

CP 1.8 Phenylalanine (Phe) Monitoring

POLICY:

1. The Genetic Disease Laboratory (GDL) will perform phenylalanine testing for individuals with hyperphenylalaninemia who are treated at California Children’s Services (CCS)-approved metabolic centers. This testing is an integral and essential component of diet monitoring, which is required to prevent disabilities resulting from this untreated disorder.
2. All specimens shall be collected and sent to GDLB according to this protocol. **Testing at GDL is done on non-holiday weekdays only; no testing is done on weekends or holidays.**
3. All specimens received by GDL will be accessioned and entered into SIS the same day they are received. They will appear on the case summary page in the “Additional Results” grid.
4. Test results will appear the second day after accession (Monday – Friday) and are available for viewing in SIS (e.g. specimens received on Tuesday before noon are accessioned and the TRF information is entered into SIS on Tuesday. If specimen is run on Tuesday, results are reviewed on Wednesday, and released to SIS during night processing. Results will be available for viewing in SIS on Thursday morning).
5. A Result Mailer will be sent to the specimen collection facility indicated on the form.

PROTOCOL:

Resp. Person	Action
GDSP Warehouse Staff	<ul style="list-style-type: none"> • Distributes <i>Test Request Form For Phe Monitoring Test</i> and pre-paid, pre-addressed mailing envelopes to Metabolic Centers upon request.
Person collecting specimen	<ul style="list-style-type: none"> • Completes all demographic fields on the <i>Test Request Form for Phe Monitoring Test</i>, following instructions on the front of the form. • Collects blood specimen on the <i>California Newborn Screening Specimen Collection Card</i>, following instructions on the back of the card. • Fills the first three circles with blood. • Dries specimen completely (horizontally to avoid pooling of blood). • Mails original white sheet of TRF along with the specimen to Genetic Disease Laboratory (GDL) via regular mail in supplied envelopes • (If a faster turnaround than noted above in policy is required, send specimen by overnight courier. Mail to: Genetic Disease Laboratory 850 Marina Bay Parkway, C227 Richmond, CA 94804 • <u>If overnight package is sent on a Friday for Saturday delivery</u>, writes the following instructions on the envelope “No Signature Required – deposit parcel in slot at shipping door at rear of building”.
GDL Staff	Accessions specimen, runs test, reviews and approves run, and enters results into SIS.
Metabolic Center ordering Phe Monitoring	Reviews results in SIS and files Result Mailer in medical record, when received.