Rady Children's Institute
Genomic Medicine

Clinical Genome Center
7910 Frost Street, Suite 240
San Diego, CA 92123
E: RCIGM_ClinicalLab@rchsd.org
P: 858 / 966-8978 **F:** 858 / 966-8092
CLIA ID# 05D2129627
CAP ID# 9487427

Place ordering provider label with patient identifier here

RCIGM Phenotype Form

Patient First Name | Last Name | Hospital MRN/Patient ID# | DOB (MM/DD/YY)

Form Completed By:_____ (MD / GC / Nurse / PA / Other) Date:_____

Indication for testing (1-2 sentences):_____

PRIMARY FEATURE

What is the primary feature:_____

Age at which the primary feature first noted:_____

Is the condition static or progressive:_____

Laboratory or imaging findings relevant to primary features:_____

FAMILY HISTORY

Family history of similar condition:_____

Other family history (or attach pedigree):_____

Consanguinity (if yes, specify):_____

History of miscarriages for the biological mother:_____

Countries of origin of both parents:_____

PREVIOUS GENETIC TESTING

Please list all genetic tests previously ordered and the results (or note if pending):_____

OTHER RELEVANT INFORMATION

Please detail any other relevant clinical information (ex: problem list, differential diagnosis, NBS results):

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RCIGM Phenotype Form

CLINICAL FEATURES (Please check all that apply. This is not a substitute for submitting clinical records)

Craniofacial
- ☐ Craniosynostosis
- ☐ Cleft lip/palate
- ☐ Other (specify): _____

Ophthalmologic
- ☐ Abnormal eye structure
- ☐ Visual impairment
- ☐ Abnormal eye movements
- ☐ Other (specify): _____

ENT
- ☐ Ear anomalies
- ☐ Hearing impairment
- ☐ Other (specify): _____

Pulmonology
- ☐ Abnormal respiratory tract structure
- ☐ Abnormal respiratory tract function
- ☐ Other (specify): _____

Cardiac
- ☐ Congenital heart defect (specify type): _____
- ☐ Cardiomyopathy (specify type): _____
- ☐ Arrhythmia (specify type): _____
- ☐ Other vascular abnormality (specify type): _____

Gastroenterology
- ☐ Failure to thrive
- ☐ Congenital GI malformation (specify): _____
- ☐ Cholestasis
- ☐ Abnormal GI motility
- ☐ Hirschsprung's Disease
- ☐ Other (specify): _____

Renal/Genitourinary
- ☐ Congenital renal anomalies
- ☐ Hypospadius
- ☐ Ambiguous genitalia
- ☐ Undescended testes
- ☐ Other renal/genitourinary abnormality (specify): _____

Hematology/Oncology
- ☐ Anemia
- ☐ Bone marrow failure
- ☐ Abnormal bleeding tendency
- ☐ Thrombophilia
- ☐ Tumor
- ☐ Other (specify): _____

Immunology
- ☐ Recurrent infections
- ☐ Laboratory findings of immunodeficiency
- ☐ Other (specify): _____

Dermatology
- ☐ Abnormal skin pigmentation (specify): _____
- ☐ Other (specify): _____

Musculoskeletal
- ☐ Myopathy
- ☐ Fractures
- ☐ Radiographic abnormalities
- ☐ Scoliosis
- ☐ Club foot
- ☐ Congenital contractures
- ☐ Other (specify): _____

Neurologic
- ☐ Developmental delay
- ☐ Developmental regression
- ☐ Autism
- ☐ Hypotonia
- ☐ Hypertonia
- ☐ Seizures
- ☐ Microcephaly
- ☐ Encephalopathy
- ☐ Abnormal brain MRI (specify): _____
- ☐ Abnormal muscle biopsy (specify): _____
- ☐ Abnormal movements (specify): _____
- ☐ Other (specify): _____

Endocrine
- ☐ Short stature
- ☐ Hypoglycemia
- ☐ Hyperglycemia
- ☐ Other (specify): _____

Metabolic
- ☐ Lactic acidosis
- ☐ Hyperammonemia
- ☐ Abnormal metabolic test (specify): _____
- ☐ Other (specify): _____

Prenatal
- ☐ IUGR
- ☐ Hydrops
- ☐ Maternal diabetes
- ☐ Abnormal ultrasound findings (specify): _____
- ☐ Other (specify): _____

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Additional Family Member Samples

Patient (Proband) Name

DOB (MM/DD/YY)

Hospital MRN/Patient ID#

Sample Collection Date (MM/DD/YY)

Instructions: To be completed if submitting samples from family members to assist with patient results. No separate results will be issued for these individuals. **Label sample tubes with at least two identifiers listed on Requisition Form.**

Additional Family Member 1

First Name Last Name DOB (MM/DD/YY) Relationship to Patient (e.g. mother)

Sex: ☐ Female ☐ Male ☐ Ambiguous

MRN

☐ Clinically Unaffected

☐ Clinically Affected Briefly list phenotype

Date Collected (MM/DD/YY) Time Collected ☐ AM ☐ PM

Sample Type: ☐ Whole blood (EDTA) ☐ DNA, Extracted from EDTA Blood in a CLIA accredited laboratory

CLIA Laboratory Name

Additional Family Member 2

First Name Last Name DOB (MM/DD/YY) Relationship to Patient (e.g. mother)

Sex: ☐ Female ☐ Male ☐ Ambiguous

MRN

☐ Clinically Unaffected

☐ Clinically Affected Briefly list phenotype

Date Collected (MM/DD/YY) Time Collected ☐ AM ☐ PM

Sample Type: ☐ Whole blood (EDTA) ☐ DNA, Extracted from EDTA Blood in a CLIA accredited laboratory

CLIA Laboratory Name

Additional Family Member 3

First Name Last Name DOB (MM/DD/YY) Relationship to Patient (e.g. mother)

Sex: ☐ Female ☐ Male ☐ Ambiguous

MRN

☐ Clinically Unaffected

☐ Clinically Affected Briefly list phenotype

Date Collected (MM/DD/YY) Time Collected ☐ AM ☐ PM

Sample Type: ☐ Whole blood (EDTA) ☐ DNA, Extracted from EDTA Blood in a CLIA accredited laboratory

CLIA Laboratory Name