Neurology Fact Sheet

Neurology is the study of the various nervous systems—central, and peripheral muscles plus the neuromuscular junction and their disorders.

There are three types of muscles in our body:

- **Skeletal muscle**: this is the type of muscle that carries out our voluntary actions, like walking, manipulating objects with our hands, blinking and breathing. Some of these actions are automated, like breathing and blinking, but can be interrupted or initiated when we want to.
- **Heart muscle**: this is the part of the heart that contracts and relaxes unintentionally day and night all life long.
- **Smooth muscle**: this is the muscle that surrounds our stomach, blood vessels and sweat glands. The action of this muscle is also unintentional, although emotions may influence its working.

Of these three types, the affected muscles in Barth syndrome are the skeletal muscle and heart muscle.

Barth syndrome manifests itself neurologically in several different ways:

- **Myopathy**—a disorder of muscle tone, weakness, hypotonia. In Barth syndrome, the amount of muscle is often diminished, although not all muscles are affected to the same degree. The muscles that move the eyes are rarely involved, neither is the diaphragm, which controls breathing. The muscles that appear to be mostly affected are leg muscles, muscles of the pelvic girdle and shoulder muscles. This affects a Barth individual's daily life and independence. Having weak muscles, or a reduced muscle mass, creates difficulty walking up stairs, lifting objects, especially above the individual's head, and also with smaller issues such as turning door handles, and opening jars. Often the reduced muscle mass of a Barth individual does not become apparent until they have reached their second decade of life. This does not mean that there is muscle wastage, but it is simply due to their body composition changing. To assist individuals with sitting and walking, different orthotic devices can be used to help with low tone. Many individuals also have a flat arch, and a device may help with balance. If it is thought necessary or helpful, the individual should be referred to a specialist who can help make the right decisions in what would be useful and how it should be used.

Due to the low muscle tone, and general weakness, most Barth syndrome individuals reach the motor development milestones in the upper normal limits of age. Many do not crawl, but they do walk, “on time” sometimes with an abnormal gait with their feet turned out. While the power of the involved muscles vary, there is no tendency to get worse, and even after a Barth individual has had a “bad” period, the muscle condition tends to go back to its original level. It is unclear why, but many of the Barth individuals also are “clumsy” and many do not place their hands out to brace themselves when falling. Many individuals also have a “nasal” speech; this is caused by a high arch palate which is common in people with low tone. Usually with a muscle disorder, the individuals' reflexes are normal or slightly decreased, however, in Barth syndrome, it has been found that some may have hyperreflexia, which suggests a dysfunction in the central nervous system, but has no functional importance.

- **Fatigue**—the majority of Barth syndrome individuals report that fatigue is an issue that affects them daily or more than once a week. Although it is difficult to quantify fatigue, it obviously does limit the individuals' function, abilities and endurance. Many children with Barth syndrome are unable to spend the whole day at school without a break in between, and older individuals have difficulty with a regular schedule. Individuals over ten have also reported that they have a specific fatigue when they are writing, and most use different methods to accommodate their needs, either using technology, or having a scribe. To cover greater distances, wheelchairs, motorized bikes and scooters can be used.
• Myalgia—refers to muscle aches and pains. This is predominantly in the calves with Barth patients, and no cause has been found. For the most part the myalgia is worse if the individual has had an active day. Although headaches are very common in the general public there seems to be an abnormally high number of Barth syndrome individuals who report having headaches; some weekly, others monthly. Many individuals indicate that although the headaches are frequent they are not particularly severe, nor do they last long. Caution should be taken when using over-the-counter medication as overuse can produce headaches. Several individuals also comment on having “other pain” not in the muscles, which is generally abdominal, in the joints or feet.

Glossary

**Contractures**: A permanent abnormal tightening or shortening of a body part, such as a muscle

**Floppy baby syndrome**: An abnormal condition of newborns and infants where there is inadequate tone of the muscles.

**Gower’s Sign**: Gower’s sign refers to an abnormal method of attempting to stand up, in which the person begins with both arms and legs on the floor and then uses the arms to push against the legs and raise up the trunk to an upright posture. Sometimes described as “climbing up the legs.”

**Hypotonia**: A condition of diminished tone of the skeletal muscles, and the reduced resistance of muscles to passive stretching.

**Lordosis**: An inward curving of the spine in the lower part of the back.

**Neurologist**: A physician who diagnoses and treats disorders of the central nervous system.

**Occupational therapist**: An individual trained to help people manage the daily activities of living, such as dressing, grooming or cooking and regaining vocational skills.

**Physical therapist**: A specialist trained in the treatment of diseases and injury through exercise and physical activities.

**Neuromuscular disorder**: A disorder involving the relationship between nerves and muscles, and especially the weakening or dysfunction of the muscles.