



**For autism, cardiology, array CGH, FISH, or mitochondrial testing  
please use specific submission forms available at [www.genedx.com](http://www.genedx.com)**

**Patient information**

First name \_\_\_\_\_ Last name \_\_\_\_\_  
 Gender:  Female  Male  Unknown  Fetus of \_\_\_\_\_  
 Date of birth (mm/dd/yy) \_\_\_\_\_  
 Mailing address \_\_\_\_\_  
 \_\_\_\_\_  
 City \_\_\_\_\_ State \_\_\_\_\_ Zip Code \_\_\_\_\_  
 Home phone \_\_\_\_\_ Work Phone \_\_\_\_\_

**Test requested**

Write gene/disease name below or check box on pages 2-6

\_\_\_\_\_

**Testing for known mutation (Go to page 2):**  
 Mutation: \_\_\_\_\_ GeneDx ID of relative \_\_\_\_\_

**If expedited testing is requested, please indicate reason:**  
 Pregnancy (gestational age \_\_\_\_\_ weeks)  Transplantation  
 Other \_\_\_\_\_

**Sample Information**

Medical record # \_\_\_\_\_ Specimen ID # \_\_\_\_\_  
 Date sample obtained (mm/dd/yy) \_\_\_\_\_

**Sample Type** (Note: only ONE specimen is required for multiple tests)  
 blood in EDTA (purple top - one tube of 1-5ml)  
 buccal brushes (must be GeneDx kits)  
 skin punch biopsy, size \_\_\_\_\_ mm  
 DNA \_\_\_\_\_ (source?) \_\_\_\_\_ (ug/ml)  
 fetal sample \_\_\_\_\_ (tissue source?)

**If other samples submitted**

Relationship to patient	Name	Sample type
_____	_____	_____
Relationship to patient	Name	Sample type
_____	_____	_____

**Reporting Address**

Physician \_\_\_\_\_ CGC \_\_\_\_\_  
 Address \_\_\_\_\_  
 \_\_\_\_\_  
 Phone \_\_\_\_\_ Fax (important) \_\_\_\_\_  
 Beeper \_\_\_\_\_ Email \_\_\_\_\_

**Second Report Address (if applicable)**

Physician \_\_\_\_\_ CGC \_\_\_\_\_  
 Address \_\_\_\_\_  
 Phone \_\_\_\_\_ Fax (important) \_\_\_\_\_  
 Beeper \_\_\_\_\_ Email \_\_\_\_\_

**Reason for testing - please complete (required):**

Diagnosis  Presymptomatic diagnosis  Carrier testing  
 Prenatal  Other \_\_\_\_\_  
 Positive control sample (no report issued) for patient/relative:  
 GeneDx ID \_\_\_\_\_ First name \_\_\_\_\_ Last name \_\_\_\_\_

**For metabolic disorders - please complete:**

Enzyme assay positive  Yes  No  Not done  
 Newborn screen positive  Yes  No

**Clinical diagnosis and family history**

Please provide relevant information below including the names or GeneDx ID# of any relatives previously tested.

Patient's karyotype if relevant \_\_\_\_\_

**Ordering Checklist**

Sample submission form (pages 1-6)  Completed payment form (page 7)  
 Informed consent (if appropriate)  Specimen tube labeled

**For office use only:**

# Testing Services for Rare Mendelian Disorders

**For autism, cardiology, array CGH, or FISH, or mitochondrial testing please use specific submission forms available at [www.genedx.com](http://www.genedx.com)**

Please check appropriate boxes below and fax only the sheets necessary

## Special services (complete box to the right)

- Mutation-specific testing
- 9011 One known familial mutation
  - 9012 Two known familial mutations
- Prenatal testing
- 902 Known familial mutation(s)
  - 9023 Maternal cell contamination studies only
- Mutation confirmations
- 9001 One known mutation identified in a research lab
  - 9002 Two known mutations identified in a research lab
- Custom deletion/duplication testing (CopyDx)
- 903 One gene or locus
- Deletion/duplication testing for a gene on the current menu
- 904 One gene or locus
- Follow-up testing for known familial deletion or duplication
- 905 One gene or locus
- DNA extraction only
- 910 One sample

## For special services please provide the information below

### Known mutation in relative (please send copy of report):

- Relative tested at GeneDx GeneDx ID/Name of relative \_\_\_\_\_
- Relative tested at another lab (**Positive control required**)
  - Positive control Included

### Required Information:

Gene or locus \_\_\_\_\_

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

Relationship to patient \_\_\_\_\_

**ExonArrayDx deletion/duplication testing is available for several genes not listed below. Please refer to and use the ExonArrayDx submission form downloadable at [www.genedx.com](http://www.genedx.com)**

**FISH testing is available for many of the disorders listed below. Please refer to and use the Molecular Cytogenetics submission form downloadable at [www.genedx.com](http://www.genedx.com)**

**Mitochondrial testing is available for many disorders. Please refer to and use the Mitochondrial submission form downloadable at [www.genedx.com](http://www.genedx.com)**

