Transgenomic®
Advancing Personalized Medicine

LABS.TRANSGENOMIC.COM

TRANSGENOMIC • FIVE SCIENCE PARK • NEW HAVEN, CT 06511
Client Services: 1.877.274.9432 • Fax: 1.855.263.8668

Test Requisition and Statement of Medical Necessity

Date of Sample Collection: __ __ / __ __ / __ __ __ __

Affix Bar Code Sticker Here

PATIENT INFORMATION

Name: Last _____ First _____ MI _____

Address: Street _____ Apt # _____

City _____ State _____ Zip _____

Telephone: Daytime _____ Evening _____

Email _____

DOB: MM/DD/YY _____

☐ Male ☐ Female ☐ Inpatient ☐ Outpatient

ORDERING PHYSICIAN

Name: Last _____ First _____ Degree _____

Institution/Hospital _____

Address: Street _____ Suite # _____

City _____ State _____ Zip _____

Telephone _____ Fax _____

Email _____

NPI # _____ Group NPI # _____

Office Contact _____

ADDITIONAL HEALTHCARE PROFESSIONAL TO RECEIVE TEST RESULTS

Name: Last _____ First _____ Degree _____

Institution/Hospital _____

Address: Street _____ Suite # _____

City _____ State _____ Zip _____

Telephone _____ Fax _____

Email _____

BILLING INFORMATION

BILL: ☐ Facility/Contract Bill ☐ Patient Insurance ☐ Self-pay ☐ Government (Canada)

For Facility Accounts
NAME OF FACILITY ACCOUNT _____

For Patient Insurance
PRIMARY INSURANCE: Please provide a legible copy of both sides of the insurance card.

SECONDARY INSURANCE: You may submit secondary insurance information when applicable. Please provide a legible copy of both sides of the insurance card.

No testing is done without the patient's or legal guardian's permission, unless otherwise requested.

Primary Insurance Company _____

Address: Street _____

City _____ State _____ Zip _____

Telephone _____

Policy Holder/Subscriber _____

Relationship to Patient _____

Policy Holder's DOB _____ Policy Holder's SSN _____

Policy # _____

Group # (if applicable) _____

Name of Employer _____

PATIENT PAYMENT: Mail payments to: Transgenomic, PO Box 83236, Woburn, MA 01813-3236

ONLINE PAYMENT: https://transgenomic.sbgvt.com/payment.php

For Self-pay
Transgenomic accepts the following credit cards: Visa, MasterCard, AMEX and Discover

☐ Yes, I plan on using my credit card to pay for testing. Please contact me directly.

TEST MENU

Please see pages 3 and 4 of this form for the full menu of tests.

FAMILY SPECIFIC TESTS /INDIVIDUAL GENE TESTS

☐ **Family Specific Test Only**

Test _____

Name of Index Case _____

Relationship to the Index Case _____

☐ **Individual Genes**

To order analysis of individual genes, please indicate the name below.

_____

REQUIRED DIAGNOSIS/ICD-9 CODE(S)

Diagnosis/ICD-9 Code(s) _____

PRESENTING SIGNS/SYMPTOMS:

_____

_____

_____

_____

_____

_____

PLEASE ATTACH CLINICAL NOTES

SIGNATURES REQUIRED ON REVERSE SIDE.

Questions? Call Transgenomic at 877.274.9432.

Internal Use Only

Date Received _____ Tech _____

Tracking # _____

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Informed Consent for Genetic Testing

I have discussed the benefits, risks and limitations of genetic testing with my healthcare provider and have had my questions answered. By signing this form, I give my consent to have my sample and relevant clinical information sent to Transgenomic for testing. I also authorize Transgenomic to disclose the test results to the ordering physician and any other provider I designate.

I UNDERSTAND THE FOLLOWING BENEFITS, RISKS AND LIMITATIONS:

1. The results of this test may indicate that you are predisposed to or have an inherited condition. Follow-up genetic counseling is available to address any questions you may have regarding the results. Your physician may recommend additional testing. You can discuss this further with your healthcare provider.

