

## **MUSCULOSKELETAL ANOMALIES IN CHARGE SYNDROME: FOR THE PHYSICIAN**

Marc S. Williams, M.D.,  
Department of Pediatrics, Gundersen Lutheran Medical Center, 1836 South Ave., LaCrosse, WI  
54601 [mwilliam@gundluth.org](mailto:mwilliam@gundluth.org) (608)782-7300 X2363

### **TYPE AND FREQUENCY OF MUSCULOSKELETAL ANOMALIES IN CHARGE SYNDROME**

- ◆ Prevalence of musculoskeletal anomalies is between 30 - 50%. This may increase as more cases of scoliosis are noted in older individuals.
- ◆ No consistent pattern of anomalies has been seen, although syndactyly of fingers or toes seems to be more frequent. Some of these patients have an atypical split hand deformity. There is often a specific palmar crease pattern with a so-called “hockey stick” distal palmar crease which is included in the minor diagnostic criteria.
- ◆ Severity of anomalies has ranged from very minor (dermatoglyphic anomalies to moderately severe (congenital hip dysplasia, syndactyly, polydactyly, clubfoot).
- ◆ A single patient is reported with absent muscles of one upper extremity. Muscle abnormalities have otherwise not been reported.
- ◆ Hypotonia, particularly of the upper body is frequent. This may be a neurologically-based problem or possibly a misinterpretation of the presence of ligamentous laxity. There have been no reported cases of a primary myopathy in CHARGE patients. Muscle biopsy would not be indicated, unless a second diagnosis is suspected.
- ◆ Scoliosis is frequent, beginning as young as 6-7 years.
- ◆ Osteoporosis may occur in adults with untreated hypogonadism.
- ◆ Information is derived from literature review of nearly 300 reported patients.

### **DIAGNOSTIC TESTS**

Careful physical examination of the musculoskeletal system is the only “test” indicated in all patients. It is important to screen for scoliosis beginning at school age.

Other diagnostic tests (X-ray, ultrasound) may be indicated based on physical findings.

X-rays obtained for other reasons (i.e. chest x-rays) should be examined carefully for definition of skeletal anatomy.

## **MEDICAL MANAGEMENT**

Treatment is anomaly specific. No differences in therapy are necessary if the patient is diagnosed with CHARGE syndrome. If surgery or sedation is necessary, anesthesia precautions are as discussed elsewhere.

## **REFERENCES**

Williams MS, Rooney BL. Limb anomalies in CHARGE association. Poster presentation 27th Annual March of Dimes Clinical Genetics Conference, 1996.

Tellier AL, Cormier-Daire V, Abadie V, et al. CHARGE Syndrome: Report of 47 cases and review. *Am J Med Genet* 1998;76:402-409.

Davenport SLH, Hefner MA, Mitchell JA. The spectrum of clinical features in CHARGE syndrome. *Clin Genet* 1986;39:298-310.

Harvey AS, Leaper PM, Bankier A. CHARGE association: clinical manifestations and developmental outcome. *Am J Med Genetics* 1991;39:48-55.

Oley CA, Baraitser M, Grant DB. A reappraisal of the CHARGE association. *J Medical Genetics* 1988;25:147-156.

## MUSCLES AND BONES IN CHARGE SYNDROME: PARENT INFORMATION

Marc S. Williams, M.D.,  
Department of Pediatrics, Gundersen Lutheran Medical Center, 1836 South Ave., LaCrosse, WI  
54601 [mwilliam@gundluth.org](mailto:mwilliam@gundluth.org) (608)782-7300 X2363

### NORMAL STRUCTURE AND FUNCTION

The musculoskeletal (MS) system consists of over 200 bones and 500 muscles. A detailed description is beyond the scope of this manual. (Besides, I can never remember them all.)

