GeneDx | Rare Genetic Disorders Test Requisition Form

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 #

Sample Information

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

❒ Blood in EDTA (5-6 mL in lavender top tube)
❒ Buccal Swab
❒ Oral Rinse (At least 30 mL of Scope oral rinse in a 50 mL centrifuge tube)
❒ DNA (>20 ug): Tissue source _____ concentration ___ (ug/ml) Vol ___(ul)
❒ Other _____(Call lab)

Patient has had a blood transfusion ❒ Yes ❒ No Date of last transfusion __/__/__
(2-4 weeks of wait time is required for mtDNA testing only) Specimens are not accepted for patients who have had allogeneic bone marrow transplants

Clinical Diagnosis: _____ **ICD-10 Codes:** _____
Age at Initial Presentation: _ **Add. ICD-10 Codes:**_____

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

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

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

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)

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 estimated 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 for the full amount stated above (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

GeneDx | Rare Genetic Disorders Test Requisition Form

an OPKO Health Company

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

Family History of Disorder/Symptoms

❒ No Known Family History
❒ Pedigree Attached
❒ Adopted

Relationship	Maternal	Paternal	Disorder/Symptoms	Age at Dx
	❒	❒		
	❒	❒		
	❒	❒		
	❒	❒		

Other clinical history or testing (summarize or attach reports)
❒ Array CGH: _____
❒ Chromosomes/FISH: _____
❒ Other relevant results (clinical or research): _____

Draw/attach pedigree and/or include additional information

Reason for testing - please complete (required):

If expedited testing is requested, please indicate reason:

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

Family Member/Carrier Testing and Special Services

Testing for known familial variant in a nuclear gene

❒ 9011 Testing for ONE known familial variant in a nuclear gene
❒ 9012 Testing for TWO known familial variants in a nuclear gene
❒ 905 Testing for ONE known familial exon-level del/dup or chromosomal microarray del/dup

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

DNA extraction only

❒ 909 One sample

Please fill out this information if selecting a test from the family member/carrier testing section:

Relative to be tested: ❒ Affected/Symptomatic ❒ Unaffected/Asymptomatic

Gene(s): _____ Variant(s): _____

Proband Name: _____

Relationship to proband: _____

Proband GeneDx Acc#: _____

❒ Proband tested at another lab. Select all that apply
- ❒ Positive control included - **Positive control is required if previous test was performed at another lab.**
- ❒ Positive control not available. Please initial to acknowledge acceptance of caveat language on a negative report _____
- ❒ Family Member Test Report included - A clear copy of the test report on the variant positive family member is recommended if previous test was performed at another lab.

Single Gene Analysis/Write-in Test Selection

❒ 906 Deletion/Duplication Analysis of 1-2 nuclear gene

❒ 703 Deletion/Duplication Analysis of 3-20 nuclear genes

If selected, write in desired gene(s) to be tested: _____

❒ Test Code: _____ Test Name: _____

❒ Test Code: _____ Test Name: _____

Many other tests are available on other requisition forms. Please visit genedx.com to find the right test for your patient.

