

CGC Genetics CLIA#: 99D1066287

IGM-UMDNJ CLIA#: 31D1085261 - CAP#: 7215375

Client Test Requisition – June 2011

* Indicates REQUIRED information

Complete this requisition for direct billing to hospitals, laboratories or clinics. Direct billing is the most efficient way to order from CGC Genetics. This can simplify the ordering process and avoid delay. Please note: CGC Genetics must bill hospitals directly for all Medicare hospital inpatient and outpatient testing.

Who should CGC Genetics contact with questions about this order?

Name _____
Phone _____
Fax _____
Email _____

Tests ordered*

Important: Write in the test code and the test name (see list on reverse)

Code _____ Name _____
Code _____ Name _____

ICD-9 Code*(Required) _____

Hospital/Laboratory Billing Information

(Hospital Billing is required for all Medicare patients – both inpatients and outpatients)

CGC Genetics Account # (if assigned) _____

CLIA #* _____

Purchase order # (if available) _____

Billing contact _____

Email _____

Phone _____

Fax _____

Hospital/Lab Name _____

Address _____

City _____ State _____

Zip _____ Country _____

Specimen Type

NOTE: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, social security no., patient ID no. These same two forms of ID should also be indicated on the test requisition.

Blood (EDTA) Buccal swab Fetal blood Amniotic Fluid

CVS Other (specify): _____

Collection date: mm /dd /yy _____

GA on US: _____ weeks _____ days

Patient Identification

Patient Name* First _____ Last _____

Patient ID # (if available) _____

S.S. # _____

Date of Birth* mm /dd /yy _____

Sex* Male Female Unknown

Ethnicity* African American Asian Caucasian Hispanic

Jewish Specify _____

Mailing Address* _____

City* _____ State* _____

Zip* _____ Country* _____

Phone #1* _____ Day Eve Cell

Phone #2 _____ Day Eve Cell

Authorized Result Report Recipients

Required Physician Information

NPI#* _____ UPIN#* _____

Name* First _____ Last _____

Address _____

City _____ State _____

Zip _____ Country _____

Phone #* _____ Day Eve Cell Fax# _____

Email* _____

Indications for testing (Check one)*

Diagnostic (symptomatic) Predictive (asymptomatic) Clinical Study

Carrier Prenatal Postnatal Other Research

Testing Authorization

I warrant that this test was ordered and is either: 1) for the purpose of diagnosing or detecting an existing disease, illness, impairment, symptom or disorder, or 2) that if is not for such purpose, I have obtained the appropriate prior written consent. This written consent was signed by the person who is the subject of the test (or if that person lacks capacity to consent, signed by the person authorized to consent for that person), and includes: a) a statement of the purpose and description of the test; b) a statement that prior to signing the consent form, the consenting person discussed with the medical practitioner ordering the test the reliability of positive or negative test results and the level of certainty that a positive test result for that disease or condition serves as a predictor of such disease; c) a statement that the consenting person was informed about the availability and importance of further testing, physician consultation and genetic counseling, and provided with written information identifying a genetic counselor or medical geneticist from whom the consenting person might obtain such counseling; d) a general description of each specific disease or condition testing for; and e) the person or persons to whom the test results may be disclosed as indicated above.

MEDICAL PRACTITIONER SIGNATURE* _____

Lab Information

Lab Name _____

CLIA# _____

Address _____

City _____ State _____

Zip _____ Country _____

Phone # _____ Day Eve Cell Fax# _____

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TEST REQUEST *

CGC Mutation Panel® • PATENT PENDING •

- 0001 Bardet-Biedl Syndrome**
130 mutations on genes: ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, MKKS, MKS1, TRIM32 and TTC8
- 0002 Congenital Deafness (Nonsyndromic)**
136 mutations on genes: ACTG1, CDH23, COCH, CRYM, DNFA5, DIAPH1, GJA1, GJB2, GJB3, GJB6, KCNQ4, MYH14, MYO1A, MYO7A, OTOA, OTOF, POU3F4, SLC26A4, SLC26A5, TECTA, TMC1 and WFS1
- 0003 Congenital Deafness (Syndromic)**
176 mutations on genes: CDH23, EYA1, GJB2, KCNE1, KCNQ1, MYO7A, PAX3, PCDH15, SIX1, SIX5, SLC26A4, USH1C, USH1G and WFS1
- 0004 Congenital Deafness (Syndromic and Nonsyndromic) Combined Panel**
312 mutations
- 0005 Craniosynostosis**
52 mutations on genes: FGFR1 (Pfeiffer), FGFR2 (Apert, Crouzon, Jackson-Weiss and Pfeiffer), FGFR3 (Muenke and Saethre-Chotzen) and RAB23 (Carpenter)
- 0006 Fraser Syndrome**
15 mutations on genes: FREM2 and FRAS1
- 0007 Metabolic Disorders**
93 mutations on genes: ACADM (MCAD), ARSA (Metachromatic leukodystrophy), ATP7B (Wilson disease), BTBD (Biotinidase deficiency), CLN2/TPP1 (Neuronal Ceroid Lipofuscinosis), CLN5 (Neuronal Ceroid Lipofuscinosis), CLN8 (Neuronal Ceroid Lipofuscinosis), CPT2 (CPT II deficiency), FAH (Tyrosinemia), G6PC (GSD I), GAA (Pompe disease or GSD II), GALC (Krabbe disease), GALT (Galactosemia), GBA (Gaucher disease), HADHA (LCHAD), HEXA (Tay-Sachs disease), HGD (Alkaptonuria), MAN2B1 (Alpha-mannosidosis deficiency), NPC1 (Niemann-Pick C disease), NPC2 (Niemann-Pick C disease), PEX1 (Zellweger disease), PEX26 (Zellweger disease), PPT1 (Neuronal Ceroid Lipofuscinosis), PYGM (McArdle or GSD V disease) and SLC37A4 (GSD I)
- 0008 Noonan Syndrome and Other Genetically Related Syndromes**
(Noonan, Costello, LEOPARD and Cardiofaciocutaneous) 80 mutations on genes: PTPN11, SOS1, RAF1, KRAS, MAP2K1, MAP2K, BRAF and HRAS
- 0009 Skeletal Dysplasia**
50 mutations on genes: FGFR3 (Achondroplasia and Thanatophoric Dysplasia), COL2A1 (Achondrogenesis type II), SLC26A2 (Achondrogenesis type IB), CRTAP (Osteogenesis Imperfecta recessive type), LEPRE1 (Osteogenesis Imperfecta recessive type), and SOX9 (Campomelic Dysplasia)
- 0010 Thrombophilia and Warfarin Pharmacogenetics**
15 mutations on genes: APOE Cys112Arg, APOE Arg158Cys, EPCR 4678G/C, Factor V Leiden Arg506Gln, Factor II G20210A, MTHFR C677T, MTHFR A1298C, PAI-1 4G/5G, PAI-1 -844 A>G, ACE Ins/Del, Beta-Fibrinogen -455G>A, Factor XIII Val34Leu, CYP2C9 and VKORC1]

