

Colorado Genetics Laboratory

Constitutional Test Requisition Form: Prenatal / Pregnancy Loss / Postnatal

Patient Information		Ordering Provider Information	
Legal Name (last, first)	Preferred Name	Ordering Provider	Provider Phone
Date of Birth (mm/dd/yyyy)	MRN	Provider Email	Provider NPI
Sex Assigned at Birth ☐ Male ☐ Female ☐ Unknown	Gender Identity	Genetic Counselor	Counselor Phone
Patient Sticker Here		Ordering Clinic/Facility	
		Clinic Address	Clinic Phone
		Clinic Fax	

Indication For Genetic Testing		
ICD-10 Codes (Required)	Other Relevant Information (Other test results, etc. not described below)	
Pre/Perinatal History ☐ Advanced Maternal Age ☐ Abnormal NIPT/NIPS, specify: _____ ☐ Cystic Hygroma ☐ Increased Nuchal Translucency ☐ Hydrops Fetalis (Nonimmune) ☐ Echogenic Bowel ☐ Aneuploidy Marker(s), specify: _____ ☐ IUGR ☐ Polyhydramnios ☐ Oligohydramnios ☐ Prematurity ☐ Reduced Fetal Movement ☐ Structural Malformations (mark in sections) ☐ Spontaneous Pregnancy Loss, gestational age: ____ wks ☐ Elective Termination for Anomalies (mark in sections) ☐ Other, specify: _____ Developmental/Behavioral Findings ☐ Autistic Behavior ☐ Intellectual Disability ☐ Global Developmental Delay ☐ Speech/Language Delay ☐ Fine Motor Delays ☐ Gross Motor Delays ☐ ADHD ☐ Hyperactivity ☐ OCD ☐ Behavioral Abnormality ☐ Specific Learning Disability ☐ Sleep Disturbance ☐ Other, specify: _____ Neurological Findings ☐ Hypotonia ☐ Hypertonia ☐ Ataxia ☐ Dystonia ☐ Chorea ☐ Seizures/Epilepsy, specify type: _____ ☐ Cerebral Palsy ☐ Encephalopathy ☐ Peripheral Neuropathy ☐ Spasticity ☐ Other, specify: _____ Hearing/Vision Differences ☐ Conductive Hearing Impairment ☐ Sensorineural Hearing Impairment ☐ Abnormality of Vision ☐ Estropia ☐ Retinitis Pigmentosa ☐ Other, specify: _____	Musculoskeletal/Growth Anomalies ☐ Failure to Thrive ☐ Short Stature ☐ Overgrowth ☐ Asymmetrical Growth, Specify: _____ ☐ Contractures, specify: _____ ☐ Clubfoot ☐ Limb Shortening, specify: _____ ☐ Skeletal Dysplasia ☐ Scoliosis ☐ Polydactyly ☐ Syndactyly ☐ Vertebral Anomaly, specify: _____ ☐ Clenched Hands ☐ Limb Fractures ☐ Other, specify: _____ Craniofacial Anomalies ☐ Aniridia ☐ Coloboma ☐ Microphthalmia/Anophthalmia ☐ Oral Cleft ☐ Craniosynostosis ☐ Dysmorphic Facial Features, specify: _____ ☐ External Ear Malformation, specify: _____ ☐ Hypertelorism ☐ Hypotelorism ☐ Micrognathia ☐ Macrocephaly ☐ Microcephaly ☐ Brachycephaly ☐ Other, specify: _____ Gastrointestinal Malformations ☐ Gastroschisis ☐ Omphalocele ☐ Hirschsprung Disease ☐ Pyloric Stenosis ☐ Tracheoesophageal Fistula ☐ Aganglionic Megacolon ☐ Absent Stomach ☐ Anal Atresia ☐ Other, specify: _____ Genitourinary Abnormalities ☐ Atypical Genitalia ☐ Hypospadias ☐ Undescended Testis ☐ Hydronephrosis ☐ Kidney Malformation, specify: _____ ☐ Renal Agenesis, specify unilateral/bilateral: _____ ☐ Urethral Malformation ☐ Ureteral Obstruction	Cardiac Abnormalities ☐ ASD ☐ VSD ☐ AV Canal Defect ☐ Coarctation of Aorta ☐ Hypoplastic Left Heart ☐ Tetralogy of Fallot ☐ Cardiomyopathy, specify: _____ ☐ Transposition of Great Vessels ☐ Ebstein Anomaly ☐ Pulmonary Valve Atresia ☐ Truncus Arteriosus ☐ Dextrocardia/situs inversus ☐ Other, specify: _____ Brain/CNS Malformations ☐ Encephalocele ☐ Anencephaly ☐ Neural Tube Defect/Spina Bifida ☐ Agenesis of the Corpus Callosum ☐ Holoprosencephaly ☐ Hydrocephalus ☐ Ventriculomegaly ☐ Lissencephaly ☐ Dandy Walker Malformation ☐ Cerebellar Hypoplasia ☐ Polymicrogyria ☐ Other, specify: _____ Pulmonary Findings ☐ Diaphragmatic Hernia ☐ CCAM ☐ Small Thoracic Cavity ☐ Pulmonary Sequestration ☐ Pleural Effusion ☐ Other, specify: _____ Endocrine Findings ☐ Delayed Puberty ☐ Precocious Puberty ☐ Abnormal Hormone Levels, specify: _____ ☐ Other, specify: _____ Metabolic/Mitochondrial Anomalies ☐ Abnormal lab results, specify: _____ Family History ☐ Recurrent Pregnancy Loss ☐ Consanguinity, specify: _____ ☐ Birth Defects, specify: _____ ☐ Genetic Disorder, specify: _____ ☐ Known Chromosome Anomaly, specify: _____ _____ _____