All single gene tests are on pages 4-6

Rare Disorders Multi-gene Panels

Test Code	Test Name	# Genes	Gene List
Dermatologic Disorders			
❒ 708	Congenital ichthyosis XomeDx*Slice*	49	*ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, ARSE, CASP14, CDSN, CERS3, CHST8, CLDN1, CSTA, CYP4F22, EBP, ELOVL4, FLG, FLG2, GJB2, GJB3, GJB4, GJB6, KDSR, KRT1, KRT10, KRT2, KRT9, LIPN, LOR, MBTPS2, NIPAL4, NSDHL, PEX7, PHGDH, PHYH, PNPLA1, POMP, PSAT1, SDR9C7, SERPINB8, SLC27A4, SNAP29, SPINK5, ST14, STS, TGM1, TGM5, VPS33B, ZMPSTE24*
❒ 707	Epidermolysis bullosa (EB) and other bullous skin disorders XomeDx*Slice*	28	*CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSG1, DSP, DST, EXPH5, FERMT1, FLG2, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, SERPINB8, TGM5*
❒ B399	Melanoma panel	9	*BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN, RB1, TP53*
Dysmorphology and Multiple Congenital Anomalies			
❒ TA46	Adams-Oliver syndrome panel	6	*ARHGAP, DLL4, DOCK6, EOGT, NOTCH1, RBPJ*
❒ TA44	Baraitser-Winter syndrome panel	2	*ACTB, ACTG1*
❒ T993	Coffin-Siris syndrome panel	8	*ARID1A, ARID1B, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SOX11*
❒ 584	Cornelia de Lange syndrome panel	7	*ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC1A, SMC3*
❒ 961	Neurofibromatosis type 1 and 2 panel	4	*NF1, NF2, SMARCB1, SPRED1*
❒ 962	Neurofibromatosis type 1 panel	2	*NF1, SPRED1*
❒ 963	Neurofibromatosis type 2 panel	2	*NF2, SMARCB1*
❒ TA06	Noonan and comprehensive RASopathies panel	25	*A2ML1, ACTB, ACTG1, BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
❒ TA39	Robinow syndrome panel	4	*DVL1, DVL3, ROR2, WNT5A*
❒ TA38	Treacher Collins syndrome panel	6	*DHODH, EFTUD2, POLR1C, POLR1D, SF3B4, TCOF1*
Endocrine Disorders			
❒ 676	Hypogonadotropic hypogonadism panel	33	*CHD7, CYP19A1, DUSP6, ESR1, FEZF1, FGF17, FGF8, FGFR1, GNRH1, GNRHR, HS6ST1, IL17RD, KAL1, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, NR0B1, NR5A1, NSMF, POLR3B, PROK2, PROKR2, PROP1, SEMA3A, SEMA3E, SOX10, SPRY4, TAC3, TACR3, WDR11*
❒ 674	Maturity-onset diabetes of the young (MODY) panel	16	*ABCC8, APPL1, BLK, CEL, GCK, GLUD1, HADH, HNF1B, HNF4A, HNF1A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1 (IPF1)*
Hematologic Disorders			
❒ 938	Congenital sideroblastic anemia panel (plus mitochondrial genome large deletion testing)	8	*ABCB7, ALAS2, GLRX5, PUS1, SLC19A2, SLC25A38, TRNT1, YARS2*
❒ J450	Diamond-Blackfan anemia panel	13	*GATA1, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7*
❒ TB47	Dyskeratosis congenita panel	12	*ACD, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53*
Immunologic Disorders			
❒ T990	Autoimmune lymphoproliferative syndrome (ALPS) panel	4	*FAS, CASP10, CASP8, FASL*
❒ T989	Chronic granulomatous disease (CGD) panel	5	*CYBA, CYBB, NCF1, NCF2, NCF4*
❒ 601	Comprehensive SCID panel	26	*ADA, AK2, ATM, CD3D, CD3E, CD3Z, CORO1A, DCLRE1C (ARTEMIS), DOCK8, FOXN1, IL2RG, IL7R, JAK3, LIG4, NHEJI, ORAI1, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RMRP, STIM1, TBX1, ZAP70*
❒ 603	B- SCID sub-panel	9	*ADA, AK2, DCLRE1C (ARTEMIS), LIG4, NHEJ1, PRKDC, RAC2, RAG1, RAG2*
❒ 602	B+ SCID sub-panel	17	*TM, CD3D, CD3E, CD3Z, CORO1A, DOCK8, FOXN1, IL2RG, IL7R, JAK3, ORAI1, PNP, PTPRC, RMRP, STIM1, TBX1, ZAP70*
❒ 678	Hyper-IgE syndrome panel	4	*DOCK8, SPINK5, STAT3, TYK2*
❒ T995	Hyper-IgM panel	4	*AICDA, CD40, CD40LG, UNG*
Neurologic Disorders			
❒ 547	Aicardi-Goutieres syndrome panel^	4	*RNASEH2A, RNASEH2B, RNASEH2C, TREX1*
❒ 526	Cerebral cavernous malformations panel	3	*CCM2, KRIT1, PDCD10*
❒ TB51	Comprehensive holoprosencephaly panel	17	*CDON, DISP1, DLL1, FGF8, FGFR1, FOXH1, GLI2, GAS1, NODAL, PTCH1, SHH, SIX3, SMAD2, STIL, TDGF1, TGIF1, ZIC2*
❒ 2371	Holoprosencephaly panel	4	*SHH, SIX3, TGIF, ZIC2*
Pulmonary Disorders			
❒ TB46	Primary ciliary dyskinesia panel	30	*ARMC4, C21ORF59 (CFAP298), CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, DNAAF1, DNAAF2, DNAAF3, DNAAF5 (HEATR2), DNAH11, DNAH5, DNAI1, DNAI2, DNAJB13, DRC1, DYX1C1 (DNAAF4), GAS8, LRRC6, PIH1D3, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10*
❒ TB48	Hermansky-Pudlak syndrome panel	10	*AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6*
❒ TB49	Surfactant dysfunction panel	5	*ABCA3, CSF2RA, CSF2RB, SFTPB, SFTPC*