2. While genetic testing is a valuable tool, it may not always give a definite answer about the genetic status of an individual. While some genetic variants are known to cause disease and others are benign, a proportion of genetic testing results are of uncertain significance.

3. No tests other than those authorized will be performed on your sample. Samples with New York State origin will be destroyed at the end of testing or no more than 60 days after the sample was taken, unless a longer period of retention is expressly authorized. If consent is given to retain your sample for test development or proficiency testing purposes, which may include genes other than those originally tested, any personal identifiers will be removed. Results will not be provided for any samples consented for use for test development or proficiency testing.

4. In rare circumstances, the laboratory may have difficulties analyzing your sample and a second sample may be requested. Genetic testing normally gives accurate information; rare sources of error include but are not limited to sample misidentification and sample contamination.

5. Genetic testing may involve emotional stress. The Genetic Information Nondiscrimination Act (GINA) of 2008 prohibits health insurance plans and employers from some discrimination based on genetic information, including the results of genetic testing. However, such genetic testing may result in life insurance, disability insurance and/or long-term care insurance discrimination that is not prohibited by law.

6. If other members of my family have had the same or similar tests, the results of this testing may suggest previously unrecognized biological relationships, such as non-paternity.

7. The results of this test will be kept confidential and will be released only to the physician(s) ordering the test or other persons authorized by you, in writing, unless otherwise required by Federal and state law.

8. The results of this test are not intended to be used as the sole means for diagnosis or management decisions.

9. By signing this consent, you give Transgenomic permission to retain the genetic information generated by this test and to contact your physician if Transgenomic learns new information about the genetic variants detected by this test that affects your reported test results. Transgenomic will make reasonable efforts to contact your physician in these instances. It is the responsibility of the patient to maintain current contact information with the healthcare provider so that the patient may be advised of any changes to their test results.

10. In the interest of advancing the understanding of these conditions, summary results from this test may be presented, for example at meetings, in publications, or on the Internet; however, no information that can identify you will ever be disclosed, unless authorized in writing by you or required by law.

11. There will be a fee for this genetic testing and you will be responsible for payment after the testing has begun, even if you decide not to receive results.

For Patient or Responsible Party Selecting the Patient Insurance Billing Option

12. I have selected the patient insurance billing option and hereby authorize Transgenomic to bill my insurance carrier. Further, I authorize Transgenomic to disclose to my insurance carrier the information on this form and any accompanying documentation provided by my healthcare provider. I authorize my health plan or insurance carrier, and other third parties involved in the administration of my plan, to disclose to Transgenomic information concerning my plan, including benefits, coverage limitations, and payments made for services. I authorize Transgenomic to release to Contractor or Carriers and any insurance carrier providing medical benefits to me and any health plan of which I am a member, any medical or other information including test results in connection with an appeal of a reimbursement denial or other reimbursement matter, if Transgenomic has made prior attempts to obtain reimbursement without the release of such information.

13. I hereby assign and authorize payment directly to Transgenomic of any benefits for the services provided. I understand that my insurance may not cover these services, or may only pay up to usual and customary rates, and that I am ultimately responsible for all costs of this test and costs of collections, including attorney fees, court costs, filing fees, and late payment fees, except where my liability is limited by contract or applicable state or Federal law.

SIGNATURES (REQUIRED)

HEALTHCARE PROFESSIONAL SIGNATURE TO AUTHORIZE TESTING AND STATEMENT OF MEDICAL NECESSITY:

I certify that the Informed Consent has been discussed with the patient or an individual legally authorized to do so on the patient's behalf (and that such form is on file), and that I obtained any other consent from the patient that is required under the laws of my state in order to perform a genetic test on a specimen. I further certify that the test ordered is medically necessary. The results of this test will be used in the medical management of the patient and/or genetic counseling of the patient and family member(s).

