Juberg-Marsidi syndrome is an extremely rare X-linked genetic disorder that is fully expressed in males only, and is apparent at birth (congenital) or during the first few weeks of life (neonatal period). Affected children exhibit severe mental retardation; delays in reaching developmental milestones (e.g., crawling, walking, etc.); muscle weakness; diminished muscle tone (hypotonia); and/or delayed bone growth as well as growth retardation, resulting in short stature. Juberg-Marsidi syndrome is an extremely rare inherited disorder that is fully expressed in males only. However, females who carry a single copy of the disease gene (heterozygotes) may exhibit some of the symptoms associated with the disorder. Approximately six affected families (kindreds) have been reported in the medical literature. Most symptoms of Juberg-Marsidi syndrome may be apparent at birth or soon thereafter. Juberg-Marsidi syndrome may be diagnosed at birth or during early infancy, based upon a thorough clinical evaluation and characteristic physical findings. Craniofacial abnormalities, low birth weight, genital malformations, and/or hearing impairment may be apparent at birth. Hearing impairment or deafness may be confirmed through a variety of specialized hearing (auditory) tests. Abnormalities in motor coordination, speech, and intellect may be monitored during infancy and childhood.