GeneDx | Rare Mendelian Disorders Test Requisition Form

an OPKO Health Company

For oncology, neurology, mitochondrial & metabolic disorders, cardiology, array CGH, FISH, whole exome sequencing or prenatal testing please use specific submission forms available at www.genedx.com/forms

Patient Information

First name Last name
Gender ❒ Male ❒ Female Date of birth (mm/dd/yy)_____
Ancestry ❒ Caucasian ❒ Eastern European ❒ Northern European
❒ Western European ❒ Native American ❒ Middle Eastern
❒ African American ❒ Asian ❒ Pacific Islander
❒ Caribbean ❒ Central/South American
❒ Ashkenazi Jewish ❒ Hispanic ❒ Other: _____

Mailing address

City State Zip code

Home phone Work phone

Email Patient's primary language if not English

Ordering Account Information

Acct # Account Name
Reporting Preference*. ❒ Care Evolve ❒ Fax ❒ Email
**If unmarked, we will use the account's default preferences or fax to new clients.*

Physician NPI #

Genetic Counselor

Street address 1

Street address 2

City State Zip code

Phone Fax (important)

Email Beeper

Send Additional Report Copies To:

Physician or GC/Acct # Fax#/Email/CE #

Physician or GC/Acct # Fax#/Email/CE #

Statement of Medical Necessity

This test is medically necessary for the diagnosis or detection of a disease, illness, impairment, symptom, syndrome or disorder. The results will determine my patient's medical management and treatment decisions. The person listed as the Ordering Provider is authorized by law to order the tests(s) requested herein. I confirm that I have provided genetic testing information to the patient and the patient has consented to genetic testing.

Signature of Physician or Other Authorized NPI Provider (required) Date

Patient Consent (sign here)

I have read the attached Informed Consent document and I give permission to GeneDx to perform genetic testing as described. I also give permission for my specimen and clinical information to be used in de-identified studies at GeneDx to improve genetic testing and for publication, if appropriate. My name or other personal identifying information will not be used in or linked to the results of any studies and publications. I also give GeneDx permission to inform me or my health care provider in the future about research opportunities, including treatments for the condition in my family. **More information is available on our website: www.genedx.com**

❒ Check this box if you are a New York state resident, and give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing.

Patient/Guardian Signature Date

Sample Information

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

Sample Type

❒ Blood in EDTA (purple top - one tube of 1-5ml)
❒ Buccal Swab
❒ Skin Punch Biopsy, size _____mm
❒ DNA _____ (source?) _____ (ug/ml)
❒ Oral Rinse (At least 30 mL of Scope oral rinse in a 50 mL centrifuge tube or GeneDx kit)

Clinical diagnosis and family history

ICD-10 Code(s): _____
Clinical Diagnosis: _____
Age at Initial Presentation: _____

Please provide relevant information below or attach detailed medical records.

Test requested

Test Code **Test Name**

_____ _____

_____ _____

Testing for known familial mutation(s)

❒ 9011 Testing for ONE known familial mutation
❒ 9012 Testing for TWO known familial mutations
❒ 905 Testing for ONE known familial exon-level del/dup
Gene(s): _____Mutation(s)_____
Proband Name: _____
Proband GeneDx Acc#: _____Relationship to proband: _____
❒ Positive control included - **Positive control is required if previous test was performed at another lab.**
❒ Family Member Test Report included - A clear copy of the test report on the mutation positive family member is recommended if previous test was performed at another lab.
❒ 906 One Gene ❒ 703 Custom Del/Dup Panel
Gene(s):_____

If expedited testing is requested, please indicate reason:

❒ Pregnancy (gestational age _____ weeks) ❒ Transplantation
❒ Other _____

Ordering Checklist:

❒ Sample submission form (pages 3-8) ❒ Completed payment form (page 2)
❒ Informed consent (if appropriate) ❒ Specimen tube, appropriately labeled with TWO identifiers

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

GeneDx | Payment Options

an OPKO Health Company

First Name _____ Last Name _____ Date of Birth (mm/dd/yy) _____

PATIENT STATUS – ONE MUST BE CHECKED: ❒ Hospital Inpatient ❒ Hospital Outpatient ❒ Not a Hospital Patient Hospital Patient Date of Discharge:_____

Payment Options

❒ Insurance Bill

Referral/Prior Authorization # _____
Please attach copy of Referral/authorization
GeneDx Benefit Investigation #_____

Insurance Carrier _____ Policy Name _____ ❒ Hold sample for Benefit Investigation (only if OOP cost is >$100)

Insurance ID # _____ Group # _____ Name of Insured _____ Date of Birth _____ Insurance Address _____ City _____ State _____ Zip _____

Relationship to Insured ❒ Child ❒ Spouse ❒ Self ❒ Other _____

Secondary Insurance Carrier Name _____ Insurance ID# _____ Group # _____ Name of Insured _____ Date of Birth _____

Relationship to Insured ❒ Child ❒ Spouse ❒ Self ❒ Other _____

Please include a copy of the front and back of the patient's insurance card (include secondary when applicable)

If you would like to expedite an assessment of your possible eligibility for GeneDx's financial assistance program (FAP), please provide the number of your household members _____ and the annual income of your household $_____. GeneDx may require additional information from you to complete an application for GeneDx's financial assistance program.