Rare Disorders Multi-gene Panels

Test Code	Test Name	# Genes	Gene List
Reproductive Disorders			
❒ T991	Neonatal 46, XY disorders of sex development (DSD) panel	19	*AR, ARX, ATRX, CHD7, CYP11A1, CYP17A1, DHCR7, DHH, DYNC2H1, HSD17B3, HSD3B2, NEK1, NR5A1, POR, SOX9, SRD5A2, SRY, STAR, WT1*
❒ 677	Premature ovarian failure panel	22	*BMP15, CYP17A1, CYP19A1, ESR1, FGFR1, FIGLA, FSHR, GDF9, KISS1, KISS1R, LHB, LHCGR, NOBOX, NR5A1, POR, PROK2, PROKR2, PSMC3IP, SEMA3A, TAC3, TACR3, WDR11*
Rheumatologic Disorders			
❒ 367	Comprehensive panel for periodic fever syndromes panel: Familial hibernian fever/TRAPS; Familial mediterranean fever; Hyper-IgD syndrome; Muckle Wells/familial cold urticaria, NOMID; Cyclic neutropenia; PAPA syndrome; Majeed syndrome^	7	*ELANE (ELA2), LPIN2, MEFV, MVK, NLRP3 (CIAS1), PSTPIP1, TNFRSF1A*
Skeletal Disorders			
❒ TA45	Abnormal mineralization panel	16	*ALPL, ANKH, AP2S1, CASR, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR*
❒ J799	Achondrogenesis panel	3	*COL2A1, SLC26A2, TRIP11*
❒ T992	Autosomal dominant osteogenesis imperfecta panel	3	*COL1A1, COL1A2, IFITM5*
❒ J804	Chondrodysplasia punctata panel	5	*AGPS, ARSE, EBP, GNPAT, PEX7*
❒ TA40	Craniosynostosis panel	30	*ALPL, ALX4, ASXL1, CDC45, CYP26B1, EFNB1, ERF, FGFR1, FGFR2, FGFR3, GLI3, IFT122, IFT43, IL11RA, MASP1, MEGF8, MSX2, P4HB, POR, RAB23, RECQL4, SEC24D, SKI, TCF12, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR35, ZIC1*
❒ TA41	Ectrodactyly/split hand-split foot malformation panel	13	*BLHHA9, CDH3, DLX5, DYNC1I (del/dup only), FGFR1, TP63, WNT10B, LBX1, BTRC, POLL, DPCD, FBXW4, 10q24(chr10:102 962, 134-103, 476, 346)*
❒ J800	FGFR-related disorders panel	2	*FGFR2, FGFR3^*
❒ T996	Hereditary multiple exostoses panel	3	*EXT1, EXT2, PTPN11*
❒ TA42	Limb abnormalities panel	71	*ANKRD11, ARHGAP31, ARID1A, ARID1B, BHLHA9, BMP2, BMPR1B, CC2D2A, CDH3, CEP290, CHSY1, DLL4, DLX5, DOCK6, DVL1, DVL3, DYNC1I1, EOGT, ESCO2, FGF10, FGF16, FGFR1, FGFR2, FGFR3, GDF5, GLI3, GNAS, HDAC4, HDAC8, HOXD13, IHH, KIF7, KMT2A, LMBR1 (including ZRS regulatory region), LRP4, MGP, MKS1, MYCN, NIPBL, NOG, NOTCH1, NSDHL, PHF6, PIGV, PTHLH, RAD21, RBPJ, RECQL4, RBM8A, ROR2, RPGRIP1L, SALL1, SALL4, SHH, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SOX11, SOX9, TBX15, TBX3, TBX5, THPO, TP63, WNT10B, WNT3, WNT5A, WNT7A and deletion/duplication coverage for 10q24*
❒ J797	Osteogenesis imperfecta panel	24	*ALPL, ANO5, B3GAT3, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, P3H1(LEPRE1), P4HB, PLOD2, PLS3, PP1B, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, WNT1*
❒ T994	Hypophosphatasia and hypophosphatemic rickets panel	9	*CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FGF23, PHEX, SLC34A3, VDR*
❒ TA43	Skeletal dysplasia panel	29	*ALPL, ARSE, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, DDR2, EBP, FGFR3, FLNB, HSPG2, INPPL1, LBR, LIFR, MMP9, MMP13, NKX3-2, NSDHL, PEX7, PTH1R, RMRP, SBDS, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4*