Two aspects of the MS system are critical: **structure** and **function**. Normal **structure** means that all of the components of the system (that is the muscles and bones) are present and in the proper relationships to one another. Normal **function** means that the bones are maintaining the normal structural integrity of the skeleton and the muscles (through contraction) are able to move the portions of the skeleton that are supposed to move (the joints). All **structural** abnormalities lead to **functional** abnormalities (although the severity of the abnormality may not be clinically significant), but not all **functional** abnormalities lead to **structural** abnormalities. An example of this would be a person who suffers a spinal cord injury. The muscles and bones are completely normal, but the muscles are unable to function because of the absence of nerve signals.

Two other important concepts are **strength** and **tone**. Most people understand strength, but tone is a harder concept to grasp. **Strength** is the ability of a muscle, or group of muscles to work against a load. It can be objectively measured. **Tone**, however, is a subjective assessment of muscle function at rest. When we are resting (that is not moving a particular muscle or group of muscles) our muscles are not completely at rest. There is a baseline level of activity that can be detected, but is not easily measured. We refer to this as **muscle tone**. Muscle tone helps with posture and maintaining normal skeletal relationships. Tone can be normal, low or high. Low tone is also called **hypotonia**. Individuals with low muscle tone are often described as floppy. This can lead to slumped posture, problems with head control, standing, etc. High tone is called **hypertonia** or **spasticity**. Individuals with spasticity feel like the muscles are constantly contracting. This can lead to abnormal joint positions and result in toe-walking, hip and knee flexion contractures, etc. An experienced physician or developmental therapist can assess Tone, but there is no way to objectively measure muscle tone. Abnormalities in muscle tone can be due to abnormalities of the **nervous system** (brain, spinal cord, spinal nerves, peripheral nerves or connections between the nerve and the muscle), or to abnormalities within the muscle itself (**myopathy**). It may be difficult to distinguish between these two causes on clinical examination.

### TYPES OF MUSCULOSKELETAL ABNORMALITIES IN CHARGE

Between 30 and 50% of patients with CHARGE are born with some type of skeletal abnormality. Severity can range from clinically nonsignificant (minor changes of the creases of the palms due to short hand bones), to quite severe (missing fingers). Several patients have been reported to have fusion of fingers or toes (syndactyly) or clefting of the hand or foot.

Low muscle tone (**hypotonia**) is very common in children with CHARGE, especially in the upper body (trunk). There have not been any patients with CHARGE known to have hypotonia due to a myopathy (abnormality of the muscles themselves). Unless new information becomes available, it is probably safe to assume that the hypotonia is due to a central nervous system abnormality (i.e. brain). Low muscle tone may have an effect on development: if the upper body is floppy, it will be more difficult to sit alone or stand. Combine weak tone with vision loss and balance problems, and you may have a child who does not walk until age 5 or 6.

Scoliosis (curvature of the spine) is common in children with CHARGE. Although scoliosis is generally thought of as a teenage problem, it has been seen in young children with CHARGE. By the teenage years, a majority of individuals with CHARGE may have some scoliosis. This may be due, at least in part, to the low muscle tone in the upper body.

## **DIAGNOSTIC TESTS**

The most important diagnostic test in very young children is a careful physical examination of the musculoskeletal system. Imaging studies (such as X-ray, ultrasound) are indicated if there is suspicion of an anomaly on physical examination. Skeletal survey (to look at all the bones of the skeleton) is not routinely indicated. Tests of muscle (muscle biopsy, electromyogram (EMG)) are generally not indicated, unless a primary muscle problem is also suspected.

In older children, regular physical exam for scoliosis is indicated. If scoliosis is suspected, the doctor may order X-rays to determine the extent of the scoliosis.

## **MANAGEMENT AND OUTCOME OF MS ANOMALIES**

Medical and/or surgical management is based on the type of anomaly. They are not managed differently whether or not the child has CHARGE. Outcome following intervention is generally good, but clearly depends on the severity of the problem and the treatment that is required. Occupational therapy (OT) and physical therapy (PT) may be helpful in dealing with low muscle tone.