New Diagnostic Approaches

- 0051 PND Plus+®** (see specific brochure)
 1. Conventional PND (aneuploidy detection + karyotype)
 2. Metabolic Disorders **see CGC Mutation Panel**
 3. Diagnostic Panel for Developmental Delay (MLPA, Prenatal)
 4. Cystic Fibrosis
 5. Fragile X Syndrome (FMR1 gene)
- 0052 Diagnostic Panel for Developmental Delay (MLPA, Prenatal)**
 - 15q24 Deletion Syndrome
 - 17q21 Microdeletion
 - 1p36 Deletion Syndrome
 - 2p16 Microdeletion
 - 9q22.3 Microdeletion
 - Cri du Chat Syndrome (5p15)
 - DiGeorge Syndrome (22q11)
 - DiGeorge Syndrome critical region II (10p15)
 - Duplication Xq28 (MECP2)
 - Langer-Giedion Syndrome (8q)
 - Microdeletion 3q29 Syndrome
 - Miller-Dieker Syndrome (17p)
 - NF1 Microdeletion Syndrome
 - Phelan-McDermid Syndrome (22q13)
 - Prader-Willi/Angelman Syndrome
 - Rubinstein-Taybi Syndrome
 - Smith-Magenis Syndrome
 - Sotos Syndrome (5q35.3)

WAGR Syndrome
Williams Syndrome
Wolf-Hirschhorn Syndrome (4p16.3)

- 0053 Psychomotor Development Delay – Option 1**
 1. Karyotype
 2. Fragile X Syndrome (FMR1 gene)
 3. Diagnostic Panel for Subtelomeric Rearrangements
 4. Diagnostic Panel for Common Microdeletions
 5. Metabolic Disorders **see CGC Mutation Panel**
- 0054 Psychomotor Development Delay – Option 2**
 1. Comparative Genomic Hybridization (CGH) deletion/duplication analysis of the genome
 2. Fragile X Syndrome (FMR1 gene)
 3. Metabolic Disorders **see CGC Mutation Panel**
- 0055 Tests for Increased Nuchal Translucency with Normal Karyotype**
 1. Noonan Syndrome and Other Genetically Related Syndromes **see CGC Mutation Panel**
 2. DiGeorge Syndrome (22q11)
 3. Spinal Muscular Atrophy
 4. 21-Hydroxylase deficiency

NEW TESTS

Gastroenterology

- 0354** Celiac disease (HLA-DQ/DR)
- 0362** Fructose intolerance (ALDOB gene)
- 0370** Lactose intolerance (MCM6 gene)
- 0373** Susceptibility to inflammatory bowel disease (Crohn disease and ulcerative colitis)

Others

- 0708** Autism related rearrangements (deletions/duplications of 15q11-13, 16p11, 22q13)
- 0710** CADASIL (gene NOTCH3, exons 2 to 6 and 11)
- 0722** Familial amyloid polyneuropathy (TTR gene, Met30)
- 1719** Sotos Syndrome (deletion/duplication analysis of NSD1 gene)
- 0132** Thrombophilia marker Antithrombin III (sequence analysis of SERPINC1 gene)
- 0139** Thrombophilia marker Protein C deficiency (sequence analysis of PROC gene)
- 0140** Thrombophilia marker Protein S deficiency (sequence analysis of PROS1 gene)
- 0624** Von Hippel-Lindau Syndrome (deletion/duplication analysis)

Cardiology

- 0101** Alström Syndrome (ALMS1 gene)
- 0102** Becker Muscular Dystrophy (DMD gene)
- 0103** CADASIL (gene NOTCH3, exons 2 to 6 and 11)
- 0104** Cardiofaciocutaneous Syndrome (frequent mutations of BRAF gene)
- 0105** Costello Syndrome (frequent mutations of HRAS gene)
- 0106** Dilated Cardiomyopathy (frequent mutations of LMNA, MYH7 and TNNT2 genes)
- 0107** Dilated Cardiomyopathy (X-linked, deletion/duplication analysis of DMD gene)
- 0108** Dilated Cardiomyopathy (X-linked, sequence analysis of TAZ gene)
- 0109** Duchenne Muscular Dystrophy (DMD gene)
- 0110** Fabry disease (GLA gene) (GLA gene)
- 0111** Fragile X Syndrome (FMR1 gene)
- 0112** Gaucher disease (mutations on GBA gene)
- 0113** Hereditary Hemochromatosis (frequent mutations of HFE gene)
- 0114** Holt-Oram Syndrome (sequence analysis of TBX5 gene)
- 0115** Hypercholesterolemia (LDLR and APOB genes)
- 0116** Hypertrophic Cardiomyopathy (MYH7, MYBPC3, TNNT2 and TNNI3 genes)
- 0117** LEOPARD Syndrome (frequent mutations of PTPN11 gene)
- 0118** Long QT Syndrome (sequence analysis of KCNE1 gene)
- 0119** Long QT Syndrome (sequence analysis of KCNH2 gene)
- 0120** Long QT Syndrome (sequence analysis of KCNQ1 gene)
- 0121** Long QT Syndrome (sequence analysis of SCN5A gene)
- 0122** Marfan Syndrome (sequence analysis of FBN1, TGFBR1 and TGFBR2 genes)
- 0123** Neurofibromatosis type I (deletion/duplication analysis of NF1 gene)
- 0124** Neurofibromatosis type I (sequence analysis of NF1 gene)
- 0125** Noncompaction of Left Ventricular Myocardium (TAZ gene, G4.5)

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TEST REQUEST *

- 0126 Noonan Syndrome (frequent mutations of PTPN11 gene)
- 0008 Noonan Syndrome and Other Genetically Related Syndromes **see CGC Mutation Panel**
- 0127 Clopidogrel (Plavix®) pharmacogenetics
- 0128 Spinal Muscular Atrophy (deletion/duplication analysis of SMN1 and SMN2 gene)
- 0129 Spinal Muscular Atrophy (sequence analysis of SMN1 gene)
- 0130 Steinert disease or Myotonic Dystrophy
- 0131 Thrombophilia marker ACE (ins/del)
- 0132 Thrombophilia marker Antithrombin III (sequence analysis of SERPINC1 gene)
- 0133 Thrombophilia marker APOE (alleles 2, 3 and 4)
- 0134 Thrombophilia marker Factor II (G20210A)
- 0135 Thrombophilia marker Factor V Leiden (R506Q)
- 0136 Thrombophilia marker FGB (-455G/A)
- 0137 Thrombophilia marker MTHFR (C677T and A1298C)
- 0138 Thrombophilia marker PAI-1 (4G/5G)
- 0139 Thrombophilia marker Protein C deficiency (sequence analysis of PROC gene)
- 0140 Thrombophilia marker Protein S deficiency (sequence analysis of PROS1 gene)
- 0010 Thrombophilia and Warfarin Pharmacogenetics **see CGC Mutation Panel**
- 0141 Tuberous Sclerosis (sequence analysis of TSC1 and TSC2 genes)
- 0142 Tuberous Sclerosis (sequence analysis of TSC1 gene)
- 0143 Tuberous Sclerosis (sequence analysis of TSC2 gene)

Endocrinology

- 0251 21-Hydroxylase deficiency (frequent mutations and deletion/duplication analysis of CYP21A2 gene)
- 0252 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation)
- 0253 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation), Prenatal
- 0001 Bardet-Biedl Syndrome **see CGC Mutation Panel**
- 0254 Hypercholesterolemia (APOB gene)
- 0255 Hypercholesterolemia (LDLR gene)
- 0256 Mitochondrial Encephalomyopathy (sequence analysis of hot-spots - MELAS syndrome)
- 0257 MODY 1 (sequence analysis of HNF4a gene)
- 0258 MODY 2 (sequence analysis of GCK gene)
- 0259 MODY 3 (sequence analysis of HNF1-a gene)
- 0260 MODY 5 (sequence analysis of HNF1-B gene)
- 0261 Multiple Endocrine Neoplasia type 2 (RET gene)
- 0262 Noonan Syndrome (frequent mutations on PTPN11 gene)
- 0263 Noonan Syndrome (frequent mutations on PTPN11 gene), Prenatal
- 0008 Noonan Syndrome and Other Genetically Related Syndromes **see CGC Mutation Panel**
- 0264 Obesity (susceptibility markers)
- 0265 Osteogenesis Imperfecta type 2,3,4 (sequence analysis of COL1A1 and COL1A2 genes)
- 0266 Osteogenesis Imperfecta (sequence analysis of COL1A1 gene)
- 0267 Osteogenesis Imperfecta (sequence analysis of COL1A2 gene)
- 0268 Short Stature (sequence analysis of SHOX gene)
- 0269 Sibutramin susceptibility (GNB3 gene)