Rare Disorders Single Gene Tests

Test Code	Test Name	Gene	Test Code	Test Name	Gene
Dermatologic Disorders - Congenital Ichthyosis					
❒ 1181	Epidermolytic ichthyosis (epidermolytic hyperkeratosis)	*KRT1, KRT10* hotspots only	❒ 119	Erythrokeratodermia variabilis	*GJB3^, GJB4^*
❒ 122	Epidermolytic ichthyosis (epidermolytic hyperkeratosis)	*KRT2* hotspots only	❒ TB14	Ichthyosis follicularis with atrichia and photophobia/keratosis follicularis spinulosa decalvans	*MBTPS2*
❒ 208	Epidermolytic PPK of Vörner	*KRT9* hotspots only	❒ 130	Syndromic palmoplantar keratoderma Vohwinkel syndrome KID syndrome^	*GJB2* (Cx26)^
Dermatologic Disorders - Connective Tissue Disorders					
❒ TB16	Prolidase deficiency	*PEPD^*	❒ 2641	Pseudoxanthoma elasticum common mutations	*ABCC6*
❒ TA86	Supravalvular aortic stenosis/autosomal dominant cutis laxa	*ELN*	❒ 2642	If negative, reflex to: full gene sequencing	
Dermatologic Disorders - Ectodermal Dysplasia (ED)					
❒ 1601E	An/hypohidrotic, X-linked	*EDA1*	❒ 157	Clouston syndrome	*GJB6* (Cx30)^
❒ TB11	An/hypohidrotic ED, autosomal dominant	*EDARADD*	❒ 306	Focal dermal hypoplasia Goltz syndrome	*PORCN*
❒ TA80	Ectodermal dysplasia Odonto-onycho-dermal dysplasia Schöpf-Schulz-Passarge syndrome	*WNT10A*	❒ 553	Incontinentia pigmenti common deletion and full gene sequencing	*IKBKG/NEMO*
			❒ 2861	Incontinentia pigmenti common deletion-females	*IKBKG/NEMO*
❒ TA50	Autosomal recessive/dominant hypohidrotic ED	*EDAR*	❒ 2862	If negative, reflex to: full gene sequencing	

