

Testing Services for Rare Mendelian Disorders

Please check appropriate boxes and fax only the sheets necessary

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
- 100F JAG1 20p12.2 deletion by FISH

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 Congenital or cyclic neutropenia, ELA2 sequencing
- 303 Congenital neutropenia, autosomal recessive (HAX1)
- Diamond-Blackfan anemia
 - 361 RPL5 sequencing
 - 362 RPL11 sequencing
 - 1061 RPL19 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)

Coffin-Lowry syndrome (RSK2)

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

Congenital ichthyoses

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

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

TEST CODE TEST NAME

- Holt-Oram syndrome (TBX5) †
 - 2361 TBX5 sequencing
 - 906 TBX5 deletion/duplication testing if sequencing is negative
 - 2363 Prenatal TBX5 test based on ultrasound abnormalities
- 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

Disorders of the immune system

- Agammaglobulinemia, X-linked (BTK)
 - 1541 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), sequencing
 - NCF1 (autosomal recessive), sequencing
 - 1433 Tier 2
 - CYBA (autosomal recessive), sequencing
 - NCF2 (autosomal recessive), sequencing
 - 1434E CYBB (X-linked) del/dup testing if sequencing negative, females only
- 312 Hyper-IgE syndrome STAT3 sequencing (mutation hotspots)
- Hyper-IgM syndrome
 - 318 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 Athabascan Indians

Ectodermal dysplasia syndromes

- X-linked hypohidrotic ED (EDA aka ED1) †
 - 1601 EDA sequencing (males)
 - 1601E EDA sequencing and deletion/duplication testing (females)
- 156 Autosomal recessive/dominant hypohidrotic ED (EDAR)
- 157 Clouston syndrome (GJB6, connexin30), sequencing
- 158 Ectrodactyly-ED-clefting (TP63, p63), sequencing
- 159 Hay-Wells syndrome (TP63, p63), sequencing
- 2863 Hypohidrotic ED with immunodeficiency (IKBK/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 3481 PLEC1 tier 2
- Epidermolysis bullosa, junctional (Herlitz / non-Herlitz)
 - 1631 Tier 1 (hotspots LAMB3, LAMC2, LAMA3)
 - 1632 LAMB3 full
 - 1633 LAMC2 full
 - 1634 LAMA3 full sequencing
 - 1636 GABEB/non-Herlitz form (COL17A1)

Specimen Requirements GeneDx prefers buccal specimens collected using a GeneDx supplied collection kit, for most tests. The exceptions to this are tests marked “†” and any CopyDx test. 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.

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TEST CODE TEST NAME

- Epidermolysis bullosa with muscular dystrophy (PLEC1)
 - 3481 PLEC1 Tier 1 sequencing
 - 3482 PLEC1 Tier 2
- Epidermolysis bullosa with pyloric atresia (JEB-PA)
 - 1641 ITGB4 sequencing 1642 ITGA6 sequencing
 - 3481 PLEC1 tier 1 3481 PLEC1 tier 2

Eye Disorders

- Aniridia, other developmental eye disorders (PAX6)
 - 131 PAX6 sequencing
 - 906 PAX6 deletion/duplication testing if sequencing is negative
- Aphthalmia, 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
- Axenhof-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
- Choroideremia (CHM)
 - 296 CHM sequencing
 - 906 CHM del/dup testing if sequencing is negative, females only
- Cone-rod dystrophy (ABCA4, CRX, PRPH2(RDS))
 - 3281 ABCA4 tier 1 3282 ABCA4 tier 2
 - 3283 Entire ABCA4 gene NOW
 - 353 CRX sequencing
 - 299 PRPH2 (RDS) sequencing
- Familial exudative vitreoretinopathy (FZD4, LRP5)
 - 3271 FZD4 sequencing
 - 3272 LRP5 sequencing
- 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 dominant
 - 353 CRX sequencing
- Leber congenital amaurosis, autosomal recessive
 - 345 RPE65 sequencing
- Retinitis pigmentosa, autosomal dominant, tiered panel (reflex testing)
 - 2971 Tier 1: Common mutations (IMPDH1, RPI1; PRPF8, PRPH2 (RDS) full, RHO full)
 - 2972 Tier 2: PRPF31 gene sequencing
 - 2973 Tier 3: PRPF3 gene sequencing
 - 2974 Tier 4: IMPDH1 full gene sequencing
 - 908 Retinitis pigmentosa panel - deletion/duplication testing
 - 353 Retinitis pigmentosa, autosomal dominant CRX sequencing
 - 295 Retinitis pigmentosa, autosomal dominant RPI1 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, autosomal recessive (ABCA4, RPE65)
 - 3281 ABCA4 tier 1 3282 ABCA4 tier 2
 - 3283 Entire ABCA4 gene NOW
 - 345 RPE65 sequencing 906 RPE65 deletion/duplication testing
- Retinitis pigmentosa, X-linked
 - 326 RP2 sequencing
 - 906 RP2 deletion/duplication testing if sequencing negative, females
- Stargardt disease (ABCA4)
 - 3281 ABCA4 tier 1 sequencing
 - 3282 ABCA4 tier 2 sequencing
 - 3283 ABCA4 full gene sequencing NOW