I represent that I am covered by insurance and authorize GeneDx, Inc. to give my designated insurance carrier, health plan, or third party administrator (collectively "Plan") the information on this form and other information provided by my health care provider necessary for reimbursement. I authorize Plan benefits to be payable to GeneDx. I understand that GeneDx will attempt to contact me if my out-of-pocket responsibility will be greater than $100 per test (for any reason, including co-insurance and deductible, or non-covered services). If GeneDx is unsuccessful in its attempts to contact me, I understand that it will be my responsibility to contact GeneDx to determine my out-of-pocket cost and to pay my out-of-pocket responsibility. I will cooperate fully with GeneDx by providing all necessary documents needed for Plan billing and appeals. I understand that I am responsible for sending GeneDx any and all of the money that I receive directly from my Plan in payment for this test. Reasonable collection and/or attorney's fees, including filing and service fees, shall be assessed if the account is sent to collection but said fees shall not exceed those permitted by state law. I permit a copy of this authorization to be used in place of the original.

Patient Signature (required)_____ Date _____

❒ Institutional Bill

GeneDx Account # _____

Hospital/Lab Name _____

Contact Name _____

Address _____

City _____ State _____ Zip Code _____

Phone _____ Fax _____

❒ Patient Bill

Amount _____

If I have insurance coverage for this testing, I am electing to be treated as a self-pay patient for this testing. As such, I agree that neither GeneDx nor I will submit a claim to my insurance for this testing.

Please bill my credit card (all major cards accepted)

❒ MasterCard ❒ Visa ❒ Discover ❒ American Express

Name as it appears on card _____

Account Number _____ Expiration date _____ CVC _____

Signature _____ Date _____

For GeneDx Use Only

First Name _____ Last Name _____ Date of Birth (mm/dd/yy) _____

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

Follow-up testing for known familial deletion or duplication
- ❑ 905 One gene or locus

DNA extraction only
- ❑ 909 One sample

ExonArrayDx: Exon-level gene-specific deletion/duplication testing *
- ❑ 906 One gene

Custom ExonArrayDx: Exon-level gene specific deletion/duplication testing (Gene(s) not on GeneDx test menu)*
- ❑ 703 One to twenty genes

* Fill in genes or gene panel to be tested: _____

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

TEST CODE TEST NAME

Alagille Syndrome (JAG1)
- ❑ 1001 Tier 1 JAG1 sequencing and deletion/duplication testing
- ❑ 1002 Tier 2 JAG1 sequencing, if Tier1 negative
- ❑ 1004 JAG1 full sequencing and deletion/duplication testing NOW

Bone marrow failure syndromes
- ❑ 104 Congenital amegakaryocytic thrombocytopenia (MPL)
- ❑ 505 X-linked Thrombocytopenia –or– X-linked Neutropenia (WAS)
- ❑ 105 Severe congenital neutropenia, autosomal dominant (ELANE aka ELA2)
- ❑ 303 Severe congenital neutropenia, autosomal recessive (HAX1)

Diamond-Blackfan anemia panel (specify concurrent or reflex ordering)
- ❑ J450 Diamond-Blackfan anemia panel (13 genes)
- ❑ 1061 RPS19 sequencing
- ❑ 361 RPL5 sequencing
- ❑ 362 RPL11 sequencing
- ❑ 906 RPS19 deletion/duplication testing

Dyskeratosis Congenita (specify concurrent or reflex testing)
- ❑ 108 DKC1 gene sequencing, X-linked
- ❑ 414 TINF2 gene exon 6 sequencing, autosomal dominant
- ❑ 107 TERC gene sequencing, autosomal dominant
- ❑ 682 TERT gene sequencing, autosomal dominant/recessive
- ❑ 906 TERC gene, deletion/duplication analysis
- ❑ 906 DKC1 gene, deletion/duplication if sequencing negative, females

- ❑ 109 Shwachman-Diamond Syndrome (SBDS)
- ❑ 938 Congenital Sideroblastic Anemia Panel (ABCB7, ALAS2, GLRX5, PUS1, SLC19A2, SLC25A38, TRNT1, YARS2, Mitochondrial genome large deletion testing)

Congenital ichthyoses
- ❑ 708 Congenital Ichthyosis XomeDxSlice. Test includes 39 genes known to cause syndromic or non-syndromic congenital ichthyosis.

Epidermolytic Ichthyosis (Epidermolytic Hyperkeratosis) (KRT1, KRT2, KRT10)
- ❑ 1181 KRT1, KRT10 hotspots
- ❑ 1182 KRT1 sequencing ❑ 1183 KRT10 sequencing
- ❑ 122 KRT2 hotspots

- ❑ 119 Erythrokeratodermia variabilis (GJB3, GJB4)
- ❑ 123 FLG (Filaggrin) Hot Spots
- ❑ 124 Keratitis-ichthyosis-deafness (KID) Syndrome (GJB2; connexin26)

Disorders involving bones and limbs
Panels for common skeletal dysplasias
- ❑ J799 Achondrogenesis Panel (COL2A1, SLC26A2, TRIP11)
- ❑ J804 Chondrodysplasia Punctata Panel (AGPS, ARSE, EBP, GNPAT, PEX7)
- ❑ 584 Cornelia de Lange Panel (HDAC8, NIPBL, RAD21, SMC1A, SMC3)
- ❑ J800 FGFR-Related Disorders Panel (FGFR2, FGFR3)
- ❑ J801 Limb Abnormalities Panel (14 genes)
- ❑ J797 Osteogenesis Imperfecta Panel (15 genes)
- ❑ J798 Short-Rib Thoracic Dysplasia (SRTD) Panel (14 Genes)

Campomelic dysplasia
- ❑ 338 SOX9 sequencing
- ❑ 906 SOX9 deletion/duplication testing if sequencing is negative

- ❑ 285 Cherubism (SH3BP2)

Duane-Radial-Ray syndrome (DRRS; SALL4) †
- ❑ 262E SALL4 sequencing and deletion/duplication testing

Grieg Cephalopolysyndactyly syndrome
- ❑ 472 GLI3 sequence (exons 2-15) and deletion/duplication analysis

