OPHTHALMOLOGY/AUDIOLOGY TEST REQUISITION FORM

GeneDx an OPKO Health Company

PATIENT INFORMATION

First Name	Last Name	
Genetic Sex ○ Male ○ Female Gender Identification (optional): _____	Date of Birth (mm/dd/yy)	
Ancestry ○ White/Caucasian ○ Hispanic ○ Black/African American ○ Native American ○ East Asian ○ South Asian ○ Middle Eastern ○ Ashkenazi Jewish ○ Other: _____		
Email		
Address		
City	State	Zip Code
Primary Phone	Is this patient deceased? ○ Yes ○ No Deceased Date: _____	

SAMPLE INFORMATION

Date Sample Collected (mm/dd/yy) (required):	Medical Record #
○ Blood ○ Buccal Swab ○ Other (specify source): _____	
Patient has had a blood transfusion ○ Yes ○ No Date of Last Transfusion: _____ (2-4 weeks of wait time is required for some testing)	
Patient has had an allogenic bone marrow transplant ○ Yes ○ No Fibroblasts are recommended for patients who had an allogenic bone marrow transplant. See www.genedx.com/specimen-requirements for details.	
○ Treatment-Related **RUSH**	Date: _____

PATIENT CONSENT FOR GENETIC TESTING, FINANCIAL AGREEMENT AND GUARANTEE:

By signing this form, I acknowledge as the patient or relative being tested that I have read the GeneDx Informed Consent document available from my healthcare provider or at genedx.com/forms, and I authorize GeneDx to perform genetic testing as ordered. I understand that, for tests that evaluate data from multiple family members concurrently, results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their healthcare providers. By my signature below, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider. For insurance billing, I understand and authorize GeneDx to bill my health insurance plan on my behalf, to release any information required for billing, and to be my designated representative for purposes of appealing any denial of benefits. I irrevocably assign to and direct that payment be made to GeneDx. I understand that my out-of-pocket costs may be different than the estimated amount indicated to me by GeneDx as part of a benefit investigation and agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. I am aware that my insurance provider may send payment directly to me for services performed by GeneDx on my behalf. I agree to endorse the insurance check and forward it to GeneDx within 30 days of receipt as payment towards GeneDx's claim. If I do not have health insurance, I agree to pay for the full cost of the genetic testing that was ordered by my healthcare provider and billed to me by GeneDx. I further understand and agree that, if I fail to make payment for genetic testing, in accordance with the payment policies of GeneDx, my account may be turned over to an external collection agency for non-payment and I agree to pay any associated collection costs, including attorney fees.

More information, including the GeneDx Notice of Privacy Policies, is available on GeneDx's website: www.genedx.com

Medicare: A completed Advance Beneficiary Notice (ABN) is required for Medicare patients. Please visit our website, www.genedx.com/billing for more information.

○ By checking this box, I confirm that I am a New York state resident, and I give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing to be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York law requires GeneDx to destroy my sample after 60 days and it cannot be used for the studies listed above.

○ Check this box if you wish to opt out of being contacted for research studies.

Signature of Patient/Guardian (required)	**Date**
Signature of Relative A	**Date**
Signature of Relative B	**Date**

ACCOUNT INFORMATION

GeneDx Account Number		**Account Name**
Phone		Fax
Address		City
State	Zip Code	Country
Ordering Provider Name		Role/Title
NPI		Phone Number
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
Additional Reporting Provider's Name		
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
SEND ADDITIONAL REPORT COPIES TO:		
Provider Name		GeneDx Acct#
Fax #/Email:		

ICD-10 CODES *(Required)*

ICD-10 Codes	
Clinical Diagnosis	Age of Onset

STATEMENT OF MEDICAL NECESSISTY

By submission of this test requisition and accompanying sample(s), I: (i) authorize and direct GeneDx to perform the testing indicated; (ii) certify that the person listed as the ordering provider is authorized by law to order the test(s) requested; (iii) certify that any custom panel and/or ordered test(s) requested on this test requisition form are reasonable and medically necessary for the diagnosis and/or treatment of a disease, illness, impairment, symptom, syndrome or disorder; (iv) the test results will determine my patient's medical management and treatment decisions of this patient's condition on this date of service; (v) have obtained this patient's and relatives', when applicable, written informed consent to undergo any genetic testing requested; and (vi) that the full and appropriate diagnosis code(s) are indicated to the highest level of specificity.