Gastroenterology

- 0351 Acute Intermittent Porphyria (HMBS gene)
- 0352 Alpha-1 Antitrypsin (genotyping)
- 0353 Carnitine Palmitoyltransferase II deficiency (mutations on CPT2 gene)
- 0354 Celiac disease (HLA-DQ/DR)
- 0356 Crigler-Najjar Syndrome (UGT1A1 gene)
- 0357 Crohn disease (NOD2 gene)
- 0358 Cystic Fibrosis (frequent mutations of CFTR gene)
- 0359 Cystic Fibrosis (sequence analysis of CFTR gene)
- 0360 Familial Adenomatous Polyposis (APC gene)
- 0361 Familial Mediterranean Fever (frequent mutations of MEFV gene)
- 0362 Fructose intolerance (ALDOB gene)
- 0363 Gastric cancer (KRAS gene)
- 0364 Gilbert Syndrome (UGT1A1 gene)
- 0365 Hepatorenal Tyrosinemia (mutations on FAH gene)
- 0366 Hereditary Hemochromatosis (frequent mutations of HFE gene)
- 0367 Hereditary Non-polyposis Colorectal Cancer, type 1 and 2, HNPCC (deletion/duplication analysis of MLH1 and MSH2 genes)
- 0368 Hereditary Non-polyposis Colorectal Cancer, type 1 and 2, HNPCC (sequence analysis of MLH1 and MSH2 genes)
- 0369 Lactose intolerance (gene MCM6)
- 0370 Microsatellite Instability in Colorectal Cancer

- 0371 Susceptibility to inflammatory bowel disease (Crohn disease and ulcerative colitis)
- 0010 Thrombophilia and Warfarin Pharmacogenetics **see CGC Mutation Panel**
- 0372 Wilson disease (ATP7B gene)

Dental Medicine

- 0501 Detection of periodontal pathogens agents (panel of 5 main bacteria)
- 0502 Detection of periodontal pathogens agents (panel of 6 bacteria)
- 0503 Periodontitis Susceptibility Test (polymorphisms on IL-1 gene)

Reproductive Medicine

- 0551 Cystic Fibrosis (frequent mutations of CFTR gene)
- 0552 Cystic Fibrosis (sequence analysis of CFTR gene)
- 0553 Premature ovarian failure (FMR1 gene)
- 0554 Y chromosome microdeletion
- 0010 Thrombophilia and Warfarin Pharmacogenetics **see CGC Mutation Panel**

Nephrology

- 0601 Alport Syndrome (sequence analysis of COL4A3 gene)
- 0602 Alport Syndrome (sequence analysis of COL4A4 gene)
- 0603 Alport Syndrome (sequence analysis of COL4A5 gene)
- 0604 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation)
- 0605 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation), Prenatal
- 0001 Bardet-Biedl Syndrome **see CGC Mutation Panel**
- 0606 Congenital Nephrotic Syndrome, Steroid-Resistant, (Autosomal Recessive) (sequence analysis of NPHS2 gene)
- 0607 Congenital Nephrotic Syndrome 1 (sequence analysis of NPHS1 gene)
- 0608 Fabry disease (GLA gene)
- 0609 Familial Mediterranean Fever (frequent mutations of MEFV gene)
- 0610 Multiple Endocrine Neoplasia type 2 (RET gene)
- 0611 Polycystic Kidney Disease (sequencing analysis Autosomal Dominant, PKD1 and PKD2 genes)
- 0612 Polycystic Kidney Disease (sequencing analysis Autosomal Dominant, PKD1 gene)
- 0613 Polycystic Kidney Disease (sequencing analysis Autosomal Dominant, PKD2 gene)
- 0614 Polycystic Kidney Disease (sequencing analysis Autosomal Recessive, PKHD1 and PKHD2 genes)
- 0615 Polycystic Kidney Disease (sequencing analysis Autosomal Recessive, PKHD1 gene)
- 0616 Polycystic Kidney Disease (sequencing analysis Autosomal Recessive, PKHD2 gene, exons 3, 32, 36, 57, 58 y 61)
- 0617 Renal Glucosuria (sequence analysis of SLC5A2 gene)
- 0618 Tuberous Sclerosis (sequence analysis of TSC1 and TSC2 genes)
- 0619 Tuberous Sclerosis (sequence analysis of TSC1 gene)
- 0620 Tuberous Sclerosis (sequence analysis of TSC2 gene)
- 0621 Von Hippel-Lindau Syndrome (deletion/duplication analysis of VHL gene)

Neurology

- 0701 Alzheimer disease (APOE; alleles 2, 3 and 4)
- 0702 Alzheimer disease (sequence analysis of exons 16 and 17 of APP gene)
- 0703 Alzheimer disease (sequence analysis of PSEN1 gene)
- 0704 Alzheimer disease (sequence analysis of PSEN2 gene)
- 0705 Amyotrophic Lateral Sclerosis (mutations on SOD1 gene)
- 0706 Antipsychotic and Antidepressive pharmacogenetics
- 0707 Autism related rearrangements (deletions/duplications of 15q11-13, 16p11, 22q13)
- 0708 Becker Muscular Dystrophy (DMD gene)
- 0709 CADASIL (gene NOTCH3, exons 2 to 6 and 11)
- 0710 Charcot-Marie-Tooth disease type 1A (microsatellite analysis)
- 0711 Charcot-Marie-Tooth disease type 1B (sequence analysis of MPZ gene)
- 0712 Charcot-Marie-Tooth disease type 1C (sequence analysis of LITAF gene)
- 0713 Charcot-Marie-Tooth disease type 1E (sequence analysis of PMP22 gene)
- 0714 Charcot-Marie-Tooth disease type 2B1 (sequence analysis of LMNA gene)
- 0715 Charcot-Marie-Tooth disease type 2E/1F (sequence analysis of NEFL gene)
- 0716 Charcot-Marie-Tooth disease type 2I/2J (sequence analysis of MPZ gene)
- 0717 Charcot-Marie-Tooth disease type 2K/4A (sequence analysis of GDAP1 gene)
- 0718 Charcot-Marie-Tooth disease X-linked (sequence analysis of GJB1 gene)