## TEST CODE TEST NAME

### Alagille Syndrome (JAG1)

- 1001 Tier 1 JAG1 sequencing and deletion/duplication testing
- 1002 Tier 2 JAG1 sequencing, if Tier 1 negative
- 1004 JAG1 full sequencing and deletion/duplication testing NOW

### Autism spectrum disorders

- Autism/macrocephaly syndrome (PTEN)
- 195 PTEN sequencing and deletion/duplication testing
- Rett syndrome / Atypical Rett syndrome (MECP2)
- 3041 MECP2 sequencing
  - 906 MECP2 deletion/duplication testing if sequencing is negative
- X-linked infantile spasm / Atypical Rett (CDKL5/STK9)
- 3051 CDKL5 sequencing
  - 906 CDKL5 deletion/duplication testing if sequencing is negative

### AutismDx panels – please use separate submission form

### Bone marrow failure syndromes

- 104 Congenital amegakaryocytic thrombocytopenia (MPL)
  - 105 Severe congenital neutropenia, autosomal dominant (ELANE aka ELA2)
  - 303 Severe congenital neutropenia, autosomal recessive (HAX1)
- Diamond-Blackfan anemia
- 361 RPL5 sequencing
  - 362 RPL11 sequencing
  - 1061 RPS19 sequencing
  - 906 RPS19 deletion/duplication testing if sequencing is negative
- Dyskeratosis congenita (specify concurrent or reflex ordering)
- 108 DKC1 gene, X-linked, sequencing
  - 414 TINF2 gene, exon 6 only, autosomal dominant, sequencing
  - 107 TERC gene, autosomal dominant, sequencing
  - 906 TERC gene, del/dup testing if negative sequencing
  - 906 DKC1 gene, del/dup testing if negative sequencing in females
- 109 Shwachman-Diamond Syndrome (SBDS)

### Congenital ichthyoses

- 114 Chanarin-Dorfman syndrome (ABHD5/CGI-58)
- Congenital recessive ichthyosis (erythrodermic)
- 1151 ALOX12B  1152 ALOXE3  1153 ICHTHYIN

## TEST CODE TEST NAME

- Epidermolytic Hyperkeratosis (KRT1, KRT10)
- 1181 KRT1, KRT10 mutation hotspots
  - 1182 KRT1 sequencing  1183 KRT10 sequencing
- 119 Erythrokeratoderma variabilis (GJB3, GJB4)
  - 120 Harlequin ichthyosis (ABCA12)
  - 122 Ichthyosis bullosa of Siemens (KRT2 mutation hotspots)
  - 123 Ichthyosis vulgaris (FLG common mutations)
  - 124 Keratitis-ichthyosis-deafness (KID) (GJB2; connexin26)
  - 125 Lamellar ichthyosis (TGM1)
  - 126 Lamellar ichthyosis type 2 (N. African) (ABCA12 hotspots)
  - 127 Netherton syndrome (SPINK5)
  - 128 Sjögren-Larsson syndrome (FALDH/ALDH3A2)

### Disorders involving bones and limbs

- Campomelic dysplasia
- 338 SOX9 sequencing
  - 906 SOX9 deletion/duplication testing if sequencing is negative
- 285 Cherubism (SH3BP2)
- Duane-Radial-Ray syndrome (DRRS; SALL4) †
- 262 SALL4 sequencing and deletion/duplication testing
  - 408 Prenatal SALL4 test based on fetal ultrasound abnormalities
- Hereditary Multiple Exostosis (EXT1/EXT2)
- 1811 EXT1 sequencing and EXT1/EXT2 deletion/duplication testing
  - 1812 EXT2 sequencing
  - 1813 EXT1+EXT2 sequencing and deletion/duplication testing NOW
- Holt-Oram syndrome (TBX5) †
- 2361 TBX5 sequencing
  - 906 TBX5 deletion/duplication testing if sequencing is negative
  - 2363 Prenatal TBX5 test based on ultrasound abnormalities
- 3272 Osteoporosis-pseudoglioma syndrome (LRP5)
  - 3272 Osteoporosis type 1, autosomal dominant (LRP5)
  - 248 Popliteal pterygium syndrome (IRF6, exon 4 only)
- Pseudoachondroplasia/multiple epiphyseal dysplasia (COMP) †
- 249 COMP sequencing
  - 906 COMP deletion/duplication testing if sequencing is negative

**Specimen Requirements** ALL tests offered by GeneDx can be performed with whole blood specimen.

As an alternative to blood, buccal specimen collection kits (supplied by GeneDx) can be used for many tests. Some exceptions are tests marked with “†” and any deletion/duplication, microarray, and non-conventional sequencing tests.

