

DNA Analysis Request Form

→ Send this Request Form (pg. 1) + Test Checklist (pg. 2) + Consent Form (pg. 3) + Client Info Fax Form (pg. 4) ←
(+ Wire Transfer Form, if paying by wire transfer)

| | | | | | |
|--|--|---------------------------------|---|---|--|
| Patient's Last Name: _____ | | First: _____ | | MI: _____ | |
| Sex: M F | | DOB: ___/___/_____ (MM/DD/YYYY) | | Hospital/ID Number: _____ | |
| Referring Physician/Geneticist/Genetic Counselor | | | Billing information (must be complete) | | |
| Name(s): _____ | | | → Is full address for bill / receipt on the Fax Form? Y N | | |
| Institution name, location: _____ | | | Phone: (____)-_____ Fax: (____)-_____ | | |
| → Is full mailing + fax info for report on the Fax Form? Y N | | | ___ Institutional billing | | |
| Signature: _____ | | | ___ Prepay: check, money order, wire transfer; 5% discount | | |
| MD Phone: (____) - _____ | | Fax: (____) - _____ | | ___ Insurance, some plans: send #, copy of card, ins. details | |
| GC Phone: (____) - _____ | | Fax: (____) - _____ | | ___ Credit card*: send #, copy of card + associated address | |
| Email(s): _____ | | | * We prefer checks to credit cards if possible, please.* | | |
| Previous DNA studies | | | Please send clinical information and prior testing results on the patient and/or family members. | | |
| YES___ NO___ (if YES, specify) | | | Individual(s) studied: _____ | | |
| Analysis performed: _____ | | | Laboratory: _____ | | |
| Approximate date of study: _____ | | | | | |
| Additional family members to be studied / Relationship | | | | | |
| 1. _____ | | 2. _____ | | | |
| Indications for testing (please include ICD-9s): _____ | | | | | |
| Ethnic background (Important for accurate interpretation) | | | | | |
| ___ African American | | ___ S European Caucasian | | ___ NW European Caucasian | |
| ___ Hispanic | | ___ Native American Indian | | ___ Mixed European Caucasian | |
| ___ Ashkenazi Jewish | | ___ Other Jewish | | ___ Asian | |
| | | | | ___ New Zealand (Maori) | |
| | | | | ___ Other: _____ | |
| Genetic disorder to be studied: _____ | | | | | |
| → → Name of disorder / test(s) <u>and</u> → → Test number(s) from our <u>List of Tests</u> : _____ | | | | | |
| Please indicate pedigree (Any consanguinity / family members related to each other?) | | | | | |
| | | | | | |
| Analysis requested (mark all that apply) | | | | | |
| ___ Diagnostic | | ___ Carrier identification | | ___ Prenatal diagnosis | |
| ___ Sequencing | | ___ Specific known mutation(s) | | ___ Other _____ | |
| | | | | ___ Panel of mutations | |
| | | | | ___ Deletion | |
| Blood specimen information (EDTA purple top tubes, room temp): Parents' carrier testing free with child's test, for some. | | | | | |
| Sample sent for Mom? Y N | | Dad? Y N | | | |
| Patient: Time/date of sample collection _____ | | Approx. volume ___ml | | Number of tubes _____ | |
| Mom: Time/date of sample collection _____ | | Approx. volume ___ml | | Number of tubes _____ | |
| Dad: Time/date of sample collection _____ | | Approx. volume ___ml | | Number of tubes _____ | |
| Prenatal information: Maternal cell contamination study (MCC, test # 7), is performed together with all prenatal testing. Send maternal blood sample unless stored maternal DNA is available from previous testing. MCC sent? Y N | | | | | |
| Amniotic fluid: ___ml | | Chorionic villi sampling: ___mg | | Cultured cells: ___ T25s | |
| LMP ___/___/___ | | Date of U/S ___/___/___ | | Twin gestation? Y N | |
| | | | | Gestation by U/S _____ | |
| | | | | Sex of fetus (if known) M F | |

Thank you for your business! Any questions?

