

## Remittance of samples for investigation of mitochondrial energy metabolism **Salzburg/Austria**

For an optimal interpretation of the results of the investigations performed, a detailed medical record is required. Analytical services offered by our facility are outlined on our web site <http://www.mito-center.org>

<b>Patient Data</b>  <input type="checkbox"/> female <input type="checkbox"/> male  name	type of specimen, date of collection
date of birth	suspected diagnosis

### **Biochemical screening of intact mitochondria**

**Specimen:** min 30 mg of native, unfrozen muscle tissue (after removal of fat and connective tissue)  
**Shipping (prior consultation with our laboratory required: Tel. +43 (0)5 7255-26281)**  
Muscle tissue should be weighed (please specify weight) and immediately placed into MITO-shipping buffer (10-fold volume), e.g. 50 mg of muscle tissue + 500 µl shipping buffer.  
Immediate shipment at 0°C (water/ice mixture) in isolated container via express carrier, taxi etc. to ensure arrival of specimen within 5 hours after collection.  
 shipment of MITO-shipping buffer is requested

### **Enzymes – mitochondrial energy metabolism**

**Specimen:** min 20 mg of frozen muscle tissue (after removal of fat and connective tissue), fibroblasts  
**Shipping** (notification of our laboratory desired):  
Send tissue specimen on dry ice (approx. 2 kg) in a thermally isolated container via express carrier.  
Fibroblasts culture with approx. 70 % confluency, filled up with culture media and sealed tightly.  
Ship samples early in the week.  
Do not ship specimens which may arrive Saturday, Sunday or holiday!

### **Genetic testing**

#### **Suspected mitochondrial syndrome:**

- MELAS    MERRF    LHON    NARP    maternal Leigh    CPEO    Kearns-Sayre    Pearson  
 Depletion Syndrome  
 POLG  
 Nuclear Genes: for detailed offer see [www.mito-center.org](http://www.mito-center.org) - Genetic testing  
Because of diversity of investigations prior consultation with Dr. Hans Mayr is essential:  
phone: +43 (0)5 7255-26281      e-mail: [h.mayr@salk.at](mailto:h.mayr@salk.at)

**Specimen:** min 10 mg of muscle tissue, fibroblasts, 10 ml urine or 2 ml EDTA-blood  
(shipments of muscle tissue and fibroblasts see „enzyme-mitochondrial metabolism“, shipment of blood at room temperature).

**Please note:** Mutations of mitochondrial DNA (mtDNA) are not always detectable in blood samples, but in muscle tissue (e.g. deletions in Kearns-Sayre or CPEO)

Date: ..... Signature: .....