**Please check appropriate boxes below and fax only the sheets necessary**

**TEST CODE TEST NAME**

Townes-Brocks syndrome (SALL1) †

- 2521 SALL1 sequencing
- 906 SALL1 deletion/duplication testing if sequencing is negative
- 2523 Prenatal SALL1 test based on fetal ultrasound abnormalities

**Disorders of the immune system**

- 1541 Agammaglobulinemia, X-linked, BTK sequencing and deletion/duplication testing
- Autoimmune lymphoproliferative syndrome (ALPS)
  - 138 ALPS1A (TNFRSF6) sequencing
  - 2611 ALPS2A (CASPI0) sequencing  2612 ALPS2B (CASP8) sequencing
- Autoimmune polyendocrinopathy/APECED (AIRE)
  - 1391 Tier 1 AIRE sequencing
  - 1392 Tier 2 AIRE sequencing, if Tier 1 negative
  - 1393 AIRE full gene sequencing NOW
- Chronic granulomatous disease (CGD)
  - 1431 Tier 1: CYBB (X-linked) and NCF1 (autosomal recessive) sequencing
  - 1433 Tier 2: CYBA and NCF2 (both autosomal recessive), sequencing
  - 1434E CYBB (X-linked) del/dup testing if sequencing negative, females only
- 312 Hyper-IgE syndrome STAT3 sequencing (mutation hotspots)
- 318 Hyper-IgM syndrome, AICDA sequencing
- 301 IRAK4 deficiency, IRAK4 sequencing
- 146 Leukocyte adhesion deficiency, ITGB2 sequencing
- Severe combined immune deficiency (SCID)
  - 352 Adenosine deaminase deficiency, ADA sequencing
  - 145 JAK3 deficiency, JAK3 sequencing
  - 147 RAG1 and RAG2 deficiency (include Omenn Syndrome) sequencing
  - 302 IL7R deficiency, IL7R sequencing
- SCID with radiation sensitivity (ARTEMIS/DCLRE1C)
  - 1501 DCLRE1C full gene sequencing and deletion/duplication testing
  - 1502 DCLRE1C exon 8 only for Athabaskan Indians

**Ectodermal dysplasia syndromes**

- X-linked hypohidrotic ED (EDA aka ED1) †
  - 1601 EDA sequencing (males)
  - 1601E EDA sequencing and deletion/duplication testing (females)
- 373 Autosomal recessive/dominant ED/Onto-onycho-dermal dysplasia, Schöpf-Schulz-Passarge Syndrome (WNT10A)
- 156 Autosomal recessive/dominant hypohidrotic ED (EDAR)
- 157 Clouston syndrome, GJB6, connexin30 sequencing
- 306 Focal dermal hypoplasia/Goltz syndrome (PORCN)
- 158 Ectrodactyly-ED-clefting, TP63 (p63) sequencing
- 159 Hay-Wells syndrome, TP63 (p63) sequencing
- 407 Prenatal p63 test based on ultrasound abnormalities
- 2863 Hypohidrotic ED with immunodeficiency, IKBKKG/NEMO sequencing

**Epidermolysis bullosa**

- 162 Epidermolysis bullosa, dystrophic (COL7A1)
- Epidermolysis bullosa, simplex (KRT5, KRT14 mutation hotspots; PLEC1)
  - 168 KRT5/KRT14 mutation hotspots
  - 3481 PLEC1 tier 1  3482 PLEC1 tier 2
- Epidermolysis bullosa, junctional (Herlitz / non-Herlitz)
  - 1631 Tier 1 (hotspots LAMB3, LAMC2, LAMA3)
  - 1632 LAMB3 full sequencing
  - 1633 LAMC2 full sequencing
  - 1634 LAMA3 full sequencing
  - 1636 GABEB/non-Herlitz form (COL17A1)
- Epidermolysis bullosa with muscular dystrophy (PLEC1)
  - 3481 PLEC1 Tier 1 sequencing
  - 3482 PLEC1 Tier 2 sequencing
- Epidermolysis bullosa with pyloric atresia (JEB-PA)
  - 1641 ITGB4 sequencing  1642 ITGA6 sequencing
  - 3481 PLEC1 tier 1  3482 PLEC1 tier 2

**Eye Disorders**

- Aniridia, other developmental eye disorders (PAX6)
  - 131 PAX6 sequencing
  - 906 PAX6 deletion/duplication testing if sequencing is negative

**TEST CODE TEST NAME**

Anophthalmia, Microphthalmia (SOX2, OTX2, VSX2)

- 132 SOX2 sequencing
- 906 SOX2 deletion/duplication testing if sequencing is negative
- 343 OTX2 sequencing
- 906 OTX2 deletion/duplication testing if sequencing is negative
- 344 VSX2 sequencing

Axenfeld-Rieger syndrome † (PITX2, FOXC1)

- 1341 PITX2 sequencing
- 906 PITX2 deletion/duplication testing if sequencing is negative
- 1342 FOXC1 sequencing
- 906 FOXC1 deletion/duplication testing if sequencing is negative

403 BEST1 related disorders (BEST1, VMD2)

- Choroideremia (CHM)
  - 296 CHM sequencing
  - 906 CHM del/dup testing if sequencing is negative

Cone-rod dystrophy (ABCA4, CRX, PRPH2(RDS))

- 387 ABCA4 sequencing
- 353 CRX sequencing
- 299 PRPH2 (RDS) sequencing

Familial exudative vitreoretinopathy (FZD4, LRP5)

- 3271 FZD4 sequencing
- 3272 LRP5 sequencing
- 906 LRP5 deletion/duplication testing if sequencing negative
- 3273 NDP sequencing in males
- 3274 NDP sequencing and deletion/duplication testing in females