Laboratory Director: Dr. Elaine B. Spector, PhD, FACMG; 303-724-3801, Elaine.Spector@UCDenver.edu

Assistant Director: Dr. Gunter Scharer, MD, FACMG; 303-724-1571, Scharer.Gunter@tchden.org

Dr. John (Pei-Wen) Chiang, PhD: 303-724-3805, Pei-Wen.Chiang@UCDenver.edu, for tests marked with an *asterisk*

Genetic Counselor / Billing: Sarina Kopinsky, MS, CGC; 303-724-1572, Sarina.Kopinsky@UCDenver.edu

CHECKLIST OF TESTS ORDERED

Each Sample Needs a Request Form + Consent Form + Client Information Fax Form + This Checklist

→→ Please Circle Name of Disorder / Test(s) and Test Number(s) ←←

Name of patient: _____

DOB: ____/____/____ (MM / DD / YYYY)

| Test # | Name of Disorder / Test | \$ |
|---|--|------|
| 1 | DNA Isolation | 75 |
| 2 | Shipping: \$50 plus all shipping costs | 50+ |
| 3 | Carrier test (for many disorders listed): 2 known mutations | 250 |
| 4 | Carrier test (for many disorders listed): 1 known mutation | 250 |
| 5 | Prenatal test (for many disorders listed): 2 known mutations | 800 |
| 6 | Prenatal test (for many disorders listed): 1 known mutation | 500 |
| 7 | MCC, Maternal Cell Contamination Study (for all prenatal testing) | 200 |
| Ashkenazi Panels + Cystic Fibrosis | | |
| 8 | Ashkenazi Comprehensive: ASH1 + ASH2 + ASH3 + CF | 590 |
| 9 | ASH1, Ashkenazi Panel 1: Tay-Sachs; Canavan; Fanconi; Familial Dysautonomia | 200 |
| 10 | ASH2, Ashk. Panel 2: Niemann-Pick; Mucopolipidosis; Bloom; Glycogen Storage Ia | 200 |
| 11 | ASH2-combo, Ashkenazi Panel 2 ordered with other ASH | 120 |
| 12 | ASH3, Ashkenazi Panel 3: Gaucher Disease | 200 |
| 13 | ASH3-combo, Ashkenazi Panel 3 ordered with other ASH | 120 |
| 100 | ASH4, Ashkenazi Panel 4: MSUD, Maple Syrup Urine Disease – COMING SOON | |
| 101 | ASH4-combo, Ashkenazi Panel 4, ordered with other ASH – COMING SOON | |
| 14 | CF, Cystic Fibrosis, CFTR-Related Disorders | 250 |
| 15 | CF-combo, Cystic Fibrosis ordered with ASH panels or with test #46, FX | 150 |
| Cardiomyopathy Disorders | | |
| 16 | ARVD comprehensive , Panels A+B ordered together | 2000 |
| 17 | ARVD Panel A: ARVD9, PKP2 | 1400 |
| 18 | ARVD Panel B: ARVD8+10+11, ordered together | 1400 |
| 19 | ARVD8, DSP, ordered alone | 525 |
| 20 | ARVD10, DSG2, ordered alone | 1000 |
| 21 | ARVD11, DSC2, ordered alone | 1060 |
| 22 | Danon Disease (LAMP2, Glycogen Storage IIB) | 650 |
| 23 | Lamin A/C (LMNA): LMNA-Related Dilated Cardiomyopathy: Autosomal Emery-Dreifuss Muscular Dystrophy; Limb-Girdle Muscular Dystrophy 1B; Familial Partial Lipodystrophy Dunnigan type; Charcot-Marie-Tooth 2B1; Hutchinson-Gilford Progeria; Mandibuloacral Dysplasia | 750 |
