

## PREFERRED SPECIMEN REQUIREMENTS

PLEASE CONTACT US WITH ADDITIONAL SPECIMEN REQUIREMENT QUESTIONS.

### WHOLE BLOOD

Collect 3 ml - 5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube), minimum 1 ml for small infants.

### DNA

Send in screw cap tube at least 5 µg -10 µg of purified DNA at a concentration of at least 20 ng/µL for NGS and Sanger tests and at least 5 µg of purified DNA at a concentration of at least 100 ng/µL for gene-centric aCGH, MLPA, and CMA tests, minimum 2 µg for limited specimens. Indicate concentration on tube label. For requests requiring more than one test, send an additional 5 µg DNA per test ordered when possible.

### SALIVA

Oragene™ or GeneFix™ Saliva Collection kit used according to manufacturer instructions.

### FETAL (CVS / AMNIOCYTES) AND OTHER CELL CULTURES

Culture and send at least two, T25 flasks of confluent cells. For sequencing or gene-centric aCGH panels, two flasks are often sufficient; however, some panels may require additional flasks (dependent on size of genes, amount of Sanger sequencing required, etc.). Multiple test requests may also require additional flasks. Please contact us for details. We strongly recommend maintaining a back-up culture. Fetal cell cultures are available at PreventionGenetics from direct amniotic fluid, chorionic villi, products of conception (POC), or other tissue via Test Code #995 (cost \$250). Collect 10 ml - 20 ml of direct amniotic fluid or 5 mg - 10 mg cleaned CVS tissue (~15-20 cleaned villi) or 2mm x 2mm x 2mm fresh tissue. CPT code 88235 for amniotic fluid/chorionic villi or 88233 for POC or tissue specimens.

### BLOOD

**DO NOT FREEZE.** During hot weather, include a frozen ice pack in the shipping container. Place a paper towel or other thin material between the ice pack and the blood tube. In cold weather include an unfrozen ice pack in the shipping container as insulation. At room temperature, blood specimen is stable for up to 48 hours. If refrigerated, blood specimen is stable for up to one week.

### DNA

DNA may be shipped at room temperature.

|            | TEST METHOD                    | WHOLE BLOOD | DNA            | SALIVA          | CELL CULTURES  | FRESH, FROZEN TISSUE | OCD-100 BUCCAL SWAB | DIRECT AMNIOTIC FLUID/CVS | OTHER              |
|------------|--------------------------------|-------------|----------------|-----------------|----------------|----------------------|---------------------|---------------------------|--------------------|
| SEQUENCING | NextGen (NGS)                  | ★           | ★              | ★               | ★              | ★                    | ■                   | ■ <sup>C</sup>            | -                  |
|            | PGxome® / PGxome Custom Panels | ★           | ★ <sup>B</sup> | ★               | ★ <sup>B</sup> | ★                    | ■                   | -                         | -                  |
|            | Sanger                         | ★           | ★              | ★               | ★              | ★                    | ■                   | ■ <sup>C</sup>            | Semen <sup>D</sup> |
| DEL / DUP  | Gene-centric aCGH              | ★           | ★              | ■               | ■              | ★                    | -                   | ■ <sup>C</sup>            | -                  |
|            | MLPA                           | ★           | ★ <sup>A</sup> | ONLY TEST #1941 | ★ <sup>A</sup> | -                    | ■                   | -                         | -                  |
|            | Chromosomal Microarray (CMA)   | ★           | ★              | ■               | ★              | ★                    | -                   | ■                         | -                  |

### EXCEPTIONS

- A - Cell cultures and DNA extracted from CVS and amniocytes not accepted for MLPA; DNA extracted from saliva (except test #1941) also not accepted.
- B - Cell cultures and DNA extracted from CVS and amniocytes acceptable for PGxome for non-ongoing pregnancies only.
- C - Direct prenatal specimen types most appropriate for targeted prenatal familial variant testing (Test Code #990), and strongly discouraged for full gene and panel tests. Back-up culture highly recommended.
- D - Semen: Collect 1-2 vials and flash freeze. Vials to be sent frozen (preferably on dry ice). Contact us for details.

