ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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

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Salt Lake City, Utah 84108-1221
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THIS IS NOT A TEST REQUEST FORM.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR EXOME SEQUENCING

Patient Name _____ **Date of Birth** _____ **Sex** ☐ F ☐ M

Physician _____ **Physician Phone** _____

Practice Specialty _____ **Physician Fax** _____

Genetic Counselor _____ **Counselor Phone** _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

What is the patient's suspected clinical diagnosis? _____

List genes that may be causative: _____

Describe ALL findings:

☐ Intellectual Disability IQ Range: _____ ☐ Mild MR ☐ Moderate MR ☐ Severe MR
☐ Autism: _____
☐ Cancer/Tumor: _____
☐ Cardiac: _____
☐ Craniofacial: _____
☐ Dermatologic: _____
☐ Dysmorphic Features: _____
☐ Gastrointestinal: _____
☐ Genital: _____
☐ Growth: _____
☐ Hematologic: _____
☐ Immunologic: _____
☐ Metabolic: _____
☐ Muscular: _____
☐ Neurologic: _____
☐ Optical: _____
☐ Otologic: _____
☐ Pulmonary: _____
☐ Skeletal: _____
☐ Urinary tract: _____
☐ Other: _____

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

Chromosome analysis ☐ Normal ☐ Abnormal ☐ Not Performed

Array CGH ☐ Normal ☐ Abnormal ☐ Not Performed

Other: _____ Method: _____ ☐ Normal ☐ Abnormal

Other: _____ Method: _____ ☐ Normal ☐ Abnormal

Other: _____ Method: _____ ☐ Normal ☐ Abnormal

Other: _____ Method: _____ ☐ Normal ☐ Abnormal

If any test results were equivocal or abnormal, please describe: _____

PATIENT HISTORY FOR EXOME SEQUENCING

Has the patient had an MRI? ☐ No ☐ Yes ☐ Unknown

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

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

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

Biological mother's name:_____ DOB:_____

Symptoms? ☐ No ☐ Yes If yes, describe:_____

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

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

Biological father's name:_____ DOB:_____

Symptoms? ☐ No ☐ Yes If yes, describe:_____

Please ATTACH the following:

1) A clinical summary report

2) A three generation medical PEDIGREE detailing all diagnoses/symptoms and age of onset in each relative

3) Array CGH results which show copy number changes that are pathogenic or have unknown significance

4) Any genetic test results that identified pathogenic mutations or variants of unknown significance

5) Any abnormal MRI results

Check the Exome Sequencing test below that you intend to order.

☐ **2006332 Exome Sequencing, Trio:** Exome sequencing is performed on the patient and his/her parents. Sensitivity may approach 45% for determining a genetic cause for the patient's symptoms. Exome sequencing on the parents allows the identification and reporting of de novo variants in genes of known or unknown function as well as phasing of identified variants.

☐ **2006336 Exome Sequencing, Proband:** Exome sequencing is performed on patient. Targeted sequencing is performed on the patient's parents only for variants related to the patient's phenotype. Sensitivity is 25% for determining a cause for the patient's symptoms. De novo variants in genes unrelated to the phenotype cannot be identified.

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

Master Label

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