Schindler disease is a rare inherited metabolic disorder characterized by the deficient activity of the lysosomal enzyme alpha-N-acetylgalactosaminidase (alpha-NAGA or alpha-galactosidase B). The enzyme defect leads to the abnormal accumulation of certain complex compounds (glycosphingolipids, glycoproteins, and oligosaccharides), which have terminal or preterminal N-acetylgalactosaminyl residues in many tissues of the body and in urine. Two major forms of Schindler disease exist - a severe form with onset in infancy (type I) and a milder form with onset in adulthood (type II). Some researchers have proposed a type III form of Schindler disease that is less severe than type I, but more severe than type II. The specific symptoms and severity of Schindler disease can vary from one person to another. Schindler disease is caused by mutations of the NAGA gene and is inherited as an autosomal recessive trait. Schindler disease affects males and females in equal numbers. The exact incidence of Schindler disease in the general population is unknown. Because cases of Schindler disease may go unrecognized or misdiagnosed, determining the disorder’s true frequency in the general population is difficult. As a group, lysosomal storage diseases are infrequent, although certain disorders may occur in specific ethnic or demographic groups at higher frequencies, about one in every 1,000-2,000 live births for Gaucher and Fabry diseases, or very infrequently (1 in 100,000 to 200,000 live births) for most of these disorders, which may be the case for alpha-N-acetylgalactosaminidase deficiency. Schindler disease was first reported in the medical literature in the late 1980s.