### KEY

- ★ PREFERRED
- ACCEPTED
- NOT ACCEPTED

### FRESH, FROZEN TISSUE

Collect 2mm x 2mm x 2mm tissue and flash freeze. Tissue to be sent frozen (preferably dry ice). If cell culture is desired, tissue should not be frozen. Contact us for additional details.

### OCD-100 BUCCAL SWAB

OCD-100 Buccal Swab used according to manufacturer instructions. Buccal swabs are most appropriate for targeted, known variant testing.

### DIRECT AMNIOTIC FLUID / CHORIONIC VILLI

Collect 10 ml -20 ml of direct amniotic fluid or 5 mg -10 mg cleaned CVS tissue (~15-20 cleaned villi). We strongly recommend maintaining a local back-up culture. Fetal cell cultures available (Test Code #995, \$250).

## SHIPPING AND HANDLING INSTRUCTIONS

Label all specimen containers with the patient's name, date of birth and/or ID number. At least two identifiers should be listed on specimen containers. Specimen deliveries are accepted Monday-Saturday for all specimen types except cell cultures, direct amniotic fluid, or direct chorionic villi. Cell culture deliveries are routinely accepted Monday-Thursday and require advance notice of arrival. If a Friday or Saturday delivery is necessary, contact us to make arrangements. Saturday delivery should be avoided when possible as prenatal specimens are not processed over the weekend. Holiday schedules will be posted on our website at least one week prior to major holidays.

**DO NOT FREEZE.** During hot weather, include a frozen ice pack in the shipping container. Place a paper towel or other thin material between the ice pack and the blood tube. In cold weather include an unfrozen ice pack in the shipping container as insulation. At room temperature, blood specimen is stable for up to 48 hours. If refrigerated, blood specimen is stable for up to one week.

### DNA

DNA may be shipped at room temperature.

Label the tube with the composition of the solute, DNA concentration as well as the patient's name, date of birth, and/or ID number. We only accept genomic DNA for testing. We do not accept products of whole genome amplification reactions or other amplification reactions.

### CELL CULTURES, DIRECT AF/CVS, AND POC

Send specimens overnight in an insulated, shatterproof container. Direct AF/CVS, POC, or tissue specimens can be sent in saline or culture media at room temperature for culturing at PreventionGenetics (Test Code #995, \$250).

### PRENATAL TESTING

Please sign Prenatal Healthcare Provider's Statement for ongoing pregnancies and contact us in advance regarding prenatal test requests. When possible, ship prenatal samples to arrive at PreventionGenetics no later than Thursday.

### DNA GENOTYPING PANEL

For quality control purposes, the Prevention Genetics DNA Genotyping Panel is performed on all clinical specimens. Genotyping results are not included in test reports.

### DNA BANKING

DNA Banking has a reduced price of \$98 for

patients if clinical testing is also being performed at PreventionGenetics. Visit our website at [www.PGDNABank.com](http://www.PGDNABank.com) for more information. For questions about PGDNABanking, contact us at (715) 387-0484, ext. 151, or email: [dnanbanking@preventiongenetics.com](mailto:dnanbanking@preventiongenetics.com).

### CONTACT US

For additional questions or concerns, contact a Client Service Representatives at (715) 387-0484, ext. 0, or our Genetic Counseling Team at option 2, or email: [clinicaldnatesting@preventiongenetics.com](mailto:clinicaldnatesting@preventiongenetics.com).

### MAILING ADDRESS

PreventionGenetics - Diagnostic Lab  
3800 S. Business Park Avenue  
Marshfield, Wisconsin 54449 USA

### TESTING KITS

Clinical testing kits with prepaid return shipping are available for U.S. Clients. Clinical testing kits are provided to International clients without the return postage. Submit requests through our website or contact a Client Service Representatives at (715) 387-0484, ext. 0.