Denver Genetic Laboratories **UCD DNA Diagnostic Laboratory** **www.DenverGenetics.org**
Complete Genetic Solutions™ **Phone: (303) 724-3801 Fax: (303) 724-3802**
Ship specimens to: Room P18-4404K, 12800 E. 19th Ave., MS 8313, Aurora, CO 80045

DNA Analysis Request Form

Complete all 4 pages: Request Form + Test Checklist + SIGNED CONSENT + Client Info Fax Form
Unlabeled specimens will not be processed.

Patient's Last Name:_____ First: _____ MI:_____

Sex: M F DOB: _____/_____/_____ (MM / DD / YYYY) Hospital/ID Number: _____

Genetic disorder to be studied: _____

→ → Name of Test(s) and Test number(s) from our List of Tests: _____

_____

Referring Physician / Geneticist / Genetic Counselor

Name(s): _____
Institution name, location: _____
Signature: _____
MD Phone: (___) - _____ Fax: (___) - _____
GC Phone: (___) - _____ Fax: (___) - _____
MD Email: _____
GC Email: _____

Billing information (must be complete)

Phone: (____)-_____ Fax: (___)-_____
→ Is SIGNEDCONSENT FORM included (Pg 3)? ___
→ Is info included for Report & Bill/Receipt (Pg 4)? __
___ Institutional billing
___ Prepay by check, 5% discount
___ Insurance (copy of card, preauth + member's address)
___ Credit card (send card info + associated address)

Previous DNA studies in patient or family

YES ___ NO ___ (if YES, specify)
Analysis performed: _____
Approximate date of study: _____

Please send clinical information and prior testing results on the patient and/or family members.

Individual(s) studied: _____
Laboratory: _____

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 __ Mixed European Caucasian
__ Hispanic __ Native American Indian __ Asian __ New Zealand (Maori)
__ Ashkenazi Jewish __ Other Jewish __ Other: _____

Please indicate pedigree – add extra page if necessary (Any consanguinity / family members related to each other?)

Analysis requested (mark all that apply) __ First test ordered __ Add-on test, stored sample __ Add-on test, new sample
__ Diagnostic __ Carrier identification __ Prenatal diagnosis __ Other _____
__ Sequencing __ Specific known mutation(s) __ Panel of mutations __ Deletion

Specimen information: Sample type: ___ Blood in purple top tube ___ DNA ___ Saliva Kit __ Other _____
Time/date of sample collection: _____ Approx. Vol. ___ml / Conc _____ Number of tubes _____

Amniotic fluid, direct ___ ml Amniotic cell culture ___ T25s CVS direct tissue, cleaned ___ mg CVS cell culture ___ T25s
DNA from amniotic fluid or cell culture ____ µl ____ (conc) DNA from direct or cultured CVS _____ µl _____ (conc)

Backup prenatal cell culture is being maintained at: _____ (location)
LMP __/__/____ Gestation _____ wks per U/S on (date) __/___/20___ Twin gestation? Y / N Sex of fetus: M / F / Unknown

Parents' carrier testing free with child's gene sequence, for some tests. Maternal cell contamination study (MCC, test #7) is performed with all prenatal tests. Send Mom's blood or DNA, unless stored sample is available from previous testing.

Paternal sample (patient's father) sent for carrier test? Y N Separate forms including CONSENT sent for Dad? Y N
Maternal sample (patient's mother) sent for carrier test? Y N Separate forms incl. CONSENT for Mom's carrier test ? Y N
Maternal sample sent for MCC? Y / N / Use stored sample Separate forms incl. CONSENT sent for Mom's MCC? Y N

Thank you for your business!

Laboratory Director: Prof. Dr. Elaine B. Spector, PhD, FACMG; 303-724-3801; Elaine.Spector@UCDenver.edu
Dr. Gunter Scharer, MD, FACMG: 303-724-1571; Scharer.Gunter@tchden.org
*Contact Dr. John Chiang, PhD, FACMG, for tests marked with an asterisk: 303-724-3805; Pei-Wen.Chiang@UCDenver.edu
Genetic Counselor: Sarina.Kopinsky@UCDenver.edu; 303-724-1572. Accountant: Lori.Gulliver@UCDenver.edu; 303-724-4670

CHECKLIST OF TESTS ORDERED

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

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

Name of patient: →→_____ **DOB: →→**___/___/_____ (MM / DD / YYYY)