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- 0719 Dejerine-Sottas disease
- 0720 Familial Amyloidotic Polyneuropathy (TTR gene, Met30)
- 0721 Familial Spastic Paraplegia type 3, SPG3 (sequence analysis of SPG3 gene)
- 0722 Familial Spastic Paraplegia type 4, SPG4 (sequence analysis of SPG4 gene)
- 0723 Friedreich Ataxia
- 0724 Huntington's disease, HD (TP-PCR)
- 0725 Leber's Hereditary Optic Neuropathy (mutation detection)
- 0726 Mitochondrial Encephalomyopathy (sequence analysis of hot-spots - MELAS syndrome)
- 0727 Myotonia Congenita AR, Thomsen disease (CLCN1)
- 0728 Myotonic Dystrophy type 2 (ZNF9 gene)
- 0729 Neurofibromatosis type I (deletion/duplication analysis of NF1 gene)
- 0730 Neurofibromatosis type I (NF1 gene)
- 0731 Neurofibromatosis type II (NF2 gene)
- 0732 Oculopharyngeal Muscular Dystrophy (PABPN1 gene)
- 0733 Parkinson disease 2, autosomal recessive juvenile (PARK 2 gene)
- 0734 Parkinson disease PARK1 (sequence analysis of SNCA gene)
- 0735 Parkinson disease PARK2 (sequence analysis of PARKIN gene)
- 0736 Parkinson disease PARK8 (sequence analysis of LRRK2 gene)
- 0737 Spinal Muscular Atrophy (deletion/duplication analysis of SMN1 and SMN2 genes)
- 0738 Spinocerebellar Ataxia Type 3 (SCA3), ATXN3 gene (TP-PCR)
- 0739 Steinert disease or Myotonic dystrophy
- 0740 Thrombophilia marker Antithrombin III (sequence analysis of SERPINC1 gene)
- 0741 Thrombophilia marker APOE (alleles 2, 3 and 4)
- 0742 Thrombophilia marker Factor II (G20210A)
- 0743 Thrombophilia marker Factor V Leiden (R506Q)
- 0744 Thrombophilia marker FGB (-455G/A)
- 0745 Thrombophilia marker MTHFR (C677T and A1298C)
- 0746 Thrombophilia marker PAI-1 (4G/5G)
- 0747 Thrombophilia marker Protein C deficiency (sequence analysis of PROC gene)
- 0748 Thrombophilia marker Protein S deficiency (sequence analysis of PROS1 gene)
- 0749 Tomaculous Neuropathy (HNPP) (dosage analysis of PMP22 gene)
- 0750 Tomaculous Neuropathy (HNPP) (microsatellite analysis)
- 0010 Thrombophilia and Warfarin Pharmacogenetics **see CGC Mutation Panel**
- 0751 Tuberous Sclerosis (sequence analysis of TSC1 and TSC2 genes)
- 0752 Tuberous Sclerosis (sequence analysis of TSC1 gene)
- 0753 Tuberous Sclerosis (sequence analysis of TSC2 gene)

Obstetrics/Gyneconology

- 0901 21-Hydroxylase deficiency (frequent mutations and deletion/duplication analysis on CYP21A2 gene)
- 0902 Achondroplasia (FGFR3 gene)
- 0903 Alpha-1 Antitrypsin (genotyping)
- 0904 Alström Syndrome (gene ALMS1)
- 0905 Angelman Syndrome (sequence analysis of UBE3A gene)
- 0001 Bardet-Biedl Syndrome **see CGC Mutation Panel**
- 0906 Becker Muscular Dystrophy (DMD gene)
- 0907 Beckwith-Wiedemann Syndrome (methylation)
- 0908 CADASIL (gene NOTCH3, exons 2 to 6 and 11)
- 0909 Comparative Genomic Hybridization (CGH) deletion/duplication analysis of the genome
- 0910 Cardiofaciocutaneous Syndrome (frequent mutations of BRAF gene)
- 0911 Charcot-Marie-Tooth disease type 1A (microsatellite analysis)
- 0912 Charcot-Marie-Tooth disease type 1B (sequence analysis of MPZ gene)
- 0913 Charcot-Marie-Tooth disease type 1C (sequence analysis of LITAF gene)
- 0914 Charcot-Marie-Tooth disease type 1E (sequence analysis of PMP22 gene)
- 0915 Charcot-Marie-Tooth disease type 2B1 (sequence analysis of LMNA gene)
- 0916 Charcot-Marie-Tooth disease type 2E/1F (sequence analysis of NEFL gene)
- 0917 Charcot-Marie-Tooth disease type 2I/2J (sequence analysis of MPZ gene)
- 0918 Charcot-Marie-Tooth disease type 2K/4A (sequence analysis of GDAP1 gene)
- 0919 Charcot-Marie-Tooth disease X-linked (sequence analysis of GJB1 gene)
- 0920 Cohen Syndrome (COH1 gene, exon 23)
- 0921 Costello Syndrome (frequent mutations of HRAS gene)
- 0005 Craniosynostosis **see CGC Mutation Panel**
- 0922 Cystic Fibrosis (frequent mutations of CFTR gene)
- 0923 Cystic Fibrosis (sequence analysis of CFTR gene)
- 0924 Detection of aneuploidies on uncultured amniotic fluid by Multiplex-PCR
- 0925 Duchenne Muscular Dystrophy (DMD gene)
- 0926 Familial Breast/Ovarian Cancer (deletion/duplication analysis of BRCA1 and BRCA2 genes)
- 0927 Familial Breast/Ovarian Cancer (sequence analysis of BRCA1 and BRCA2 genes)