Glaucoma (CYP1B1, MYOC, OPTN)

- Primary congenital glaucoma
  - 330 CYP1B1 sequencing
- Primary open-angle glaucoma / juvenile open-angle glaucoma
  - 329 MYOC sequencing
- Primary open-angle glaucoma / Normal tension glaucoma
  - 346 OPTN sequencing

Leber congenital amaurosis, autosomal recessive. Tiered panel (reflex testing)

- 2980 Tier 1: Common mutations (CEP290, GUCY2D, AIPL1, CRB1, RPE65)
- 2981 Tier 2: CRB1 exons 1-6, 8, 10-12 only
- 2982 Tier 3: RPE65 exons 2-3, 6-7, 11-14 only
- 2983 Tier 4: GUCY2D exons 3-11, 14, 16-19 only
- 2984 Tier 5: AIPL1 exons 1, 3, 5

Leber congenital amaurosis, autosomal dominant. Tiered panel (reflex testing)

- 412 Tier 1: IMPDH1 full gene sequencing
- 353 Tier 2: CRX full gene sequencing

Leber congenital amaurosis, comprehensive panel (CEP290, GUCY2D, CRB1, RPE65, AIPL1, IMPDH1, CRX)

- 376 CEP290 gene: IVS26+1655A>G mutation only
- 377 Entire GUCY2D gene NOW
- 378 Entire CRB1 gene NOW
- 345 Entire RPE65 gene NOW
- 379 Entire AIPL1 gene NOW
- 412 Entire IMPDH1 gene NOW
- 353 Entire CRX gene NOW

Lenz microphthalmia syndrome (BCOR)

- 370 BCOR LMS: P85L mutation only

Norrie disease (NDP)

- 3273 NDP sequencing in males
- 3274 NDP sequencing and deletion/duplication testing in females

Oculofaciocardiodental syndrome (BCOR; females only)

- 3691 BCOR Tier 1: mutation hotspots and deletion/duplication testing
- 3692 BCOR Tier 2: Rest of gene sequencing if Tier 1 is negative
- 3693 BCOR full gene sequencing and deletion/duplication testing NOW

Retinitis pigmentosa, autosomal dominant, tiered panel (reflex testing)

- 2971 Tier 1: Common mutations (IMPDH1, RPI, PRPF8, PRPH2 (RDS) full, RHO full)

- 2975 Tier 2: PRPF31 gene sequencing and deletion/duplication testing
- 2974 Tier 3: IMPDH1 rest of gene sequencing

Retinitis pigmentosa, autosomal dominant, additional genes

- 2973 Retinitis pigmentosa, autosomal dominant, PRPF3 gene sequencing
- 353 Retinitis pigmentosa, autosomal dominant CRX sequencing
- 403 Retinitis pigmentosa, autosomal dominant BEST1 sequencing

**Specimen Requirements** ALL tests offered by GeneDx can be performed with whole blood specimen.

As an alternative to blood, buccal specimen collection kits (supplied by GeneDx) can be used for many tests. Some exceptions are tests marked with “†” and any deletion/duplication, microarray, and non-conventional sequencing tests.

**Please check appropriate boxes and fax only the sheets necessary**

**TEST CODE TEST NAME**

- Retinitis pigmentosa, autosomal dominant, individual genes
- 412 Retinitis pigmentosa, autosomal dominant IMPDH1 sequencing
  - 295 Retinitis pigmentosa, autosomal dominant RPI sequencing
  - 298 Retinitis pigmentosa, autosomal dominant RHO sequencing
  - 299 Retinitis pigmentosa, autosomal dom. PRPH2 (RDS) sequencing
  - 300 Retinitis pigmentosa, autosomal dominant PRPF8 sequencing
- Retinitis pigmentosa panel (7 genes), autosomal recessive/sporadic RP
- 368 USH2A, EYS, ABCA4, PDE6A, PDE6B, RPE65, CRB1 sequencing
  - 908 Autosomal recessive RP panel - deletion/duplication testing
  - 417 CNGA1 sequencing
- Retinitis pigmentosa, X-linked
- 326 RP2 sequencing
  - 906 RP2 deletion/duplication testing if sequencing negative, females
- Stargardt disease (ABCA4)
- 387 ABCA4 sequencing
- X-linked juvenile retinoschisis
- 2571 RSI sequencing
  - 906 RSI del/dup testing if sequencing is negative, females only

**Familial hyperparathyroid syndromes/Endocrine neoplasias**

- 169 Autosomal dominant hypocalcemia (CASR)
  - 170 Familial hypocalciuric hypercalcemia (CASR)
  - 171 Familial isolated hypoparathyroidism (CASR)
- Hyperparathyroidism-jaw tumor syndrome or parathyroid carcinoma or familial isolated hyperparathyroidism (HRPT2)
- 1731 Tier 1 HRPT2 sequencing
  - 1732 Tier 2, if Tier 1 negative
  - 1733 HRPT2 full gene sequencing NOW
- Multiple Endocrine Neoplasia Type I (MEN1, Menin)
- 176 MEN1 sequencing
  - 904 MEN1 deletion/duplication testing if sequencing is negative
- 177 Multiple endocrine neoplasia Type 2A or familial medullary thyroid carcinoma, RET sequencing
  - 178 Multiple endocrine neoplasia type 2B, RET sequencing
  - 180 Neonatal severe primary hyperparathyroidism, CASR sequencing