Name of Disorder / Test	Test#	$$
DNA Isolation	1	75
Shipping: $50 plus all shipping costs	2	50+
Specific mutations (for most genes listed): **1-2 mutations**	3	250
Test #3P, 1-2 mutations: **PARENTS FREE** with child's gene sequencing	3P	$0
Specific mutations (**ANY GENE** not on our list): **1-2 mutations NEW!**	4	350
Prenatal test: 2 known mutations (also order test #7)	5	800
Prenatal test: 1 known mutation (also order test #7)	6	500
MCC, Maternal Cell Contamination Study (for all prenatal tests)	7	350
Ashkenazi Panels + Cystic Fibrosis (Expanded Panels in Development)		
ASH-Temp-NEW, Jewish mutations, 17 disorders: – NEW!! Tay-Sachs (+Tay-Sachs enzyme); Canavan; Fanconi; Familial Dysautonomia; Niemann-Pick; Mucolipidosis; Bloom; Gaucher; Glycogen Storage 1a; Usher 1F; Usher 3; Joubert 2; Nemaline Myopathy; Familial Hyperinsulinism; MSUD; MSUD 3; Cystic Fibrosis (39 mutations). **3 TUBES OF BLOOD: PURPLE + GREEN +RED**	#000	**1100** –5% = **1045**
ASH4: MSUD, Maple Syrup Urine Disease, ordered alone	100	350
CF, Cystic Fibrosis, CFTR-Related Disorders, 23-mutation panel, ordered alone	14	250
CF-combo, 23-mutation panel, **discount** when ordered with FX (#46)	15	150
ASH Comprehensive, More Disorders/More Genes in development		
ASH Gene SEQUENCING, in development		
ASH-Temp-OLD: 9 disorders**, Discontinued, Order #000 instead.**	00	~~590~~
Ashkenazi-Comprehensive-OLD: ASH-Temp-OLD (Discontinued) + ASH4	8	~~915~~
Ashkenazi Mutations: Original Panels ASH1, ASH2 & ASH3 discontinued.		
Cardiomyopathy Disorders		
ARVD comprehensive, Panels A+B ordered together	16	2000
ARVD Panel A: ARVD9, *PKP2*	17	1400
ARVD Panel B: ARVD8+10+11, ordered together	18	1400
ARVD8, *DSP*, ordered alone	19	525
ARVD10, *DSG2*, ordered alone	20	1000
ARVD11, *DSC2*, ordered alone	21	1060
Danon Disease (*LAMP2*, Glycogen Storage IIB)	22	650
Lamin A/C (*LMNA*): LMNA-Related Dilated Cardiomyopathy; Aut. Emery-Dreifuss MD; Limb-Girdle MD 1B; Famil. Partial Lipodystrophy – Dunnigan; Charcot-Marie-Tooth 2B1; Hutchinson-Gilford Progeria; Mandibuloacral Dysplasia	23	750
Clotting Disorders		
FV, Factor V (five) Leiden Thrombophilia	24	150
MTHFR-AV, A233V: C677T (Thermolabile Variant)	25	175
MTHFR-EA, E429A: A1298C	26	75
PT, Prothrombin *G20210* Thrombophilia (Factor II, FII or F2)	27	125
Deafness / Hearing Loss		
Connexin 26, *GJB2*-Related DFNB1, sequence	29	450
Connexin 30, *GJB6*-Related DFNB1, common deletion	30	350
Pendred Syndrome, *SLC26A4*	31	1100
Waardenburg Comprehensive, all 4 genes ordered together	32	2400
Waardenburg syndrome 1, 3, CDHS: *PAX3*, ordered alone	33	650
Waardenburg syndrome 2, Tietz: *MITF*, ordered alone	34	1000
Waardenburg syndrome 4: *SOX10*, ordered alone	35	400
Waardenburg-Shah syndrome, *EDNRB*, ordered alone	36	600
Disorders of Sex Development		
AIS Panel (Androgen Insensitivity Syndrome): ***AR+SRY+WT1*** ordered together	37	1500
AR: Androgen Receptor, sequence	38	1000
SRY: XY Gonadal Dysgenesis, Y-linked	39	250
WT1-Related Disorders: Denys-Drash; Frasier; Wilms Tumor; Nephrotic Syndr.	40	750
FGFR, Fibroblast Growth Factor Receptor Genes		
FGFR1, full sequence (Test is in development)	128	
FGFR2, full sequence (Test is in development)	129	
FGFR3		
FGFR3, full sequence (Test is in development)	130	
Achondroplasia; Hypochondroplasia: Sequence exons 7, 10, 13, 15	41	450
Thanatophoric Dysplasia, Types I and II: Sequence exons 7, 10, 13, 15, 19	42	450
Muenke Syndrome: Sequence exon 7 for P250R mutation	43	250
Crouzon Syndrome with Acanthosis Nigricans: Sequence exon 10 for A391E	44	250
Saddan: Sequence exon 15 for K650M	45	250
FMR1: Fragile X Syndrome; **FXTAS**, Adult-Onset Tremor Ataxia Syndrome; **FXPOI**, FX-Related Primary Ovarian Insufficiency (Note discount, #15, CF w. FX)	FX #46	$300
Iron Storage Disorder, **Hereditary Hemochromatosis:** *C282Y, H63D, S65C*	47	150
Limb / Heart Disorders		
SALL1, Townes-Brock Syndrome	48	950
SALL4, Duane Radial Ray Syndrome	49	950
TBX5, Holt-Oram Syndrome	50	950
Metabolic Disorders		
3MCC Panel, 3-Methylcrotonyl-CoA Carboxylase Def: 3MCC A+B ordered together	52	1500
3MCC-A (*3MCCC1*), ordered alone	53	900
3MCC-B (*3MCCC2*), ordered alone	54	800
ANT, Antiquitin, Pyridoxine-Dependent Neonatal Seizures, *ALDH7A1*– **NEW PRICE !!**	55	680
Biotinidase Deficiency, sequence *BTD* gene – NEW !!	126	450
GA1, Glutaric Acidemia Type 1, *GCD*	56	525
GA2 Comprehensive, GA2, Glutaric Acidemia Type 2, all three genes	57	2400
GA2 (MADD), *ETFDH* (also known as *ETF-QO*)	58	1000
GA2 (MADD), *ETFA*	59	850
GA2 (MADD), *ETFB*	60	550
GA3, Glutaric Acidemia Type 3, *C7orf10* – NEW !!	102	1100