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

HOXD13-Associated Limb Abnormalities
- ❑ 503 HOXD13 sequencing
- ❑ 906 HOXD13 deletion/duplication testing if sequencing is negative

- ❑ 3272 Osteoporosis-pseudoglioma syndrome (LRP5)
- ❑ 3272 Osteopetrosis type 1, autosomal dominant (LRP5)
- ❑ 248 Popliteal pterygium syndrome (IRF6, exon 4 only)

Pallister Hall Syndrome
- ❑ 4711 Tier 1 GLI3 sequence analysis of exons 13-15
- ❑ 4712 Tier 2 GLI3 sequence analysis of remaining exons (2-12) and del/dup analysis

Pseudoachondroplasia/multiple epiphyseal dysplasia (COMP) †
- ❑ 249 COMP sequencing
- ❑ 906 COMP deletion/duplication testing if sequencing is negative

Triphalangeal Thumb Polydactyly
- ❑ 502 ZRS sequence analysis (intron 5 of LMBR1 gene)
- ❑ 906 ZRS deletion/duplication analysis (intron 5 of LMBR1 gene) if sequencing is negative

Townes-Brocks syndrome (SALL1) †
- ❑ 2521 SALL1 sequencing
- ❑ 906 SALL1 deletion/duplication testing if sequencing is negative

Disorders of the immune system
- ❑ 154 Agammaglobulinemia, X-linked, BTK sequencing and deletion/duplication testing

Autoimmune lymphoproliferative syndrome (ALPS)
- ❑ 138 ALPS1A–FAS (TNFRSF6) sequencing
- ❑ 2611 ALPS2A (CASP10) 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) (specify concurrent or reflex ordering)
- ❑ 1434 CYBB sequencing (X-linked)
- ❑ 1435 NCF1 exon 2 only (recessive)
- ❑ 1436 CYBA sequencing (recessive)
- ❑ 1437 NCF2 sequencing (recessive)
- ❑ 906 CYBB (X-linked) deletion/duplication if sequencing negative, females

Specimen Requirements ALL tests offered by GeneDx can be performed with whole blood specimen.
As an alternative to blood, buccal specimen or mouthwash 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.

First Name Last Name Date of Birth (mm/dd/yy)

TEST CODE TEST NAME

Hyper-IgE syndrome (specify concurrent or reflex ordering)
- ❑ 678 Hyper-IgE Syndromes Panel (STAT3, DOCK8, TYK2 and SPINK5 gene sequencing + DOCK8 deletion/duplication analysis)
- ❑ 312 STAT3 sequence analysis, selected exons (dominant)
- ❑ 3122 STAT3 sequence analysis, remaining exons (dominant)
- ❑ 3123 STAT3 (Full gene sequencing, dominant)
- ❑ 736 DOCK8 sequencing and deletion/duplication testing (recessive)
- ❑ 679 DOCK8 (Full gene sequencing, recessive)
- ❑ 906 DOCK8 deletion/duplication testing (recessive)

Immunodeficiency Syndrome with Hyper-IgM
- ❑ 669 CD40LG sequencing; Type 1 (X-linked)
- ❑ 318 AICDA sequencing; Type 2
- ❑ 668 CD40 sequencing; Type 3
- ❑ 670 UNG sequencing; Type 5

❑ 301 IRAK4 deficiency, IRAK4 sequencing
❑ 146 Leukocyte adhesion deficiency, ITGB2 sequencing

Severe combined immune deficiency (SCID)
- ❑ 601 Comprehensive SCID Panel, 26 genes
  - ❑ 602 B+ SCID Sub-panel, 17 genes
  - ❑ 603 B- SCID Sub-panel, 9 genes

SCID with radiation sensitivity (ARTEMIS/DCLRE1C)
- ❑ 1501 DCLRE1C full gene sequencing and deletion/duplication testing
- ❑ 1502 DCLRE1C exon 8 only for Athabascan Indians

Severe combined immune deficiency (SCID)
- ❑ 492 X-linked SCID, IL2RG sequencing
- ❑ 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

Wiskott Aldrich Syndrome (X-linked)
- ❑ 505 WAS gene sequencing
- ❑ 906 WAS gene deletion/duplication testing for females

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)
❑ 617 Autosomal hypohidrotic/anhidrotic ED (EDARADD)
❑ 157 Clouston syndrome, GJB6, connexin30 sequencing
❑ 306 Focal dermal hypoplasia/Goltz syndrome (PORCN)
❑ 158 TP63 Select Exons Sequencing
❑ 1581 TP63 Remaining Exons Sequencing

Epidermolysis bullosa

❑ 707 XomeDxSlice – Epidermolysis Bullosa (EB) and other bullous skin disorders
Test includes ALL of the known genes for Dystrophic, Simplex, Junctional and Hemidesmosomal EB (COL7A1, COL17A1, KRT5/KRT14, LAMA3/LAMB3/LAMC2, PLEC1, ITGA6/ITGB4) and 17 additional genes (MMP1, DSP, CD151, FERMT1, NID1, GRIP1, TGM5, PKP1, DST, EXPH5, CHST8, CSTA, DSG1, DSG2, DSG3, DSG4, ITGA3)

❑ 162 Epidermolysis bullosa, dystrophic (COL7A1)

Epidermolysis bullosa, simplex (KRT5, KRT14 hotspots; PLEC1)
- ❑ 168 KRT5/KRT14 hotspots

Eye Disorders

Achromatopsia
- ❑ 513 CNGB3 sequencing
- ❑ 514 CNGA3 sequencing

Aniridia
- ❑ 491 PAX6 sequencing and deletion/duplication PAX6/DCDC1/ELP4/WT1

Anophthalmia, Microphthalmia
- ❑ 132 SOX2 sequencing
- ❑ 906 SOX2 deletion/duplication testing if sequencing is negative
- ❑ 343 OTX2 sequencing
- ❑ 906 OTX2 deletion/duplication testing if sequencing is negative
- ❑ 509 RAX sequencing
- ❑ 516 STRA6 sequencing
- ❑ 604 FOXE3 sequencing
- ❑ 344 VSX2 sequencing

