ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform exome sequence testing.
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_____ /_____/_____Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)
[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] Other _____

What is the patient's suspected clinical diagnosis?_____

Describe ALL findings:
[ ] Intellectual Disability IQ Range_____ [ ] Mild MR [ ] Moderate MR [ ] Severe MR
[ ] Autism_____
[ ] Neurologic _____
[ ] Muscular_____
[ ] Dysmorphic features _____
[ ] Growth _____
[ ] Skeletal _____
[ ] Craniofacial _____
[ ] Cardiac _____
[ ] Urinary tract _____
[ ] Genital _____
[ ] Optical _____
[ ] Otologic _____
[ ] Immunologic _____
[ ] Dermatologic_____
[ ] Metabolic _____
[ ] Pulmonary_____
[ ] Gastrointestinal_____
[ ] Hematologic _____
[ ] Cancer/ Tumor_____
[ ] 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_____

Has the patient had an MRI? [ ] No [ ]Yes [ ] Unknown
If yes, was it abnormal? [ ] No [ ]Yes [ ] Unknown

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PATIENT HISTORY FOR EXOME SEQUENCING (Cont.)

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**

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

2006332 Exome Sequencing Symptom Guided Analysis: Exome sequencing is performed on the patient and his/her parents. Sensitivity is 30% for determining a genetic cause for the patient's symptoms. Full exome sequencing on the parents allows the identification and reporting of de novo variants in genes of known or unknown function.

2006336 Exome Sequencing Symptom Guided Analysis, Patient Only: Exome sequencing is performed on patient and targeted sequencing only for variants related to the patient's phenotype is performed on the patient's parents. Sensitivity is 10% 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