

# Congenital SMA with Arthrogryposis

**This is one of the rarest forms of spinal muscular atrophy. It is present at birth, and children exhibit severe contractures of the joints, resulting in limb deformity; spinal curvature; deformities of the chest wall; difficulties breathing; abnormally small jaw; and upper eyelid droop (ptosis).**

## Diagnosis

Diagnosis is by a combination of clinical observation; blood tests that reveal an increased level of creatine kinase (which appears in the blood when muscle tissue is being broken down); distinctive abnormalities on muscle biopsy; characteristic electromyography and nerve conduction abnormalities; and genetic testing.

## Treatment

There are no cures for any of the forms of spinal muscular atrophy. The treatments involve addressing the symptoms and attempting to improve quality of life.

Medical treatment may be necessary for recurrent pneumonia and other respiratory infections. Surgery may be necessary for spinal curvature and severe contractures. Physical therapy, occupational therapy, and other types of rehabilitation programs may help individuals achieve the highest level of functioning possible.

## Prognosis

The prognosis for spinal muscular atrophy is variable. Life expectancy is dependent on the degree of respiratory impairment present. Because of the slow progression of symptoms, individuals with Types III or Kennedy syndrome may have normal life spans.

## Prevention

There is no way to prevent spinal muscular atrophy. However, genetic counselling is crucial so that parents can make informed decisions about having children. In general, when a family has already had a child with SMA, each subsequent pregnancy has a 25 percent chance of producing another child with SMA. Prenatal testing is available. Parents must then decide whether to use the information to help them prepare for the arrival of a baby with SMA or to terminate the pregnancy.

## Parental Concerns

Caring for a child with SMA can be very challenging and emotionally draining. Support groups, respite care, and help to support other siblings in the family can be important adjunct measures.

Extracted from Gale Encyclopaedia of Children's Health.



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