Name of Disorder / Test	Test#	$$
HCS, Holocarboxylase Synthetase Deficiency	61	725
Homocystinuria (HCU) due to **CBS** Deficiency, sequence *CBS* gene	62	1200
LCHAD, Long Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency, common mut.	63	250
MCAD, Medium Chain Acyl-CoA Dehydrogenase Deficiency, common mutation	64	250
MCAD, Medium Chain Acyl-CoA Dehydrogenase Deficiency, full sequence	65	1000
MMA, Methylmalonic Acidemia		
MMA Panel 1: MUT + A + B (Tests #67 + #68 + #69 ordered together)	66	1500
MMA-MUT, ordered alone, Methylmalonic Acidemia	67	960
MMA-A, ordered alone, Methylmalonic Acidemia	68	480
MMA-B, ordered alone, Methylmalonic Acidemia	69	640
MMA-CHC, Cobalamin C, CblC, ordered alone, Methylmalonic Acidemia	71	320
MCEE, Epimerase, ordered alone, Methylmalonic Acidemia	72	320
NKH Tier 1, Non-Ketotic Hyperglycinemia/GlycineEnceph/Neonatal Sz, ***AMT+GLDC***	73	2400
NKH, sequence *AMT* only	74	800
NKH, sequence *GLDC* only	75	1600
NKH Tier 2, Non-Ketotic Hyperglycinemia, sequence ***GCSH***	76	400
Propionic Acidemia A+B	77	1800
Propionic Acidemia due to *PCCA* Deficiency	78	1100
Propionic Acidemia due to *PCCB* Deficiency	79	700
SPR, Sepiapterin Reductase Deficiency	80	400
Trimethylaminuria, *TMAU* (after pos. Biochem result) **– NEW!!**	81	600
VLCAD, Very Long Chain Acyl-CoA Dehydrogenase Deficiency, sequence *ACADVL*	82	725
Mitochondrial Disorders – NEW !!		
POLG1-Related disorders: Polymerase gamma, assoc. with mitochondrial disease	103	1500
Pigmentation Disorders		
CHS, Chediak-Higashi Syndrome, sequence *CHS1* (also called *LYST*)	104	1750
c-KIT: Piebaldism; Mast Cell Leukemia; Gastrointestinal Stromal Tumor	105	900
HPS Comprehensive, sequence **8 genes**, ordered together: **HPS1–HPS8 NEW!!**	84	3500
HPS1, Hermansky-Pudlak Syndrome Type 1, common PR mutation only	83	250
HPS3, Hermansky-Pudlak Syndrome Type 3, common PR mutation only	3	250
HPS3, Hermansky-Pudlak Syndrome Type 3, common Ashkenazi mutation only	3	250
OCA-OA Panel: Max price for all OCA1-4 + OA1 testing needed by patient		2500
OCA1, Oculo-Cutaneous Albinism, Type 1a/1b, sequence *TYR*	87	1000
OCA2, Oculo-Cutaneous Albinism, Type 2, sequence *P-gene*	88	1500
OCA2, Oculo-Cutaneous Albinism, Type 2, *P-gene* deletion	89	350
OCA3, Oculo-Cutaneous Albinism, Type 3, sequence *TYRP1*	90	1000
OCA4, Oculo-Cutaneous Albinism, Type 4, sequence *MATP*	91	1000
OA1, X-linked Ocular Albinism, sequence *GPR143*	92	800
Syndromes – Various Other		
Aicardi-Goutieres Syndrome – NEW !!		
AGS Comprehensive: Aicardi-Goutieres Syndrome, all **5 genes** ordered together	93	2300
AGS Tier 1: *TREX1* + *RNASEH2B*, #96 + #97 together (65% of mutations)	94	900