TEST CODE TEST NAME

Anterior segment dysgenesis of the eye
- ❑ 491 PAX6 sequencing and deletion/duplication PAX6/DCDC1/ELP4/WT1
- ❑ 604 FOXE3 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 (VMD2)

Bothnia retinal dystrophy
- ❑ 4242 RLBP1 BRD: R234W mutation only

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

Cone and cone-rod dystrophies
- ❑ 379 AIPL1 sequencing
- ❑ 468 Cone rod dystrophy panel: ABCA4, PRPH2 (RDS)
- ❑ 506 CERKL sequencing
- ❑ 513 CNGB3 sequencing
- ❑ 514 CNGA3 sequencing
- ❑ 353 CRX sequencing
- ❑ 476 GUCA1A sequencing
- ❑ 467 GUCY2D exon 13 only

Congenital nystagmus, X-linked
- ❑ 432 FRMD7 sequencing

Congenital stationary night blindness, autosomal dominant
- ❑ 298 RHO sequencing
- ❑ 589 GNAT1 sequencing

Congenital stationary night blindness, autosomal recessive
- ❑ 489 TRPM1 sequencing
- ❑ 588 GRM6 sequencing
- ❑ 589 GNAT1 sequencing
- ❑ 517 Tier 1 SAG: c.926delA mutation only
- ❑ 518 Tier 2 SAG rest of gene sequencing
- ❑ 590 CABP4 sequencing
- ❑ 427 RDH5 sequencing

Congenital stationary night blindness, X-linked
- ❑ 431 NYX sequencing
- ❑ 587 CACNA1F sequencing

Enhanced S-Cone Syndrome
- ❑ 586 NR2E3 sequencing

Familial exudative vitreoretinopathy (FZD4, LRP5, NDP, TSPAN12)
- ❑ 3271 FZD4 sequencing
- ❑ 3272 LRP5 sequencing
- ❑ 906 LRP5 deletion/duplication testing if sequencing is negative
- ❑ 3273 NDP sequencing in males
- ❑ 3274 NDP sequencing and deletion/duplication testing in females
- ❑ 3275 TSPAN12 sequencing

Fundus albipunctatus
- ❑ 427 RDH5 sequencing
- ❑ 4241 RLBP1 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

❑ 649 Glycogen storage disease type V (GSD V) (PYGM)

Goldmann-Favre Syndrome
- ❑ 586 NR2E3 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
- ❑ 2985 Tier 6: RPGRIP1 (entire gene)

Leber congenital amaurosis, autosomal dominant. Tiered panel (reflex testing)
- ❑ 412 Tier 1: IMPDH1 full gene sequencing
- ❑ 353 Tier 2: CRX full gene sequencing

Specimen Requirements ALL tests offered by GeneDx can be performed with whole blood specimen. As an alternative to blood, buccal specimen or mouthwash 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.

First Name Last Name Date of Birth (mm/dd/yy)

Please check appropriate boxes and fax only the sheets necessary

TEST CODE TEST NAME

Leber congenital amaurosis, comprehensive panel (CEP290, GUCY2D, CRB1, RPE65, AIPL1, IMPDHI, CRX, RPGRIP1)
- ❑ 376 CEP290 gene: IVS26+1655A>G mutation only
- ❑ 377 Entire GUCY2D gene
- ❑ 378 Entire CRB1 gene
- ❑ 345 Entire RPE65 gene
- ❑ 379 Entire AIPL1 gene
- ❑ 412 Entire IMPDH1 gene
- ❑ 353 Entire CRX gene
- ❑ 2985 Entire RPGRIP1 gene

Lenz microphthalmia syndrome (BCOR)
- ❑ 370 BCOR LMS: P85L mutation only

Newfoundland rod-cone dystrophy
- ❑ 4243 RLBP1 NFRCD: IVS4+2 T>C and c.141G>A (K47K) mutations 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

Oguchi disease
- ❑ 517 Tier 1 SAG: c.926delA mutation only
- ❑ 518 Tier 2 SAG rest of gene sequencing

Retinitis pigmentosa, autosomal dominant, tiered panel (reflex testing)
- ❑ 2971 Tier 1: Common mutations (IMPDH1, RP1, 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
- ❑ 412 Retinitis pigmentosa, autosomal dominant IMPDH1 sequencing
- ❑ 295 Retinitis pigmentosa, autosomal dominant RP1 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/sporadic RP
- ❑ 368 Nine gene panel: ABCA4, CERKL, CNGA1, CRB1, EYS, PDE6A, PDE6B, RPE65, USH2A sequencing
- ❑ 908 Autosomal recessive RP panel - deletion/duplication testing
- ❑ 506 CERKL sequencing
- ❑ 417 CNGA1 sequencing

Retinitis pigmentosa, X-linked
- ❑ 326 RP2 sequencing
- ❑ 906 RP2 deletion/duplication testing if sequencing negative, females

Retinitis punctata albescens
- ❑ 4241 RLBP1 sequencing

❑ 474 Septo-optic dysplasia (HESX1)

Stargardt panel: Stargardt disease, fundus flavimaculatus, Stargardt-like macular dystrophy, other maculopathies
- ❑ 466 ABCA4, PRPH2 (RDS), and ELOVL4

Stargardt-like macular dystrophy, autosomal dominant
- ❑ 2990 Tier 1: ELOVL4 mutations hot spot
- ❑ 2991 Tier 2: ELOVL4 remaining exons

