ARUP LABORATORIES

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

EXOME SEQUENCING PATIENT HISTORY FORM

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity (check all that apply)

☐ African American ☐ Asian ☐ Hispanic ☐ Native American

☐ Ashkenazi Jewish ☐ White ☐ Middle Eastern ☐ Other: _____

Suspected clinical diagnosis? _____

Potentially causative genes? _____

Describe ALL findings:

☐ Intellectual disability IQ Range: _____ ☐ Mild ID ☐ Moderate ID ☐ Severe ID

☐ Autism: _____

☐ Cancer/Tumor: _____

☐ Cardiac: _____

☐ Craniofacial: _____

☐ Dermatologic: _____

☐ Dysmorphic: _____

☐ Gastrointestinal: _____

☐ Genital: _____

☐ Growth: _____

☐ Hematologic: _____

☐ Immunologic: _____

☐ Metabolic: _____

☐ Muscular: _____

☐ Neurologic: _____

☐ Optic: _____

☐ Otologic: _____

☐ Pulmonary: _____

☐ Skeletal: _____

☐ Urinary tract: _____

☐ Other: _____

Has the patient undergone previous genetic testing? ..... ☐ No ☐ Yes ☐ Unknown

Please include a copy of all equivocal or abnormal genetic test results.

Chromosome analysis	☐ Normal	☐ Abnormal	☐ Not Performed	
Prenatal genomic microarray	☐ Normal	☐ Abnormal	☐ Not Performed	Performing lab _____
Postnatal genomic microarray	☐ Normal	☐ Abnormal	☐ Not Performed	Performing lab _____

EXOME SEQUENCING PATIENT HISTORY FORM

Other Test:_____ Method:_____ ☐ Normal ☐ Abnormal

Other Test:_____ Method:_____ ☐ Normal ☐ Abnormal

Other Test:_____ Method:_____ ☐ Normal ☐ Abnormal

Other Test:_____ Method:_____ ☐ Normal ☐ Abnormal

Has the patient had an MRI or CT ..... ☐ No ☐ Yes ☐ Unknown

If yes, was it abnormal? ..... ☐ No ☐ Yes ☐ Unknown

If abnormal, please enclose a copy of MRI/ CT.

Mother's sample is strongly recommended for result interpretation of all exome sequencing tests:

Date of sample collection:_____ Not Available ☐ Will be sent at later date _____

☐ Biological mother's name: Symptoms? _____ DOB:_____

☐ No ☐ Yes Has the mother been examined by a doctor? If yes, describe: _____

_____

Father's sample is strongly recommended for result interpretation of all exome sequencing tests:

Date of sample collection:_____ Not Available ☐ Will be sent at later date _____

☐ Biological father's name: Symptoms? _____ DOB:_____

☐ No ☐ Yes Has the father been examined by a doctor? If yes, describe: _____

_____

Please include the following:

☐ **Clinical summary**

☐ **Three generation medical PEDIGREE**

☐ **Genomic microarray results**

☐ **All abnormal or equivocal genetic test results**

☐ **Abnormal MRI/CT or imaging results**

Check the test below that you intend to order.

☐ **2006332 Exome Sequencing, Trio:** Exome sequencing is performed on the patient, his/her parents, and up to two other affected family members. A diagnosis is determined in ~45% of patients. Parental testing allows identification of de novo variants and phasing of variants.

☐ **2006336 Exome Sequencing, Proband:** Exome sequencing is performed on patient only. Targeted sequencing, for variants of interest in the proband, is performed on parents. A diagnosis is determined in ~35% of patients when parental samples are submitted and in only ~20% without. De novo variants are typically not identifiable.

Master Label

For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.

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