| Clotting Disorders | | |
| 24 | FV, Factor V (five) Leiden Thrombophilia | 150 |
| 25 | MTHFR-AV, A233V: C677T (Thermolabile Variant) | 175 |
| 26 | MTHFR-EA, E429A: A1298C | 75 |
| 27 | PT, Prothrombin G20210 Thrombophilia (Factor II, FII or F2) | 125 |
| 28 | Warfarin / Coumadin Dosage Test (test discontinued) | |
| Deafness / Hearing Loss | | |
| 29 | Connexin 26, GJB2-Related DFNB1, sequence | 450 |
| 30 | Connexin 30, GJB6-Related DFNB1, common deletion | 350 |
| 31 | Pendred Syndrome, SLC26A4 | 1100 |
| 32 | Waardenburg Comprehensive , all 4 genes ordered together | 2400 |
| 33 | Waardenburg syndrome 1, 3, CDHS: PAX3, ordered alone | 650 |
| 34 | Waardenburg syndrome 2, Tietz: MITF, ordered alone | 1000 |
| 35 | Waardenburg syndrome 4: SOX10, ordered alone | 400 |
| 36 | Waardenburg-Shah syndrome, EDNRB, ordered alone | 600 |
| Disorders of Sex Development | | |
| 37 | AIS Panel (Androgen Insensitivity Syndrome): AR+SRY+WT1 ordered together | 1500 |
| 38 | AR: Androgen Receptor, sequence | 1000 |
| 39 | SRY: XY Gonadal Dysgenesis, Y-linked | 250 |
| 40 | WT1-Related Disorders: Denys-Drash; Frasier; Wilms Tumor; Nephrotic Syndr. | 750 |
| FGFR3, Fibroblast Growth Factor Receptor 3 | | |
| 41 | Achondroplasia; Hypochondroplasia: Sequence exons 7, 10, 13, 15 | 450 |
| 42 | Thanatophoric Dysplasia, Types I and II: Sequence exons 7, 10, 13, 15, 19 | 450 |
| 43 | Muenke Syndrome: Sequence exon 7 for P250R mutation | 250 |
| 44 | Crouzon Syndrome with Acanthosis Nigricans: Sequence exon 10 for A391E | 250 |
| 45 | Saddan: Sequence exon 15 for K650M | 250 |
| 46 | FX, FMR-1: Fragile X Syndrome: FXTAS: Adult-Onset Tremor Ataxia Syndrome; POF: Premature Ovarian Failure → Note discount price for test #15, CF-combo ordered with FX, \$150 ← | 300 |
| 47 | Iron Storage Disorders: Hereditary Hemochromatosis | 150 |
| Limb / Heart Disorders | | |
| 48 | SALL1, Townes-Brock Syndrome | 950 |
| 49 | SALL4, Duane Radial Ray Syndrome | 950 |
| 50 | TBX5, Holt-Oram Syndrome | 950 |
| Metabolic Disorders | | |
| 52 | 3MCC Panel , 3-Methylcrotonyl-CoA Carboxylase Def: 3MCC A+B ordered together | 1500 |
| 53 | 3MCC-A (3MCCC1), ordered alone | 900 |
| 54 | 3MCC-B (3MCCC2), ordered alone | 800 |
| 55 | ANT, Antiquitin, Pyridoxine-Dependent Neonatal Seizures, ALDH7A1 | 1500 |
| 56 | GA I, Glutaric Acidemia Type 1, GCD | 525 |
| 102 | GA III, Glutaric Acidemia Type 3, C7orf110 – COMING SOON | |