X-linked juvenile retinoschisis
- ❑ 2571 RS1 sequencing
- ❑ 906 RS1 deletion/duplication if sequencing negative, females

Familial hyperparathyroid syndromes/Endocrine neoplasias

❑ 170 Familial hypocalciuric hypercalcemia (CASR)

Hyperparathyroidism-jaw tumor syndrome or parathyroid carcinoma or familial isolated hyperparathyroidism (HRPT2)
- ❑ 1731 Tier 1 HRPT2 sequencing
- ❑ 1732 Tier 2, if Tier1negative
- ❑ 173 HRPT2 full gene sequencing NOW

Multiple Endocrine Neoplasia Type 1 (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 ex10, 11, 13 and 14

❑ 178 Multiple endocrine neoplasia type 2B, RET ex15 and 16

TEST CODE TEST NAME

Hearing loss

❑ J806 Hearing Loss Test (131 genes)

Pendred syndrome/DFNB4 Nonsyndromic hearing loss
- ❑ 572 SLC26A4 gene 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

Neurodevelopmental intellectual disability disorders

Angelman/Angelman-Like Syndrome
- ❑ 374 UBE3A Sequencing
- ❑ 375 SLC9A6 Sequencing
- ❑ 566 Methylation-MLPA (UPD, deletions, imprinting errors)

Autism/macrocephaly syndrome (PTEN)
- ❑ 195 PTEN sequencing and deletion/duplication testing

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

Cornelia de Lange syndrome (NIPBL, SMC1A)
- ❑ 568 NIPBL sequencing of select exons
- ❑ 569 NIPBL sequencing of remaining exons
- ❑ 906 NIPBL deletion/duplication
- ❑ 570 SMC1A full sequencing
- ❑ 906 SMC1A deletion/duplication

Prader-Willi syndrome
- ❑ 595 Methylation-MLPA (UPD, deletions, imprinting errors)

Rett syndrome / Atypical Rett syndrome (MECP2)/ASD
- ❑ 549 Rett/Atypical Rett syndromes (MECP2 seq & del/dup)

Rubinstein-Taybi syndrome (CREBBP) †
- ❑ 2921 CREBBP Tier 1 mutation hotspots and deletion/duplication 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)/ASD
- ❑ 3051 CDKL5 sequencing
- ❑ 906 CDKL5 deletion/duplication testing sequencing is negative

Neurofibromatosis
- ❑ J660 NF Type 1 (NF1 seq & del/dup only)
- ❑ 962 NF1 panel: NF1 and SPRED1 sequencing and deletion/duplication testing
  - ❑ 534 Reflex to Noonan syndrome and RASopathies panel (sequencing of 15 genes) if 962 is negative
- ❑ 963 NF2 panel: NF2 and SMARCB1 sequencing and deletion/duplication testing
- ❑ 961 Combined NF panel: NF1, SPRED1, NF2, and SMARCB1 sequencing and deletion/duplication testing
- ❑ J660 NF type 1 (NF1 seq & del/dup only)
- ❑ 816 Legius syndrome (SPRED1 seq only)

Noonan, LEOPARD, Cardiofaciocutaneous, and Costello syndromes and related RASopathies

❑ 534 Noonan Syndrome and RASopathies Panel (15 genes): ACTB, ACTG1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1, SPRED1

Individual gene testing -
- ❑ 191 HRAS sequencing
- ❑ 192 PTPN11 sequencing
- ❑ 389 SHOC2 (S2G mutation only)
- ❑ 535 CBL/NRAS sequencing
- ❑ 815 RIT1 sequencing

Other hereditary skin disorders

Birt-Hogg-Dubé syndrome (FLCN)
- ❑ 197 FLCN sequencing
- ❑ 906 FLCN deletion/duplication testing if sequencing is negative

Carney complex (PRKAR1A)
- ❑ 198 PRKAR1A sequencing
- ❑ 906 PRKAR1deletion/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 or mouthwash 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.

First Name Last Name Date of Birth (mm/dd/yy)

Please check appropriate boxes and fax only the sheets necessary

TEST CODE TEST NAME

Cowden Syndrome (PTEN)/(BRRS)/ASD
- ❑ 195 PTEN sequencing and deletion/duplication testing

❑ 201 Darier Disease (ATP2A2)
❑ B399 Familial cutaneous malignant melanoma (CDKN2A, CDK4)
❑ 512 Ferguson-Smith disease/Multiple Self-Healing Squamous Epithelioma (TGFBR1)
Gorlin Syndrome (PTCH1)
- ❑ 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

❑ 693 Ichthyosis Follicularis with Atrichia and Photophobia / Keratosis Follicularis Spinulosa Decalvans (MBTPS2)
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 Palmoplantar Keratoderma (incl. Vohwinkel syndr.) (GJB2, connexin 26)

Other keratin disorders

❑ 208 Epidermolytic PPK of Vörner (KRT9 hotspots)
Pachyonychia congenita
- ❑ 2091 KRT16, KRT6a hotspots
- ❑ 2092 KRT17, KRT6b hotspots

❑ 2111 Steatocystoma multiplex (KRT17 hotspots)
❑ 2131 White sponge nevus (KRT4, KRT13 hotspots)
Non-epidermolytic Palmoplantar Keratoderma (NEPPK), Unna-Thost disease
- ❑ 2121 KRT16 hotspots
- ❑ 1182 KRT1 sequencing

Periodic fever syndromes

❑ 367 Comprehensive panel 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 fever panel if 2 or more genes of the Periodic Fever Panel have been previously tested at GeneDx
❑ 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
- ❑ 324 SDHD sequencing
- ❑ 323 SDHC sequencing
- ❑ 555 TMEM127 sequencing

❑ 454 SDHAF2 targeted testing (G78R mutation only)