Rare Disorders Single Gene Tests

Test Code	Test Name	Gene	Test Code	Test Name	Gene
Dermatologic Disorders - Epidermolysis Bullosa					
❒ TA53	Epidermolysis bullosa, dystrophic	*COL7A1*	❒ 168	Epidermolysis bullosa, simplex	*KRT5, KRT14* hotspots only
❒ 1631	Epidermolysis bullosa, junctional type	*LAM5* hotspots only			
Dermatologic Disorders - Other Skin/Nail/Hair/Mucosal Disorders					
❒ TA79	Bloom syndrome	*BLM*	❒ 2091	Pachyonychia congenita	*KRT16, KRT6a* hotspots only
❒ TA54	Darier disease	*ATP2A2*	❒ 2092	Pachyonychia congenita	*KRT17, KRT6b* hotspots only
❒ TA55	Hailey-Hailey disease	*ATP2C1*	❒ 2131	White sponge nevus	*KRT4, KRT13* hotspots only
❒ TB15	Haim-Munk syndrome Papillon-Lefevre syndrome	*CTSC*			
Dermatologic Disorders - Pigmentary Disorders					
❒ 189	Hermansky-Pudlak syndrome: Ashkenazi splice mutation	*HPS3*	❒ 188	Hermansky-Pudlak syndrome: Puerto Rican mutations	*HPS1^, HPS3*
Dermatologic Disorders - Skin Cancers					
❒ 2071	Peutz-Jeghers syndrome	*STK11*	❒ 205	Gorlin syndrome	*PTCH1*
❒ 714	Birt-Hogg-Dube syndrome	*FLCN*	❒ 713	Hereditary leiomyomatosis and renal cell cancer	*FH*
❒ 715	Carney complex	*PRKAR1A*	❒ 195	Cowden syndrome Bannayan-Riley-Ruvalcaba syndrome Macrocephaly/ASD	*PTEN*
Dysmorphology & Multiple Congenital Anomalies					
❒ 491	Aniridia WAGR	*PAX6*	❒ TB27	Oral-facial-digital syndrome type 1	*OFD1(CXORF5)*
❒ 1004	Alagille syndrome	*JAG1*	❒ 2923	Rubinstein-Taybi syndrome	*CREBBP*
❒ 315E	Branchiootorenal syndrome	*EYA1*	❒ 415E	Simpson-Golabi-Behmel syndrome	*GPC3*
❒ TB21	CHARGE syndrome	*CHD7*	❒ 2511	Smith-Magenis syndrome	*RAI1*
❒ 550	Coffin-Lowry syndrome	*RPS6KA3 (RSK2)*	❒ 406	Sotos syndrome	*NSD1*
❒ TA58	Cohen syndrome	*VPS13B*	❒ TA62	Van der Woude syndrome	*IRF6*
❒ TB26	Craniofrontonasal dysplasia	*EFNB1*	❒ 358	Velocardiofacial syndrome DiGeorge syndrome	*TBX1^*
❒ TA52	Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome and TP63-related disorders	*TP63*	❒ TB04	Kabuki syndrome	*KMT2D*
❒ TA63	Feingold syndrome	*MYCN*	❒ TB20	Hirschsprung disease	*RET*
❒ J660	Neurofibromatosis type 1	*NF1*			
Endocrine Disorders					
❒ 402	17-alpha hydroxylase/17,20-lyase deficiency	*CYP17A1^*	❒ 332	Von Hippel-Lindau syndrome	*VHL*
❒ TA56	Allgrove (Triple-A) syndrome	*AAAS*	❒ 719	Multiple endocrine neoplasia, type 1	*MEN1*
❒ TA57	Androgen insensitivity syndrome	*AR*	❒ TB03	Pendred syndrome DFNB4 Nonsyndromic hearing loss	*SLC26A4*
❒ TB19	Autoimmune polyendocrinopathy APECED	*AIRE*	❒ 1771	Multiple endocrine neoplasia, types 2A and 2B	*RET^*
❒ 721	Hyperparathyroidism-jaw tumor syndrome	*CDC73*	❒ TA94	Septo-optic dysplasia	*HESX1*
Hematologic Disorders - Dyskeratosis Congenita (DKC)					
❒ 108	DKC, X-linked	*DKC1^*	❒ 682	DKC, autosomal dominant/recessive	*TERT^*
❒ 107	DKC, autosomal dominant	*TERC^*	❒ 414	DKC, autosomal dominant (exon 6 sequencing only)	*TINF2^*
Hematologic Disorders - Bone Marrow Failure Syndromes					
❒ TA47	Congenital amegakaryocytic thrombocytopenia	*MPL*	❒ TA97	X-linked thrombocytopenia –or– X-linked neutropenia	*WAS*
❒ 109	Shwachman-Diamond syndrome	*SBDS^*			

