



Patient First Name: \_\_\_\_\_ MI \_\_\_\_\_ Last Name \_\_\_\_\_  
 Gender:  Male  Female  Unknown  
 \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  
 Date of Birth: (mm/dd/yy)  
 Patient Address: Street \_\_\_\_\_ Apt # \_\_\_\_\_  
 \_\_\_\_\_  
 City/State/Zip Code \_\_\_\_\_  
 \_\_\_\_\_  
 ( ) ( )  
 Patient Phone: Home \_\_\_\_\_ Work Phone \_\_\_\_\_

**Reporting Address**

Physician/CGC \_\_\_\_\_  
 Address \_\_\_\_\_  
 \_\_\_\_\_  
 ( ) ( )  
 Phone \_\_\_\_\_ Fax (\*Important) \_\_\_\_\_  
 Beeper \_\_\_\_\_  
 Email \_\_\_\_\_

**Duplicate Report Address**

Physician/CGC \_\_\_\_\_  
 Address \_\_\_\_\_  
 \_\_\_\_\_  
 ( ) ( )  
 Phone \_\_\_\_\_ Fax (\*Important) \_\_\_\_\_  
 Beeper \_\_\_\_\_  
 Email \_\_\_\_\_

\_\_\_\_\_  
 SUBMITTER'S PATIENT IDENTIFIER(S):  
 \_\_\_\_\_  
 \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  
 Date Sample Obtained (mm/dd/yy)

**Sample Type** (NOTE, only ONE specimen is required for multiple tests)

- buccal brushes (must be GeneDx kits)
- blood in EDTA (lavender top - single tube of 1-5mL)
- skin punch biopsy, size \_\_\_\_\_mm
- DNA from \_\_\_\_\_ (tissue type?)  
(concentration: \_\_\_\_\_ µg/mL)
- fetal tissue: Specify type \_\_\_\_\_

**History: Please provide any applicable information**

Clinical diagnosis or family history, including any reason to be considered for preferential priority (scheduled surgery, pregnancy management, etc.).

Relationship to & name of other relatives submitted.

Gestational age if currently pregnant:

**Tests: Write or check on pages 2 - 3**

- I would like expedited testing
- Reason:  Pregnancy  Transplantation  \_\_\_\_\_

**Payment: Please Complete Page 4**

**Ordering Checklist**

- Sample Submission form (pages 1-3)
- Completed Payment Option form (page 4)
- Signed Informed Consent form (if appropriate)
- Specimen Tube, appropriately labeled

**For GenomeDx Oligo Microarray CGH Analysis, Use Separate GenomeDx Sample Submission Form**

# Testing Services for Rare Mendelian Disorders

For GenomeDx Oligo Microarray CGH Analysis, Use Separate GenomeDx Sample Submission Form

## Test Code Test Name

### Alagille Syndrome (JAG1)

- I001 Tier 1 Sequencing
- I002 Tier 2 Sequencing, if Tier 1 negative
- I003 CopyDx<sup>SM</sup>, if appropriate
- I004 Full Gene Sequencing NOW

### Bone Marrow Failure Syndromes

- I04 Congenital Amegakaryocytic Thrombocytopenia (MPL)
- I05 Congenital or Cyclic Neutropenia (ELA2)
- I06 Diamond-Blackfan Anemia (RPS19)
- I07 Dyskeratosis Congenita, Autosomal (hTR/TERC)
- I08 Dyskeratosis Congenita, X-linked (DKC1)
- I09 Shwachman-Diamond Syndrome (SBDS)

### Coffin-Lowry Syndrome (RSK2)

- I101 Tier 1 Sequencing
- I102 Tier 2 Sequencing, if Tier 1 negative
- I103 CopyDx<sup>SM</sup> (females only)
- I104 Full Gene Sequencing NOW

