



GeneDx, Inc.

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Submission (Requisition) Form

Date Sample Obtained ____/____/____ mm/dd/yy

- Sample Type [] buccal brushes (must be GeneDx kits)
 [] blood in EDTA (lavender top – single tube of 1-5mL)
 [] skin punch biopsy, size _____mm
 [] DNA [concentration: _____µg/mL]
 [] fetal tissue: Specify type _____

Patient Name _____, _____, _____
Last name First name MI

Submitter's Patient Identifier(s) _____

Date of Birth ____/____/____ mm/dd/yy

Gender []Male []Female []Unknown

Patient Address _____ Apt _____
Number and Street
City ST Zipcode

Patient Phone Home () _____ - _____
Work () _____ - _____ ext _____

TESTS REQUESTED: Mark on PAGE 2 of this form

REPORTING ADDRESS

Physician/CGC _____
Address _____

Phone () _____ - _____
Fax *Important () _____ - _____
Beeper () _____ - _____
Email _____@_____

ADDRESS FOR DUPLICATE REPORT

Phone () _____ - _____
Fax () _____ - _____
Beeper () _____ - _____
Email _____@_____

PAYMENT: Submit the PAYMENT OPTIONS form

History: Please provide any applicable information

- Clinical diagnosis or family history, including any reason to be considered for preferential priority (scheduled surgery, pregnancy management, etc.).
- Relationship to & Name of other relatives submitted.
- Gestational age if pregnancy:
- ICD9 codes, if insurance submission is requested:

Tests Available

Periodic Fever Syndromes

- Familial Mediterranean Fever (MEFV)
- Familial Hibernian Fever / TRAPS (TNFRSF1A)
- Hyper- IgD Syndrome (MVK)
- Muckle-Wells /Familial Cold Urticaria/ NOMID (CIAS1)

Ectodermal Dysplasia syndromes

- X-linked hypohidrotic ED (EDA1 aka ED1, EDA)
- Autosomal rec/dom hypohidrotic ED (EDAR)
- Clouston syndrome (GJB6, connexin30)
- Ectrodactyly-ED-Clefting (TP63, p63)
- Hay-Wells (TP63, p63)

Congenital Ichthyoses

- Lamellar ichthyosis (TGM1)
- Congenital Recessive Ichthyosis (erythrodermic)
 - ALOX12B* *ALOXE3*, if 12B is neg *Both now*
- Lamellar ichthyosis Type 2 (N. African type) (ABCA12)
- Harlequin ichthyosis (ABCA12)
- Sjögren-Larsson syndrome (FALDH)
- Erythrokeratoderma variabilis (GJB3, GJB4)
- Epidermolytic hyperkeratosis (KRT1, KRT10)
- Ichthyosis bullosa of Siemens (KRT2e)
- Vohwinkel syndrome (GJB2; connexin26)
- Keratitis-Ichthyosis-Deafness (KID) (GJB2; connexin26)
- Netherton syndrome (SPINK5)
- Chanarin-Dorfman Syndrome (CGI-58/ABHD5)

Other Keratin Disorders

- Pachyonychia congenita
 - KRT16, KRT6a (PC1) KRT17, KRT6b (PC2)
- Epidermolytic PPK of Vörner (KRT9)
- Unna-Thost disease (KRT1, KRT16)
- White sponge nevus (KRT4, KRT13)
- Steatocystoma multiplex (KRT17)

Epidermolysis Bullosa

- Epidermolysis Bullosa, Simplex (KRT5, KRT14)
- Epidermolysis Bullosa, Dystrophica (COL7A1)
- Epidermolysis Bullosa, Junctional (Laminin 5)
 - Tier 1 (hot spots), if negative: LAMB3 full LAMC2 full*
 - With Pyloric Atresia (JEB-PA) (ITGB4)*

Disorders of the Immune System

- Autoimmune Lymphoproliferative Syn (ALPS; TNFRSF6)
- Chronic Granulomatous Disease
 - X-linked (CYBB) and common recessive (NCF1)
 - Other autosomal recessive (NCF2, CYBA)
- Severe Combined Immunodeficiency (autosomal recessive)
 - RAG1 and RAG2 deficiency (incl. Omenn Syndrome)
 - Jak3 Deficiency (JAK3)
 - X-linked agammaglobulinemia (BTK)
 - Leukocyte Adhesion Deficiency (ITGB2)
 - Autoimmune Polyendocrinopathy /APECED (AIRE)
 - Tier 1 only* *Tier 2, if tier 1 neg* *Both tiers now*

Hermansky-Pudlak Syn (HPS1 and/or HPS3)

