g., congenital Finnish nephrosis), full gene sequence
_____ ☐ OPA1 (optic atrophy 1) (e.g., optic atrophy), full gene sequence
_____ ☐ PCDH15 (protocadherin-related 15) (e.g., Usher syndrome, type 1), full gene sequence
_____ ☐ PKD1 (polycystic kidney disease 1 [autosomal dominant]) (e.g., polycystic kidney disease), full gene sequence
_____ ☐ PLCE1 (phospholipase C, epsilon 1) (e.g., nephrotic syndrome type 3), full gene sequence
_____ ☐ SCN1A (sodium channel, voltage-gated, type 1, alpha subunit) (e.g., generalized epilepsy with febrile seizures), full gene sequence
_____ ☐ SCN5A (sodium channel, voltage-gated, type V, alpha subunit) (e.g., familial dilated cardiomyopathy), full gene sequence
_____ ☐ SLC12A1 (solute carrier family 12 [sodium/potassium/chloride transporters], member 1) (e.g., Bartter syndrome), full gene sequence
_____ ☐ SLC12A3 (solute carrier family 12 [sodium/chloride transporters], member 3) (e.g., Gitelman syndrome), full gene sequence
_____ ☐ SPG11 (spastic paraplegia 11 [autosomal recessive]) (e.g., spastic paraplegia), full gene sequence
_____ ☐ SPTBN2 (spectrin, beta, non-erythrocytic 2) (e.g., spinocerebellar ataxia), full gene sequence
_____ ☐ TMEM67 (transmembrane protein 67) (e.g., Joubert syndrome), full gene sequence
_____ ☐ TSC2 (tuberous sclerosis 2) (e.g., tuberous sclerosis), full gene sequence
_____ ☐ USH1C (Usher syndrome 1C [autosomal recessive, severe]) (e.g., Usher syndrome, type 1), full gene sequence
_____ ☐ VPS13B (vacuolar protein sorting 13 homolog B [yeast]) (e.g., Cohen syndrome), duplication/deletion analysis
_____ ☐ WDR62 (WD repeat domain 62) (e.g., primary autosomal recessive microcephaly), full gene sequence

Indication/Rationale for Testing:

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81408 Molecular Pathology Level 9

Units

_____ ☐ ABCA4 (ATP-binding cassette, sub-family A [ABC1], member 4) (e.g., Stargardt disease, age-related macular degeneration), full gene sequence
_____ ☐ ATM (ataxia telangiectasia mutated) (e.g., ataxia telangiectasia), full gene sequence
_____ ☐ CDH23 (cadherin-related 23) (e.g., Usher syndrome, type 1), full gene sequence
_____ ☐ CEP290 (centrosomal protein 290kDa) (e.g., Joubert syndrome), full gene sequence
_____ ☐ COL1A1 (collagen, type I, alpha 1) (e.g., osteogenesis imperfecta, type I), full gene sequence
_____ ☐ COL1A2 (collagen, type I, alpha 2) (e.g., osteogenesis imperfecta, type I), full gene sequence
_____ ☐ COL4A1 (collagen, type IV, alpha 1) (e.g., brain small-vessel disease with hemorrhage), full gene sequence
_____ ☐ COL4A3 (collagen, type IV, alpha 3 [Goodpasture antigen]) (e.g., Alport syndrome), full gene sequence
_____ ☐ COL4A5 (collagen, type IV, alpha 5) (e.g., Alport syndrome), full gene sequence
_____ ☐ DMD (dystrophin) (e.g., Duchenne/Becker muscular dystrophy), full gene sequence
_____ ☐ DYSF (dysferlin, limb girdle muscular dystrophy 2B [autosomal recessive]) (e.g., limb-girdle muscular dystrophy), full gene sequence
_____ ☐ FBN1 (fibrillin 1) (e.g., Marfan syndrome), full gene sequence
_____ ☐ ITPR1 (inositol 1,4,5-trisphosphate receptor, type 1) (e.g., spinocerebellar ataxia), full gene sequence
_____ ☐ LAMA2 (laminin, alpha 2) (e.g., congenital muscular dystrophy), full gene sequence
_____ ☐ LRRK2 (leucine-rich repeat kinase 2) (e.g., Parkinson disease), full gene sequence
_____ ☐ MYH11 (myosin, heavy chain 11, smooth muscle) (e.g., thoracic aortic aneurysms and aortic dissections), full gene sequence
_____ ☐ NEB (nebulin) (e.g., nemaline myopathy 2), full gene sequence
_____ ☐ NF1 (neurofibromin 1) (e.g., neurofibromatosis, type 1), full gene sequence
_____ ☐ PKHD1 (polycystic kidney and hepatic disease 1) (e.g., autosomal recessive polycystic kidney disease), full gene sequence
_____ ☐ RYR1 (ryanodine receptor 1, skeletal) (e.g., malignant hyperthermia), full gene sequence
_____ ☐ RYR2 (ryanodine receptor 2 [cardiac]) (e.g., catecholaminergic polymorphic ventricular tachycardia, arrhythmogenic right ventricular dysplasia), full gene sequence or targeted sequence analysis of > 50 exons
_____ ☐ USH2A (Usher syndrome 2A [autosomal recessive, mild]) (e.g., Usher syndrome, type 2), full gene sequence
_____ ☐ VPS13B (vacuolar protein sorting 13 homolog B [yeast]) (e.g., Cohen syndrome), full gene sequence
_____ ☐ VWF (von Willebrand factor) (e.g., von Willebrand disease types 1 and 3), full gene sequence

Indication/Rationale for Testing:

BlueCross BlueShield of New Mexico

81479 Molecular Pathology, Unlisted

Units

_____ ☐ Description:

Indication/Rationale for Testing:

_____

Practitioner Name Printed

_____

Practitioner Signature NPI Number Date