- 0928 Familial Hemochromatosis (frequent mutations of HFE gene)
- 0929 Fetal Rh (from Amniotic fluid ONLY)
- 0930 Fragile X Syndrome (FMR1 gene)
- 0006 Fraser Syndrome **see CGC Mutation Panel**
- 0931 Holt-Oram Syndrome (sequence analysis of TBX5 gene)
- 0932 HPV genotyping (by PCR, greater sensitivity than hybrid capture)
- 0933 Hypocondroplasia (FGFR3 gene)
- 0934 Leber's Hereditary Optic Neuropathy (sequence analysis of hot-spots)
- 0935 LEOPARD Syndrome (frequent mutations of PTPN11 gene)
- 0936 Marfan Syndrome (sequence analysis of FBN1, TGFB1 and TGFB2 genes)
- 0007 Metabolic Disorders **see CGC Mutation Panel**
- 0937 Neurofibromatosis type I (deletion/duplication analysis of NF1 gene)
- 0938 Neurofibromatosis type I (NF1 gene)
- 0939 Noonan Syndrome (frequent mutations of PTPN11 gene)
- 0940 Noonan Syndrome (frequent mutations on PTPN11 gene), Prenatal
- 0008 Noonan Syndrome and Other Genetically Related Syndromes **see CGC Mutation Panel**

Mutation Panel

- 0941 Oculopharyngeal Muscular Dystrophy (PABPN1 gene)
- 0942 Prader-Willi/Angelman Syndrome (methylation)
- 0943 Premature ovarian failure (FMR1 gene)
- 0944 Pycnodysostosis (gene CTSK)
- 0945 Silver-Russell Syndrome (methylation analysis of H19 gene)
- 0009 Skeletal Dysplasia **see CGC Mutation Panel**
- 0946 Spinal Muscular Atrophy (deletion/duplication analysis of SMN1 and SMN2 genes)
- 0947 Spinal Muscular Atrophy (sequence analysis of SMN1 gene)
- 0948 Steinert disease or Myotonic Dystrophy
- 0010 Thrombophilia and Warfarin Pharmacogenetics **see CGC Mutation Panel**
- 0949 Tomaculous Neuropathy (HNPP) (dosage analysis of PMP22 gene)
- 0950 Tomaculous Neuropathy (HNPP) (microsatellite analysis)
- 0052 Panel of Syndromes associated with Developmental Delay (MLPA, Prenatal) **See CGC New Diagnostic Approaches**

Ophthalmology

- 1151 Alström Syndrome (gene ALMS1)
- 1152 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation)
- 1153 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation), Prenatal
- 0001 Bardet-Biedl Syndrome **see CGC Mutation Panel**
- 1154 Leber's Hereditary Optic Neuropathy (sequence analysis of hot-spots)
- 1155 Retinitis Pigmentosa

Otorhinolaryngology

- 1201 Congenital Deafness (Mitochondrial frequent mutations)
- 1202 Congenital Deafness (MLPA deletion/duplication analysis of GJB2, GJB6, GJB3, POU3F4 and WFS1 genes)
- 1203 Congenital Deafness (MLPA deletion/duplication analysis of GJB2, GJB6, GJB3, POU3F4 and WFS1 genes) AND (Nonsyndromic) DFNB1 and DFNA3 (GJB2/Connexin 26 sequencing)
- 1204 Congenital Deafness (Nonsyndromic X-linked) DFN3 (POU3F4 gene)
- 1205 Congenital Deafness (Nonsyndromic) DFNB1 (GJB6 sequencing)
- 1206 Congenital Deafness (Nonsyndromic) DFNB1 (GJB6, 2 deletion)
- 1207 Congenital Deafness (Nonsyndromic) DFNB1 and DFNA3 (GJB2/Connexin 26 sequencing)
- 1209 Congenital Deafness (Nonsyndromic) DFNB9 (OTOF gene sequencing)
- 0002 Congenital Deafness (Nonsyndromic) **see CGC Mutation Panel**
- 0003 Congenital Deafness (Syndromic) **see CGC Mutation Panel**
- 0004 Congenital Deafness (Syndromic and Nonsyndromic) **see CGC Mutation Panel**
- 1210 Usher Syndrome (mutations on MYO7A, CDH23, PCDH15, USH1C and USH1G genes)
- 1211 Waardenburg Syndrome (PAX3 gene)