Signature of Provider (required)	Date

PAYMENT OPTIONS *(Select One)*

○ **INSURANCE BILL** *(select all that applies)*

○ Commercial
○ Medicaid
○ Medicare
○ Tricare

FOR ALL INSURANCE CARDS PROVIDE FRONT AND BACK COPY OF CARD(S)

Patient Status
○ Hospital outpatient ○ Hospital inpatient; Date of Discharge _____
○ Not a hospital patient

Name of Insurance Carrier	Insurance ID#:
Relationship to Insured ○ Self ○ Spouse ○ Child ○ Other: _____	Policy Holder's Name Policy Holder's Date of Birth
Referral/Prior Authorization # (please attach)	GeneDx Benefit Investigation #

Secondary Insurance Type:

Insurance Carrier	Insurance ID #	Subscriber Name	Date of Birth

Relationship to Insured:
○ Self ○ Spouse ○ Child ○ Other: _____

○ **PATIENT BILL**

Amount Due: _____

If Patient Bill is selected, I am electing to be treated as a self-pay patient for this testing. I agree that neither GeneDx nor I will submit a claim to my insurance for this testing, if I have insurance. GeneDx will send an invoice to the patient listed above.

Authorized Patient/Guardian Signature

○ **INSTITUTIONAL BILL**

GeneDx Account #

Hospital/Lab Name

Place Sticker/Stamp Here

CLINICAL INFORMATION

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

Current Diagnosis: _____ Age of Onset: _____

○ Unilateral ○ Bilateral Disease Consanguinity: ○ Yes ○ No

Intraocular Pressure: _____ ERG Results: _____

Audiogram (dB): Left_____ Right_____

Eye/Vision Abnormalities

- ○ Abnormality of Vision
- ○ Aniridia
- ○ Anophthalmia
- ○ Astigmatism
- ○ Blue sclerae
- ○ Cataracts
- ○ Coloboma
- ○ Corneal arcus
- ○ Ectopia lentis
- ○ Esotropia
- ○ External ophthalmoplegia
- ○ Hyperopia
- ○ Hypoplasia of the fovea
- ○ Keratoconus/Anterior Lenticonus
- ○ Microphthalmia
- ○ Myopia
- ○ Nystagmus
- ○ Optic Atrophy
- ○ Photophobia
- ○ Ptosis
- ○ Retinal detachment
- ○ Retinitis pigmentosa
- ○ Strabismus
- ○ Visual impairment

Craniofacial/Dysmorphism

- ○ Abnormal facial shape (Dysmorphic features)
- ○ External ear malformation
- ○ Macrocephaly
- ○ Microcephaly

Developmental/Behavioral

- ○ Absent speech
- ○ Delayed fine motor development
- ○ Delayed gross motor development
- ○ Delayed speech & language development
- ○ Failure to thrive
- ○ Incoordination
- ○ Intellectual disability

Renal

- ○ Renal cysts
- ○ Other renal: _____

Hearing Impairment

- ○ Abnormal newborn screen: _____
- ○ Aminoglycoside-induced hearing loss
- ○ Conductive hearing impairment
  - ○ bilateral ○ unilateral
- ○ Enlarged Vestibular Aqueduct
- ○ Hearing impairment, mixed or unknown
  - ○ bilateral ○ unilateral
- ○ Morphological Abnormality of the Inner Ear
- ○ Sensorineural hearing impairment
  - ○ bilateral ○ unilateral
- ○ Tinnitus

Hematologic or Immunologic Issues

- ○ Recurrent infections
- ○ Recurrent otitis media

Neurological Findings

- ○ Vocal cord paresis

Skin/Hair Findings

- ○ Allergic dermatitis
- ○ Anhidrosis/Hypohidrosis
- ○ Cutaneous photosensitivity
- ○ Dermatitis
- ○ Hypopigmentation of the skin
- ○ Ichthyosis
- ○ Skin fragility/blistering
- ○ Sparse hair

