Aicardi syndrome typically begins as involuntary muscle spasms between four months and four years of age. Other symptoms may include epilepsy, intellectual disability, profound muscle weakness (hypotonia), an abnormally small head (microcephaly), abnormally small eyes (microphthalmia), an incomplete development of the retina and nerve in the back of the eye (colobomas), and/or abnormalities of the ribs and/or spinal column. Children of all ages with Aicardi syndrome have significant delay in motor development. Aicardi syndrome can be life-threatening during childhood due to complications from upper respiratory infections. Aicardi syndrome usually affects only females. In very rare cases, males with Klinefelter syndrome (47,XXY) may have Aicardi syndrome. It has been estimated that there are between 300 and 500 cases of Aicardi syndrome worldwide. There do not appear to be any differences based on ethnicity or gender. It is usual to have an MRI of the brain. This study makes pictures of the brain to look for a small or missing corpus callosum and other problems with the formation of the brain. Individuals with Aicardi syndrome should have a test to look at the brain waves (EEG) to diagnose and treat seizures. An ophthalmologist should look into the eyes at the retina. In Aicardi syndrome, this almost always reveals small cream-colored cavities (lucunae) within the retina.