Rare Disorders Single Gene Tests

Test Code	Test Name	Gene	Test Code	Test Name	Gene
Hematologic Disorders - Other					
❒ 2341	Hereditary angioedema (HAE) type I/II	*SERPING1 (C1NH)*	❒ 388	Hereditary angioedema type III exon 9/Thr328 mutation only	*F12^*
Immunologic Disorders					
❒ 2862	Ectodermal dysplasia with immunodeficiency Incontinentia pigmenti	*IKBKG/NEMO^*	❒ TA48	Severe congenital neutropenia, autosomal dominant	*ELANE (ELA2)*
❒ TA69	IRAK4 deficiency	*IRAK4*	❒ TA70	Severe congenital neutropenia, autosomal recessive	*HAX1*
❒ 154	X-linked Agammaglobulinemia	*BTK*			
Neurological Disorders					
❒ TA81	Angelman Angelman-like syndrome	*SLC9A6*	❒ 549	Rett/atypical Rett syndromes	*MECP2*
❒ TB12	Erythromelalgia Paroxysmal extreme pain disorder Small fiber neuropathy Congenital insensitivity to pain	*SCN9A*	❒ 548	X-linked early infantile epileptic encephalopathy Atypical Rett syndrome West syndrome	*CDKL5*
❒ TA60	Congenital insensitivity to pain and anhidrosis	*NTRK1*	❒ 552	X-linked hydrocephalus, X-linked spastic paraplegia MASA CRASH syndrome	*L1CAM*
❒ TA78	Tyrosine hydroxylase deficiency	*TH*			
Pulmonology Disorders					
❒ T829	Cystic fibrosis/congenital bilateral absence of the vas deferens	*CFTR*			
Renal Disorders					
❒ TA64	Alport syndrome	*COL4A5*	❒ TA59	Dent disease X-linked recessive nephrolithiasis	*CLCN5*
❒ TA71	Branchiootic syndrome 3	*SIX1*	❒ T422	Polycystic kidney disease, deletion/duplication only	*PKD1/PKD2/TSC2*
❒ TA73	Dent disease 2 Lowe syndrome	*OCRL*	❒ TB29	Renal-Coloboma syndrome Papillorenal syndrome	*PAX2*
Reproductive Disorders - Disorders of Sexual Differentiation					
❒ 339	Adrenal hyperplasia POR deficiency	*POR^*	❒ 259	XY gonadal dysgenesis	*SRY^*
❒ TA89	X-linked adrenal hypoplasia congenita	*NR0B1 (DAX1)*			
Reproductive Disorders - Infertility					
❒ 522	FMR1-associated premature ovarian failure, CGG repeat analysis only	*FMR1*			
Rheumatologic Disorders					
❒ 215	Familial Hibernian fever TRAPS exons 2-5 sequencing only	*TNFRSF1A*	❒ 216	Hyper-IgD syndrome (MVK) exons 8 and 10 sequencing only	*MVK*
❒ 214	Familial Mediterranean fever exons 2,3 and 10 sequencing only	*MEFV*	❒ 217	Muckle-Wells Familial cold urticaria NOMID exon 3 sequencing only	*CIAS1*
Skeletal Disorders					
❒ TA74	Campomelic dysplasia	*SOX9*	❒ 472	Grieg cephalopolysyndactyly syndrome	*GLI3*
❒ 225	Cartilage-hair hypoplasia and associated disorders	*RMRP^*	❒ TB13	KBG syndrome	*ANKRD11*
❒ 285	Cherubism	*SH3BP2^*	❒ TA61	Pseudoachondroplasia Multiple epiphyseal dysplasia	*COMP*
❒ 282E	Chondrodysplasia punctata, X-linked	*ARSE*	❒ 1861E	X-linked dominant hypophosphatemia	*PHEX*
❒ TB31	Familial hypocalciuric hypercalcemia	*CASR*	❒ TB22	Holt-Oram Syndrome	*TBX5*

