GENETICS CENTER
211 South Main Street, Orange, CA 92868

PHONE: 714-288-3500
FAX: 714-288-3510
www.geneticscenter.com

GC _____ DR _____
Depart _____
Verify By _____

Highlighted fields are required

MOLECULAR GENETICS REQUISITION

PATIENT INFORMATION

Patient Name: (Last, First)_____
Date of Birth: MM_____/DD_____/YY_____
Gender: ☐ Male ☐ Female ☐ Unknown
Ethnic Background (Select all that apply):

☐ African American ☐ Hispanic
☐ Asian ☐ Native American Indian
☐ Ashkenazi Jewish ☐ Other Jewish
☐ European Caucasian ☐ Other (please specify):_____

REFERRING PHYSICIAN

Physician: _____
Phone: _____
Fax:_____
Email: _____

ADDITIONAL REPORT RECIPIENTS

Physician: _____
Phone:_____
Fax:_____
Email: _____

SAMPLE INFORMATION

Date of Collection: MM_____/DD_____/YY_____
Hospital: _____
Accession #: _____
Sample Type: (Please select one)

☐ Blood ☐ Bone Marrow ☐ Other:_____
☐ Cord Blood ☐ Amniotic Fluid
☐ Tissue (specify source): _____
☐ DNA (specify source): _____
(DNA concentration): _____ ug/ul
☐ Tumor section:_____

INDICATION FOR STUDY

☐ Autism spectrum disorder ☐ Cognitive impairment
☐ Developmental delay ☐ Dysmorphic features
☐ Failure to thrive ☐ Short stature
☐ Family history of cognitive impairment
☐ Suspected thrombophilia
☐ Carrier screening for _____
☐ Congenital malformation (specify) _____
☐ Other _____

TARGETED ANALYSIS

Gene Name:_____
Name of Proband:_____ (please provide proband's test results)
Variant:_____
Transcript:_____

GC Lab #:_____
Relationship to relative:_____
Positive Control Sample: ☐ Sample Provided
☐ Sample Not Available ☐ Results Provided

Note: A copy of the familial result including transcript number and/or positive control are required for this testing. Please contact our lab for a collection kit.

☐ **Alport Syndrome** *(COL4A3, COL4A4, COL4A5)*

☐ **Angelman Syndrome Methylation Analysis**

☐ **Angelman Syndrome Methylation w/ reflex to FISH for 15q11 microdeletion**

☐ **Aortopathy Panel** *(ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, GLI3, MED12, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2)*

Array CGH (Microarray):

- ☐ 180K Oligonucleotide/SNP Array
- ☐ 60K Oligonucleotide Array
- ☐ Prenatal Targeted Oligo/SNP Array
- ☐ Targeted Parental Array (please include child's results)
- ☐ Chromosome Analysis with reflex to Array CGH

☐ **Ataxia Telangiectasia** *(ATM)*

☐ **Autosomal Dominant Polycystic Kidney Disease** *(PKD1, PKD2)*

☐ **Bannayan-Riley-Ruvalcaba Syndrome** *(PTEN)*

☐ ***Birt-Hogg-Dube Syndrome*** *(FLCN)*

CFTR-related Disorders:

- ☐ Cystic Fibrosis Targeted Mutation Panel
- ☐ Cystic Fibrosis CFTR gene sequence analysis
- ☐ Cystic Fibrosis CFTR gene deletion & duplication analysis

☐ **Congential Central Hypoventilation Syndrome** *(PHOX2B)*

☐ **Costello Syndrome** *(HRAS)*

Craniosynostosis Syndromes:

- ☐ Craniosynostosis Panel *(FGFR1, FGFR2, FGFR3, TWIST1)*
- ☐ Apert Syndrome
- ☐ Crouzon Syndrome
- ☐ Crouzon Syndrome with Acanthosis Nigricans
- ☐ Muenke Syndrome
- ☐ Non-Syndromic Craniosynostosis
- ☐ Pfeiffer Syndrome
- ☐ Saethre-Chotzen Syndrome

☐ **Denys-Drash Syndrome** *(WT1)*

☐ **DNA Extraction**

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Last:_____ **First:**_____ **DOB:**_____

MOLECULAR GENETICS REQUISITION *(page 2)*

Duchenne Muscular Dystrophy:

- ☐ DMD Deletion/duplication analysis
- ☐ DMD Sequencing

☐ **Early On-Set Familial Alzheimer Disease Panel** *(APP, PSEN1, PSEN2)*

☐ **Familial Adenomatous Polyposis (FAP) -related disorder** *(APC)*

☐ **Familial Hypercholesterolemia (FH) Panel** (APOB, LDLR, LDLRAP1,PCSK9)

☐ **Familial Mediterranean Fever** *(MEFV)*

☐ **Fragile X DNA Analysis**

Hearing Loss:

- ☐ Hearing Loss Panel (Connexin 26 and 30, *mt-RNR-1* and *mt-TS1*)
- ☐ Connexin 26 and 30 Targeted Mutation Analysis
- ☐ Connexin 26 Targeted Mutation Analysis
- ☐ Connexin 26 - *GJB2* Full Gene Sequence Analysis
- ☐ Connexin 30 Targeted Mutation Analysis
- ☐ Mitochondrial DNA Hearing Loss Panel *(mt-RNR1, mtTS1)*

☐ **HFE-related Hemochromatosis**

☐ **Hereditary Hemorrhagic Telangiectasia** *(ACVRL1, ENG, GDF2, RASA1, SMAD4)*

☐ **Hereditary Mismatch Repair Deficiency Syndrome** *(MLH1, MSH2, PMS2, MSH6)*

☐ **Huntington Disease** *(HTT)*

☐ **Legius/NF1-like Syndrome** *(SPRED1)*

☐ **Li-Fraumeni Syndrome** *(TP53)*

☐ **Marfan Syndrome** *(FBN1)*

☐ **MECP2 Sequencing (Rett Syndrome)**

☐ **Multiple Endocrine Neoplasia, Type 1** *(MEN1)*

☐ **Multiple Endocrine Neoplasia, Types 2A & 2B** *(RET)*

☐ **Myotonic Dystrophy DNA Analysis**

☐ **Neurofibromatosis, Type 1** *(NF1)*

☐ **Neurofibromatosis, Type 2** *(NF2)*

☐ **Nevoid Basal Cell Carcinoma Syndrome (Gorlin Syndrome)** *(PTCH1, SUFU)*

☐ **Noonan Panel** *(BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, R1T1, SHOC2, SOS1, SOS2)*

☐ **Osteogenesis Imperfecta** *(COLIA1, COLIA2)*

☐ **Pallister-Hall Syndrome** *(GL13)*

☐ **Pancreatitis Panel** *(CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1)*

☐ **Paternity/Identity Testing**

☐ **Peutz-Jeghers Syndrome** *(STK11)*

☐ **Prader-Willi Methylation Analysis**

☐ **Prader-Willi Methylation Analysis with reflex to FISH for 15q11 microdeletion**

☐ **PTEN-related disorder** *(PTEN)*

☐ **RASA1-related disorder** *(RASA1)*

☐ **Rasopathy Panel** *(BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, R1T1, SHOC2, SOS1, SOS2, SPRED1)*

☐ **Retinoblastoma** *(RB1)* (peripheral blood only)

☐ **Schwannomatosis** *(SMARCB1)*

☐ **Simpson-Golabi-Behmel Syndrome** *(GPC3)*

Skeletal Dysplasias:

- ☐ Achondroplasia
- ☐ Hypochondroplasia
- ☐ Achondroplasia/Hypochondroplasia Panel
- ☐ Thanatrophoric Dysplasia (types I and II)

☐ **Sotos Syndrome** *(NSD1)*

Spinal Muscular Atrophy (SMA):

- ☐ Carrier Testing (*SMNI* with intron 7 c.*3+80T>G SNP)
- ☐ Diagnostic Testing *(SMN1 & SMN2* with intron 7 c.*3+80T>G SNP*)*

☐ **Stickler Syndrome Panel** *(COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3)*

Thrombophilia/Obstetric Complication Panel:

- ☐ Thrombophilia Panel (Factor II and Factor V)
- ☐ Prothrombin (Factor II) Mutation Analysis
- ☐ Factor V Leiden Mutation Analysis

☐ **Tuberous Sclerosis** *(TSC1 & TSC2)*

☐ **Twin Zygosity**

☐ **Von Hippel-Lindau** *(VHL)*

☐ **Wilms Tumor** *(WT1)*

☐ **Other (please specify):**

_____

_____

_____

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PID

INFORMED CONSENT FOR MOLECULAR TESTING

I, _____, hereby authorize samples of blood or other specimens to be collected from me and/or members of my family for genetic testing for (name of disease) _____, using a molecular (DNA) test. In addition, if prenatal diagnosis is involved, I authorize fetal cells obtained by amniocentesis, chorionic villus sampling (CVS), cord blood, etc. to be used. I hereby give permission to collect blood, buccal and/or saliva samples from my minor children, named below, to be used for molecular testing for the disease listed above.

Child's Name	Date of Birth	Sex
_____	_____	_____
_____	_____	_____

I understand that:

The blood, saliva, buccal and/or fetal samples will be used for the purpose of attempting to determine if I or members of my family are carriers of the disease gene, or are affected with, or at increased risk to someday be affected with this genetic disease. It is highly recommended that you seek pre- and post-test genetic counseling to discuss the benefits, risks and limitations of this test.

Possible Results:
Your personal and family health history, other relevant laboratory tests, results of physical examination, and the clinical impression of your doctor should all be taken into consideration when interpreting the results of this test. Only final test results will be provided.

Testing may yield one of the following possible results:

- *Positive:* a mutation is found in a gene that is associated with a particular hereditary genetic condition. This may allow you to make informed decisions about your/your child's health as well as provide information for future family planning.
- *Negative:* no currently relevant mutations are identified in the genes tested. The likelihood of having a mutation in the genes tested is greatly reduced.
- *Variant of Unknown Significance*: a variant is identified, but it is currently unknown if the variant is associated with the condition that is being tested for.