| Test # | Name of Disorder / Test | \$ |
|-----------------------------------|--|------|
| 57 | GA II comprehensive , GA2, Glutaric Acidemia Type 2, all three genes | 2400 |
| 58 | GA II (MADD), ETFDH (also known as ETF-QO) | 1000 |
| 59 | GA II (MADD), ETFA | 850 |
| 60 | GA II (MADD), ETFB | 550 |
| 61 | HCS, Holocarboxylase Synthetase Deficiency | 725 |
| 62 | Homocystinuria due to CBS Deficiency, sequence CBS gene | 1200 |
| 63 | LCHAD, Long Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency, common mut. | 250 |
| 64 | MCAD, Medium Chain Acyl-CoA Dehydrogenase Deficiency, common mutation | 250 |
| 65 | MCAD, Medium Chain Acyl-CoA Dehydrogenase Deficiency, full sequence | 1000 |
| 66 | MMA, Methylmalonic Acidemia, Panel 1: MUT + A + B | 1500 |
| 67 | Methylmalonic Acidemia, MMA-MUT, ordered alone | 960 |
| 68 | Methylmalonic Acidemia, MMA-A, ordered alone | 480 |
| 69 | Methylmalonic Acidemia, MMA-B, ordered alone | 640 |
| 70 | MMA, Methylmalonic Acidemia, Panel 2: C + E | 500 |
| 71 | Methylmalonic Acidemia, MMA-CHC, ordered alone | 320 |
| 72 | Methylmalonic Acidemia, MCEE, ordered alone | 240 |
| 73 | NKH Tier 1 , Non-Ketotic Hyperglycinemia, AMT + GLDC | 2400 |
| 74 | NKH, sequence AMT only | 800 |
| 75 | NKH, sequence GLDC only | 1600 |
| 76 | NKH Tier 2 , Non-Ketotic Hyperglycinemia, sequence GCSH | 400 |
| 103 | POLG1-related disorders, sequence POLG1 – COMING SOON | |
| 77 | Propionic Acidemia A+B | 1800 |
| 78 | Propionic Acidemia due to PCCA Deficiency | 1100 |
| 79 | Propionic Acidemia due to PCCB Deficiency | 700 |
| 80 | SPR, Sepiapterin Reductase Deficiency | 400 |
| 81 | Trimethylaminuria, TMAU (coming soon, not available now) | 500 |
| 82 | VLCAD, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, sequence ACADVL | 725 |
| Pigmentation Disorders | | |
| 104 | CHS, Chediak-Higashi Syndrome, sequence CHS1 (also called LYST) | 1750 |
| 105 | c-KIT: Piebaldism; Mast Cell Leukemia; Gastrointestinal Stromal Tumor | 900 |
| 84 | HPS comprehensive , sequence HPS1+HPS2+HPS3+HPS4 ordered together | 3000 |
| 85 | HPS1, Hermansky-Pudlak Syndrome Type 1, sequence | 1700 |
| 83 | HPS1, Hermansky-Pudlak Syndrome Type 1, common mutation only | 250 |
| 116 | HPS2, Hermansky-Pudlak Syndrome Type 2, sequence | 1700 |
| 117 | HPS3, Hermansky-Pudlak Syndrome Type 3, sequence | 1700 |
| 86 | HPS4, Hermansky-Pudlak Syndrome Type 4, sequence | 1700 |
| | HPS 5/6/7/8, Hermansky-Pudlak Syndrome Types 5-8, sequence | |
| | OCA-OA Panel: Max price for all OCA1-4 + OA1 testing needed by patient | 2500 |
| 87 | OCA1, Oculo-Cutaneous Albinism, Type 1a/1b, sequence TYR | 1000 |
| 88 | OCA2, Oculo-Cutaneous Albinism, Type 2, sequence P-gene | 1500 |
| 89 | OCA2, Oculo-Cutaneous Albinism, Type 2, P-gene deletion | 350 |
| 90 | OCA3, Oculo-Cutaneous Albinism, Type 3, sequence TYRP1 | 1000 |
| 91 | OCA4, Oculo-Cutaneous Albinism, Type 4, sequence MATP | 1000 |
| 92 | OA1, X-linked Ocular Albinism, sequence GPR143 | 800 |
| Syndromes – Various other | | |
| Aicardi-Goutieres Syndrome | | |
| 93 | AGS Comprehensive: Aicardi-Goutieres Syndrome, all 4 genes ordered together | 1650 |
| 94 | AGS Tier 1 , AGS 1,5+2: TREX1 + RNASEH2B together (65% of mutations) | 900 |
| 95 | AGS Tier 2 , AGS 3+4: RNASEH2C + RNASEH2A ordered together | 900 |
| 96 | AGS Type 1.5: TREX1-Related Aicardi-Goutieres Syndrome, ordered alone | 450 |
| 97 | AGS2, RNASEH2B-Related, Type 2, ordered alone | 650 |
| 98 | AGS3, RNASEH2C-Related, Type 3, ordered alone | 500 |
| 99 | AGS4, RNASEH2A-Related, Type 4, ordered alone | 600 |
| 106 | Joubert syndr. , comprehensive, CEP290 + RPGRIP1L + TMEM67 – SOON | |
| 107 | CEP290, Joubert syndrome (this gene is also included in test # 114, LCA) | 1800 |
| 108 | RPGRIP1L, Joubert syndrome – COMING SOON | |
| 109 | TMEM67, Joubert syndrome (for now, free with CEP290) | |
| 110 | PTEN-Related Disorders , sequence PTEN: Macrocephaly / Autism Syndrome; PTEN Hamartoma Tumor Syndrome (PHTS); VACTERL Association with Hydrocephalus | 950 |
| 111 | Rubinstein-Taybi Syndrome , comprehensive, CREBBP + EP300 | 2500 |
| 112 | CREBBP ordered alone, Rubinstein-Taybi Syndrome | 1500 |
| 113 | EP300 ordered alone, Rubinstein-Taybi Syndrome | 1500 |
| Vision Loss / Blindness | | |
| 114 | LCA Panel, Leber Congenital Amaurosis , sequence all 14 genes: AIPL1, CEP290, CRB1, CRX, GUCY2D, IMPDH1, LCA5, LRAT, RD3, RDH12, RPE65, RFGRI1, SPATA7, TULP1 | 2500 |
| 115 | adRP, Retinitis Pigmentosa, autosomal dominant panel , sequence all 18 genes: CA4, CRX, FSCN2, GUCATB, IMPDH1, NR2E3, NRL, PRPF3, PRPF8, PRPF31, PRPH2/RDS, RDH12, RHO, ROM1, RP1, RPP9, SEMA4A, TOPORS | 2500 |

