

Table 2. Other Forms of Severe Spinal Muscular Atrophy Not Linked to *SMN* Gene

SMA Variants	Inheritance/Linkage/Gene	Clinical Presentation	Reference
Scapulooperoneal spinal muscular atrophy	Autosomal dominant 12q24.1-q24.31	Congenital absence of muscles, progressive weakness of scapulooperoneal and laryngeal muscles	(13, 14)
Pontocerebellar hypoplasia with spinal muscular atrophy	Autosomal recessive	Onset 0-6 mo, cerebellar and brainstem hypoplasia, absent dentate nucleus, neuronal loss in basal ganglia, cortical atrophy	(15-19)
X-linked infantile spinal muscular atrophy with arthrogryposis	X-linked Xp11.3-q11.2	Onset at birth or infancy, contractures, death less than 2 y	(20, 21)
Spinal muscular atrophy with respiratory distress type 1	Autosomal recessive 11q13.2-q13.4 IGHMBP2	Onset within the first 3 mo of life, eventration of the right or both hemidiaphragms, finger contractures, pes equines foot deformities	(22, 23)

NOTE: SMA = spinal muscular atrophy.