"Joubert syndrome is an autosomal recessive genetic disorder that affects the area of the brain that controls balance and coordination. This condition is characterized by a specific finding on an MRI called a ""molar tooth sign"" in which the cerebellar vermis of the brain is absent or underdeveloped and the brain stem is abnormal. The most common features of Joubert syndrome are lack of muscle control (ataxia), abnormal breathing patterns (hyperpnea), sleep apnea, abnormal eye and tongue movements and low muscle tone. The prevalence of Joubert syndrome has been estimated to be 1/258,000 but is probably an underestimate of the true prevalence, which may be closer to 1/100,000. The diagnosis of Joubert syndrome is based on physical symptoms and the ""molar tooth sign"" as seen on an MRI. Molecular genetic testing is available for the four genes that have been shown to cause Joubert syndrome in about 40% of cases. Carrier testing and prenatal diagnosis are available if one of these gene mutations has been identified in an affected family member."