### Congenital Ichthyoses

- I14 Chanarin-Dorfman Syndrome (CGI-58/ABHD5)
- Congenital Recessive Ichthyosis (Erythrodermic)
  - I151 ALOX12B
  - I152 ALOXE3, if ALOX12B is negative
- I18 Epidermolytic Hyperkeratosis (KRT1, KRT10)
- I19 Erythrokeratoderma Variabilis (GJB3, GJB4)
- I20 Harlequin Ichthyosis (ABCA12)
- I22 Ichthyosis Bullosa of Siemens (KRT2e)
- I23 Ichthyosis Vulgaris (FLG)
- I24 Keratitis-Ichthyosis-Deafness (KID) (GJB2; connexin26)
- I25 Lamellar Ichthyosis (TGM1)
- I26 Lamellar Ichthyosis Type 2 (N.African type) (ABCA12)
- I27 Netherton Syndrome (SPINK5)
- I28 Sjögren-Larsson Syndrome (FALDH)

### Developmental Eye Disorders

- Aniridia, other diagnoses (PAX6)
  - I311 Sequencing
  - I312 CopyDx<sup>SM</sup>, if appropriate
- Anophthalmia, Microphthalmia (SIX6)
  - I321 Sequencing
  - I322 CopyDx<sup>SM</sup>, if appropriate
- Anophthalmia, Microphthalmia (SOX2)
  - I331 Sequencing
  - I332 CopyDx<sup>SM</sup>, if appropriate
- Axenfeld-Rieger Syndrome
  - I341 PITX2 Sequencing
  - I343 CopyDx<sup>SM</sup>, if appropriate
  - I342 FOXC1 Sequencing
  - I344 CopyDx<sup>SM</sup>, if appropriate
- X-linked Retinoschisis (XLRSL)
  - I2571 Sequencing
  - I2572 CopyDx<sup>SM</sup> (females only)

### Disorders involving Bones and Limbs

- I285 Cherubism (SH3BP2)
- I262 Duane-Radial-Ray Syndrome (DRRS; SALL4)
- Hereditary Multiple Exostosis (EXT1/EXT2)
  - I1811 EXT1 only
  - I1812 EXT2, if EXT1 negative
  - I1813 Both EXT1 & EXT2 NOW
- Holt-Oram Syndrome (TBX5)
  - I2361 Sequencing
  - I2362 CopyDx<sup>SM</sup>, if appropriate
  - I2363 Prenatal TBX5 test based on ultrasound abnormalities
- I248 Popliteal Pterygium Syndrome (IRF6)
- I249 Pseudoachondroplasia/Mult. Epiphyseal Dysplasia (COMP) †
- Townes-Brocks Syndrome (SALL1)
  - I2521 Sequencing
  - I2522 CopyDx<sup>SM</sup>, if appropriate
  - I2523 Prenatal SALL1 test based on ultrasound abnormalities

### Disorders of the Immune System

- Agammaglobulinemia, X-linked (BTK)
  - I541 Sequencing
  - I542 CopyDx<sup>SM</sup> Analysis, (females only)
- Autoimmune Lymphoproliferative Syndrome (ALPS)
  - I138 ALPS1A (TNFRSF6)
  - I2611 ALPS2A (CASPI0)
  - I2612 ALPS2B (CASP8)
- Autoimmune Polyendocrinopathy/APECED (AIRE)
  - I391 Tier 1 Sequencing
  - I392 Tier 2 Sequencing, if Tier 1 negative
  - I393 Full Gene Sequencing NOW
- Chronic Granulomatous Disease (CGD)
  - I434 CYBB (p91) X-linked, Sequencing
  - I435 NCF1, exon 2 (p47), Common Autosomal Recessive
  - I436 CYBA (p22) Autosomal Recessive, Sequencing
  - I437 NCF2 (p67) Autosomal Recessive, Sequencing
  - I432 CYBB (p91) CopyDx (female add-on if negative I434)