Healthcare Professional Signature: _____ Date: _____

PATIENT/RESPONSIBLE PARTY SIGNATURE TO AUTHORIZE TESTING AND VERIFY INFORMED CONSENT:

I authorize my physician and other medical personnel to provide information to Transgenomic concerning my medical history, and I authorize Transgenomic to disclose the results of my testing and any related health and personal information to my physician. I have read the Informed Consent for genetic testing and understand its content. I have had the opportunity to ask questions about this form and have had any questions answered. I authorize Transgenomic to release to Contractor or Carriers and any insurance carrier providing medical benefits to me and any health plan of which I am a member, any medical or other information including test results in connection with an appeal of a reimbursement denial or other reimbursement matter, if Transgenomic has made prior attempts to obtain reimbursement without the release of such information.

OPTIONAL: ☐ Yes ☐ No After my testing is complete, I consent to have Transgenomic remove personal identifiers from my sample and retain it indefinitely for test development or proficiency testing purposes which may include genes other than those originally tested.

Responsible Party Signature: _____ Date: _____

Print Name: _____ Relationship to Patient: _____

Note: Genetic testing on children less than 18 years of age requires that the ordering physician obtain an informed consent from a parent or legal guardian.

✓			Blood	Saliva	Muscle
	Please select each test requested. If ordering multiple tests, please number the tests to signify the order in which they should be completed.				
	Mitochondrial Genome Disorders See online catalog for specific mutations included in each panel.				
	MITOWGA-NuclearMitome REF	Mitochondrial Whole Genome Analysis (mtDNA) reflex to Comprehensive Sequence Analysis of 448 Nuclear Mitochondrial Genes	●		
	MITO-WGA	Mitochondrial Whole Genome Analysis with Haplotyping (Muscle tissue is preferred when deletions indicative of KSS or CPEO are suspected. See Test Catalog for details.)	●	●	●
	MITO-PT-DEL	Mitochondrial Point Mutation Deletion Assay (Including Aminoglycoside-induced Deafness, LHON, MELAS, MERRF, NARP, CPEO, KSS, Pearson's Syndrome; Muscle tissue is preferred when deletions indicative of KSS or CPEO are suspected. See Test Catalog for details.)	●	●	●
	MITO-DEL	Mitochondrial DNA Deletion Assay (Including CPEO, KSS, Pearson's Syndrome; Muscle tissue is preferred when deletions indicative of KSS or CPEO are suspected. See Test Catalog for details.)	●	●	●
	DFNS	Aminoglycoside-induced Nonsyndromic Deafness	●	●	●
	LHON	Leber's Hereditary Optic Neuropathy	●	●	●
	MELAS	Mitochondrial Encephalomyopathy, Lactic Acidosis and Stroke-like Episodes	●	●	●
	MERRF	Myoclonus Epilepsy with Ragged Red Fibers	●	●	●
	NARP	Neurogenic Weakness, Ataxia, Retinitis Pigmentosa and Leigh's Syndrome	●	●	●
	RCD	Reversible Cox Deficiency	●	●	●
	CYTB	Cytochrome b (*MT-CYB*)	●	●	●
	Nuclear Mitochondrial Genes Analysis of individual genes can be ordered by noting the gene(s) on the first page.				
	NuclearMitome	Comprehensive Sequence Analysis of 448 Nuclear Mitochondrial Genes	●		
	AD-PEO PANEL	Autosomal Dominant Progressive External Opthalmoplegia Panel (*C10orf2*, *OPA1*, *POLG, POLG2, SLC25A4*)	●	●	●
	COMPLEX I PANEL	Complex I Deficiency Panel *(NDUFA1, NDUFA2, NDUFA7, NDUFAF2, NDUFAF3, NDUFAF4, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2*)	●	●	●
	COMPLEX III PANEL	Complex III Deficiency Panel (*BCS1L*) – Bjornstad Syndrome, GRACILE Syndrome	●	●	●
