University *of* Pittsburgh Medical Center
Molecular Diagnostics Division

MOLECULAR GENETIC TESTS

Laboratory Mailing Address: Univ. of Pittsburgh Dept. of Pathology
Division of Molecular Diagnostics
Room S780 Scaife Hall
3550 Terrace Street
Pittsburgh, PA 15213-2500

Division Office: (412) 648-8519 **FAX:** (412) 383-9594

IMPRINT PATIENT IDENTIFICATION PLATE HERE

Instructions:

1. Complete **all** information as requested. Type or print **clearly**.
2. Send the lab a copy of this requisition form with the specimen.
3. Specimen(s) will only be accepted Monday through Friday; see "Specimen Requirements" for weekend storage suggestions.

Specimen Types/Requirements (Please check the sample type submitted):

☐ **Peripheral Blood:** 5-10 ml of fresh anticoagulated (purple top/ EDTA preferred; yellow or black/citrate tubes may also be used). If storage is necessary, refrigerate at 4°C. If 5 ml can not be collected, send what you can obtain. Blood may be shipped at ambient temperature, although take care to protect from freezing during the winter.

☐ **Buccal Swabs:** Please contact the laboratory for specific instructions on how to collect adequate buccal cells (cheek brushes) for analysis.

☐ **Other Tissue or Tumor Specimens:** Frozen or fresh tissue may be used. A minimum of 2 x 2 x 2 mm is required (5 x 5 x 5 mm is preferred). The preferred approach is to snap freeze tissue directly or as cell pellets at -20 or -70°C and mail with sufficient dry ice to prevent thawing before arrival at our laboratory. Less desirable, but often successful, is to send samples to the lab in saline, chilled as described for CVS tissue above.

☐ **Paraffin Sections:** 10 sections on glass slides. More sections may be required if the tissue is small. Please call the lab if you have questions.

☐ **Cultured Cells:** Cultured cells (amniocytes, skin fibroblasts, EBV-transformed lines) may be sent at ambient temperature in culture flasks completely filled with media. One near-confluent T-75 flask is ideal; two T-25 flasks are also acceptable. Please keep backup cells growing in another laboratory until you receive results from us in case of sample loss in shipment for analysis.

☐ **Amniotic Fluid or Chorionic Villus (CVS):** We prefer cultured cells. Direct CVS samples may be submitted if previously arranged with the laboratory. CVS (10-50 mg dissected free of maternal tissue) and other tissue for same day or overnight delivery should be placed in sterile isotonic saline or tissue culture medium and sent to the lab chilled on wet ice. Facilities are available through our Clinical Cytogenetics Laboratory at additional charge for primary culture of amniotic or CVS cells if necessary.

Sample Information Collection Date: Collection Time: ICD-9 code (required):

Patient Identification:

Name: SS#: Sex: ☐ M ☐ F Birth date:

Ordering Facility: Ethnicity:

Facility Address: Phone#: ( )

FAX#: ( )

Requesting Physician Name (Required):

Physician Address: Phone#: ( )

FAX# ( )

Name of Person Responsible for Payment: Phone# ( )

Billing Address:

Insurance Information:

Please attach a clinical summary, family history and pedigree. For family studies, one specimen from each family member to be tested should be submitted (please list any known mutations that exist in the family, if applicable).

Please check box ("X") indicating test(s) requested:

☐ Factor II (Prothrombin 20210G>A) Analysis
☐ Factor V Leiden (*F5* gene)
☐ Fragile X Syndrome (*FMR1* gene)
  ☐ Southern blot (used for screening) ☐ PCR analysis (if needed)
☐ Hemochromatosis (*HFE* gene, C282Y and H63D variants)
☐ Hereditary Pancreatitis (*PRSS1* gene, R122H, N29I, A16V mutations)
☐ **Hereditary Paraganglioma^ (Can order each gene separately – see below)
  ☐ *SDHD* ☐ *SDHB* ☐ *SDHC* (Method: Sequencing**)

A separate clinical information sheet for paraganglioma testing is available on our web site and will be faxed for completion if not submitted with sample.

☐ Huntington Disease (*HTT* gene): Informed consent required for presymptomatic testing)
☐ **Isovaleric Acidemia (Isovaleryl-CoA-Dehydrogenase, IVD) gene sequencing
☐ **Malignant Hyperthermia (RYR1 gene sequence analysis – 17 high risk regions)
☐ **MCAD Deficiency (Medium Chain Acyl CoA Dehydrogenase), ACADM gene exon 11 - detects common and other mutations
☐ **Multiple Endocrine Neoplasia, MEN 2A and 2B (*RET* oncogene sequencing)
☐ Mitochondrial Mutation Panel (Can order each group separately)
  ☐ MELAS ☐ MERRF ☐ NARP
☐ MTHFR, 677C>T Thermolabile variant
☐ **RETT Syndrome (MECP2 gene sequence analysis)
☐ Spinal Muscular Atrophy, autosomal recessive (types I, II, III)
☐ Spinal Muscular Atrophy, X-linked form (Kennedy's disease, androgen receptor gene)
☐ *SPINK1* (Serine peptidase inhibitor, Kazal type 1), pancreatitis
☐ **Von Hippel Lindau Disease (*VHL* gene sequence analysis)
☐ Other (Please state genetic test requested): ______________
☐ **Is there a known mutation in the family? Indicate gene and mutation.

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Note: It is the responsibility of the patient's physician to obtain proper informed consent for genetic testing.

** check box at end of menu if testing for a KNOWN mutation (usually applies to gene sequencing assays, but may be relevant for others)

PCR tests are performed pursuant to a license from Roche Molecular Systems.

Revised 11/03/2011