Mucolipidosis IV is a rare metabolic disorder characterized by intellectual disability; severe impairment in the acquisition of skills requiring the coordination of muscular and mental activities (psychomotor retardation); diminished muscle tone (hypotonia); clouding (opacity) of the clear portion of the eyes through which light passes (cornea); and progressive degeneration of the nerve-rich membrane lining the eyes (retinal degeneration). Mucolipidosis IV is inherited as an autosomal recessive genetic trait and caused by mutations in the MCOLN1 gene. Mucolipidosis IV is a rare inherited metabolic disorder that affects males and females in equal numbers. The disorder was first identified in 1974 and as of 2010, 70 cases have been reported in the medical literature. The precise incidence is unknown, but is estimated to be approximately 1:40,000. About 70% of those diagnosed are of Ashkenazi Jewish ancestry. Mucolipidosis IV may be suspected based upon a thorough clinical examination, a detailed patient history, and a variety of specialized tests. Individuals with mucolipidosis IV present with iron deficiency anemia, high serum gastrin levels and characteristic findings on brain MRI examinations. In most cases, an electron microscope is used to visualize characteristic lysosomal storage bodies in fibroblasts obtained from biopsied tissue of the skin and/or the delicate membrane that lines the eyes (conjunctiva). Molecular genetic testing for mutations in the MCOLN1 gene is available to confirm the diagnosis.