

# Phenotypic Checklist – Pediatric

The accurate interpretation and reporting of genetic results relies on the provision of pertinent clinical and family history information. To help ensure the most clinically appropriate interpretation of results, please complete this clinical information form. Testing cannot be initiated without a reason for referral.

## Please Check All That Apply

### Perinatal History

- Prematurity
- IUGR
- Oligohydramnios
- Polyhydramnios
- Other fetal anomaly: \_\_\_\_\_
- None specified

### Developmental/Behavioral

- Fine motor delay
- Gross motor delay
- Speech delay
- Learning disability
- Intellectual disability
- Autistic features
- Autism spectrum disorder
- Oppositional-defiant disorder
- Psychiatric illness:
  - Bipolar disorder
  - Anxiety
  - Depression
  - ADHD
  - Other: \_\_\_\_\_

### Craniofacial

- Cleft lip
- Cleft palate
- Coloboma
- Craniosynostosis
- Dysmorphic facial features
- Macrocephaly
- Microcephaly
- Hearing loss
- Ear malformation
- Cataracts

### Cardiac

- Atrial septal defect
- AVSD/AV canal defect
- Coarctation of the aorta
- Hypoplastic left heart
- Tetralogy of Fallot
- Ventricular septal defect
- Dilated aortic root
- Cardiomyopathy: \_\_\_\_\_
- Arrhythmia

### Cutaneous

- Hyperpigmentation
- Hypopigmentation
- Cafe au lait spots
- Eczema

### Gastrointestinal

- Gastroschisis
- Hirschprung disease
- Omphalocele
- Pyloric stenosis
- Tracheoesophageal fistula
- Meconium ileus
- Anal atresia
- Diaphragmatic hernia

### Genitourinary

- Ambiguous genitalia
- Hydronephrosis
- Kidney anomaly: \_\_\_\_\_
- Hypospadias

### Growth

- Short stature
- Tall stature
- Obesity
- Failure to thrive
- Delayed puberty
- Precocious puberty

### Musculoskeletal

- Contractures (arthrogryposis)
- Club foot
- Diaphragmatic hernia
- Limb anomaly
- Polydactyly (hands)
- Polydactyly (feet)
- Syndactyly (hands)
- Syndactyly (feet)
- Vertebral anomaly

### Neurological

- Ataxia
- Cerebral palsy
- Dystonia
- Hypotonia
- Hypertonia
- Developmental regression
- Muscle weakness
- Neural tube defect
- Seizures
- Abnormal brain MRI: \_\_\_\_\_

### Family History

- Parents with  $\geq 2$  miscarriages
- Relative with known chromosome abnormality: \_\_\_\_\_
- Other relatives with clinical history similar to patient (please describe) \_\_\_\_\_

### Other (not on list)

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

As a participant in the International Collaboration for Clinical Genomics (ICCG), this clinical cytogenetics laboratory contributes submitted clinical information and test results to a HIPAA compliant, de-identified public database as part of the NIH's effort to improve understanding of the relationship between genetic changes and clinical symptoms. Confidentiality is maintained.

Patients may request to opt out of this scientific effort by 1) checking the box below or 2) calling the laboratory at 800.710.0624 and asking to speak with a laboratory genetic counselor. Please call with any questions.

Mark here to indicate refusal for inclusion in these efforts by checking this box. If the box is not marked, data will be anonymized and submitted.

### Testing Notes

CombiMatrix will attempt to perform all tests ordered. If the sample size is insufficient to do so, CombiMatrix will perform testing according to a predefined standing order contingency plan. If no standing order contingency plan exists, CombiMatrix will promptly notify the client and discuss what testing can and cannot be performed. If CombiMatrix is unable to make contact with a client the same day (the day the sample was received) to determine what testing is desired, CombiMatrix will run only the tests that are possible based on the sample received. To set up a standing order contingency plan, please call our Genetic Counseling Services at 949.255.0921.

If testing other than the options listed on this form is desired, please contact one of CombiMatrix's genetic counselors to discuss whether it is possible to accommodate your request prior to sending the sample. If prior arrangements are not made, CombiMatrix cannot guarantee the ability to provide the requested testing.

### Patient Information

Please place the completed label from the Test Requisition Form with the patient information here and return this form with the test sample:

MM	DD	YY	
Specimen Collection Date			Last Name
Patient DOB			First Name