Include additional clinical information:

_____

_____

_____

_____

_____

_____ _____

Signature of Provider (required) Date

OPHTHALMOLOGY/AUDIOLOGY TEST REQUISITION FORM

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REASON FOR EXPEDITED TESTING (REQUIRED)

○ Pregnancy (gestational age____weeks) ○ Transplantation ○ Other:_____

TARGETED VARIANT TESTING AND SPECIAL SERVICES

Individual to be tested: ○ Affected/Symptomatic ○ Unaffected/Asymptomatic

○ Known Familial Variant(s) in a Nuclear Gene	○ Targeted Mosaic Variant Testing **(Insurance Billing NOT Accepted; Patient Bill or Institutional Bill MUST be selected on page 1)**
○ Known Familial Copy Number variant(s)	○ Known mtDNA Variant(s) Testing
○ Confirmation of Variant Identified in Research Lab	

Proband Name: _____ Relationship to Proband: _____ Proband GeneDx Accession #:_____

Non-GeneDx Test: ○ Family member test report included (recommended if previous test was performed at another lab)
○ Positive control included/will be sent - **Positive control is recommended if previous test was performed at another lab.**
○ Positive control not available (caveat language will be included on a negative report)

Variant Information (please fill out the below information if family member report is not included)

Number of Variants:_____

Gene: _____ Coding DNA (c.): _____ Amino Acid (p.): _____ Transcript (NM#): _____

Gene: _____ Coding DNA (c.): _____ Amino Acid (p.): _____ Transcript (NM#): _____

Copy Number Variants (CNV(s) require coordinates and genome build or transcript # and exon #)

Number of Variants:_____

Gene(s):_____ Exon #:_____ Coordinates:_____ Genome Build: _____

Gene(s):_____ Exon #:_____ Coordinates:_____ Genome Build: _____

CUSTOM DEL/DUP TESTING

○ 906	Deletion/Duplication Analysis of ONE nuclear gene	○ 703	Deletion/Duplication Analysis of 2-20 nuclear genes

Write in desired gene(s) to be tested:_____

WRITE-IN TEST SELECTION

○ Test Code:_____	Test Name: _____

HISTORY

FAMILY HISTORY

○ No Known Family History ○ Pedigree Attached ○ Adopted

RELATIONSHIP TO INDIVIDUAL TO BE TESTED	MATERNAL	PATERNAL	RELEVANT HISTORY	AGE AT DX
	○	○		
	○	○		
	○	○		

TESTING HISTORY

○ Test report included (recommended)

Other relevant results (clinical, laboratory/biochemical or research):_____

OPHTHALMOLOGY/AUDIOLOGY TEST REQUISITION FORM

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HEARING LOSS TESTS

TEST CODE	# GENES	GENE LIST
○ J806	149	*ABHD12, ACTB, ACTG1, ADCY1, AIFM1, ALMS1, ANKH, ATP6V1B1, BDP1, BSND, CABP2,CACNA1D, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CHD7, CIB2, CLDN14, CLIC5,CLPP, CLRN1, COCH, COL2A1, COL11A1, COL11A2, COL4A3, COL4A4, COL4A5,COL4A6, CRYM, DCDC2, DFNA5, DIABLO, DIAPH1, DIAPH3, DNMT1, DSPP, EDN3,EDNRB, ELMOD3, EPS8, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FGFR1, FGFR2,FGFR3, FOXI1, GATA3, GIPC3, GJA1, GJB2, GJB3, GJB6, GPR98, GPSM2, GRHL2,GRXCR1, HARS, HARS2, HGF, HOMER2, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10,KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LRTOMT, MARVELD2, MCM2, MIR96, MITF,MSRB3, MT-RNR1^, MT-TL1^, MT-TS1^, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A,NDP, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15,PDZD7, DFNB59, PMP22, PNPT1, POU3F4, POU4F3, POLR1D, PRPS1, PTPRQ, RDX,S1PR2, SALL1, SEMA3E, SERPINB6, SIX1, SIX5, SLC17A8, SLC26A4, SLC26A5, SLC33A1,SLITRK6, SMPX, SNAI2, SOX10, SOX2, STRC (del/dup only), SYNE4, TBC1D24, TBX1^, TCOF1, TECTA,TIMM8A, TFAP2A, TJP2, TMC1, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C,USH1G, USH2A, WFS1, WHRN*
○ TA49	2	*GJB2/GJB6* common deletions
○ 130	1	*GJB2 (Cx26)^*
○ 157	1	*GJB6 (Cx30)^*
○ 572	1	*SLC26A4^*