**Hereditary rickets**

- 184 Autosomal dominant hypophosphataemia (FGF23)
  - 185 Autosomal recessive vitamin D-dependent rickets (CYP27B1)
  - 314 Autosomal recessive hypophosphatemic rickets (DMP1)
- X-linked dominant hypophosphatemia (PHEX)
- 186I PHEX sequencing in males
  - 186IE PHEX sequencing and deletion/duplication testing in females

**Inborn errors of metabolism**

- 380 6-pyruvyl-tetrahydropterin synthase deficiency (PTS)
  - 354  $\beta$ -ketothiolase deficiency (ACAT1)
  - 294 Biotinidase deficiency (BTD)
  - 334 Carnitine palmitoyltransferase deficiency type II (CPT2)
  - 382 Classic Citrullinemia (ASS1)
  - 274 cobalamin C deficiency (MMACHC)
  - 381 Dihydropteridone reductase (DHPR) deficiency (QDPR)
  - 411 Ethylmalonic Encephalopathy (ETHE1)
- Fabry disease (GLA)
- 232I GLA sequencing
  - 906 GLA deletion/duplication testing if sequencing is negative, females
- Fumarate hydratase deficiency (FH) (see also hereditary leiomyomatosis)
- 2843 FH sequencing
- 349 Galactosemia / Galactosyltransferase deficiency (GALT)
  - 399 Glutaric aciduria type I (GCDH)
- Glutaric aciduria II / Multiple acyl-CoA dehydrogenase deficiency (MADD)
- 278 ETFA  279 ETFB  280 ETFDH sequencing
  - 293 ETFA / E TFB / ETFDH tiered testing
- 287 Glycogen storage disease II (Pompe disease) (GAA)
  - 230 GTP cyclohydrolase I deficiency (GCH1)<sup>†</sup> (see dopa-responsive dystonia)
- HMG CoA lyase deficiency (HMGCL)
- 321I HMGCL full gene sequencing
  - 3212 HMGCL sequence exon 2 only (Saudi/Spanish mutation)
  - 3213 Sequence rest of HMGCL gene, (if 3212 negative)

**TEST CODE TEST NAME**

- 320 Holocarboxylase synthetase deficiency (HLCS)
  - 331 Homocystinuria (CBS)
  - 351 Isobutyryl CoA dehydrogenase deficiency (ACAD8) Isovaleric acidemia (IVD)
    - 319I Full sequencing
    - 3192 Sequence exon 9 only (includes common A282V mutation)
    - 3193 Rest of IVD (if 3192 negative)
- LCHAD/trifunctional protein deficiency (HADHA/HADHA and HADHB)
- 271I HADHA Tier 1 (common mutation; c.1528G>C)
  - 2712 HADHA Full sequencing  272 HADHB Full sequencing
- Lowe syndrome (OCRL)
- 335 Lowe syndrome, OCRL full sequencing
  - 906 OCRL deletion/duplication testing if sequencing negative, females only
- 404 Malonyl-CoA Decarboxylase Deficiency (MLYCD)
- MCAD deficiency (ACADM)
- 2682 Full gene sequencing NOW
  - 268I Sequence exon 11 only (includes common K329E mutation)
  - 2683 Rest of ACADM
- 3-Methylcrotonyl CoA carboxylase deficiency
- 288I Tier 1: MCCC2  2882 Tier 2: MCCC1, if necessary
  - 2883 MCCC1, MCCC2 both NOW
- Methylmalonic acidemia (MUT, MMAA, MMAB)
- 275I Reflex testing: MUT, MMAA, MMAB, if necessary
  - 2752 MUT full sequencing  276 MMAA  277 MMAB
  - MUT, MMAA, MMAB all NOW: 2752, 276, 277
  - 2753 MUT sequence exon 2 only (Hispanic mutations)
  - 2754 MUT, rest of gene, after 2753, if necessary
- 243I Tier 1 Mucopolipidosis type IV (MCOLN1) (Ashkenazi mutations only)
  - 2432 Tier 2 Mucopolipidosis type IV (MCOLN1) (sequence analysis)
- Niemann Pick disease (NPD)
- 263I NPD type A/B, SMPD1 full gene sequencing
  - 2632 NPD type A/B (SMPD1) Ashkenazi Jewish mutations
  - 246 NPD type C1 (NPC1)  247 NPD type C2 (NPC2/HE1)
  - 355 NPD Type C1 reflex testing (NPC1, NPC2 if necessary)
- Ornithine transcarbamylase deficiency (OTC)
- 313 OTC sequencing (males)
  - 313E OTC sequencing and deletion/duplication testing (females)
- 273 Phenylalanine hydroxylase (PAH)
  - 287 Pompe disease/glycogen storage disease type II (GAA)
- Propionic acidemia
- 290I Tier 1: PCCB  2902 Tier 2: PCCA, if necessary
  - 2903 PCCA, PCCB both NOW
- 365 Primary/systemic carnitine deficiency (SLC22A5)
  - 269 SCAD deficiency (ACADS)
  - 383 Short/Branched Chain Acyl-CoA Dehydrogenase Deficiency (ACADSB)
- Smith-Lemli-Opitz syndrome (DHCR7)
- 2502 DHCR7 sequencing
  - 2503 Prenatal DHCR7 test based on ultrasound abnormalities
- Tyrosinemia type I (FAH)
- 366I FAH full sequencing  3662 Sequencing exon 12 only
  - 3663 FAH rest of the gene (if 3662 negative)
- 270 VLCAD deficiency (ACADVL)