Sex differentiation disorders

❑ 339 Adrenal hyperplasia, POR deficiency (POR)
❑ 402 17-alpha hydroxylase/17,20-lyase deficiency (CYP17A1)
5-alpha reductase deficiency (SRD5A2)
- ❑ 469 SRD5A2 sequencing

Androgen Insensitivity Syndrome (AR) †
- ❑ 220 AR sequencing

❑ 340 Aromatase deficiency (CYP19A1)
Campomelic dysplasia (SOX9)
- ❑ 338 SOX9 sequencing
- ❑ 906 SOX9 deletion/duplication testing if sequencing is negative

TEST CODE TEST NAME

XY gonadal dysgenesis
- ❑ 341 NR5A1/SF-1 sequencing
- ❑ 259 SRY sequencing
- ❑ 422 DHH sequencing
- ❑ 906 NROB1/DAX1 gene duplication testing

Other genetic disorders

❑ 547 Aicardi-Goutieres syndrome (TREX1, RNASEH2A, RNASEH2B, RNASEH2C sequencing)
❑ 219 Allgrove (Triple-A) syndrome (AAAS)
Alport syndrome (COL4A5)
- ❑ 281 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 Branchiootic syndrome 3 (SIX1)
Branchiootorenal syndrome 3 (EYA1)
- ❑ 315E 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

Cerebral Cavernous Malformations (CCM) †
- ❑ 526 Cerebral cavernous malformations (KRIT1, CCM2, PDCD10 sequencing and deletion/duplication testing)
- ❑ 4181 KRIT1 Tier 1 sequencing (exons 14, 16, and 18)
- ❑ 4182 KRIT1 Tier 2 sequencing (rest of KRIT1) + deletion/duplication testing (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)

❑ 413 Chuvash Polycythemia (VHL)
❑ 227 Cohen syndrome (VPS13B) ❑ 2271 Finnish mutation only
❑ 650 Congenital indifference to pain (SCN9A)
❑ 239 Congenital insensitivity to pain and anhidrosis (NTRK1)
Craniofrontonasal dysplasia (EFNB1)
- ❑ 3251 EFNB1 sequencing
- ❑ 906 EFNB1 deletion/duplication testing if sequencing negative, females

❑ 229 Dent disease, X-linked recessive nephrolithiasis (CLCN5)
❑ 906 CLCN5 deletion/duplication testing if sequencing negative, females
Dopa-responsive dystonia (GCH1, TH) †
- ❑ 230 GCH1 sequencing
- ❑ 906 GCH1 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

Grieg Cephalopolysyndactyly syndrome
- ❑ 472 GLI3 sequence (exons 1-15) and deletion/duplication analysis

Hereditary angioedema
- ❑ 2341 Type I/II SERPING1 (C1NH) 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: 2, 3, 5, 6, 9, 10, 12, 13, and 17
- ❑ 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

Hypogonadotropic hypogonadism (HH) / Kallmann syndrome
- ❑ 676 HH sequencing and deletion/duplication panel, 14 genes

Specimen Requirements ALL tests offered by GeneDx can be performed with whole blood specimen. As an alternative to blood, buccal specimen or mouthwash 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.

First Name | Last Name | Date of Birth (mm/dd/yy)

Please check appropriate boxes and fax only the sheets necessary

TEST CODE TEST NAME

- ❑ 2401 KAL1 gene sequencing
- ❑ 906 KAL1 deletion/duplication testing if sequencing is negative, females
- ❑ 2402 FGFR1 gene sequencing

❑ 238 Inclusion body myopathy (GNE; M712T only)
❑ 650 Inherited erythromelalgia (SCN9A)

Juvenile Polyposis syndrome (JPS) (including JPS-HHT)
- ❑ 536 JPS Tier 1 SMAD4 sequencing + SMAD4 and BMPR1A deletion/duplication
- ❑ 537 JPS Tier 2 BMPR1A sequencing
- ❑ 538 SMAD4/BMPR1A deletion/duplication testing ONLY

Kabuki syndrome (KS)
- ❑ 583 KMT2D sequencing

❑ 673 KBG syndrome (ANKRD11)

Legius syndrome
- ❑ 816 SPRED1 sequencing
- ❑ 906 SPRED1 deletion/duplication testing

Li-Fraumeni Syndrome/Li-Fraumeni Like Syndrome
- ❑ 559 TP53 sequencing
- ❑ 906 TP53 deletion/duplication testing if sequencing is negative

Maturity-onset diabetes of the young (MODY)
- ❑ 674 MODY panel: GCK, HNF1A, HNF1B, HNF4A, PDX1

Nemaline myopathy, autosomal recessive
- ❑ 244 Nemaline myopathy (ACTA1) †
- ❑ 245 Nemaline myopathy (NEB; Askenazi Jewish mutation)

Oral-facial-digital syndrome type 1 (OFD1, aka CXORF5)
- ❑ 3641 Tier 1 OFD1 sequencing
- ❑ 3642 Tier 2 OFD1 sequencing
- ❑ 906 OFD1 deletion/duplication testing if sequencing is negative

Pallister Hall Syndrome
- ❑ 4711 Tier 1 GLI3 sequence analysis of exons 13-15
- ❑ 4712 Tier 2 GLI3 sequence analysis of remaining exons (1-12) and deletion/duplication analysis

❑ 650 Paroxysmal extreme pain disorder (SCN9A)

Premature ovarian failure (POF)
- ❑ 522 FMR1 CGG repeat analysis
- ❑ 677 POF sequencing panel: BMP15, CYP17A1, CYP19A1, FIGLA, FSHR, GDF9, LHCGR, NOBOX, NR5A1, POR, PSMC3IP

