

## SPECIMEN REQUIREMENTS AND TURNAROUND TIMES (TAT)

### PREVENTIONGENETICS PREFERRED SPECIMEN TYPES

PLEASE CONTACT US WITH ADDITIONAL SPECIMEN REQUIREMENT QUESTIONS.

STAT TAT (8-10 calendar days) available at a 25% surcharge for Sanger Sequencing and aCGH. Cannot be guaranteed for aCGH.

### 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™ 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. Contact us for details. We strongly recommend maintaining a local back-up culture; PreventionGenetics does not culture cells.

TEST METHOD		WHOLE BLOOD	DNA	SALIVA	CELL CULTURES	FRESH, FROZEN TISSUE	BUCCAL SWABS	DIRECT AMNIOTIC FLUID/CVS	OTHER	TURN AROUND TIME (TAT)
SEQUENCING	NextGen (NGS)	★	★	★	★	★	■	■ <sup>C</sup>	—	28 days
	PGxome™/ PGxome Custom Panels	★	★ <sup>B</sup>	★	★ <sup>B</sup>	★	—	—	—	6 weeks
	Sanger	★	★	★	★	★	■	■ <sup>C</sup>	Semen <sup>D</sup>	18 days
DEL / DUP	Gene-centric aCGH	★	★	■	■	★	—	■ <sup>C</sup>	—	28 days
	MLPA	★	★ <sup>A</sup>	ONLY TEST #1941	★ <sup>A</sup>	—	—	—	—	20 days
	Chromosomal Microarray (CMA)	★	★	■	★	★	—	■	—	20 days

### 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). Contact us for additional details.

### BUCCAL SWABS

Buccal swabs are most appropriate for targeted, known variant testing. Collect 3-6 buccal swabs for targeted,

known variant testing and 10-20 buccal swabs for sequencing of full gene(s).

### DIRECT AMNIOTIC FLUID/CHORIONIC VILLI

Collect 10 ml -20 ml of direct amniotic fluid or 5mg -10mg cleaned CVS tissue (~15-20 cleaned villi). We strongly recommend maintaining a local back-up culture.

## SHIPPING & HANDLING INSTRUCTIONS

Please 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. We accept specimen deliveries 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, please contact us to make arrangements. Saturday delivery should especially be avoided when possible as prenatal specimens are not processed over the weekend. Holiday schedules will be posted on our home page at least one week prior to major holidays.

### 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. 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 AND DIRECT AF/CVS

We are NOT able to culture cells. Send specimens overnight in an insulated, shatterproof container.

### ADDRESS

Diagnostic Lab  
PreventionGenetics  
3800 S. Business Park Ave.  
Marshfield, WI 54449  
USA

### TESTING KITS

Clinical testing kits with prepaid return shipping are available for U.S. Clients. We are able to provide clinical testing kits to International clients without the return postage. To order test kits, submit requests through our electronic order form (see website) or contact our Client Service Representatives at (715) 387-0484, ext. 0.

### 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. PreventionGenetics does not culture cells.

### DNA GENOTYPING PANEL

For quality control purposes, the PreventionGenetics 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 for information about the process and forms. For questions related to PGDNABanking, contact our DNA Banking Director at (715) 387-0484, ext. 151, or email: dnabanking@preventiongenetics.com.

### CONTACT US

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