AGS Type 1: *TREX1*-Related, recessive and dominant forms, ordered alone	96	450
AGS2, *RNASEH2B*-Related, Type 2, ordered alone	97	650
AGS Tier 2, AGS 3+4: *RNASEH2C* + *RNASEH2A* ordered together	95	900
AGS3, *RNASEH2C*-Related, Type 3, ordered alone	98	500
AGS4, *RNASEH2A*-Related, Type 4, ordered alone	99	600
AGS Tier 3, AGS 5: *SAMHD1*-Related, Type 5, ordered alone **– NEW !!**	123	800
PTEN-Related Disorders, sequence *PTEN*: Macrocephaly / Autism Syndrome; PTEN Hamartoma Tumor Syndrome (PHTS); VACTERL Association with Hydrocephalus	110	950
Rubinstein-Taybi Syndrome, comprehensive, *CREBBP* + *EP300* ordered together	111	2500
CREBBP ordered alone, Rubinstein-Taybi Syndrome	112	1500
EP300 ordered alone, Rubinstein-Taybi Syndrome	113	1500
Vision Loss / Blindness / Eye Diseases		
ABCA4-Related Disorders, sequence *ABCA4*: Stargardt; arRP (autosomal recessive Retinitis Pigmentosa); Age-Related Macular Degeneration; CORD3	124	1750
Achromatopsia: *CNGA3*, Achromatopsia 2	119	595
CNGB3, Achromatopsia 3	120	850
GNAT2, Achromatopsia 4	121	595
PDE6C, Achromatopsia 5 (also part of #125)	126	850
Aniridia/ Anophthalmia: *PAX6* sequence **– NEW !!**	148	650
CEP290 sequence: **Joubert syndrome**; Bardet-Biedl; Meckel; Senior-Loken; LCA	107	1800
CSNB, Congenital Stationary Night Blindness – NEW !!		
CSNB, **X-Linked Panel**, ***NYX*** + ***CACNA1F*** ordered together	132	1200
CSNB, **X-Linked, Complete**, sequence ***NYX*** only	133	400
CSNB, **X-Linked, Incomplete**, sequence ***CACNA1F*** only	134	950
CSNB, **Non-X-Linked** Panel, **9 genes**: Aut.Dominant/Recessive/Oguchi	135	2500
adCone, Cone/Rod Dystrophy, CORD, **autosomal dominant**, **10** genes: *AIPL1, CRX, GUCA1A, GUCY2D, PITPNM3, PROM1, PRPH2, RIMS1, SEMA4A, UNC119*	122	2500
arCone, Cone/Rod Dystrophy, CORD, **autosomal recessive**, **10** genes: *ABCA4, ADAM9, CACNA2D4, CERKL, CNGB3, KCNV2, PDE6C, RAX2 (RAXL1), RDH5, RPGRIP1*	125	2500
LCA Panel, Leber Congenital Amaurosis, **17** genes*: AIPL1,* ***CABP4****, CEP290, CRB1, CRX, GUCY2D, IMPDH1, IQCB1, LCA5, LRAT,* ***OTX2****, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1*	114	2500
LCA, common **Jewish** mutation of the ***LCA5*** gene **– NEW !!**	3	250
adRP, Retinitis Pigmentosa, autosomal dominant, **21 genes**: *ASCC3L1, CA4, CRX, FSCN2, GUCA1B, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF8, PRPF31, PRPH2/RDS, RDH12, RHO, ROM1, RP1, RP9, SEMA4A, TOPORS, VMD2(BEST1)*	115	2500
Expanded panels in development: **arRP; Meckel; Joubert; Usher; Bardet-Biedl**		
Senior-Loken Syndrome, 4 genes: *CEP290, IQCB1,* ***NPHP1, NPHP4*** **– NEW !!**	150	2500