Renal-Coloboma Syndrome / Papillorenal Syndrome
- ❑ 5211 PAX2 Tier 1 sequencing
- ❑ 5212 PAX2 Tier 2 sequencing (rest of PAX2)
- ❑ 5213 PAX2 full gene sequencing NOW
- ❑ 906 PAX2 deletion/duplication testing

Simpson-Golabi-Behmel Syndrome (SGBS)
- ❑ 415 GPC3 sequencing (males)
- ❑ 415E GPC3 sequencing and deletion/duplication testing (females)

❑ 650 Small fiber neuropathy (SCN9A)

Sotos Syndrome
- ❑ 406 NSD1 sequencing and deletion/duplication testing

Spinal muscular atrophy with respiratory distress, type 1 (IGHMBP2)
- ❑ 342 IGHMBP2 sequencing

❑ 401 Supravalvular aortic stenosis / autosomal dominat cutis laxa (ELN)

Treacher Collins Syndrome (TCOF1)
- ❑ 653 TCOF1 sequencing
- ❑ 906 TCOF1 deletion/duplication testing if sequencing is negative

Usher syndrome panel (9 genes)
- ❑ 585 9 genes panel: MYO7A, USH1C, CDH23, PCDH15, USH1G, USH2A, GPR98, DFNB31, and CLRN1 sequencing
- ❑ 908 9 genes Usher syndrome panel, deletion/duplication testing

Van der Woude syndrome (IRF6)
- ❑ 253 IRF6 sequencing

Velocardiofacial syndrome / DiGeorge syndrome (TBX1)
- ❑ 358 TBX1 sequencing

X-linked Adrenal Hypoplasia Congenita (AHC)
- ❑ 416 NR0B1 sequencing

X-linked hydrocephalus, X-linked spastic paraplegia, MASA, CRASH syndrome (L1CAM)
- ❑ 2551 L1CAM sequencing
- ❑ 906 L1CAM deletion/duplication testing

GeneDx
an OPKO Health Company

Rare Mendelian Disorders Clinical Information Form

Account # Account Name

First Name Last Name Date of Birth (mm/dd/yy)

Clinical Diagnosis: _____ Age of Onset: _____

Clinical diagnosis: _____

ICD-10 codes: _____

PLEASE ATTACH DETAILED MEDICAL RECORDS, CLINICAL SUMMARY, PICTURES AND FAMILY HISTORY.
CLINICAL INFORMATION IS CRUCIAL FOR ACCURATE INTERPRETATION OF RESULTS.

Please check all that apply.

Perinatal history
- ❒ Prematurity
- ❒ IUGR
- ❒ Oligohydramnios
- ❒ Polyhydramnios
- ❒ Cystic hygroma/increased NT

Growth
- ❒ Failure to thrive
- ❒ Growth retardation/short stature
- ❒ Overgrowth
- ❒ Macrocephaly
- ❒ Microcephaly

Physical/Cognitive Development
- ❒ Fine motor delay
- ❒ Gross motor delay
- ❒ Speech delay
- ❒ Intellectual disability/MR
  IQ: _____
- ❒ Learning disability
- ❒ Developmental regression

Behavioral
- ❒ Autism spectrum disorder
- ❒ Autistic features
- ❒ Obsessive-compulsive disorder
- ❒ Stereotypic behaviors
- ❒ Other psychiatric symptoms

Craniofacial/Ophthalmalogic/Auditory
- ❒ Cataracts
- ❒ Cleft lip/palate
- ❒ Coloboma of eye
- ❒ CPEO (opthalmoplegia)
- ❒ Ptosis
- ❒ Blindness
- ❒ Optic atrophy
- ❒ Retinitis pigmentosa
- ❒ Hearing loss
- ❒ Ototoxicity (aminoglycoside-induced)
- ❒ External ear malformation
- ❒ Other visual abnormality type:_____
- ❒ Facial dysmorphism - please describe:
  _____
  _____

Cardiac/congenital heart malformations
- ❒ ASD
- ❒ VSD
- ❒ Coarctation of aorta
- ❒ Hypoplastic left heart
- ❒ Tetralogy of Fallot
- ❒ Cardiomyopathy
- ❒ Arrhythmia/conduction defect
- ❒ Other:_____

Cancer/Malignancy
- ❒ Age of onset:_____
- ❒ Tumor type:_____
- ❒ Location(s):_____
- ❒ Affected relatives: _____
  _____

Skin, Hair, and Nail Abnormalities
- ❒ Abnormal connective tissue: _____
  _____
- ❒ Abnormal nails: _____
- ❒ Abnormal pigmentation:_____
- ❒ Axillary and/or inguinal freckling
- ❒ Blistering
- ❒ Hypopigmentation/hyperpigmentation type: _____
- ❒ Ichthyosis
- ❒ Skin tumors/Malignancies
- ❒ Other: _____

Brain malformations/abnormal imaging
- ❒ Agenesis of the corpus callosum
- ❒ Holoprosencephaly
- ❒ Lissencephaly
- ❒ Cortical dysplasia
- ❒ Heterotopia
- ❒ Hydrocephalus
- ❒ Brain atrophy
- ❒ Periventricular leukomalacia
- ❒ Hemimegalencephaly
- ❒ Abnormalities of basal ganglia
- ❒ Other: _____

Neurological/Muscular
- ❒ Ataxia
- ❒ Chorea
- ❒ Dystonia
- ❒ Hypotonia
- ❒ Hypertonia
- ❒ Seizures type: _____
- ❒ Spasticity
- ❒ Exercise intolerance/easy fatigue
- ❒ Muscle weakness
- ❒ Stroke/stroke-like episodes
- ❒ Recurrent headache/migraine