TEST CODE TEST NAME

- X-linked juvenile retinoschisis
 - 2571 RSI sequencing
 - 906 RSI deletion/duplication 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
 - 906 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 (DMPL1)
- X-linked dominant hypophosphatemia (PHEX)
 - 1861 PHEX sequencing in males
 - 1861E PHEX sequencing and deletion/duplication testing in females

Hermansky-Pudlak syndrome (HPS1 and/or HPS3)

- 188 HPS1 and HPS3 Puerto Rican mutations
- 189 HPS3 Ashkenazi splice mutation

Inborn errors of metabolism

- 354 β-ketothiolase deficiency (ACAT1)
- 294 Biotinidase deficiency (BTD)
- 334 Carnitine palmitoyltransferase deficiency type II (CPT2)
- 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)
- 295 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 (GCHI)† (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)
- 359 Infantile Parkinsonism, autosomal rec. (tyrosine hydroxylase deficiency)
- 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 and HADHB)
 - 2711 HADHA Tier 1 (common mutation; c.1528G>C)
 - 2712 HADHA Full sequencing
 - 272 HADHB Full sequencing

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- 335 Lowe syndrome full sequencing (OCRL)
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
 - 2883 MCCC1, MCCC2 both NOW
- Methylmalonic acidemia (MUT, MMAA, MMAB)
 - 2751 Reflex testing: MUT, MMAA, MMAB, if necessary
 - 2752 MUT Full gene 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 Mucopolipidosis 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/HEI)
 - 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
 - 2903 PCCA, PCCB both NOW
- 269 SCAD deficiency (ACADS)
- Smith-Lemli-Opitz syndrome (DHCR7)
 - 2502 DHCR7 sequencing
 - 2503 Prenatal DHCR7 test based on ultrasound abnormalities
- 270 VLCAD deficiency (ACADVL)

Noonan, cardiofaciocutaneous, and Costello syndromes

- Cardiofaciocutaneous (CFC) syndrome, tiered panel (reflex testing) (BRAF, MAP2K1/MAP2K2, KRAS)
 - 1901 Tier 1: BRAF exons 6, 11, 12, 14, 15 only
 - 1904 Tier 2: MAP2K1/MAP2K2 genes
 - 1905 Tier 3: KRAS gene
 - 1902 Tier 4: Rest of BRAF gene
- Cardiofaciocutaneous (CFC) syndrome, comprehensive panel
 - 1903 Entire BRAF gene NOW
 - 1902 Entire MAP2K1/MAP2K2 genes NOW
 - 1905 Entire KRAS gene NOW
- 191 Costello syndrome (HRAS)
- Noonan syndrome tiered panel (reflex testing)
 - 1921 Tier 1: PTPN11, exons 3, 8/9, 13 only
 - 1922 Tier 2: Rest of PTPN11 plus RAF1 (3 exons)
 - 1924 Tier 3: SOS1, exons 7, 11, 17 only
 - 1925 Tier 4: KRAS gene
 - 1926 Tier 5: Rest of SOS1 gene
- Noonan syndrome comprehensive panel
 - 192 Entire PTPN11 gene NOW
 - 193 Entire SOS1 gene NOW
 - 1925 Entire KRAS gene NOW
 - 1906 RAF1, exons 7, 14, 17 only NOW
- Noonan syndromes, individual tests
 - 1921 PTPN11, exons 3, 8/9, 13 only
 - 1923 Rest of PTPN11 gene
 - 1924 SOS1, exons 7, 11, 17 only
 - 1926 Rest of SOS1 gene
 - 1906 RAF1, Exons 7, 14, 17 only
 - 1907 Rest of RAF1 gene
 - 1925 Entire KRAS gene

TEST CODE TEST NAME

- Noonan syndrome—prenatal testing based on ultrasound findings
 - 1908 Comprehensive prenatal Noonan syndrome panel (PTPN11; RAF1 exons 7, 14, 17; SOS1 exons 7, 11, 17; KRAS)
 - 192 Entire PTPN11 gene NOW
- LEOPARD Syndrome (PTPN11, RAF1)
 - 2421 Tier 1: PTPN11, exons 7, 12, 13 only
 - 2422 Tier 2: Rest of PTPN11 plus RAF1 (3 exons)