	COMPLEX IV PANEL	Complex IV Panel (*COX10, SCO1, SCO2, SURF1*)	●	●	●
	MITO-DEP PANEL	mtDNA Depletion Syndrome, Hepatocerebral Form, Panel (*POLG*, *DGUOK, MPV17*)	●	●	●
	MITO-DEP/DEL PANEL	mtDNA Depletion Syndrome and Multiple Deletion Panel (*POLG*, *DGUOK, MPV17, C10orf2, OPA1, POLG2, SLC25A4, TK2, TYMP)*	●	●	●
	Gene-specific Tests: **POLG**	POLG-Related Disorders (PEO, Alpers' Disease, Ataxia Neuropathy spectrum, mtDNA Depletion)	●	●	●
	Gene-specific Tests: **POLG2**	AD-PEO, Multiple Deletions	●	●	●
	Gene-specific Tests: **C10orf2**	AD-PEO, Multiple Deletions, mtDNA Depletion	●	●	●
	Gene-specific Tests: **OPA1**	Optic atrophy, AD-PEO, Multiple Deletions	●	●	●
	Gene-specific Tests: **SLC25A4**	AD-PEO, Multiple Deletions	●	●	●
	Gene-specific Tests: **TYMP**	Mitochondrial neurogastrointestinal encephalopathy (MNGIE)	●	●	●
	Epilepsy & Seizure-related Disorders				
	Comprehensive EP Panel	Comprehensive Epilepsy Evaluation NGS Panel for 377 genes that have been reported to cause epilepsy and seizure related disorders	●		
	EP-WES Reflex	Comprehensive Epilepsy Evaluation NGS Panel reflexes to WES. To order this test, please contact our Customer Service Department or visit our website at http://www.transgenomic.com/labs/WES.	●		
	FSP	Male Febrile Seizure Panel (Sequencing of *SCN1A, SCN1B, SCN2A, SCN9A, GABRD, GABRG2 and Large Del/Dup analysis of SCN1A*)	●	●	
	FFSP	Female Febrile Seizure Panel (Male Febrile Seizure Panel plus sequencing of *PCDH19*)	●	●	
	Gene-specific Tests: **SCN1A**	*SCN1A* **CHECK ONE:** ☐ REF ☐ COM ☐ SEQ ☐ DEL/DUP See explanation below.	●	●	
	Gene-specific Tests: **SCN1B**	*SCN1B*	●	●	
	Gene-specific Tests: **SCN2A**	*SCN2A*	●	●	
	Gene-specific Tests: **SCN9A**	*SCN9A*	●	●	
	Gene-specific Tests: **GABRD**	*GABRD*	●	●	
	Gene-specific Tests: **GABRG2**	*GABRG2*	●	●	
	Gene-specific Tests: **MERRF**	Myoclonus Epilepsy with Ragged Red Fibers	●	●	●
	Gene-specific Tests: **POLG**	Alpers' disease	●	●	●
	Gene-specific Tests: **PCDH19**	*PCDH19* Epilepsy limited to females with mental retardation	●	●	
	Whole Exome Sequencing				
	WES	Whole Exome Sequencing for the affected individual. To order this test, please contact our Customer Service Department or visit our website at http://www.transgenomic.com/labs/WES.	●		
	WES-TRIO	Whole Exome Sequencing – TRIO To order this test, please contact our Customer Service Department or visit our website at http://www.transgenomic.com/labs/WES.	●		
	HDSA-WES COM	HDSA and Whole Exome Sequencing run simultaneously. HDSA to detect copy number changes, uniparental disomy, and loss of heterozygosity/regions of homozygosity and WES to detect sequence variants. To order this test, please contact our Customer Service Department or visit our website at http://www.transgenomic.com/labs/WES.	●		
	Other Tests				
	CPT2	CPT2 Deficiency	●	●	
	VLCAD	VLCAD Deficiency (*ACADVL*)	●	●	
	SCN9A	Primary Erythermalgia, Small Fiber Neuropathy, Paroxysmal Extreme Pain Disorder and Congenital Indifference to Pain	●	●	
	LPIN1	Autosomal Recessive Recurrent Myoglobinuria (Sequencing and Large Del/Dup analysis)	●	●	

Refer to the Transgenomic Test Catalog for technical details at: http://labs.transgenomic.com.
WES – Whole Exome Sequencing
NGS – Next Generation Sequencing
REF – The recommended approach: negative sequencing tests will reflex to DEL/DUP analysis. If a variant of unknown significance alone is detected, Transgenomic will contact the provider to see if further testing is desired.
COM – Comprehensive testing in which SEQ and DEL/DUP are run in parallel.
SEQ – Sequencing only
DEL/DUP – Testing for large deletions and duplications by MLPA.

