Pseudo-Hurler polydystrophy (mucolipidosis type III) is a rare genetic metabolic disorder characterized by a defective enzyme known as UPD-N-acetylglucosamine-1-phosphotransferase. This defective enzyme ultimately results in the accumulation of certain complex carbohydrates (mucopolysaccharides) and fatty substances (mucolipids) in various tissues of the body. The symptoms of this disorder are similar, but less severe than those of I-cell disease (mucolipidosis type II) and may include progressive joint stiffness, curvature of the spine (scoliosis), and/or skeletal deformities of the hands (e.g., claw-hands). Growth delays accompanied by deterioration of the hip joints typically develop in children with pseudo-Hurler polydystrophy. Additional symptoms may include clouding of the corneas of the eyes, mild to moderate coarseness of facial features, mild mental retardation, easy fatigability, and/or heart disease. Pseudo-Hurler polydystrophy is inherited as an autosomal recessive trait. A diagnosis of pseudo-Hurler polydystrophy may be suspected based upon a thorough clinical evaluation, a detailed patient history and identification of characteristic findings. A variety of specialized tests may confirm a diagnosis. These tests include detecting elevated lysosomal enzyme activity in serum or decreased enzyme levels in white blood cells or cultured connective tissue cells (fibroblasts).