All sequencing tests include del/dup analysis unless indicated by a ^ or otherwise noted

Many other tests are available on other requisition forms. Please visit genedx.com to find the right test for your patient.

Did you Remember to...?

- ❒ Label specimen tube appropriately with TWO identifiers
- ❒ Get a signature for medical necessity and patient consent
- ❒ Fill out sample submission form (pages 3 - 6)
- ❒ Complete clinical information (page 7)
- ❒ Complete payment form (page 1)

GeneDx
an OPKO Health Company

Rare Genetic Disorders Clinical Information Form

Account # Account Name

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

PLEASE ATTACH DETAILED MEDICAL RECORDS

Clinical Diagnosis:_____ **ICD-10 Codes:**_____ **Age at Initial Presentation:**_____

❒ **Parent/Carrier testing (Circle One: Asymptomatic/Symptomatic)**

Perinatal History
❒ Prematurity
❒ IUGR
❒ Oligohydramnios
❒ Polyhydramnios
❒ Cystic hygroma/increased NT

Growth
❒ Failure to thrive (%ile:_____)
❒ Growth retardation/short stature (%ile:_____)
❒ Overgrowth (%ile:_____)
❒ 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
❒ Blue/gray sclerae
❒ Cataracts
❒ Cleft lip/palate
❒ Coloboma of eye
❒ CPEO (opthalmoplegia)
❒ Glaucoma
❒ 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
❒ Atrial septal defect
❒ Ventricular septal defect
❒ 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
❒ Connective tissue abnormalities:_____
_____
❒ Abnormal hair or nails: _____
❒ Abnormal pigmentation
❒ Hypopigmentation/hyperpigmentation:_____
❒ Axillary and/or inguinal freckling
❒ Blistering
❒ Ichthyosis/hyperkeratosis
❒ 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
❒ Eosinophilic esophagitis
❒ Gastrointestinal reflux
❒ Recurrent vomiting
❒ Chronic diarrhea
❒ Constipation
❒ Chronic intestinal pseudo-obstruction
❒ Hirschsprung disease
❒ Hepatic failure
❒ Elevated transaminases

Skeletal/Limb abnormalities
❒ Abnormal ribs (TC ratio: _____; Specify:_____)
❒ Contractures
❒ Club foot
❒ Fractures (#_____; Area:_____)
❒ Limb anomaly (Specify:_____)
❒ Polydactyly (Specify:_____)
❒ Syndactyly (Specify:_____)
❒ Scoliosis
❒ Vertebral anomaly (Specify: _____)
❒ 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

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

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 certain changes in DNA affecting the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these 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 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.

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 by 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 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. In addition, I or my/my child's health care providers may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find current information about the clinical interpretation of my/my child's variant(s).

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.

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, the limitations of genetic testing, as well as information about how specimens and information are stored and used.