ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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 RAPID MENDELIAN GENES SEQUENCING PANEL, TRIO

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

Physician _____ Physician Email _____

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 / indication for testing? _____

List specific genes of interest: _____

Describe ALL findings:

☐ Prenatal Findings
Decreased fetal movement? ☐ No ☐ Yes Polyhydramnios? ☐ No ☐ Yes ☐ Other: _____

☐ Prematurity? ☐ No ☐ Yes (list gestational age at delivery: _____)

☐ Growth: _____
Failure to thrive ☐ No ☐ Yes IUGR ☐ No ☐ Yes Macrocephaly ☐ No ☐ Yes Microcephaly ☐ No ☐ Yes
Overgrowth ☐ No ☐ Yes

☐ Body wall defect: _____

☐ Cancer/tumor: _____

☐ Cardiac: _____
Arrhythmia ☐ No ☐ Yes Bradycardia ☐ No ☐ Yes

☐ CNS (structural brain malformations): _____

☐ Craniofacial: _____

☐ Dermatologic: _____

☐ Dysmorphic features: _____

☐ Gastrointestinal: _____

☐ Genital: _____

☐ Hematologic: _____

☐ Immunologic: _____

☐ Limb abnormalities: _____

☐ Metabolic: _____
Acidosis ☐ No ☐ Yes Hyperammonemia ☐ No ☐ Yes Liver Function Defect ☐ No ☐ Yes

☐ Muscular: _____

☐ Neurologic: _____
Abnormal movements ☐ No ☐ Yes Ataxia ☐ No ☐ Yes Central apnea ☐ No ☐ Yes Seizures ☐ No ☐ Yes
Cranial nerve defects ☐ No ☐ Yes Hypertonia/spasticity ☐ No ☐ Yes Hypotonia ☐ No ☐ Yes

☐ Opthalmologic: _____

☐ Otologic: _____

☐ Pulmonary: _____
Diaphragmatic defect ☐ No ☐ Yes Laryngotracheal abnormalities ☐ No ☐ Yes Lung malformation ☐ No ☐ Yes
Respiratory failure ☐ No ☐ Yes

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☐ Skeletal: _____

Hyperextensibility ☐ No ☐ Yes Joint contractures ☐ No ☐ Yes Vertebral anomalies/scoliosis ☐ No ☐ Yes

☐ Urinary tract: _____

☐ Other: _____

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

Chromosome analysis ☐ Normal ☐ Abnormal ☐ Not performed

Genomic microarray ☐ 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

If abnormal, describe: _____

Mother's Sample

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

Biological mother's name: _____ DOB: _____

Symptoms? ☐ No ☐ Yes, describe: _____

Father's Sample

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

Biological father's name: _____ DOB: _____

Symptoms? ☐ No ☐ Yes, describe: _____

Please ATTACH the following:

1) Completed consent form for patient
2) Clinical summary report
3) Three generation medical PEDIGREE detailing all diagnoses/symptoms and age of onset in each relative
4) Genomic microarray results showing copy number changes that are pathogenic or have unknown significance
5) Any genetic test results that identified pathogenic mutations or variants of unknown significance
6) Any abnormal MRI results
7) Any abnormal Echo/Ultrasound/X-ray or metabolic test results

Master Label

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

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