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Patient Information (page 2)

Legal Name		Patient Sticker Here
Date of Birth	MRN	

Prenatal Testing Orders

Gestational Age by U/S	LMP	Gravida, Para G ___ P ___
Fetal Sex ☐ M ☐ F ☐ Unk	Multiples (Presumed) ☐ Di/Di ☐ Mo/Di ☐ Mo	Donor Use ☐ Ovum ☐ Sperm
Test Options Most Common ☐ STAT Aneuploidy FISH with 5-cell chromosome analysis and chromosomal microarray (CMA). If aneuploidy FISH abnormal, CMA will be canceled and standard chromosome analysis will be performed. ☐ aAFP with reflex to ACHE (amniotic fluid samples only) ☐ Maternal Cell Contamination (MCC) if needed* (submit maternal blood specimen) ☐ Culture and freeze ☐ Add reflex: Cancel freeze if FISH is abnormal ☐ Culture and Send Out (Complete Send Out Section, see bottom right)		
Other Combinations ☐ Standard chromosome analysis Add FISH: ☐ STAT Aneuploidy FISH ☐ Sex Chromosome Evaluation (X/Y/SRY/STS/Yqh) ☐ Chromosomal microarray ☐ Add 5-cell chromosome analysis ☐ Add STAT Aneuploidy FISH Add Specialty FISH: ☐ 22q11.2 Microdeletion ☐ 7q11.2 Williams Syndrome (ELN) ☐ 15q11.2 Prader-Willi/Angelman Syndromes ☐ Xp22.21 Ichthyosis (STS) ☐ Sex Chromosome Evaluation (X/Y/SRY/STS/Yqh)		

*Recommended for female fetus or unknown sex. MCC performed if results are normal.

Pregnancy Loss/Autopsy Testing Orders

Gestational Age by U/S	LMP	Gravida, Para G ___ P ___
Fetal Sex ☐ M ☐ F ☐ Unk	Multiples (Presumed) ☐ Di/Di ☐ Mo/Di ☐ Mo	Donor Use ☐ Ovum ☐ Sperm
Test Options ☐ Chromosomal microarray (CMA) with reflex to pregnancy loss FISH** if CMA failure ☐ Add Send Out of extracted DNA (Complete Send Out Section, see bottom right) ☐ Maternal Cell Contamination (MCC) if needed* (submit maternal blood specimen)		

*Recommended for female fetus or unknown sex. MCC performed if results are normal.
 **Pregnancy loss FISH: 13, 15, 16, 18, 21, 22, X, Y chromosomes

Postnatal Testing Orders

Test Options Most Common ☐ Chromosomal microarray (CMA) and 5-cell chromosome analysis	
Preliminary Tests (must also select a diagnostic test) ☐ STAT Aneuploidy FISH Specialty FISH: ☐ 22q11.2 Microdeletion ☐ 7q11.2 Williams Syndrome (ELN) ☐ 15q11.2 Prader-Willi/Angelman Syndromes ☐ Xp22.21 Ichthyosis (STS) ☐ Sex Chromosome Evaluation (X/Y/SRY/STS/Yqh)	
Diagnostic Tests ☐ Chromosomal microarray (CMA) ☐ Standard chromosome analysis ☐ High-Resolution chromosome analysis (Only known familial chromosome abnormality)	
Add-on Options ☐ Send Out (complete Send Out section, see right)	
Family Testing ☐ Targeted Chromosomal Microarray (TCMA) Proband: _____ Proband's DOB: _____ CGL Accession/Case No.: _____	