## Test Code Test Name

- I146 Leukocyte Adhesion Deficiency (ITGB2)
- Severe Combined Immunodeficiency [SCID]
  - I145 JAK3 Deficiency (JAK3)
  - I147 RAG1 and RAG2 Deficiency (incl. Omenn Syndrome)
- SCID with Radiation Sensitivity (ARTEMIS/DCLRE1C)
  - I501 Entire gene Sequencing
  - I502 Exon 8 only for Athabaskan Indians

### Ectodermal Dysplasia Syndromes

- X-linked Hypohidrotic ED (EDA aka ED1) †
  - I601 Sequencing
  - I602 CopyDx<sup>SM</sup> (females only)
- I56 Autos. Rec./Dom. Hypohidrotic ED (EDAR)
- I57 Clouston Syndrome (GJB6, connexin30)
- I58 Ectrodactyly-ED-Clefting (TP63, p63)
- I59 Hay-Wells Syndrome (TP63, p63)

### Epidermolysis Bullosa

- I62 Epidermolysis Bullosa, Dystrophic (COL7A1)
- Epidermolysis Bullosa, Junctional (Laminin 5)
  - I631 Tier 1 (hot spots LAMB3, LAMC2, LAMA3)
  - I632 LAMB3 full, if Tier 1 negative
  - I633 LAMC2 full, if Tier 1 and LAMB3 negative
  - I634 LAMA3 full Sequencing, if others negative
- I635 Epidermolysis Bullosa With Pyloric Atresia (JEB-PA) (ITGB4)
- I68 Epidermolysis Bullosa, Simplex (KRT5, KRT14)
- I636 GABEB/non-Herlitz form (COL17A1)

### Familial Hyperparathyroid Syndromes/Endocrine Neoplasias

- I69 Autosomal Dominant Hypocalcemia (CASR)
- I170 Familial Hypocalciuric Hypercalcemia (CASR)
- I171 Familial Isolated Hypoparathyroidism (CASR)
- Hyperparathyroidism-Jaw Tumor Syndrome or Parathyroid carcinoma or Familial Isolated Hyperparathyroidism (HRPT2)
  - I731 Tier 1
  - I732 Tier 2, if Tier 1 negative
  - I733 Entire HRPT2 gene NOW
- Multiple Endocrine Neoplasia Type 1 (MEN1, Menin)
  - I761 Sequencing
  - I762 CopyDx<sup>SM</sup>, if appropriate
- I177 Multiple Endocrine Neoplasia Type 2A or Familial Medullary Thyroid Carcinoma (RET)
- I178 Multiple Endocrine Neoplasia Type 2B (RET)
- I180 Neonatal Severe Primary Hyperparathyroidism (CASR)

### Hereditary Rickets

- I184 Autos. Dom. Hypophosphataemia (FGF23)
- I185 Autos. Rec. Vit. D Dependent Rickets (CYP27B1)
- X-linked Dominant Hypophosphatemia (PHEX)
  - I1861 Sequencing
  - I1862 CopyDx<sup>SM</sup> (females only)

### Hermansky-Pudlak Syndrome (HPS1 and/or HPS3)

- I188 HPS1 and HPS3 Puerto Rican mutations
- I189 HPS3 Ashkenazi splice mutation

