



Further Inform Neurogenetic Disorders

Issues with the Bones, Joints and Muscles in Lowe Syndrome

Poor muscle tone (known as hypotonia) means that babies are floppy after birth. Muscle tone in Lowe syndrome generally improves with age, but normal motor tone or strength are rarely achieved.

Decreased motor tone also results in delayed motor milestones. Walking independently occurs in approximately 25% of boys between the ages of 3 and 6 and in 75% between the ages of 6 to 13. Some never walk and require the use of a wheelchair for mobility.

This poor muscle tone causes issues associated with feeding difficulties in infancy as well as later in life. In infancy, poor head control and difficulties sucking and swallowing may present issues in feeding. Later in life, aspiration of food and the inability to cough it up effectively due to weaker muscles can cause lung-related issues such as pneumonia or lung disease. Poor abdominal muscle tone also increases the risk for chronic constipation and the development of hernias.

Decreased motor tone in the trunk of the body increases the risk of developing scoliosis (curvature of the spine), present in approximately 50% of individuals with Lowe syndrome. Individuals with Lowe syndrome are most at risk of developing scoliosis from their early teenage years through adulthood. Cases of scoliosis in Lowe syndrome can vary from mild (and treatable with a brace) to severe (where it causes back pain, reduces lung capacity and significantly affects mobility).

Some individuals with Lowe syndrome may suffer with an abnormally rounded back (kyphosis). This may be caused by repeated vertebral compression fractures due to low bone density or arthritis. Treatments for this include body braces and physical therapy.

Hypermobile joints may result in joint dislocation, especially of the hips and knees.

Many children with Lowe syndrome have a history of rickets or soft bones. This could be for a number of reasons. For good bone-mineralization, the body needs calcium, phosphate and active vitamin D, which is formed by the kidneys. Therefore, many Lowe patients need supplementation of phosphate and active vitamin D to help keep their bones healthy. Apart from this, hypotonia and its subsequent reduced muscle tension may be a cause of weak bones due to inactivity.

Due to the reduced bone density mentioned above, some individuals with Lowe syndrome will suffer from bone fractures. These fractures will usually involve bones in the upper leg around 6 years old when they are learning to walk.

Methods of improving these bone-related issues include early diagnosis and treatment of Vitamin D deficiency and low bone density, as well as regular exercise.