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 906 FH Tier 2 sequencing
 - 906 FH deletion/duplication testing if sequencing is negative
- Incontinentia pigmenti (IKBKG/NEMO)
 - 2861 Tier 1: Common deletion assay
 - 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
- 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

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

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Sex determination disorders

- 339 Adrenal hyperplasia, POR deficiency (POR)
- Androgen Insensitivity Syndrome (AR) †
 - 220 AR sequencing
 - 220F AR deletion in Xq12 by FISH
 - 220I 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)
- XY female gonadal dysgenesis (SRY)
 - 259 SRY sequencing
 - 259F FISH for SRY deletion

Other disorders

- 218 Alexander disease (GFAP)
- 219 Allgrove (Triple-A) syndrome (AAAS)
- Alport syndrome (COL4A5)
 - 281I COL4A5 sequencing
 - 906 COL4A5 del/dup testing if sequencing negative, females only
 - 317 Branchiootic syndrome 3 (SIX1)
- Branchiootorenal syndrome 3 (EYA1)
 - 315 EYA1 sequencing and deletion/duplication testing
- 225 Cartilage-hair hypoplasia and associated disorders (RMRP)
- Bannayan-Riley-Ruvalcaba syndrome (PTEN) † (see also Cowden syn.)
 - 195 PTEN sequencing and deletion/duplication testing
- CHARGE syndrome (CHD7)
 - 226I 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) 227I Finnish mutation only
- Craniofrontonasal dysplasia (EFNB1)
 - 325I 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 TH sequencing
- Feingold syndrome (MYCN)
 - 260 MYCN sequencing
 - 906 MYCN deletion/duplication testing if sequencing is negative
- Hereditary angioedema (C1NH)
 - 234I C1NH sequencing and deletion/duplication testing
- Hirschsprung disease (RET)
 - 235I RET sequencing
 - 906 RET deletion/duplication testing if sequencing is negative
- Holoprosencephaly (SHH, ZIC2, SIX3, TGIF) †
 - 237I Sequencing and deletion/duplication testing
 - 2373 Prenatal test based on ultrasound abnormalities
- 238 Inclusion body myopathy (GNE; M712T only)
- 239 Insensitivity to pain and anhidrosis (NTRK1)
- Kallmann syndrome:
 - 240I KALI gene sequencing
 - 906 KALI del/dup testing if sequencing is negative, females only
 - 240F KALI deletion in Xp22.31 by FISH
 - 2402 FGFR1 gene
- Nemaline myopathy, autosomal recessive
 - 244 Nemaline myopathy (ACTA1) †
 - 245 Nemaline myopathy (NEB; Askenazi Jewish mutation)

TEST CODE TEST NAME

- Rett syndrome / atypical Rett syndrome (MECP2)
 - 304I MECP2 sequencing
 - 906 MECP2 deletion/duplication testing if sequencing is negative
 - 304F MECP2 Xq28 deletion by FISH
- Rubinstein-Taybi syndrome (CREBBP) †
 - 292I CREBBP Tier 1 mutation hotspots and deletion/duplication testing
 - 2922 CREBBP Rest of gene sequencing if Tier 1 negative
 - 292F Rubenstein-Taybi 16p13.3 deletion by FISH
- Spinal muscular atrophy with respiratory distress, type 1 (IGHMBP2)
 - 342 IGHMBP2 sequencing
- Smith-Magenis syndrome (RAI1)
 - 251I FISH for common SMS deletion
 - 251I Sequencing and intragenic deletion/duplication testing
- 363 Transthyretin amyloidosis/familial amyloid cardiomyopathy (TTR)
- Van der Woude syndrome (IRF6)
 - 253 IRF6 sequencing
- X-linked hydrocephalus, X-linked spastic paraplegia, MASA, CRASH syndrome (LICAM)
 - 255I Sequencing (male)
 - 255IE Sequencing and deletion/duplication testing (female)
 - 2553 Prenatal LICAM test based on ultrasound abnormalities

ExonArrayDx deletion/duplication testing for genes not found in the list above - please use separate ExonArrayDx submission form downloadable at www.genedx.com

Special services (complete box below)

- Mutation-specific testing
 - 901I One known familial mutation
 - 9012 Two known familial mutations
- Prenatal testing
 - 902 Known familial mutation(s)
 - 9023 Maternal cell contamination studies only
- Mutation confirmations
 - 900I 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 (CopyDx) 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

Required Information:

Gene or locus _____

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 _____