### Inborn Errors of Metabolism

- I274 Cobalamin C Deficiency (MMACHC)
- Fabry Disease (GLA)
  - I2321 Sequencing
  - I2322 CopyDx<sup>SM</sup> (females only)
- LCHAD/Trifunctional Protein deficiency (HADHA and HADHB)
  - I2711 HADHA Tier 1 (Common Mutation)
  - I2712 HADHA (Full Sequencing)
  - I272 HADHB
- MCAD deficiency (ACADM)
  - I2681 Sequencing exon 11 only
  - I2682 Full Gene Sequencing
- Methylmalonic Acidemia (MUT, MMAA, MMAB)
  - I2751 MUT Sequencing Exon 2, if appropriate
  - I2752 MUT Full Sequencing
  - I276 MMAA
  - I277 MMAB
- I243 Mucopolidosis Type IV (MCOLN1)
- Niemann-Pick disease (NPD)
  - I2631 NPD Type A/B (SMPD1) Entire Gene
  - I2632 NPD Type A/B (SMPD1) Ashkenazi Jewish mutations
  - I246 NPD Type C1 (NPC1)
  - I247 NPD Type C2 (NPC2/HE1)
- I273 Phenylalanine hydroxylase (PAH)
- I269 SCAD deficiency (ACADS)
- Smith-Lemli-Opitz Syndrome (DHCR7)
  - I2501 Sequencing
  - I2502 Prenatal DHCR7 test based on ultrasound abnormalities
- I270 VLCAD deficiency (ACADVL)

**Specimen Requirements** GeneDx prefers buccal specimens collected using a GeneDx supplied collection kit, for most tests. The exceptions to this are tests marked “†”. These tests require a 1-5ml whole blood specimen. Please note that ALL tests offered by GeneDx can be performed with a whole blood specimen.

**Test Code    Test Name**

**Noonan, Cardio-Facio-Cutaneous and Costello Syndrome**

- Cardio-Facio-Cutaneous (CFC) Syndrome, Tiered Panel (BRAF, MAP2K1/MAP2K2, KRAS)
  - I901 Tier 1: BRAF Exons 6, 11, 12, 14, 15 only
  - I904 Tier 2: MAP2K1/MAP2K2
  - I905 Tier 3: KRAS Gene
  - I902 Tier 4: Rest of BRAF Gene
- I903 Entire BRAF Gene NOW
- I91 Costello syndrome (HRAS)
- Noonan Syndrome Tiered Panel (Reflex Testing)
  - I921 Tier 1: PTPN11, exons 3, 8/9, 13 only
  - I922 Tier 2: Rest of PTPN11 plus RAF1 (3 exons)
  - I924 Tier 3: SOS1, exons 7, 11, 17 only
  - I925 Tier 4: KRAS
  - I926 Tier 5: Rest of SOS1
- I92 Entire PTPN11 Gene NOW
- I931 Tier 1: SOS1, exons 7, 11, 17
- I932 Tier 2: SOS1, Rest of SOS1
- I905 Entire KRAS Gene NOW
- I906 Tier 1: RAF1 gene (3 exons only)
- I907 Tier 2: RAF1 gene rest (if tier 1 negative)

**Other Hereditary Skin Disorders**

- Bannayan-Riley-Ruvalcaba Syndrome (PTEN)
  - I951 Sequencing
  - I952 CopyDx<sup>SM</sup>, if appropriate
- I97 Birt-Hogg-Dubé Syndrome (FLCN)
- I98 Carney Complex (PRKAR1A)
- Cowden Syndrome (PTEN)
  - 2001 Sequencing  2002 CopyDx<sup>SM</sup>, if appropriate
- 201 Darier Disease (ATP2A2)
- Familial Cutaneous Malignant Melanoma
  - 2021 CDKN2A/p16  2022 CDK4
- 205 Gorlin Syndrome (PTCH) †
- 206 Hailey-Hailey Disease (ATP2C1)
- Incontinentia Pigmenti (IKBKG/NEMO)
  - 2861 Tier 1: Common Deletion
  - 2862 Tier 2: Entire Gene Sequencing, if T1 negative
- Peutz-Jeghers Syndrome (STK11)
  - 2071 Sequencing  2072 CopyDx<sup>SM</sup>, if appropriate
- 264 Pseudoxanthoma Elasticum (PXE; ABCC6)
  - 2641 Tier 1: Common Mutation
  - 2642 Tier 2: Entire Gene Sequencing
- Hereditary Leiomyomatosis and Renal Cell Carcinoma (FH)
  - 2841 Tier 1  2842 Tier 2, if T1 negative
- I30 Syndromic PPK (incl. Vohwinkel syndrome) (GJB2, connexin 26)