- HPS1 and HPS3 Puerto Rican mutations*
- HPS3 Ashkenazi splice mutation*

Bone Marrow Failure Syndromes

- Cong. amegakaryocytic thrombocytopenia (MPL)
- Shwachman-Diamond Syndrome (SBDS)
- Congenital and cyclic neutropenia (ELA2)
- Diamond-Blackfan anemia (RPS19)
- Dyskeratosis Congenita, X-linked (DKC1)
- Dyskeratosis Congenita, Autosomal (hTR)

Other Hereditary Skin Disorders

- Gorlin syndrome (PTCH)
- Cowden syndrome (PTEN)
- Bannayan-Riley-Ruvalcaba syndrome (PTEN)
- Peutz-Jeghers syndrome (STK11)
- Carney Complex (PRKAR1A)
- Birt-Hogg-Dube Syndrome (FLCN)
- Darier Disease (ATP2A2)
- Hailey-Hailey Disease (ATP2C1)
- Familial Cutaneous Malignant Melanoma
 - CDKN2A/ p16* *CDK4* *Both genes*

Developmental Eye Disease

- Aniridia, other diagnoses (PAX6)
- Anophthalmia, microphthalmia (SOX2)
- Anophthalmia, microphthalmia (SIX6)
- CHARGE Syndrome (CHD7)

Hereditary Rickets

- X-linked dominant hypophosphataemia (PHEX)
- Autosomal dom. hypophosphataemia (FGF23)
- Auto rec. Vit. D dependent rickets (CYP27B1)

Familial Hyperparathyroid Syndromes/Endocrine Neoplasias

- Multiple endocrine neoplasia type 1 (MEN1, Menin)
- Multiple endocrine neoplasia type 2A or
- Familial Medullary Thyroid Carcinoma (RET)
- Multiple endocrine neoplasia 2B (RET)
- Hyperparathyroidism-Jaw Tumor Syndrome or Parathyroid carcinoma or Familial Isolated Hyperparathyroidism (HRPT2)
 - Tier 1* *Tier 2* *Entire HRPT2 gene now*
- Familial Hypocalciuric Hypercalcemia (CASR)
- Neonatal Severe Primary Hyperpara (CASR)
- Autosomal Dominant Hypocalcemia (CASR)
- Familial Isolated Hypoparathyroidism (CASR)

Alagille Syndrome (JAG1)

- Tier 1* *Tier 2 if Tier 1 is negative*

Coffin-Lowry syndrome (RSK2)

- Tier 1* *Tier 2 if Tier 1 is negative*

Hereditary multiple exostoses (EXT1 /EXT2)

- EXT1 only* *EXT2 if EXT1 neg* *Both now*

Noonan Syndrome (PTPN11)

- Exons 3,8/9,13 only* *Rest of gene (if tier 1 neg)*
- Entire PTPN11 gene now (tier 1 + tier 2)*
- Prenatal test for Noonan based on ultrasound*

Other Disorders

- Alexander Disease (GFAP)
- Allgrove (Triple-A) Syndrome (AAAS)
- Androgen Insensitivity Syndrome (AR)
- Cartilage-Hair Hypoplasia and assoc. (RMRP)
- Cardio-Facio-Cutaneous Syndrome (BRAF)
- CHARGE Syndrome (CHD7)
- Costello Syndrome (HRAS)
- Dent Disease/ X-linked rec nephrolithiasis (CLCN5)
- Dopa-Responsive Dystonia (GCH1)
- Fabry Disease (GLA)
- Hereditary angioedema (C1INH)
- Hirschsprung Disease (RET)
- Holt-Oram (TBX5)
- Holoprosencephaly (SHH, ZIC2, SIX3, TGIF)
- Inclusion Body Myopathy (GNE; *M712T only*)
- Insensitivity to pain and anhidrosis (NTRK1)
- Kallman Syndrome: *KAL1 gene* *FGFR1 gene*
- LEOPARD Syndrome (PTPN11, exons 7,12,13)
- Mucopolidosis Type IV (MCOLN1)
- Nemaline myopathy (ACTA1)
- Nemaline myopathy, auto recessive (*Nebulin gene; Ashkenazi Jewish mutation only*)
- Popliteal Pterygium Syndrome (IRF6)
- Pseudoachondroplasia/Mult Epiphyseal Dys (COMP)
- Smith-Lemli-Opitz Syndrome (DHCR7)
- Smith-Magenis Syndrome (RAI1)
- VanderWoude Syndrome (IRF6)
- X-linked Hydrocephalus (L1CAM)
- X-linked Retinoschisis (XLR5)
- XY Female Gonadal Dysgenesis (SRY sequencing)

Alternatively, the following special services are available for families with specific previously identified mutations. Check one per specimen.

- Confirmation of mutation identified elsewhere
- Carrier Detection in relatives
- Prenatal Diagnosis

Required Info: Gene Name _____

Mutation(s) _____

Relative's Name or GeneDx Accession #, if applicable:
