



For autism, cardiology, array CGH, FISH, or mitochondrial testing please use specific submission forms available at www.genedx.com

Patient information

First name _____ Last name _____
 Gender: Female Male Unknown

 Date of birth (mm/dd/yy) _____
 Mailing address _____

 City _____ State _____ Zip Code _____
 Home phone _____ Work Phone _____

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?)

Reporting Address

Physician _____ CGC _____

 Address _____

 Phone _____ Fax (important) _____

 Beeper _____ Email _____

Second Report Address (if applicable)

Physician _____ CGC _____

 Address _____

 Phone _____ Fax (important) _____

 Beeper _____ Email _____

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 _____

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

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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

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

Mitochondrial testing is available for many disorders. Please refer to and use the Mitochondrial submission form downloadable at 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
- 107 Dyskeratosis congenita, autosomal hTR/TERC sequencing
- Dyskeratosis congenita, X-linked (DKC1)
- 108 DKC1 sequencing
 - 906 DKC1 del/dup testing if sequencing is negative, females only
- 109 Shwachman-Diamond Syndrome (SBDS)

Congenital ichthyoses

- 114 Chanarin-Dorfman syndrome (ABHD5/CGI-58)
- Congenital recessive ichthyosis (erythrodermic)
- 1151 ALOXI2B 1152 ALOXE3 1153 ICHTHYIN
- Epidermolytic Hyperkeratosis (KRT1, KRT10)
- 1181 KRT1, KRT10 mutation hotspots
 - 1182 KRT1 sequencing 1183 KRT10 sequencing

TEST CODE TEST NAME

- 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)

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) †
- 2621 SALL4 sequencing and deletion/duplication testing
- 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
- Townes-Brocks syndrome (SALL1) †
- 2521 SALL1 sequencing
 - 906 SALL1 deletion/duplication testing if sequencing is negative
 - 2523 Prenatal SALL1 test based on ultrasound abnormalities

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

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Please check appropriate boxes below and fax only the sheets necessary

TEST CODE TEST NAME**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 (CASPB) 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/DCLREIC)
 - 1501 DCLREIC full gene sequencing and deletion/duplication testing
 - 1502 DCLREIC 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/Odonto-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
- 2863 Hypohidrotic ED with immunodeficiency, IKBK/G/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
- 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

TEST CODE TEST NAME

- 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
- 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)
 - 2974 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
 - 2974 Entire IMPDH1 gene NOW
 - 353 Entire CRX gene NOW
- Lenz microphthalmia syndrome (BCOR)
 - 370 BCOR LMS: P85L mutation only
- 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
- Retinitis pigmentosa, autosomal dominant, individual genes
 - 294 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
- Retinitis pigmentosa, X-linked
 - 326 RP2 sequencing
 - 906 RP2 deletion/duplication testing if sequencing negative, females

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TEST CODE TEST NAME

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 hyperparathyroidism (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 1 (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)

 1861 PHEX sequencing in males 1861E PHEX sequencing and deletion/duplication testing in females**Inborn errors of metabolism** 380 6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency 354 β-ketothiolase deficiency (ACAT1) 294 Biotinidase deficiency (BTD) 334 Carnitine palmitoyltransferase deficiency type II (CPT2) 382 Classic Citrullinemia (ASS1) 274 cobalamin C deficiency (MMACHC)

Fabry disease (GLA)

 2321 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 / ETFB / ETFDH tiered testing 287 Glycogen storage disease II (Pompe disease) (GAA) 230 GTP cyclohydrolase I deficiency (GCH1)† (see dopa-responsive dystonia)

HMG CoA lyase deficiency (HMGCL)

 3211 HMGCL full gene sequencing 3212 HMGCL sequence exon 2 only (Saudi/Spanish mutation) 3213 Sequence rest of HMGCL gene, (if 3212 negative) 320 Holocarboxylase synthetase deficiency (HLCs) 331 Homocystinuria (CBS) 351 Isobutyryl CoA dehydrogenase deficiency (ACAD8)

Isovaleric acidemia (IVD)

 3191 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)

 2711 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)**TEST CODE TEST NAME**

MCAD deficiency (ACADM)

 2682 Full gene sequencing NOW 2681 Sequence exon 11 only (includes common K329E mutation) 2683 Rest of ACADM

3-Methylcrotonyl CoA carboxylase deficiency

 2881 Tier 1: MCCC2 2882 Tier 2: MCCC1, if necessary 2883 MCCC1, MCCC2 both NOW

Methylmalonic acidemia (MUT, MMAA, MMAB)

 2751 Reflex testing: MUT, MMAA, MMAB, if necessary 2752 MUT full sequencing 276 MMAA 277 MMAB 2755 MUT, MMAA, MMAB all NOW 2753 MUT sequence exon 2 only (Hispanic mutations) 2754 MUT, rest of gene, after 2753, if necessary 243 Mucopolidosis type IV (MCOLN1) (Ashkenazi mutations only)