Pediatrics/Clinical Genetics

- 1651 21-Hydroxylase deficiency (frequent mutations and deletion/duplication analysis of CYP21A2 gene)
- 1652 Achondroplasia (FGFR3 gene)
- 1653 Alpha-1 Antitrypsin (genotyping)
- 1654 Alstrom Syndrome (ALMS1 gene)
- 1655 Angelman Syndrome (sequence analysis of UBE3A gene)
- 1656 Autism related rearrangements (deletions/duplications of 15q11-13, 16p11, 22q13)

CGC Genetics CLIA#: 99D1066287

IGM-UMDNJ CLIA#: 31D1085261 - CAP#: 7215375

Client Test Requisition – June 2011

* Indicates REQUIRED information

TEST REQUEST *

- 1657 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation)
- 1658 Bardet-Biedl Syndrome (BBS1 gene, M390R mutation), Prenatal
- 0001 Bardet-Biedl Syndrome **see CGC Mutation Panel**
- 1659 Becker Muscular Dystrophy (DMD gene)
- 1660 Beckwith-Wiedemann Syndrome (methylation)
- 1661 Cardiofaciocutaneous Syndrome (frequent mutations of BRAF gene)
- 1662 Celiac disease (HLA-DQ/DR)
- 1663 Charcot-Marie-Tooth disease type 1A (microsatellite analysis)
- 1664 Charcot-Marie-Tooth disease type 1B (sequence analysis of MPZ gene)
- 1665 Charcot-Marie-Tooth disease type 1C (sequence analysis of LITAF gene)
- 1666 Charcot-Marie-Tooth disease type 1E (sequence analysis of PMP22 gene)
- 1667 Charcot-Marie-Tooth disease type 2B1 (sequence analysis of LMNA gene)
- 1668 Charcot-Marie-Tooth disease type 2E/1F (sequence analysis of NEFL gene)
- 1679 Charcot-Marie-Tooth disease type 2I/2J (sequence analysis of MPZ gene)
- 1670 Charcot-Marie-Tooth disease type 2K/4A (sequence analysis of GDAP1 gene)
- 1671 Charcot-Marie-Tooth disease X-linked (sequence analysis of GJB1 gene)
- 1672 Cohen Syndrome (COH1 gene, exon 23)
- 1673 Comparative Genomic Hybridization (CGH) deletion/duplication analysis of the genome
- 1674 Congenital Deafness (MLPA deletion/duplication analysis of GJB2, GJB6, GJB3, POU3F4 and 1 genes)
- 1675 Congenital Deafness (Nonsyndromic) DFNA3 (sequence analysis of GJB6 gene)
- 1676 Congenital Deafness (Nonsyndromic) DFNB1 (sequence analysis of GJB6 gene)
- 1677 Congenital Deafness (Nonsyndromic) DFNB1 and DFNA3 (sequence analysis of GJB2/Connexin 26 gene)
- 1678 Congenital Deafness (Nonsyndromic) DFNB9 (sequence analysis of OTOF gene)
- 1679 Congenital Deafness (X-Linked sequence analysis of POU3F4 gene)
- 0002 Congenital Deafness (Nonsyndromic) **see CGC Mutation Panel**
- 0003 Congenital Deafness (Syndromic) **see CGC Mutation Panel**
- 0004 Congenital Deafness (Syndromic and Nonsyndromic) **see CGC Mutation Panel**
- 1681 Congenital Nephrotic Syndrome (sequence analysis NPHS2 gene)
- 1682 Congenital Nephrotic Syndrome 1 (sequence analysis NPHS1 gene)
- 1683 Costello Syndrome (frequent mutations of HRAS gene)
- 1684 Crohn disease (NOD2 gene)
- 1685 Cystic Fibrosis (frequent mutations of CFTR gene)
- 1686 Cystic Fibrosis (sequence analysis of CFTR gene)
- 1687 Duchenne Muscular Dystrophy (DMD gene)
- 1688 Familial Mediterranean Fever (frequent mutations of MEFV gene)
- 1689 Fragile X Syndrome (FMR1 gene) (FMR1 gene)
- 0006 Fraser Syndrome **see CGC Mutation Panel**
- 1690 Fructose intolerance (ALDOB gene)
- 1691 Holt-Oram Syndrome (sequence analysis of TBX5 gene)
- 1692 Hypochondroplasia (FGFR3 gene)
- 1693 LEOPARD Syndrome (frequent mutations of PTPN11 gene)
- 1694 Long QT Syndrome (sequence analysis of KCNE1 gene)
- 1695 Long QT Syndrome (sequence analysis of KCNH2 gene)
- 1696 Long QT Syndrome (sequence analysis of KCNQ1 gene)
- 1697 Long QT Syndrome (sequence analysis of SCN5A gene)
- 1698 Marfan Syndrome (sequence analysis of FBN1, TGFBR1 and TGFBR2 genes)
- 1699 Mitochondrial Congenital Deafness (frequent mutations)
- 1700 Mitochondrial Encephalomyopathy (sequence analysis of hot-spots - MELAS syndrome)
- 1701 MODY 1 (sequence analysis of HNF4a gene)
- 1702 MODY 2 (sequence analysis of GCK gene)
- 1703 MODY 3 (sequence analysis of HNF1-a gene)
- 1704 MODY 5 (sequence analysis of HNF1-B gene)
- 1705 Neurofibromatosis type I (deletion/duplication analysis of NF1 gene)
- 1706 Neurofibromatosis type I (NF1 gene)
- 1707 Noonan Syndrome (frequent mutations of PTPN11 gene)
- 1708 Noonan Syndrome (frequent mutations of PTPN11 gene), Prenatal
- 0008 Noonan Syndrome and Other Genetically Related Syndromes **see CGC Mutation Panel**
- 1709 Oculopharyngeal Muscular Dystrophy (PABPN1)
- 1710 Osteogenesis Imperfecta type 2,3,4 (sequence analysis of COL1A1 and COL1A2 genes)
- 1711 Osteogenesis Imperfecta (sequence analysis of COL1A1 gene)
- 1712 Osteogenesis Imperfecta (sequence analysis of COL1A2 gene)
- 1713 Prader Willi/Angelman Syndrome (methylation)
- 1714 Pycnodysostosis (gene CTSK)
- 1715 Renal Glucosuria (sequence analysis of SLC5A2 gene)
- 1716 Saethre-Shotzen Syndrome (sequence analysis of TWIST1 gene)
- 1717 Short Stature (sequence analysis of SHOX gene)