Specimen Information and Requirements

Collection Date	Collection Time
Prenatal/Autopsy (rec volume) ☐ Chorionic Villi (25 mg) ☐ Amniotic Fluid (25 ml) ☐ Fetal Urine (25 ml) ☐ PUBS (3 ml in NaHep) ☐ Products of Conception (1 cm ³) ☐ Placenta (1 cm ³) ☐ Fetal/Autopsy Tissue (1 cm ³)^ specify: _____	Postnatal (rec volume) ☐ Peripheral Blood (2-4 ml in NaHep), if CMA ordered also send 2-4 ml in EDTA. ☐ Peripheral Blood for MCC (3 ml in EDTA) ☐ Cord Blood (2-4 ml in NaHep), if CMA ordered also send 2-4 ml in EDTA. ☐ Buccal Swab (call for kit) ☐ Tissue Biopsy (3-4mm ³ in transport media)

^Acceptable tissues: kidney, lung, gonad, fascia lata. **Whole fetuses are NOT accepted.**
 All pregnancy loss/autopsy/tissue specimens should be in sterile transport media.
 Additional specimen requirements on website.

Specimen Information and Requirements

Payor ☐ Institution/Facility Bill ☐ Provider/Clinic Bill ☐ Patient Self-Pay*** ☐ Patient Insurance	Patient Self-Pay Details ***Payment is required prior to testing. Self-pay patients receive a discount on test costs. Email or fax the payment card number, expiration date, and name on the card to CGL. Email: CGLClientServices@olucdenver.onmicrosoft.com
Patient Insurance Details Insurance Provider: _____ Insurance ID: _____ Group No.: _____ Policy Holder (Last, First): _____ Date of Birth: _____ Billing Address: _____	

Consent

Genetic testing results may have significant implications for a patient's physical wellbeing and mental health. Thorough discussion between the ordering provider and the patient (or their legal guardian) on the risks, benefits, and limitations of testing should be performed before ordering. Chromosomal microarray (CMA) has additional nuances to consider and points for consenting are provided on the next page. It is recommended to provide the patient with that consent page for their information.

While it is assumed that the ordering provider has reviewed the risks, benefits, and limitations of the ordered tests with the patient before submitting any specimen to CGL, it is recommended that the provider or patient confirm consent to testing. Please make elections below:

☐ Check this box if the patient wishes to OPT OUT having de-identified data used for research.

Date	Relationship to Patient ☐ Ordering Provider ☐ Self ☐ Legal Guardian
Consenting Individual (Print)	Consenting Individual Signature

Specimen Send Out Details

A completed test requisition for the recipient laboratory MUST be sent to CGL before shipping can occur. Charges from the recipient laboratory CANNOT be billed to CGL.	
Recipient Laboratory	Test(s) Requested
Lab Address	Specimen to Send: ☐ Cultured amniocytes ☐ Cultured chorionic villi ☐ Uncultured amniocytes ☐ Uncultured chorionic villi ☐ Extracted DNA ☐ Other: _____
Lab Phone	Volume/Specimen Requirements
Lab Fax	Lab Contact Person
FedEx Billing # or send prepaid label	Other Information

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Provide this page to the patient/legal guardian and discuss these points before ordering chromosomal microarray.

Consent for Chromosomal Microarray (CMA)

Purpose and Benefits of Testing

- The purpose of a chromosomal microarray (CMA) is to look for extra or missing pieces of DNA. These duplications or deletions are referred to as Copy Number Variants (CNVs). CMA is able to detect CNVs which may be too small to see by standard chromosome analysis and provides more accurate information on the size of a duplication or deletion and the gene content.
- While all humans have small CNVs, most do not cause illness. However, depending on what genes are included in the duplication or deletion, some may cause significant genetic conditions and health concerns in the individual. Finding a genetic diagnosis by CMA can be useful in understanding an individual's current and future symptoms, forming a specific care plan, and knowing if other family members could also be at risk for the same illness.
- CMA is recommended by multiple national medical boards as a possible first step in genetic testing for individuals who have autism spectrum disorder, intellectual disability, and/or congenital malformations. Individuals with other symptoms may also benefit from CMA testing based on the recommendation of their provider.
- Regardless of results, consultation with a genetic counselor to review and understand CMA testing and results is strongly recommended. Your provider can refer you to a local genetic counselor.
- Patients or providers with additional questions on the details of CMA testing, limitations, risks, and benefits can contact the Colorado Genetics Laboratory (CGL) genetic counselor line at 303-724-7075. Please note formal genetic counseling for a patient about specific genetic testing and results is not provided by CGL and a clinical genetic counseling session is recommended.

