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 (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


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  (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.