Sialidosis, also known as mucolipidosis type I, is a rare inherited metabolic disorder characterized by a deficiency of the enzyme neuraminidase (sometimes referred to as sialidase). Deficiency of neuraminidase results in the abnormal accumulation of toxic materials in the body. Sialidosis is divided into two types (i.e., type I and type II). Sialidosis type I usually becomes apparent during the second decade of life with the development of sudden involuntary muscle contractions (myoclonus), distinctive red spots (cherry-red macules) in the eyes, and sometimes additional neurological findings. Sialidosis type II is usually more severe than sialidosis type I. Type II often begins during infancy or later during childhood and is characterized by cherry-red macules, mildly coarse facial features, skeletal malformations and mild cognitive impairment. Sialidosis is inherited as an autosomal recessive trait. Sialidosis affects males and females in equal numbers. The exact incidence of sialidosis in the general population is unknown. One estimate places the incidence at 1 in 4.2 million individuals in the Australian population. Another estimate placed the incidence at 1-4 individuals per 200,000 of the general population. Because rare disorders like sialidosis often go unrecognized or misdiagnosed, determining the true frequency of sialidosis in the general population is difficult.