- 1718 Silver-Russell Syndrome (methylation analysis of H19 gene)
- 0009 Skeletal Dysplasia **see CGC Duplication Panel**
- 1719 Sotos Syndrome (deletion/duplication analysis of NSD1 gene)
- 1720 Sotos Syndrome (NSD1 gene)
- 1721 Spinal Muscular Atrophy (deletion/duplication analysis of SMN1 and SMN2 gene)
- 1722 Spinal Muscular Atrophy (sequence analysis of SMN1 gene)
- 1723 Steinert disease or Myotonic Dystrophy
- 1724 Subtelomeric rearrangements by MLPA
- 0010 Thrombophilia and Warfarin Pharmacogenetics **see CGC Mutation Panel**
- 1725 Tomaculous Neuropathy (HNPP) (dosage analysis of PMP22 gene)
- 1726 Tomaculous Neuropathy (HNPP) (microsatellite analysis)
- 1727 Tuberous Sclerosis (sequence analysis of TSC1 and TSC2 genes)
- 1728 Tuberous Sclerosis (sequence analysis of TSC1 gene)
- 1729 Tuberous Sclerosis (sequence analysis of TSC2 gene)
- 1730 Usher Syndrome (mutations on MYO7A, CDH23, PCDH15, USH1C and USH1G genes)
- 1731 Waardenburg Syndrome (mutations on PAX3 gene)
- 0052 Panel of Syndromes associated with Developmental Delay (MLPA, Prenatal) **See CGC New Diagnostic Approaches**

Pharmacogenomics

- 1951 Antipsychotic and Antidepressive pharmacogenetics
- 1952 Clopidogrel (Plavix®) pharmacogenetics
- 1953 Cardiology Panel for pharmacogenetics
- 1954 Drug metabolism (CYP2D6, CYP2C9, CYP2C19, CYP3A4 and NAT2 genes)
- 1955 Resistance to Imatinib (c-KIT)
- 1956 Resistance to Imatinib due to BCR/ABL mutations
- 1957 Resistance to Methotrexate (MTHFR)
- 1959 Susceptibility to Cetuximab (KRAS)
- 1960 Susceptibility to Irinotecan (UGT1A1)
- 1961 Susceptibility to Sibutramin (GNB3)
- 1962 Susceptibility to Warfarin (CYP2C9 and VKORC1)
- 1963 Tamoxifen pharmacogenetics

Pulmonology/Pneumology

- 2051 Alpha-1 Antitrypsin (genotyping)
- 2052 Cystic Fibrosis (frequent mutations of CFTR gene)
- 2053 Cystic Fibrosis (sequence analysis of CFTR gene)
- 2054 Drug Metabolism (CYP2D6, CYP2C9, CYP2C19, CYP3A4 and NAT2 genes)
- 0010 Thrombophilia and Warfarin Pharmacogenetics **see CGC Mutation Panel**
- 2055 Xenobiotics Metabolism (GSTM1, GSTT1 and NAT2 genes)

Family Testing

- 2101 Familial mutation assessment
- 2102 Familial mutation assessment with primary test performed at another lab
- 2103 Familial mutation assessment with primary test performed at another lab, Prenatal

Type of Analysis	Type of Sample	Amount
CGC Mutation Panel	DNA	500 ng
	Peripheral Blood – L	3-5 mL
Molecular Diagnosis	DNA	500 ng
	Peripheral Blood – L	3-5 mL
Molecular Diagnosis (prenatal testing)	DNA from fetus +	500 ng
	DNA from mother	
Molecular Diagnosis (expression analysis)	RNA	1000 ng
	Peripheral Blood (PAX gene tubes)	3 mL
Cytogenetics Analysis	Non stained cytogenetics slides	3 slides per culture
	Fixed cell suspension	1 tube per culture
	Peripheral Blood (green top tube with Sodium Heparin) (for conventional karyotyping and FISH)	3-5 mL

Shipping: Send specimen overnight at room temperature (must arrive less than 24 hrs after collection). Ship **Monday through Thursday** only.

Tube Type: L - Lavender top tube with EDTA

Note: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, social security no., patient ID no. These same two forms of ID should also be indicated on the test requisition.