*Saliva is an accepted sample type for sequencing only.

See the next page for our expanded Developmental Delay and Intellectual Disabilities Test menu.

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	AUTISM SPECTRUM DISORDERS AND INTELLECTUAL DISABILITY TESTS		ACCEPTED SAMPLES	
√	*Please select each test requested. If ordering multiple tests, please number the tests to signify the order in which they should be completed.*		Blood	Saliva
	Autism Spectrum Disorders (ASD) / Intellectual Disability (ID) / Multiple Anomalies (MA)			
	HDSA	Postnatal High Density SNP Array *Indicate the reason for testing:* ☐ *Autism Spectrum Disorder* ☐ *Developmental Delay* ☐ *Dysmorphic Features* ☐ *Multiple Congenital Abnormalities* ☐ *Seizure Disorder* ☐ *Failure to Thrive* ☐ *Neuromuscular Disorders* ☐ *Angelman Syndrome* ☐ *Prader-Willi Syndrome* ☐ *Russell-Silver Syndrome* ☐ *Rett Syndrome* ☐ *Other*_____ ☐ *Mitochondrial Disease*	•	•
	KARYO	G-banded Karyotype Analysis	•	
	ASD/ID/MA NGS PANEL	Disorders: Aarskog syndrome, Angelman syndrome, Autism susceptibility, Bardet-Biedl syndrome, Cardiofaciocutaneous syndrome, CHARGE syndrome, Cockayne syndrome, Coffin-Lowry syndrome, Cohen syndrome, Cornelia de Lange syndrome, Costello syndrome, DMD/BMD, Gorlin syndrome, Kabuki syndrome, Legius syndrome, LEOPARD syndrome, Lesch-Nyhan syndrome, Lissencephaly, MED 12-related disorders, Microcephaly, Neurofibromatosis type 1, Noonan syndrome, Opitz syndrome, Phelan-McDermid syndrome, Pitt-Hopkins syndrome, PTEN-related syndromes, Rett syndrome, Rubinstein-Taybi syndrome, Smith-Lemli-Opitz syndrome, Smith Magenis syndrome, Sotos syndrome, Tuberous sclerosis, X-linked hydrocephalus, X-linked mental retardation. Genes: *ARX, ASPM, AP1S2, ATRX, AVPR1A, BDNF, BRAF,CASK, CBL, CDKL5, CHD7, CNTNAP2, CREBBP, DCX, DHCR7, DMD, EHMT1, ERCC6, ERCC8, FGD1, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, GABRB3, HDAC8, HOXA1, HPRT1, HRAS, KDM5C, KRAS, L1CAM, PAFAH1B1 (LIS1), MAP2K1, MAP2K2, MBD5, MECP2, MED12, MEF2C, MET, MID1, MKKS, MLL2, NF1, NHS, NIPBL, NLGN3, NLGN4, NRAS, NRXN1, NSD1, OPHN1, PAFAH1B1 (LIS1), PCDH19, PHF6, PNKP, PQBP1, PTCH1, PTEN, PTPN11, RAB39B, RAD21, RAF1, RAI1, RELN, RPGRIP1L, RPS6KA3, SCN1A, SHANK2, SHANK3, SHOC2, SLC2A1, SLC64A, SLC9A6, SMC1A, SMC3, SOS1, SPRED1, TCF4, TSC1, TSC2, TUBA1A, UBE3A, VPS13B, ZEB2*	•	
	Comprehensive ASD/ ID/MA Evaluation	Postnatal High Density SNP Array & Autism Spectrum Disorders / Intellectual Disabilities / Multiple Anomalies NGS Panel run simultaneously.	•	
	FRAGX	Fragile X (*FMR1*) **CHECK ONE:** ☐ Male ☐ Female	•	
	PTEN	Autism with Macrocephaly, Cowden Syndrome **CHECK ONE:** ☐REF ☐COM ☐SEQ ☐DEL/DUP See explanation below.	•	
	Rett, Angelman and Prader-Willi Syndrome			