Niemann Pick disease (NPD)

 2631 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

 2901 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)

 3661 FAH full sequencing 3662 Sequencing exon 12 only 3661 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)

Noonan syndrome – prenatal testing based on ultrasound findings

 357 Comprehensive Noonan syndrome prenatal panel

Genes: PTPNI1, SOS1, RAF1, KRAS, BRAF, MAP2K1/MAP2K2, HRAS, SHOC2 (S2G mutation)

 194 Entire PTPNI1 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

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TEST CODE TEST NAME

- Coffin-Lowry syndrome (RSK2)
 1101 RSK2 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 (IKBK/ NEMO)
 2861 Tier 1: Common deletion assay
 2862 Tier 2: IKBK 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
- 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)

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)
- 214 Familial Mediterranean fever (MEFV)
 215 Familial Hibernian fever/ TRAPS (TNFRSF1A)
 216 Hyper-IgD Syndrome (MVK)
 217 Muckle-Wells/familial cold urticaria/NOMID (CIAS1)

TEST CODE TEST NAME

- 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
- 341 XY sex reversal (NR5A1/SF-1)
 259 XY female gonadal dysgenesis, SRY sequencing

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, females only
- Bannayan-Riley-Ruvalcaba syndrome (PTEN) † (see also Cowden syn.)
 195 PTEN sequencing and deletion/duplication testing
- 372 Bloom Syndrome (BLM)
 317 Branchiootid 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
 2263 Prenatal CHD7 test based on ultrasound abnormalities
- Chondrodysplasia punctata, X-linked (ARSE)
 282 ARSE sequencing (males)
 282E ARSE sequencing and deletion/duplication testing (females)
- 227 Cohen syndrome (COH1) 2271 Finnish mutation only
- Craniofrontonasal dysplasia (EFNB1)
 3251 EFNB1 sequencing
 906 EFNB1 del/dup testing if sequencing negative, females only
- 229 Dent disease, X-linked recessive nephrolithiasis (CLCN5)
- Dopa-responsive dystonia (GCHI, TH) †
 230 GCHI sequencing
 906 GCHI deletion/duplication testing if sequencing is negative
 359 Infantile Parkinsonism (TH deficiency) - TH sequencing
- Feingold syndrome (MYCN)
 260 MYCN sequencing
 906 MYCN deletion/duplication testing if sequencing is negative
- Hereditary angioedema
 2341 Type I/II C1NH sequencing and deletion/duplication testing
 388 Type III F12 sequencing of exon 9 (Thr328 mutation)
- Hermansky-Pudlak syndrome (HPS1 and HPS3)
 188 HPS1 and HPS3 Puerto Rican mutations
 189 HPS3 Ashkenazi splice mutation
- Hirschsprung disease (RET)
 2351 RET sequencing of select exons
 2352 RET sequencing of remaining exons if select exons negative
 906 RET deletion/duplication testing if sequencing is negative
- Holoprosencephaly (SHH, ZIC2, SIX3, TGIF) †
 2371 Sequencing and deletion/duplication testing
 2373 Prenatal test based on ultrasound abnormalities

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TEST CODE TEST NAME

TEST CODE TEST NAME

- 238 Inclusion body myopathy (GNE; M712T only)
- 239 Insensitivity to pain and anhidrosis (NTRK1)
- Kallmann syndrome:
 - 2401 KAL1 gene sequencing
 - 906 KAL1 del/dup testing if sequencing is negative, females only
 - 2402 FGFR1 gene
- Nemaline myopathy, autosomal recessive
 - 244 Nemaline myopathy (ACTA1) †
 - 245 Nemaline myopathy (NEB; Askenazi Jewish mutation)
- Oral-facial-digital syndrome type I (OFD1, aka CXORF5)
 - 3641 Tier 1 OFD1 sequencing
 - 3642 Tier 2 OFD1 sequencing
 - 906 OFD1 deletion/duplication testing if sequencing is negative
- 401 Supravalvular aortic stenosis / autosomal dominant cutis laxa (ELN)
- Spinal muscular atrophy with respiratory distress, type I (IGHMBP2)
 - 342 IGHMBP2 sequencing
- 363 Transthyretin amyloidosis/familial amyloid cardiomyopathy (TTR)
- Usher syndrome type 2
 - 4051 Tier 1: USH2A 2299delG mutation only
 - 4061 Tier 2: USH2A sequencing and deletion/duplication testing
- Van der Woude syndrome (IRF6)
 - 253 IRF6 sequencing
- Velocardiofacial syndrome / DiGeorge syndrome (TBX1)
 - 358 TBX1 sequencing
- X-linked hydrocephalus, X-linked spastic paraplegia, MASA, CRASH syndrome (LICAM)
 - 2551 Sequencing (male)
 - 2551E Sequencing and deletion/duplication testing (female)
 - 2553 Prenatal LICAM test based on ultrasound abnormalities

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 _____