MOUNT SINAI SCHOOL OF MEDICINE

Department of Genetics and
Genomic Sciences
Mount Sinai School of Medicine
One Gustave L. Levy Place
Box 1497
New York, NY 10029-6574

Tel: 212-241-7518
Fax: 212-241-0139

Mount Sinai Genetic Testing Laboratory – Molecular Genetics Laboratory

Informed Consent for Autism Spectrum Disorder Sequencing Panel on Peripheral Blood

I/my child, ______________________________________, hereby request DNA based testing for autism spectrum disorders (ASDs). I have received verbal and/or written information from my physician or from a genetic counselor that described, in words that I understood, the nature of the genetic testing that I (or my child) am about to undergo.

I understand that blood will be drawn from me and/or my child. I understand that the specimens will be used for determining if I and/or my child carry a mutation(s) in a gene(s) within the panel that has been shown to cause ASD (Please see table of genes tested below).

ASD genes to be sequenced

Gene	Associated abnormal phenotype	Gene	Associated abnormal phenotype
NRXN1	Pitt-Hopkins-like syndrome, ASD	*PTCHD1*	ASD, ID
NSD1	Sotos syndrome	*ARX*	ASD, ID, epilepsy
AHI1	Joubert syndrome	*IL1RAPL1*	ASD, ID, schizophrenia
CNTNAP2	Cortical Dysplasia-Focal Epilepsy Syndrome, Pitt-Hopkins-like syndrome	*OTC*	OTC deficiency
TSC1	Tuberous Sclerosis	*KDM5C*	ASD, ID
PTEN	ASD + macrocephaly, Cowden syndrome, etc.	*OPHN1*	ASD, ID, cerebellar hypoplasia
SHANK2	ASD, intellectual disability (ID)	*PCDH19*	ASD, ID, epilepsy
DHCR7	Smith-Lemli-Opitz syndrome	*UPF3B*	ASD, ID, schizophrenia
CACNA1C	Timothy syndrome	*GRIA3*	ASD, ID
UBE3A	Angelman syndrome	*GPC3*	Simpson-Golabi-Behmel syndrome
TSC2	Tuberous Sclerosis	*SLC9A6*	ASD, ID, Christianson syndrome, Angelman-like
SHANK3	ASD, ID	*FMR1*	Fragile X syndrome
NLGN4X	ASD, ID	*SLC6A8*	Creatine deficiency syndrome
AP1S2	ASD, ID, Fried syndrome	*MECP2*	Rett syndrome
CDKL5	Variant Rett syndrome, Angelman-like	*RAB39B*	ASD, ID

DNA errors detectable by this test panel of 30 different autism genes may account for 5-7% of autism. Mutations in other genes responsible for ASDs have either not yet been identified or have been identified only recently and have not yet been included in this test. In addition, certain types of mutations such as small insertions and deletions of DNA sequence as well as large genomic rearrangements may not be identified by this testing. I understand that a negative result does not rule out a genetic cause of ASD in me/my child.

The nature of DNA testing has been explained to me and the accuracy of the test and its limitations have been detailed. I understand that while results obtained from DNA testing are usually highly accurate, infrequent errors may occur. The likelihood of this occurring has been estimated to be less than 1%. 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 and this test may detect non-paternity.

I give consent to have my specimen be used anonymously by the laboratory for the purposes of quality control or for research related to genetic disease. Please check the box below to consent. If you do not consent your sample will be discarded within 2 months of completion of the testing.

☐ I agree to have my sample used anonymously for research by the laboratory.______
Initials

I understand that this testing may yield results that are of unknown clinical significance and that parental or other relatives blood samples may also be tested to determine whether a specific finding was inherited. In addition, I understand that some mutations may be inherited and still be causative or contributory in my or my child's phenotype due to incomplete penetrance of the mutation or variable expressivity in the phenotypes associated with the mutation.

The results of my/or my child's test will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a clinical geneticist.

I have had the opportunity to have all of my questions answered. If I am signing this form on behalf of a minor for whom I am the legal guardian, I am satisfied that I have received enough information to sign on his or her behalf. I understand that this consent is being obtained in order to protect my right to have all of my questions answered before testing. I also understand that the results of this testing will become part of my medical record and may only be disclosed to individuals who have legal access to this record or to individuals who I designate to receive this information.

______________________________ ______________
Signature of Person Being Tested (or guardian) Date

______________________________ ______________
Witness Date