These test results could have clinical or reproductive implications for you and/or your family members, which should be discussed with the appropriate healthcare provider. If you have a positive test result, you are recommended to discuss the result with your healthcare provider as well as receive genetic counseling to discuss the risk of your children and/or biological relatives inheriting the same mutation(s).

The US Genetic Information Nondiscrimination Act (GINA) of 2008 (Public Law 110-233) prohibits discrimination on the basis of genetic information with respect to health insurance and employment. However, GINA does not apply to life insurance, disability insurance, or long-term care insurance, which may be governed by state law. For information on GINA, visit http://www.genome.gov/10002328.

Participation in molecular testing is completely voluntary, and the results are confidential. Because of the complexity of DNA based testing and the important implications of the test, upon request, the results will be reported to me only through my physician, genetic counselor, or other health care specialist whom I have designated. The results will only be released to other medical professionals or other parties including insurance carriers with my written consent. Genetics Center is fully in compliance with all Health Insurance Portability and Accountability Act (HIPAA) and other relevant regulations.

Prenatal Samples:
In order to perform accurate prenatal diagnosis, blood samples may be required from the affected individual in the family, both parents of the fetus, and possibly from other family members. We request the submission of both a direct and a cultured fetal specimen (amniotic fluid or CVS) for each prenatal study. The final report for a fetal analysis will be sent only after the confirmation study is complete. This is a time consuming process and may take weeks prior to achieving results. Sometimes a definite diagnosis may not be made, and the results could be non-conclusive.

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Test Limitations:
An error in the diagnosis may occur if the true biological relationships of the family members involved in this study are not as I have stated. For example, nonpaternity means that the father of an individual is not the person stated to be the father. This test may detect nonpaternity, and it may be necessary to report this finding to the individual who requested testing.

Any incorrect diagnosis in a family member can lead to an inaccurate diagnosis for other related individuals. Generally, genetic testing is complex and are being improved and expanded continuously. This test is not considered research, but is considered diagnostic. It is possible that there are mutations or genetic aberrations that this testing technology is unable to detect. Knowledge of genetic information may improve over time, or new information may become available in the future, that could impact the interpretation of my results. There may be additional mutations and/or genes that other tests could cover and/or will be known in the future as genetic testing evolves. Testing utilizes specialized methods and materials, thus there is always a small possibility that the test may not work properly, or that an error will occur.

I understand that the DNA analysis performed at Genetics Center for this disease is specific only with respect to it. A negative result in no way guarantees my health or the health of my current children or fetus. The accuracy of DNA analysis is entirely dependent on the clinical diagnosis made, and Genetics Center cannot be responsible for erroneous clinical diagnosis made by others.

Other:
I understand that my genetic sample is not being banked. The laboratory does not return DNA samples or raw test data to individuals or physicians. However, in some cases it may be possible for the laboratory to reanalyze my remaining DNA (if available) upon request. The request for additional studies must be ordered by my referring physician/counselor, and there will be an additional fee.

Once my test result is completed, an aliquot of my DNA may be made anonymous (name and all other identifiers removed) and used for quality control or research purposes. No compensation will be given for any invention(s) resulting from the use of my DNA in research and development. You may refuse to have your specimen used in this way, and your refusal will in no way affect the present testing results. Please indicate your consent or denial below. If left blank, it will be assumed that you consent to the use of your DNA sample as described above.

☐ I consent to the use of my DNA for quality control or research purposes

☐ I *do not* consent to the use of my DNA for quality control or research purposes

Patient Consent:
My signature below acknowledges that my doctor, genetic counselor, or other health care specialist has explained the limitations and benefits of molecular testing to me and I have had the opportunity to ask questions I might have regarding this test. I have read this entire document, and I voluntarily give my consent for sample collection and genetic testing and acknowledge that I am ultimately responsible for payment.

Signature:_____ **Date**:_____

Witnessed By:_____

BILLING/INSURANCE INFORMATION
(Attach copy of insurance card front and back)

☐ Hospital/Institution ☐ HMO/PPO ☐ Patient/Insurance ☐ Medicare ☐ Payment Enclosed

Insurance Co._____

Billing Address_____

City, State, Zip_____

GENETICS CENTER

California HMO Medical Group Name_____

Name of Insured_____

Test Preauthorization no._____

Relationship to Patient: Self Spouse Child Other_____

Insured's Employer_____

Policy no._____ Group no._____

I hereby authorize Genetics Center to furnish my designated insurance carrier such information concerning my laboratory tests that is necessary for reimbursement. I also authorize benefits to be paid directly to Genetics Center. I understand that my insurance coverage is a contract between me and my insurance carrier, and I am responsible for any amount not paid by my insurance (including co-pays, unmet deductibles, lack of coverage, etc). The charges for these services are ultimately my responsibility. I permit a copy of this authorization to be used in place of the original.

Patient or Parent (or Guardian) Signature:_____ **Date:**_____