**Mitochondrial disorders – please use separate submission form****Noonan, LEOPARD, cardiofaciocutaneous, and Costello syndromes and related disorders**

- 356 Comprehensive resequencing array for Noonan syndrome and related disorders [PTPNI1, SOS1, RAF1, KRAS, BRAF, MAP2K1/MAP2K2, HRAS, SHOC2 (S2G mutation)]
- Individual gene testing -
- 191 Entire HRAS gene
  - 192 Entire PTPNI1 gene
  - 193 Entire SOS1 gene
  - 1906 RAF1 select exons (7, 14, 17 only)
  - 1925 Entire KRAS gene
  - 1903 Entire BRAF gene
  - 1904 Entire MAP2K1/MAP2K2 genes
  - 389 SHOC2 (S2G mutation only)

**Specimen Requirements** ALL tests offered by GeneDx can be performed with whole blood specimen.

As an alternative to blood, buccal specimen collection kits (supplied by GeneDx) can be used for many tests. Some exceptions are tests marked with “†” and any deletion/duplication, microarray, and non-conventional sequencing tests.

Please check appropriate boxes and fax only the sheets necessary

**TEST CODE TEST NAME**

Noonan syndrome – prenatal testing based on ultrasound findings

- 357 Comprehensive Noonan syndrome prenatal panel  
Genes: PTPN11, SOS1, RAF1, KRAS, BRAF, MAP2K1/MAP2K2, HRAS, SHOC2 (S2G mutation)
- 194 Entire PTPN11 gene only

**Neurodevelopmental / mental retardation disorders**

Angelman/Angelman-Like Syndrome

- 374 UBE3A Sequencing
- 375 SLC9A6 Sequencing

Autism/macrocephaly syndrome (PTEN)

- 195 PTEN sequencing and deletion/duplication testing

Coffin-Lowry syndrome (RSK2)

- 1101RSK2 Tier 1 sequencing
- 1102 RSK2 Tier 2 sequencing, if Tier 1 negative
- 906 RSK2 del/dup testing if sequencing negative, females only
- 1104 Full RSK2 gene sequencing NOW

Rett syndrome / Atypical Rett syndrome (MECP2)

- 3041 MECP2 sequencing
- 906 MECP2 deletion/duplication testing if sequencing is negative

Rubinstein-Taybi syndrome (CREBBP) †

- 2921 CREBBP Tier 1 mutation hotspots and del/dup testing
- 2922 CREBBP Rest of gene sequencing if Tier 1 negative

Smith-Magenis syndrome (RAI1)

- 2511 Sequencing and intragenic deletion/duplication testing

X-linked infantile spasm / Atypical Rett (CDKL5/STK9)

- 3051 CDKL5 sequencing
- 906 CDKL5 deletion/duplication testing if sequencing is negative

**Other hereditary skin disorders**

197 Birt-Hogg-Dubé syndrome (FLCN)

Carney complex (PRKARIA)

- 198 PRKARIA sequencing
- 906 PRKARIA deletion/duplication testing if sequencing is negative

Cowden Syndrome (PTEN) † (see also BRRS)

- 195 PTEN sequencing and deletion/duplication testing

201 Darier Disease (ATP2A2)

Familial cutaneous malignant melanoma

- 2021 CDKN2A/p16 and CDK4 (exon 2)
- 2022 CDKN2A/p16 only

Gorlin Syndrome (PTCH) †

- 205 Sequencing and deletion/duplication testing
- 206 Hailey-Hailey disease (ATP2C1)

Hereditary leiomyomatosis and renal cell carcinoma (FH)

- 2841 FH Tier 1 sequencing  2842 FH Tier 2 sequencing
- 906 FH deletion/duplication testing if sequencing is negative

Incontinentia pigmenti (IKBKG/NEMO)

- 2861 Tier 1: Common deletion assay for females only
- 2862 Tier 2: IKBKG full gene sequencing if tier 1 negative

Peutz-Jeghers syndrome (STK11)

- 2071 Sequencing and deletion/duplication testing

Pseudoxanthoma elasticum (PXE;ABCC6)

- 2641 Tier 1: Common mutations
- 2642 Tier 2: Full gene sequencing if T1 negative