Laboratory Director: Prof. Elaine Spector, PhD, FACMG;
Tel: 303-724-3801; Fax: 303-724-3802; Elaine.Spector@UCDenver.edu;
www.UCDenver.edu/DNALab

Consent Form

| | | | | | |
|---|--|---------------------------------|--|--------------|--|
| Patient's Last Name: _____ | | First Name: _____ | | MI: _____ | |
| Hospital/ID Number: _____ | | DOB ____/____/____ (MM/DD/YYYY) | | Sex: M__ F__ | |
| If patient is a minor: Guardian's Name(s) and relationship to patient: _____ | | | | | |
| Patient's full mailing address + zip, including name(s) to use with this address _____ | | | | | |
| Phone, H: _____ W: _____ ext. ____ Mobile/Pager: _____ (Pls. circle preferred #) | | | | | |
| Email address(es): _____ | | | | | |
| → → I request DNA analysis for: _____ (genetic condition) → → TEST NUMBER(S): _____ The intended purpose is: <input type="checkbox"/> Diagnostic <input type="checkbox"/> Carrier identification <input type="checkbox"/> Prenatal diagnosis <input type="checkbox"/> Other <input type="checkbox"/> Sequencing <input type="checkbox"/> Specific known mutation(s) <input type="checkbox"/> Panel of mutations <input type="checkbox"/> Deletion | | | | | |
| <p>I give my consent to have my sample(s) sent to the UCD DNA Diagnostic Laboratory for DNA testing for the above-designated genetic condition(s) / test number(s). I have discussed the principles, benefits and risks of this testing with a physician / geneticist / genetic counselor, and I have had my questions answered. I understand the following benefits, risks and limitations:</p> <ol style="list-style-type: none"> While DNA testing is a valuable diagnostic tool, it may not always give a definite answer about the genetic status of an individual. More specific information will be reported with the results of the test. Results will be sent to the referring healthcare provider / facility. This DNA test is specific <i>only for the condition(s)/ test(s) named above.</i> While mutation analysis often gives precise information, there are several possible sources of error. These include but are not limited to clinical misdiagnosis of the condition, sample misidentification, incorrect paternity identification, and sample contamination. The test is complex. It is not FDA approved. It uses some reagents produced for research purposes only. There is always a possibility that a diagnostic error may occur. Also, the laboratory may have difficulties analyzing my sample and a second sample may be requested. In the unlikely event that the test fails to produce a result, a repeat test will usually be offered at no extra charge. The test may reveal previously unrecognized biological relationships, such as non-paternity. DNA tests may also reveal a genetic condition in another family member. After the DNA testing of my sample is completed, the DNA may be used for medical research or test development. → → Please check here YES__ NO__ ← ← Refusal to permit use of my sample for research will not affect this test procedure. I am free to withdraw this consent at any time without prejudice to future care. I can withdraw my consent by contacting the laboratory director. → → I understand there will be a fee for this DNA testing _____ (signature) ← ← DNA testing may involve emotional stress and may result in discrimination (insurance- or work-related). The results of this testing will be treated in the standard manner to ensure medical confidentiality. The laboratory is obligated to release test results to my insurance provider or other payer if the provider / payer asks for them in order to pay for the test. Follow-up genetic counseling is available. I can contact the laboratory director, Dr. Elaine Spector, PhD, FACMG, at (303) 724-3801, Elaine.Spector@UCDenver.edu, for information about the test or genetic counseling. I can decide not to receive the results of the test, but I will still be responsible for the cost of the test. In the event of physical injury resulting from this procedure the University of Colorado Denver School of Medicine is not able to offer financial compensation or to absorb the cost of medical treatment. However, necessary facilities, emergency treatment and professional services will be available just as they are to the community generally. Any disputes that may arise in relation to the DNA testing shall be governed by the laws, rules and regulations of the State of Colorado, as are now in effect or as may be later amended or modified, without reference to the choice of law or rules of any state. I submit to the exclusive jurisdiction and venue of any court having subject matter jurisdiction located in the City and County of Denver, State of Colorado, including the United States District Court for the District of Colorado, in the event of any litigation concerning the DNA testing, regardless of where this consent is executed or where I reside. | | | | | |
| Name of Physician / Geneticist / Genetic Counselor: _____ | | | | | |
| Statement by Physician / Geneticist / Genetic Counselor: I have explained DNA testing to this person. I have addressed the limitations outlined above and have answered his / her questions. Signature: _____ Date: _____ | | | | | |
| Patient or Legal Guardian who is signing consent, Printed Name: _____ Signature: _____ Date: _____ | | | | | |
| Person who is witnessing the consent, Printed Name: _____ Signature of Witness: _____ Date: _____ | | | | | |