OPHTHALMOLOGY XPANDED PANELS

TEST CODE	TEST NAME	# GENES	TEST CODE	TEST NAME	# GENES
○ J894	Nystagmus Xpanded (Proband only or Trio)	~825	○ J905	Retinal Dystrophy Xpanded (Proband only or Trio)	~875

BIOLOGICAL PARENT SAMPLE INFORMATION

***ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS.**

Mother:	○ Not available	○ To be sent within 3 weeks*	○ At GeneDx	
First Name	Last Name	DOB	○ Asymptomatic	○ Symptomatic
Father:	○ Not available	○ To be sent within 3 weeks*	○ At GeneDx	
First Name	Last Name	DOB	○ Asymptomatic	○ Symptomatic
Other: Relationship:	○ Not available	○ To be sent within 3 weeks*	○ At GeneDx	
First Name	Last Name	DOB	○ Asymptomatic	○ Symptomatic

OPHTHALMOLOGY MULTI-GENE PANELS

TEST CODE	TEST NAME	# GENES	GENE LIST
○ J957	Anophthalmia and Microphthalmia Panel	23	*ALDH1A3, BCOR, BMP4, BMP7, COX7B, CRYBA4, FOXE3, GDF6, HCCS, MITF, NAA10, NDUFB11, OTX2, PAX6, PRSS56, RAX, SALL1, SHH, SIX6, SOX2, STRA6, TENM3, VSX2 (CHX10)*
○ J958	Cataracts Panel	86	*ABCA3, ADAMTSL4, AGK, AKR1E2, ALDH18A1, BCOR, BEST1, BFSP1, BFSP2, CHMP4B, COL11A1, COL2A1, COL4A1, COL4A2, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, CYP27A1, CYP51A1, EPG5, EPHA2, FAM126A, FOXC1, FOXE3, FTL, FYCO1, FZD4, GALK1, GCNT2, GFER, GJA1, GJA3, GJA8, HMX1, HSF4, JAM3, LIM2, LSS, LONP1, MAF, MIP, MIR184, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PITX2, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RECQL4, RGS6, RNLS, RRAGA, SC5D, SIL1, SIPA1L3, SIX6, SLC16A12, SLC33A1, TBC1D20, TDRD7, TFAP2A, TMEM70, UNC45B, VIM, VSX2, WDR87, WFS1, WRN*
○ J956	Cone-Rod Dystrophy Panel	31	*ABCA4, ADAM9, AIPL1, BEST1, C8ORF37, CABP4, CACNA1F, CDH3, CDHR1, CEP290, CERKL, CNGA3, CNGB3, CRX, DRAM2, ELOVL4, GUCA1A, GUCY2D, PAX6, PITPNM3, POC1B, PROM1, PRPH2 (RDS), RAB28, RAX2 (QRX), RDH5, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5*
○ J959	Congenital Stationary Night Blindness Panel	12	*CABP4, CACNA1F, CHM, GNAT1, GRM6, NYX, PDE6B, RDH5, RHO, RPE65, SAG,* and *TRPM1*
○ J955	Familial Exudative Vitreoretinopathy (FEVR) Panel	4	*FZD4, LRP5, NDP, TSPAN12*
○ J960	Glaucoma Panel	37	*ADAMTS10, ASB10 (GLC1F), BEST1, BMP4, COL4A1, COL8A2, CREBBP, CYP1B1, FBN1, FOXC1, FOXE3, GJA1, ISPD, LMX1B, LTBP2, MAF, MYOC, NTF4, OPA1, OPA3, OPTC, OPTN, PAX6, PIK3R1, PITX2, PITX3, POMT1, PRSS56, PXDN, RPS19, RRM2B, SBF2, SH3PXD2B, SIX6, TBK1, TMEM126A, TTR, WDR36.*
○ TB48	Hermansky-Pudlak Syndrome Panel	10	*AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6*
○ 577	Progressive External Ophthalmoplegia (PEO)/ Optic Atrophy Nuclear Gene Panel	44	*ACO2, AUH, C12orf65, CLPB, DGUOK, DNA2, DNAJC19, DNM1L, EARS2, FH, GYG2, ISCA2, MFF, MFN2, MGME1, MTFMT, MTO1, MTPAP, NARS2, NDUFAF3, NR2F1, OPA1, OPA3, PDHX, PDSS1, POLG, POLG2, RNASEH1, RRM2B, SLC19A2, SLC19A3, SLC25A4, SLC25A46, SPG7, SUCLA2, TAC01, TIMM8A, TK2, TMEM126A, TSFM, TWNK, TYMP, VARS2, WFS1*
○ 466	Stargardt Disease Panel	3	*ABCA4, ELOVL4* and *PRPH2 (RDS)*
○ TA02	Stickler Syndrome Panel	6	*COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2*
○ T006	Usher Syndrome Panel	10	*ADGRV1 (GPR98), CDH23, CLRN1, DFNB31 (WHRN), MYO7A, PCDH15, USH1C, USH1G, USH2A* and *PDZD7*
○ TH12	Leber Hereditary Optic Neuropathy (LHON) Panel		

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

OPHTHALMOLOGY/AUDIOLOGY TEST REQUISITION FORM

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

OPHTHALMOLOGY SINGLE GENE TESTS

TEST CODE	TEST NAME/GENE NAME	DISORDER LIST
○ TA84	*AIPL1*	Autosomal Dominant Cone-Rod Dystrophy Autosomal Recessive Leber Congenital Amaurosis
○ 3693	*BCOR*	Oculofaciocardiodental Syndrome
○ 370	*BCOR P85L* Variant	Microphthalmia Lenz Syndrome
○ TA87	*BEST1*	Best Vitelliform Macular Dystrophy Autosomal Recessive Bestrophinopathy Adult-onset Foveomacular Vitelliform Dystrophy Autosomal Dominant Retinitis Pigmentosa Autosomal Dominant Vitreoretinochoroidopathy
○ TB09	*CABP4*	Autosomal Recessive Congenital Stationary Night Blindness
○ TB06	*CACNA1F*	X-Linked Congenital Stationary Night Blindness
○ TA98	*CERKL*	Autosomal Recessive Cone-Rod Dystrophy Autosomal Recessive Retinitis Pigmentosa
○ 376	*CEP290^*	Autosomal Recessive Leber Congenital Amaurosis
○ TA66	*CHM*	Choroideremia
○ TA90	*CNGA1*	Autosomal Recessive Retinitis Pigmentosa
○ TB01	*CNGA3*	Achromatopsia; Autosomal Recessive Cone-Rod Dystrophy
○ TA99	*CNGB3*	Achromatopsia; Autosomal Recessive Cone-Rod Dystrophy
○ TA83	*CRB1*	Autosomal Recessive Leber Congenital Amaurosis
○ TA76	*CRX*	Autosomal Dominant Cone-Rod Dystrophy Autosomal Dominant Macular Degeneration Autosomal Dominant Retinitis Pigmentosa
○ TB18	*FOXC1*	Axenfeld-Rieger Syndrome Peter's Anomaly Rieger Syndrome Iris Hypoplasia
○ 604	*FOXE3^*	Anterior Segment Dysgenesis Developmental Eye Disorders
○ TA93	*FRMD7*	X-Linked Congenital Nystagmus
○ TB08	*GNAT1*	Axenfeld-Rieger Syndrome Peter's Anomaly Rieger Syndrome
○ TA95	*GUCA1A*	Autosomal Dominant Cone-Rod Dystrophy Autosomal Dominant Retinitis Pigmentosa
○ TA82	*GUCY2D*	Autosomal Recessive Leber Congenital Amaurosis
○ 189	*HPS3*	Hermansky-Pudlak Syndrome: Ashkenazi Splice Mutation
○ 188	*HPS1^, HPS3*	Hermansky-Pudlak Syndrome: Puerto Rican Mutations
○ TA88	*IMPDH1*	Autosomal Dominant Cone-Rod Dystrophy Autosomal Dominant Retinitis Pigmentosa
○ TB25	*LCA* Tier 6 *RPGRIP1*	Autosomal Recessive Leber Congenital Amaurosis
○ TB05	*NR2E3*	Enhanced S-Cone Syndrome; Goldmann-Favre Syndrome Autosomal Dominant Retinitis Pigmentosa
○ TA92	*NYX*	Autosomal Recessive Retinitis Pigmentosa
○ TB17	*PITX2*	Axenfeld-Rieger Syndrome Peter's Anomaly Rieger Syndrome
○ TB24	*PRPF3*	Autosomal Dominant Retinitis Pigmentosa
○ TA68	*PRPH2 (RDS)*	Autosomal Dominant Cone-Rod Dystrophy Autosomal Dominant Macular Degeneration Autosomal Dominant Retinitis Pigmentosa
○ TB50	*RB1*	Hereditary Retinoblastoma
○ TA91	*RDH5*	Autosomal Recessive Retinitis Pigmentosa
○ TA67	*RHO*	Congenital Stationary Night Blindness Autosomal Dominant Retinitis Pigmentosa
○ TB28	*RLBP1*	Bothnia Retinal Dystrophy; Fundus Albipunctatus Newfoundland Rod-Cone Dystrophy Retinitis Punctata Albescens
○ TA75	*RPE65*	Autosomal Recessive Leber Congenital Amaurosis
○ TA72	*RP2*	X-Linked Retinitis Pigmentosa
○ 2571	*RS1^*	Juvenile X-Linked Retinoschisis
○ TB02	*SAG*	Autosomal Recessive Congenital Stationary Night Blindness
○ TA96	*TRPM1*	Autosomal Recessive Congenital Stationary Night Blindness

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

INFORMED CONSENT

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

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 is Trio/Duo-based genetic testing?

For some genetic tests, including samples from the biological parents and/or other biological relatives along with the patient's sample can help with the interpretation of results. These tests are often referred to as "trio tests" since they typically include samples from the patient and both parents. Samples from relatives should be submitted with the patient's sample. Clinical information must be provided for the patient and any relative who submits a sample.

I understand that GeneDx will use the relative sample(s) when needed for the interpretation of my/my child's test results. The patient report may include clinical and genetic information about a relative when it is relevant to the interpretation of the results. Relatives do not receive an independent analysis of data nor a separate 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 complete gene lists, 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.

Specimen Retention

After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring heath care providers unless specific prior arrangements have been made.

I understand that samples from residents of New York State will not be included in the de-identified research studies described in this authorization and will not be retained for more than 60 days after test completion, unless specifically authorized by my selection. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language. No tests other than those authorized shall be performed on the biological sample.

Database Participation

De-identified heath history and genetic information can help health care providers and scientists understand how genes affect human health. Though {I/my child} may not personally benefit, sharing this information helps health care providers to provide better care for their patients and researchers to make discoveries. GeneDx shares this type of information with health care providers, scientists and health care databases. No personal identifying information will be shared, as it will be replaced with a unique code.

Even though only a code is used for the reporting to the database, there is a risk that {I/my child} could be identified based on the genetic and health information that is shared. GeneDx believes that this is unlikely, though the risk is greater if I have already shared {my/my child's} genetic or health information with public resources, such as genealogy websites.

Recontact for Research Participation

Separate from the above, GeneDx may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases and to develop new treatments. If there are opportunities to participate in research relevant to the disorder in {my/my child's} family, and if I have consented for recontact, GeneDx may allow my healthcare provider to be recontacted for research purposes, such as the development of new testing, drug development, or other treatment modalities. In some situations, such as if my health care provider is not available, I may be contacted directly.

Any research that results in medical advances, including new products, tests or discoveries, may have potential commercial value and may be developed and owned by GeneDx or the collaborating researchers. If any individuals or corporations benefit financially from these studies, no compensation will be provided to {my/my child} or {my/my child's} heirs.