130 Syndromic PPK (incl. Vohwinkel syndr.) (GJB2, connexin 26)

**Other keratin disorders**

208 Epidermolytic PPK of Vörner (KRT9 hotspots)

Pachyonychia congenita

- 2091 KRT16, KRT6a hotspots (PC1)
- 2092 KRT17, KRT6b hotspots (PC2)
- 2111 Steatocystoma multiplex (KRT17 hotspots)
- 2131 White sponge nevus (KRT4, KRT13 hotspots)
- 266 Dowling-Degos disease (KRT5)
- 267 Naegeli-Franceschetti-Jadassohn syndrome (NFJS; KRT14)

Non-epidermolytic PPK (Unna-Thost)

- 2122 KRT1 full gene sequencing
- 2121 KRT16 mutation hotspots  2123 KRT16 full gene sequencing
- 265 Transgradient non-epidermolytic PPK (Greither) (KRT5)

**TEST CODE TEST NAME****Periodic fever syndromes**

- 367 Comprehensive resequencing array for Periodic Fever Syndromes: Familial Hibernian Fever/TRAPS; Familial Mediterranean Fever; Hyper-IgD Syndrome; Muckle Wells/Familial Cold Urticaria, NOMID; Cyclic neutropenia; PAPA Syndrome; Majeed syndrome (MEFV, TNFRSF1A, MVK, NLRP3 (CIAS1), ELANE (ELA2), PSTPIP1, and LPIN2)
- 400 Rest of resequencing array if 2 or more genes of the Periodic Fever Panel have been previously tested
- 214 Familial Mediterranean fever (MEFV) Exons 2,3 and 10 only
- 215 Familial Hibernian fever/ TRAPS (TNFRSF1A) Exons 2-5 only
- 216 Hyper-IgD Syndrome (MVK) Exons 8 and 10 only
- 217 Muckle-Wells/familial cold urticaria/NOMID (CIAS1) Exon 3 only
- Pyogenic sterile arthritis, pyoderma gangrenosum, acne (PAPA) (PSTPIP1)
  - 2101 Tier 1 (Exons 10,11)  2102 Tier 2 (rest), if Tier 1 negative

**Pheochromocytoma and related cancer syndromes**

von Hippel-Lindau syndrome (VHL)

- 332 VHL sequencing and deletion/duplication testing

Hereditary paraganglioma-pheochromocytoma syndrome

- 322 SDHB sequencing
- 906 SDHB/C/D deletion/duplication testing
- 323 SDHD sequencing
- 324 SDHC sequencing

**Sex differentiation disorders**

339 Adrenal hyperplasia, POR deficiency (POR)

Androgen Insensitivity Syndrome (AR) †

- 220 AR sequencing
- 2201 Prenatal AR test based on ultrasound abnormalities
- 340 Aromatase deficiency (CYP19A1)

Campomelic dysplasia (SOX9)

- 338 SOX9 sequencing
- 906 SOX9 deletion/duplication testing if sequencing is negative
- 3383 Prenatal SOX9 test based on ultrasound abnormalities
- 341 XY gonadal dysgenesis (NR5A1/SF-1 sequencing)
- 259 XY gonadal dysgenesis (SRY sequencing)
- 409 Prenatal SRY test based on ultrasound abnormalities
- 422 XY gonadal dysgenesis (DHH sequencing)
- 906 XY gonadal dysgenesis (NROB1/DAX1 testing for gene duplication)

**Other genetic disorders**

218 Alexander disease (GFAP)

219 Allgrove (Triple-A) syndrome (AAAS)

Alport syndrome (COL4A5)

- 2811 COL4A5 sequencing
- 906 COL4A5 del/dup testing if sequencing negative
- Bannayan-Riley-Ruvalcaba syndrome (PTEN) † (see also Cowden syn.)
  - 195 PTEN sequencing and deletion/duplication testing

372 Bloom Syndrome (BLM)

317 Branchiootoc syndrome 3 (SIX1)

Branchiootorenal syndrome 3 (EYA1)

315 EYA1 sequencing and deletion/duplication testing

225 Cartilage-hair hypoplasia and associated disorders (RMRP)

CHARGE syndrome (CHD7)

- 2261 CHD7 sequencing
- 906 CHD7 deletion/duplication testing if sequencing is negative
- 2262 Prenatal CHD7 test based on ultrasound abnormalities

Cerebral Cavemous Malformations (CCM) †

- 4181 KRIT1 Tier 1 sequencing (exons 13, 15, and 17)
- 4182 KRIT1 Tier 2 sequencing (rest of KRIT1) + deletion/duplication testing for KRIT1/CCM2/PDCD10
- 419 CCM2 sequencing
- 420 PDCD10 sequencing
- 906 KRIT1/CCM2/PDCD10 deletion/duplication testing ONLY

Chondrodysplasia punctata, X-linked (ARSE)

- 282 ARSE sequencing (males)
- 282E ARSE sequencing and deletion/duplication testing (females)

**Specimen Requirements** ALL tests offered by GeneDx can be performed with whole blood specimen.

As an alternative to blood, buccal specimen collection kits (supplied by GeneDx) can be used for many tests. Some exceptions are tests marked with “†” and any deletion/duplication, microarray, and non-conventional sequencing tests.

