Schwartz-Jampel syndrome (SJS) is a rare genetic disorder characterized by abnormalities of the skeletal muscles, including muscle weakness and stiffness (myotonic myopathy); abnormal bone development (bone dysplasia); permanent bending or extension of certain joints in a fixed position (joint contractures); and/or growth delays resulting in abnormally short stature (dwarfism). Affected individuals may also have small, fixed facial features and various abnormalities of the eyes, some of which may cause impaired vision. The range and severity of symptoms may vary from person to person. Two types of the disorder have been identified that may be differentiated by age of onset and other factors. SJS type 1, which is considered the classical form of the disorder, may become apparent during early to late infancy or childhood. SJS type 2, a more rare form of the disorder, is typically recognized at birth (congenital). Most researchers now believe that SJS type 2 is actually the same disorder as Stuve-Wiedemann syndrome and not a form of SJS. (For more information on Stuve-Wiedemann syndrome see the Related Disorders section of this report.) SJS (SJS) types 1 and 2 are rare disorders that appear to affect males and females in equal numbers. More than 85 cases have been reported in the medical literature, including individuals affected by the classical (type 1) and the more severe neonatal form (type 2) of the disorder. SJS type 2 appears to be most common in individuals of United Arab Emirates descent. Depending upon the form of the disorder present, associated symptoms and findings may be recognized at birth or may become apparent during infancy or within the second year of life.