**Other Keratin Disorders**

- 208 Epidermolytic PPK of Vörner (KRT9)
- Pachyonychia Congenita
  - 2091 KRT16, KRT6a (PC1)
  - 2092 KRT17, KRT6b (PC2)
- 211 Steatocystoma Multiplex (KRT17)
- 213 White Sponge Nevus (KRT4, KRT13)
- 266 Dowling-Degos Disease (KRT5)
- 267 Naegeli-Franceschetti-Jadassohn syndrome (NFJS; KRT14)
- Non-Epidermolytic PPK (Unna-Thost)
  - 2121 KRT1 Entire Gene  2122 KRT16 Hotspots
  - 2123 KRT16 Entire Gene
- 265 Transgradient Non-Epidermolytic PPK (Greither) (KRT5)

**Periodic Fever Syndromes**

- 214 Familial Mediterranean Fever (MEFV)
- 215 Familial Hibernian Fever/ TRAPS (TNFRSF1A)
- 216 Hyper- IgD Syndrome (MYK)
- 217 Muckle-Wells/Familial Cold Urticaria/NOMID (CIAS1)
- Pyogenic Sterile Arthritis, Pyoderma Gangrenosum, Acne (PAPA) (PSTPI1)
  - 2101 Tier 1 (Exons 10,11)
  - 2102 Tier 2 (rest), if Tier 1 negative

**Test Code    Test Name**

**Other Disorders**

- 218 Alexander Disease (GFAP)
- 219 Allgrove (Triple-A) Syndrome (AAAS)
- Alport Syndrome (COL4A5)
  - 2811 Sequencing  2812 CopyDx Analysis (females only)
- 283 Alzheimer Disease, Early-onset (APP, selected exons)
- Androgen Insensitivity Syndrome (AR) †
  - 2201 Sequencing
  - 2202 Prenatal AR test based on ultrasound abnormalities
- 225 Cartilage-Hair Hypoplasia and Assoc. Disorders (RMRP)
- CHARGE Syndrome (CHD7)
  - 2261 Sequencing  2262 CopyDx<sup>SM</sup>, if appropriate
  - 2263 Prenatal CHD7 test based on ultrasound abnormalities
- 282 Chondrodysplasia punctata, X-linked (ARSE)
- 227 Cohen Syndrome (COH1)
- 229 Dent Disease, X-linked Recessive Nephrolithiasis (CLCN5)
- Dopa-Responsive Dystonia (GCH1) †
  - 2301 Sequencing  2302 CopyDx<sup>SM</sup>, if appropriate
- Feingold Syndrome (MYCN)
  - 2601 Sequencing  2602 CopyDx<sup>SM</sup>, if appropriate
- Hereditary Angioedema (C1INH)
  - 2341 Sequencing  2342 CopyDx<sup>SM</sup>, if appropriate
- Hirschsprung Disease (RET)
  - 2351 Sequencing  2352 CopyDx<sup>SM</sup>, if appropriate
- Holoprosencephaly (SHH, ZIC2, SIX3, TGIF) †
  - 2371 Sequencing  2372 CopyDx<sup>SM</sup>, if appropriate
  - 2373 Prenatal test based on ultrasound abnormalities
- 238 Inclusion Body Myopathy (GNE; M712T only)
- 239 Insensitivity to Pain and Anhidrosis (NTRK1)
- Kallmann Syndrome:
  - 2401 KALI gene  2402 FGFR1 gene
- LEOPARD Syndrome (PTPN11, RAF1)
  - 2421 Tier 1: PTPN11, exons 7, 12, 13 only
  - 2422 Tier 2: Rest of PTPN11 plus RAF1 (3 exons), if Tier 1 neg
- 244 NemaLine Myopathy (ACTA1) †
- 245 NemaLine Myopathy, Auto Recessive (NEB; Askenazi Jewish Mutation)
- Rubinstein-Taybi Syndrome (CREBBP)
  - 2721 Tier 1  2722 Tier 2, if T1 negative
- Smith-Magenis Syndrome (RAI1)
  - 2511 Sequencing  2512 CopyDx<sup>SM</sup>, if appropriate
- 253 VanderWoude Syndrome (IRF6)
- X-linked Hydrocephalus, X-linked Spastic Paraplegia, MASA, CRASH Syndrome (L1CAM)
  - 2551 Sequencing  2552 CopyDx<sup>SM</sup>, (female only)
  - 2553 Prenatal L1CAM test based on ultrasound abnormalities
- 259 XY Female Gonadal Dysgenesis (SRY sequencing)