Gastrointestinal
- ❒ Gastroschisis/omphalocele
- ❒ Pyloric stenosis
- ❒ Tracheoesophageal fistula
- ❒ Delayed gastric emptying
- ❒ Eosinophilic esophagitis
- ❒ Gastrointestinal reflux
- ❒ Recurrent vomiting
- ❒ Chronic diarrhea
- ❒ Constipation
- ❒ Chronic intestinal pseudo-obstruction
- ❒ Hirschsprung disease
- ❒ Hepatic failure
- ❒ Elevated transaminases

Skeletal/Limb abnormalities
- ❒ Contractures
- ❒ Club foot
- ❒ Polydactyly
- ❒ Syndactyly
- ❒ Scoliosis
- ❒ Vertebral anomaly
- ❒ Other: _____

Genitourinary abnormalities
- ❒ Ambiguous genitalia
- ❒ Hypospadias
- ❒ Hydronephrosis
- ❒ Undescended testis
- ❒ Kidney malformation
- ❒ Renal agenesis
- ❒ Renal tubulopathy
- ❒ Other: _____

Endocrine
- ❒ Diabetes mellitus: ❒ Type I ❒ Type II
- ❒ Hypothyroidism
- ❒ Hypoparathyroidism
- ❒ Pheochromocytoma/paraganglioma

Metabolic
- ❒ Ketosis
- ❒ Lactic acidemia/high CSF lactate
- ❒ Elevated pyruvate
- ❒ Elevated alanine
- ❒ Organic aciduria
- ❒ Low plasma carnitine
- ❒ CPK abnormalities

Hemotologic/Immunologic
- ❒ Recurrent fever
- ❒ Anemia/neutropenia/pancytopenia
- ❒ Immunodeficiency: Type:_____
- ❒ Other: _____

Other testing (summarize or attach reports):
- ❒ Chromosomes/FISH: _____
- ❒ Array CGH:_____
- ❒ Fragile X syndrome:_____
- ❒ Muscle biopsy: _____
- ❒ Other relevant results (clinical or research):
  _____
  _____
  _____

Additional relevant clinical info: _____
_____
_____
_____
_____

Informed Consent

Account # Account Name

First Name Last Name Date of Birth (mm/dd/yy)

I understand that my health care provider has ordered the following genetic testing for {me/my child}:_____.

General Information About Genetic Testing

What is genetic testing?

DNA provides instructions for our body's growth and development. Genes are distinct sequences of DNA, and are arranged on chromosomes. The DNA in a gene contains instructions for making proteins, which determine things like growth and metabolism as well as traits like eye color and blood type. Genetic disorders are caused by harmful changes in DNA or from changes in the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these harmful changes in chromosomes or the DNA. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic and results may require additional diagnostic testing.

The purpose of this test is to see if I, or my child, may have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the chance that I, or my child, will develop or pass on a genetic disorder in the future. 'My child' can also mean my unborn child, for the purposes of this consent.

Additional information about the specific test being ordered is available from my health care provider or I can go to the GeneDx website, www.genedx.com. This information includes the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, and the limitations of genetic testing.

If {I/my child} already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I will inform the laboratory of this information.

What could I learn from this genetic test?

The following describes the possible results from the test:

1) Positive: A positive result indicates that a genetic variant has been identified that explains the cause of {my/my child's} genetic disorder or indicates that {I/my child} am at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

2) Negative: A negative result indicates that no disease-causing genetic variant was identified for the test performed. It does not guarantee that {I/my child} will be healthy or free from genetic disorders or medical conditions. If {I/my child} test negative for a variant known to cause the genetic disorder in other members of {my/my child's} family, this result rules out a diagnosis of the same genetic disorder in {me/my child} due to this specific change.

3) Inconclusive/Variant of Uncertain Significance (VUS): A finding of a variant of uncertain significance indicates that a genetic change was detected, but it is currently unknown whether that change is associated with a genetic disorder either now or in the future. A variant of uncertain significance is not the same as a positive result and does not clarify whether {I/my child} is at increased risk to develop a genetic disorder. The change could be a normal genetic variant or it could be disease-causing. Further analysis may be recommended, including testing both parents and other family members. Detailed medical records or information from other family members also may be needed to help clarify results.

4) Unexpected results: In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering this test. For example, this test may tell me about the risk for another genetic condition {I/my child} is not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. This information may be disclosed to the ordering health care provider if it likely impacts medical care.

Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information GeneDx used to interpret {my/my child's} results. Providers can contact GeneDx at any time to discuss the classification of an identified variant.

For tests that evaluate data from multiple family members, my spouse, or partner concurrently, results may be included in a single comprehensive report.

What are the risks and limitations of this genetic test?

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology.
- Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in {my/my child's} family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results. In some cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. This includes non-paternity (the stated father of an individual is not the biological father) and consanguinity (the parents of an individual are related by blood). It may be necessary to report these findings to the health care provider who ordered the test.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to: mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, or the presence of change(s) in such a small percentage of cells that the change(s) may not be detectable by the test (mosaicism).
- This test does not have the ability to detect all of the long-term medical risks that {I/my child} might experience. The result of this test does not guarantee my health or the health of my child/fetus. Other diagnostic tests may still need to be done, especially when only a genetic screening test has been performed previously.
- Occasionally, an additional sample may be needed if the initial specimen is not adequate.

Patient Confidentiality and Genetic Counseling

It is recommended that I receive genetic counseling before and after having this genetic test. I can find a genetic counselor in my area here: www.nsgc.org. Further testing or additional consultations with a health care provider may be necessary.

To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to me, to other health care providers involved in {my/my child's} diagnosis and treatment, or to others as entitled by law. The United States Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, I understand that I can visit www.genome.gov/10002077.

International Specimens

If {I/my child} reside outside the United States, I attest that by providing a sample for testing, I am not knowingly violating any export ban or other legal restriction in the country of {my/my child's} residence.