

# Phenotypic Checklist – Prenatal

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

### Primary Indication for Testing

- Abnormal screening results:
  - Abnormal NIPT result (please include report): \_\_\_\_\_
  - Abnormal serum screen: \_\_\_\_\_
  - Increased NT/cystic hygroma
- Advanced maternal age
- Fetal abnormality

### Growth

- Hydrops
- IUGR
- Macrosomia
- Molar pregnancy
- Oligohydramnios
- Placental abnormality
- Polyhydramnios
- Single umbilical artery/2 vessel cord
- Other: \_\_\_\_\_

### Gastrointestinal

- Anal atresia
- Absent stomach
- Duodenal atresia (double bubble sign)
- Echogenic bowel
- Gastroschisis
- Omphalocele
- Tracheoesophageal fistula
- Other: \_\_\_\_\_

### Craniofacial

- Cleft lip and/or cleft palate
- Hypertelorism
- Hypotelorism
- Macrocephaly
- Microcephaly
- Micrognathia
- Pierre Robin sequence
- Other: \_\_\_\_\_

### Pulmonary

- CCAM
- Diaphragmatic hernia
- Eventration of diaphragm
- Pleural effusion
- Pulmonary sequestration
- Small thoracic cavity
- Other: \_\_\_\_\_

### Cardiac

- Aortic atresia
- Atrial septal defect (ASD)
- AV canal defect
- Coarctation of aorta
- Ebstein anomaly
- Echogenic intracardiac focus
- Hypoplastic left heart
- Hypoplastic right heart
- Pericardial effusion
- Pulmonary atresia
- Tetralogy of Fallot
- Transportation of the great vessels
- Truncus arteriosus
- Ventricular septal defect (VSD)
- Other: \_\_\_\_\_

### Neurological

- Abnormal gyri/Lissencephaly
- Agenesis of the corpus callosum
- Cerebellar hypoplasia
- Choroid plexus cyst(s)
- Dandy Walker (posterior fossa abn.)
- Decreased fetal movement
- Holoprosencephaly
- Open neural tube defect (ONTD)
  - Anencephaly
  - Spina bifida
- Structural brain anomaly
- Ventriculomegaly/hydrocephaly
- Other: \_\_\_\_\_

### Musculoskeletal

- Contractures (arthrogryposis)
- Club Foot
- Limb anomaly (lower)
- Limb anomaly (upper)
- Polydactyly (feet)
- Polydactyly (hands)
- Rocker-bottom feet
- Scoliosis
- Shortened long bones
- Skeletal dysplasia
- Syndactyly (feet)
- Syndactyly (feet)
- Vertebral anomaly

### Genitourinary

- Ambiguous genitalia
- Hydronephrosis
- Megacystis
- Pyelectasis
- Polycystic kidneys
- Renal agenesis
- Urethral/ureteral obstruction

### Family History

- Patient has had  $\geq 2$  miscarriages
- Previous pregnancy with chromosome abnormality: \_\_\_\_\_
- Family history of known chromosome abnormality/genetic condition: \_\_\_\_\_
- Other (please describe) \_\_\_\_\_

### Other

\_\_\_\_\_  
 \_\_\_\_\_  
 \_\_\_\_\_

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 contact CombiMatrix's Director of Genetic Counseling Services at 800.710.0624 option 3.

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 _____