Possible Results

- CGL will report CNVs with potential clinical relevance. CNVs known to be Benign or Likely Benign are not typically reported because they are not predicted to impact a person's health.
- Clinically significant CNVs will be reported to you and your doctor. These include, but are not limited to:
 - Pathogenic/Likely Pathogenic:* These duplications or deletions are known (or strongly suspected) to cause a genetic condition and related health issues in the patient. Details on expected symptoms based on other reported patients will be provided.
 - Variant of Uncertain Significance (VUS):* These duplications or deletions are not common in healthy or sick people, so current science is not able to say if the CNV is the reason for the patient's symptoms. VUSs usually do not indicate a diagnosis and may not change a patient's care plan
 - Risk for Autosomal Recessive Condition:* These CNVs are not problematic on their own but may be one of several changes leading to the patient's symptoms. CGL reports these CNVs so your provider can consider additional testing for a specific condition.
 - Absence of Heterozygosity (AOH):* Sometimes, large areas of the DNA are identical (homozygous). While large identical stretches are not necessarily problematic, they can sometimes indicate an underlying genetic change that CMA cannot identify. These areas are reported so that your provider can focus additional testing efforts, if relevant.
- For Pathogenic, Likely Pathogenic, and VUS results, the testing of biological parents or other family members may be recommended to know if the CNV was inherited from a parent or occurred for the first time in the patient (*de novo*). This information may help clarify the clinical significance of the CNV and risk to other family members.
- The interpretation of CMA results can be dependent on accurate medical information shared with the laboratory. **It is recommended to submit an accurate and complete clinical description of symptoms and family history with the specimen for best results.**

Risks and Limitations of Testing

- CMA does not detect every possible genetic condition and is often just one part of the genetic testing strategy to diagnose an affected person. Therefore, a negative (normal) CMA result does NOT mean the patient has no genetic condition, only that a diagnosis cannot be made from CMA alone. In pregnancy a negative CMA for a fetus does not guarantee a healthy newborn. Details on the technical limitations of CMA can be found on the CGL website.
- CMA is considered a highly reliable test for its scope of evaluation. However, as with all types of testing, inaccurate results may occur. Such situations are rare but may be caused by mislabeled specimens, damage in specimen transportation to the laboratory, inaccurate medical information provided to the laboratory, technical errors, or other reasons. An additional specimen may be requested in such situations to achieve more accurate results.
- Unexpected results unrelated to the patient's symptoms are possible and may be reported in some situations. **Some people find such unexpected results to be especially distressing, while others find the information medically valuable.** Unexpected results may include, but are not limited to:
 - A genetic condition may be found that was unrelated to the patient's symptoms, such as a significantly increased risk for cancer, etc. Family members may be found to also be at risk of the identified condition.
 - Testing results may reveal the biological parents of the patient being tested are closely related to each other.
 - Testing of parental specimens may reveal cases of non-paternity/misattributed parentage. Such situations may limit the laboratory's ability to determine the inheritance of a CNV. However, CGL does not provide formal paternity testing and therefore **will NOT include information about misattributed parentage on testing reports.**

Information Security and Research

- CGL follows all HIPAA requirements to safeguard the privacy of your medical information. Your results, medical information, and acknowledgement that testing has been performed at CGL will only be provided to providers involved in your care, to you, and to others you expressly provide consent to have the information, or as required by law. Federal law prohibits unauthorized disclosure of your private medical information and genetic results.
- CGL typically retains extracted DNA from samples with positive (abnormal) CMA results indefinitely. Extracted DNA from negative CMAs are typically held for 6 months before discard. These de-identified samples may be used for research, test development and improvement, training, quality assurance, and other laboratory functions.
- In some cases, this extracted DNA can be sent to other laboratories for additional testing if your provider deems it relevant. You have the right to request that CGL immediately discard any remaining specimen after testing is complete. You may also request any remaining specimen be returned to you after testing, though this request must be received at the time of testing.
- De-identified health history and genetic results can help expand scientific and medical understanding of how our DNA affects health. Sharing anonymized data with researchers and genetic databases improves the ability to make discoveries, identify/name rare disorders, and improve the effectiveness of genetic testing over time. CGL may contribute the findings of genetic tests performed to such databases or scientific literature (through research papers), though all names, birthdates, and patient identifiers are removed and replaced with a unique code to protect patient identity. However, there is a small risk that a patient may be identified by nature of the rare genetic finding they have. While this risk is believed to be very low, it may be increased if someone has already shared their genetic information with a public resource, like ancestry websites or other entities.