**Please check appropriate boxes and fax only the sheets necessary**

TEST CODE	TEST NAME
<input type="checkbox"/>	413 Chuvash Polycythemia (VHL)
<input type="checkbox"/>	227 Cohen syndrome (COH1) <input type="checkbox"/> 227I Finnish mutation only
	Craniofrontonasal dysplasia (EFNB1)
<input type="checkbox"/>	325I EFNB1 sequencing
<input type="checkbox"/>	906 EFNB1 del/dup testing if sequencing negative, females only
<input type="checkbox"/>	229 Dent disease, X-linked recessive nephrolithiasis (CLCN5)
<input type="checkbox"/>	906 CLCN5 del/dup testing if sequencing negative, females only
	Dopa-responsive dystonia (GCHI, TH) †
<input type="checkbox"/>	230 GCHI sequencing
<input type="checkbox"/>	906 GCHI deletion/duplication testing if sequencing is negative
<input type="checkbox"/>	359 Infantile Parkinsonism (TH deficiency) - TH sequencing
	Feingold syndrome (MYCN)
<input type="checkbox"/>	260 MYCN sequencing
<input type="checkbox"/>	906 MYCN deletion/duplication testing if sequencing is negative
	Hereditary angioedema
<input type="checkbox"/>	234I Type I/II C1NH sequencing and deletion/duplication testing
<input type="checkbox"/>	388 Type III F12 sequencing of exon 9 (Thr328 mutation)
	Hermansky-Pudlak syndrome (HPS1 and HPS3)
<input type="checkbox"/>	188 HPS1 and HPS3 Puerto Rican mutations
<input type="checkbox"/>	189 HPS3 Ashkenazi splice mutation
	Hirschsprung disease (RET)
<input type="checkbox"/>	235I RET sequencing of select exons
<input type="checkbox"/>	2352 RET sequencing of remaining exons if select exons negative
<input type="checkbox"/>	906 RET deletion/duplication testing if sequencing is negative
	Holoprosencephaly (SHH, ZIC2, SIX3, TGIF) †
<input type="checkbox"/>	237I Sequencing and deletion/duplication testing
<input type="checkbox"/>	2373 Prenatal test based on ultrasound abnormalities
<input type="checkbox"/>	238 Inclusion body myopathy (GNE; M712T only)
<input type="checkbox"/>	239 Insensitivity to pain and anhidrosis (NTRK1)
	Kallmann syndrome:
<input type="checkbox"/>	240I KAL1 gene sequencing
<input type="checkbox"/>	906 KAL1 del/dup testing if sequencing is negative, females only
<input type="checkbox"/>	2402 FGFR1 gene
	Nemaline myopathy, autosomal recessive
<input type="checkbox"/>	244 Nemaline myopathy (ACTA1) †
<input type="checkbox"/>	245 Nemaline myopathy (NEB; Askenazi Jewish mutation)
	Oral-facial-digital syndrome type I (OFD1, aka CXORF5)
<input type="checkbox"/>	364I Tier 1 OFD1 sequencing
<input type="checkbox"/>	3642 Tier 2 OFD1 sequencing
<input type="checkbox"/>	906 OFD1 deletion/duplication testing if sequencing is negative
<input type="checkbox"/>	40I Supravalvular aortic stenosis / autosomal dominant cutis laxa (ELN)
	Simpson-Golabi-Behmel Syndrome (SGBS)
<input type="checkbox"/>	415 GPC3 sequencing (males)
<input type="checkbox"/>	415E GPC3 sequencing and deletion/duplication testing (females)
	Sotos Syndrome
<input type="checkbox"/>	406 NSD1 sequencing and deletion/duplication testing
	Spinal muscular atrophy with respiratory distress, type I (IGHMBP2)
<input type="checkbox"/>	342 IGHMBP2 sequencing
<input type="checkbox"/>	363 Transthyretin amyloidosis/familial amyloid cardiomyopathy (TTR)
	Usher syndrome type 2
<input type="checkbox"/>	405I Tier 1: USH2A 2299delG mutation only
<input type="checkbox"/>	4052 Tier 2: USH2A sequencing and deletion/duplication testing
	Van der Woude syndrome (IRF6)
<input type="checkbox"/>	253 IRF6 sequencing
	Velocardiofacial syndrome / DiGeorge syndrome (TBX1)
<input type="checkbox"/>	358 TBX1 sequencing
	X-linked Adrenal Hypoplasia Congenita (AHC)
<input type="checkbox"/>	416 NROB1 sequencing
	X-linked hydrocephalus, X-linked spastic paraplegia, MASA, CRASH syndrome (LICAM)
<input type="checkbox"/>	255I Sequencing (male)
<input type="checkbox"/>	255IE Sequencing and deletion/duplication testing (female)
<input type="checkbox"/>	2553 Prenatal LICAM test based on ultrasound abnormalities (male)
<input type="checkbox"/>	2553E Prenatal LICAM test based on ultrasound abnormalities (female)

# 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: (Must also complete Section 2, credit card info)

*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 \_\_\_\_\_