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Consent Form – Important

Note: If Consent Form is absent, incomplete or unsigned, our policy is to extract DNA and wait for the paperwork before running the test. Cancellation policy: $75 for DNA extraction.

Patient's Last Name: _____ **First Name:** _____ **MI:** ____

Hospital/ID Number: _____ **DOB** ____/____/_____ (MM / DD / YYYY) **Sex: M__ F__**

Guardian's Name(s) and relationship to patient (if patient is a minor): _____

Patient's full mailing address + zip (Please include 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:**

__ Diagnostic __ Carrier identification __ Prenatal diagnosis __ Other

__ Sequencing __ Specific known mutation(s) __ Panel of mutations __ Deletion

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:

1. 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.
2. This DNA test is specific ***only for the condition(s)/ test(s) named above.***
3. 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, & sample contamination.
4. 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.
5. The test may reveal previously unrecognized biological relationships, such as non-paternity. DNA tests may also reveal a genetic condition in another family member.
6. **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.
7. **→ → I understand there will be a fee for this DNA testing** _____ **(signature) ← ←**
8. 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.
9. 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 a referral to genetic counseling.
10. I can decide not to receive the results of the test, but I will still be responsible for the cost of the test.
11. 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.
12. 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.

A. 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: _____

B. Patient or Legal Guardian who is signing consent, Printed Name: _____

Signature of Consent: _____ **Date:** _____

C. Person who is witnessing the consent, Printed Name: _____

Signature of Witness: _____ Date: _____

UCD DNA Diagnostic Laboratory
Director: Prof. Dr. Elaine B. Spector, PhD, FACMG
Phone: 303-724-3801; Fax: 303-724-3802
www.DenverGenetics.org

CLIENT INFORMATION FAX FORM

→ → WE USE THIS CLIENT INFO PAGE AS THE FAX COVER SHEET TO SEND RESULTS BY FAX. ← ←

Complete this page to receive: A) Reports by Fax; B) Reports by Mail; C) Bill / Receipt

New Clients: WELCOME! Please complete the NEW CLIENT INFORMATION PAGE (complete once per client)

A) We will FAX REPORTS to names shown below.

PATIENT'S NAME: _____

→ → **Non-USA?** Please add exact, complete **dialing codes for country and city**. Thanks. ← ←

	FAX FROM DENVER GENETICS TO CLIENT:	Fax Date	# of Pages	
1 Name/Title:				
Institution:				
Phone:				
Fax:				
2 Name/Title:				
Institution:				
Phone:				
Fax:				
3 Name/Title:				
Institution:				
Phone:				
Fax:				

B) Exact mailing address for REPORT

ZIP _____

Country: _____

C) Exact mailing address for BILL/RECEIPT

ZIP _____

Country: _____

Ship samples to: Room P18-4404K, 12800 E. 19th Avenue, MS 8313, Aurora, CO 80045
Phone: (303) 724-3801 (main lab number); E-mail: Elaine.Spector@UCDenver.edu
Dr. Gunter Scharer, MD, FACMG; 303-724-1571; Scharer.Gunter@tchden.org
Contact Dr. John Chiang, PhD, FACMG; 303-724-3805; Pei-Wen.Chiang@UCDenver.edu, for tests marked *
Genetic counselor: Sarina.Kopinsky@UCDenver.edu; 303-724-1572 ~ Billing: Lori.Gulliver@UCDenver.edu; 303-724-4670