

## Metabolic Muscle Biopsies

A muscle biopsy is a surgical procedure in which one or more small pieces of muscle tissue are removed for further microscopic or biochemical examination. The procedure, often used in the diagnosis of a neuromuscular disorder, is considered "minor" surgery and is usually performed under local anesthetic.

A doctor is likely to order a muscle biopsy after looking at preliminary tests which may include blood tests, performing an electromyogram (EMG) and physical examination, and determining that your symptoms may indicate an underlying neuromuscular disorder.

To do the biopsy, the surgeon will make an incision over the quadriceps (thigh) muscle. The incision is usually 1-1 ½ inches long. The area is first frozen with a numbing medication called lidocaine (similar to what most dentists use). This part can be mildly uncomfortable. Once the numbing medicine has taken effect, however, you should not feel any pain. During the procedure, you may be aware of pressure around the surgical site but this should not be painful. The amount of muscle tissue taken is approximately the size of an eraser head on the end of a pencil. It does not leave you with any weakness. The incision is closed with dissolvable stitches. It may feel sore for a few days after the procedure. Usually plain Tylenol is sufficient to manage this pain.

### **Planning for the biopsy**

- You will need to make sure you have someone to drive you home from the biopsy (the procedure will take 30-45 minutes)
- If you are taking **coenzyme Q10, carnitine, creatine, riboflavin, thiamine, or alpha-lipoic acid**, please stop taking them for one month prior to the biopsy. Once you have the biopsy, you may resume taking these supplements.
- Avoid taking **aspirin** for 7 days prior to the procedure as it may increase bleeding.
- If you are taking **coumadin, warfarin, fragmin or plavix** please notify the muscle biopsy clinic immediately
- Plan to be at VGH at least 15 minutes prior to the procedure and proceed to the **Diamond Health Sciences Centre, 3<sup>rd</sup> floor, Minor Surgical Procedures Clinic** to check in.

### **After care**

- Minimize weight-bearing for 24 hours.
- The surgeon will apply tapes across the incision and then place a clear, plastic dressing called an Opsite on top. You can remove this dressing in three days. Leave the tapes on the incision until they peel off on their own. A little bit of bleeding and swelling are to be expected. If you are bleeding a lot from the site, see your family physician or go to the nearest hospital emergency room.
- You may shower the day after the surgery. Do not have a bath for five days. You want to avoid immersing the incision in water.
- The local anesthetic will wear off one hour after the procedure.
- The stitches will dissolve on their own. They are buried under your skin.
- Refrain from heavy exercise for 2 weeks after the surgery.

## **Analyzing the Sample**

When the muscle samples are sent to a laboratory for analysis, the technicians cut them into many thin sections for examination. Using different tests on different sections, they look at the tissue's overall appearance, chemical activities in the tissue, and the presence or absence of critical proteins. The information these tests provide helps determine whether or not you have a mitochondrial disorder.

Histology tests (histo means tissue) employ chemical stains to see the muscle's overall appearance and the structure of the muscle cells. This analysis can yield information about mitochondrial abnormalities.

Histochemistry uses stains to detect chemical activities in the cells, including the actions of specific enzymes and metabolic processes.

Immunohistochemistry uses antibodies to detect the presence or absence of proteins. This analysis can show whether the mitochondria are missing enzymes.

The lab may also use electron microscopy to get a very high magnification view of the cellular structure, which can confirm structural abnormalities of the mitochondria.

Finally, based on the results of your muscle histopathology testing, your physician may request mitochondrial DNA testing which can be performed on a portion of the muscle sample. This test may detect a genetic mutation (change) in the mitochondria. Although a blood sample is usually adequate for testing of nuclear DNA (the DNA inherited from both your parents), a muscle sample is needed to test for mitochondrial DNA mutations. Your physician and clinic nurse will discuss this testing with you at your appointment.

The surgeon usually takes the muscle sample in four to five separate pieces. However, the total amount taken is no larger than an eraser head on the end of a pencil. Having at least four muscle samples from the biopsy gives the lab an adequate amount of tissue to work with.

## **Getting Results**

The analysis of a muscle biopsy sample is a very tedious and labor-intensive process in which many sections of the muscle must be cut, many different types of procedures performed, and the results carefully analyzed. The full set of results can take up to one year.

The results will be sent to the Adult Metabolic Diseases Clinic. Results will be discussed with you at your scheduled one year follow-up appointment, unless abnormal results are reported before that date. You will be contacted by the Adult Metabolic Diseases Clinic to discuss the results with your treating physician.