*Alternatively, the following special services are available for families with specific previously identified mutations. Check one per specimen.*

- 900 Confirmation of mutation identified elsewhere
- 901 Carrier Detection in Relatives
- 902 Prenatal Diagnosis

**Required Information:**

Gene Name \_\_\_\_\_

Mutation(s) \_\_\_\_\_

Relative's Name or GeneDx Accession #, if applicable: \_\_\_\_\_

# Payment Options

## I. Institutional Billing Information:

\_\_\_\_\_  
PO#/Department Code

\_\_\_\_\_  
Hospital/Lab Name

\_\_\_\_\_  
Contact Name

\_\_\_\_\_  
Address

\_\_\_\_\_  
City State Zip Code

\_\_\_\_\_  
Phone Fax

INSTITUTIONAL BILLING ADDRESS STAMP

## 2. Payment by credit card

*The full amount of the test fee is charged at the time of sample submission.*

\_\_\_\_\_  
Name as it appears on card

\_\_\_\_\_  
Billing address

\_\_\_\_\_  
City State Zip Code

\_\_\_\_\_  
Phone

Mastercard  Visa  Discover  American Express

\_\_\_\_\_  
Account Number

\_\_\_\_\_  
Expiration date 3/4 Digit Security Code

Please bill my credit card in the amount of \$\_\_\_\_\_ for diagnostic laboratory tests performed by GeneDx, Inc.

\_\_\_\_\_  
Signature (Required) Date

## 3. Payment by check or money order:

*Minimum of 75% of the cost of the test is required at the time of sample submission\*, with the remainder of the fee billed at the time of test completion.*

Check or money order enclosed in the amount of \$\_\_\_\_\_.

**\* For patients from outside the United States, 100% of the fee is due at the time of sample submission**

## 4. Insurance Billing:

*GeneDx cannot bill Medicare. GeneDx is not a participating member with any Medicaid/MediCal program.*

**GeneDx does not bill Insurance Companies directly unless all of the following is submitted:**

- Credit card information (complete part 2) to which any outstanding balance may be billed;
- An authorization number or letter of agreement from the insurance company.
  - The letter of agreement should be directed to GeneDx
  - detail the reimbursement rate
  - the name of the department or individual to whom the bill will be sent (including address, phone and fax numbers)
  - the patient's name and policy number.
- Copy of both sides of the insurance card.
- ICD9 codes (to be provided by physician) \_\_\_\_\_

I UNDERSTAND THAT I AM RESPONSIBLE IN ALL CASES FOR ALL FEES NOT COVERED BY INSURANCE.

\_\_\_\_\_  
Signature (Required)

## Note

IF YOU plan to apply on your own to your insurance carrier for reimbursement of your expenses for this test, the following information may be helpful in the case that GeneDx is requested by the carrier to prepare supporting documentation for you to use in your insurance claim:

\_\_\_\_\_  
Insurance Carrier  
Is this a Blue Cross/Blue Shield Plan?  YES  NO

\_\_\_\_\_  
Subscriber Name  
Is this a Medicaid plan?  YES  NO

\_\_\_\_\_  
Subscriber DOB