	Rett/Atypical Rett/ Angelman NGS Panel	Disorders: Angelman syndrome, Angelman-like syndrome, Atypical Rett syndrome, Classic Rett syndrome, Pitt-Hopkins sydrome, X-linked MR Christianson type, Autoimmune lymphoproligerative syndrome. Genes: *ARX, CDKL5, CNTNAP2, FOXG1, MECP2, MEF2C, NRXN1, SLC9A6, TCF4, UBE3A*	•	
	Chromosome 15 Methylation	Methylation analysis of the Angelman/Prader-Willi critical region of chromosome 15q.11.2-q13.	•	
	MECP2	Rett & Angelman Syndrome **CHECK ONE:** ☐REF ☐COM ☐SEQ ☐DEL/DUP See explanation below.	•	
	UBE3A	Angelman Syndrome	•	•
	FISH	FISH 15q11q-13 (Angelman/Prader-Willi) *Requires a green top tube (available upon request).*	•	
	BDNF	Val66Met polymorphism, modifier of Rett Syndrome	•	
	CDKL5	Atypical Rett, seizures, developmental delay **CHECK ONE:** ☐REF ☐COM ☐SEQ ☐DEL/DUP See explanation below.	•	
	Angelman Syndrome Evaluation	Chromosome 15 methylation analysis and Postnatal High Density SNP Array run simultaneously. If negative, Rett/Atypical Rett/Angelman NGS Panel.	•	
	PWS Evaluation	Chromosome 15 methylation analysis and Postnatal High Density SNP Array run simultaneously.	•	•
	Noonan/RASopathy Disorders			
	Noonan/RASopathy NGS Panel	Disorders: Noonan syndrome, Noonan-like syndrome, LEOPARD syndrome, Costello syndrome, Cardiofaciocutaneous syndrome (CFC), Autoimmune lymphoproliferative syndrome. Genes: *PTPN11, RAF1, SOS1, KRAS, BRAF, MAP2K1, MAP2K2, HRAS, SHOC2, NRAS, CBL*	•	
	Craniosynostosis			
	Craniosynostosis NGS Panel	Disorders: Achondroplasia, Antley-Bixler syndrome, Apert syndrome, Kallmann syndrome, LADD, Muenke syndrome, Pfeiffer syndrome, Radioulnar synostosis, Rhizomelic limb shortening,Robinow-Sorauf syndrome, Saethre-Chotzen syndrome, Beare-Stevenson syndrome, CATSHL (tall stature, hearing loss), Crouzon syndrome, Hypochondroplasia, Jackson-Weiss syndrome. Genes: *FGFR1, FGFR2, FGFR3, TWIST*	•	
	Duchenne/Becker Muscular Dystrophy			
	DMD Reflex Panel	Tier 1: Postnatal High Density SNP Array Tier 2: DMD gene sequencing	•	

SPECIMEN REQUIREMENTS:
Collections kits available at no charge. To order kits, visit http://www.transgenomic.com/labs/neurology/order-test-kits

Blood:
- 4 ml in a EDTA (purple top) tube shipped at room temperature
- FISH analysis requires a sodium heparin (green top) tube
- Newborn minimum blood requirement: 1-2 ml

Saliva:
- Full Oragene saliva collection tube shipped at room temperature

Muscle tissue, fresh or frozen (ship overnight on dry ice):
- Adults: 40-50 mg
- Children: 25 mg

Abbreviations:
NGS – Next Generation Sequencing
ASD/ID/MA – Autism Spectrum Disorder/Intellectual Disability/Multiple Anomalies
DMD – Duchenne Muscular Dystrophy

Refer to the Transgenomic Test Catalog for technical details at: http://labs.transgenomic.com.
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