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 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: _____

**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 _____

# PATIENT HISTORY FOR EXOME SEQUENCING

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:_____ DOB:_____

Symptoms? ☐ No ☐ Yes 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:_____ DOB:_____

Symptoms? ☐ No ☐ Yes 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 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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