Aspartylglycosaminuria is a very rare genetic disorder that is concentrated among persons of Finnish decent, but is also found, even more rarely, in other populations around the world. It is an inborn error of metabolism, and one of the lysosomal storage diseases. It becomes apparent after the infant is a few months old. Major symptoms may include coarse facial features, spine and eye deformities, behavior problems and mental retardation. Aspartylglycosaminuria occurs as a result of deficient activity of a particular enzyme, leading to the accumulation of metabolic products in the body. Aspartylglycosaminuria is a lysosomal storage disease characterized by normal development during the first months of life after which abnormal development begins to occur. Diarrhea and infections that keep reoccurring are noticed. After the first few years facial features begin to get coarse which continues during the following years. The skeleton may become deformed and the ocular lens may develop crystalline deposits. Mental deterioration may begin to occur after age five and behavior problems are common. Lung, heart and blood problems tend to occur in later years. The patient may show mental retardation uneven development of the head and face with sagging cheeks, a wide nose and broad face. The spine may be twisted (scoliosis) and the neck may be unusually short. Adult stature is usually below normal. Aspartylglycosaminuria is a rare disorder that affects males and females in equal numbers. However, in Finland where the majority of cases are reported, there are an estimated 130 cases in 4.5 million persons. In the rest of the world, the condition is extremely rare and affects persons of various heritages. Treatment of Aspartylglycosaminuria is symptomatic and supportive. Genetic counseling may be of benefit for families.