

**DEPARTMENT OF PATHOLOGY AND LABORATORY MEDICINE  
MOLECULAR GENETICS LABORATORY****DNA TESTING PROTOCOL****CATHERINE A. STOLLE, PH.D., DIRECTOR****SPECIMEN REQUIREMENTS**

**\*Blood:** 3-5 ml of blood in EDTA (purple top tubes). Transfer or ship to the laboratory at ambient temperature within 24 hours. Blood samples may be refrigerated prior to delivery if shipment is delayed. Do not freeze.

**\*Cultured Cells** (including skin fibroblasts, amniocytes, or chorionic villi cells): 2 nearly confluent T-25 flasks filled with media. Please keep backup cultures growing in your laboratory until results of testing have been received. Please send a maternal blood sample (EDTA tube) with fetal samples for maternal contamination studies.

**\*Frozen Tissue:** Fresh surgical specimens or biopsy material (~1 cm<sup>3</sup>) may be frozen directly or in OCT (used for surgical frozen sections) at -20°C or -70°C and transferred to the laboratory on dry ice.

**\*Other Specimens:** High molecular weight genomic DNA, cheek epithelial cells, or other samples containing DNA may be acceptable. Contact the laboratory for specific instructions regarding such samples.

**NB: All samples must be labeled with the patient's full name, date and time of draw, and phlebotomist's (or collector's) name.**

**DOCUMENTATION**

Each specimen must be accompanied by a **Physician's Test Request**. **Informed Consent** is desirable. A **Financial Responsibility form** should accompany samples not originating within CHOP. Please provide the clinical indicator(s) for testing, and the type of testing requested. In addition, the following clinical information is specifically requested:

Alpha / Beta Thalassemia:	Hemoglobin electrophoresis results (Hb A <sub>2</sub> and F quantitation); CBC results (MCV); results of iron studies, if available.
CADASIL	Disease status (symptomatic / asymptomatic); results of skin punch biopsy on symptomatic patients; <b>NB: Informed Consent is mandatory.</b>
DMD / BMD:	On affected individual: CPK results, muscle biopsy, dystrophin analysis report and/or DNA results and clinical history.
Friedreich's ataxia:	Clinical description of patient /affected family member or relationship of consultand to affected family member.
Sickle Cell Disease/Trait:	Hemoglobin electrophoresis results on parents.
von Hippel-Lindau:	Type/location of tumors (uni- or bi-lateral). Family history. Results of previous molecular analysis, if available. Clinical criteria form is available upon request.

**SHIPPING AND CONTACT INFORMATION**

Samples should be shipped by overnight carrier to arrive Tuesday-Friday by 4:00pm. There is no one in the laboratory on evenings or weekends to receive samples. Other arrangements can be made by calling Dr. Stolle.

**NB: Sender is responsible for shipping charges. Samples marked "Bill To Recipient" will be declined.**

**\*Shipping Address:** Catherine A. Stolle, Ph.D.  
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Abramson Research Center, Room 714G  
3615 Civic Center Boulevard  
Philadelphia, PA 19104-4318  
215-590-8736 (Office) or 215-590-4404 (Lab)