Tyrosine hydroxylase deficiency is a rare genetic disorder characterized by a wide spectrum of disease ranging from a mild movement disorder at one end to a life-threatening, neurological disorder at the other. The symptoms of the disorder can vary widely from person to another, even among members of the same family. Common symptoms include an uncoordinated or clumsy manner of walking (abnormal gait) and dystonia. Dystonia is a general term for a group of muscle disorders generally characterized by involuntary muscle contractions that force the body into abnormal, sometimes painful, movements and positions (postures). Dystonia in tyrosine hydroxylase deficiency usually affects the legs. Additional symptoms that may occur include tremors, eye abnormalities, and a tendency of affected children to walk on their tiptoes. The severe form of tyrosine hydroxylase deficiency causes symptoms at a very young age (first months of life). The symptoms generally do not resemble those of a movement disorder, but rather give the impression of a severe, diffuse brain disorder. Mild and moderate forms of tyrosine hydroxylase deficiency show dramatic improvement when treated with levodopa. Levodopa is an amino acid that is converted to dopamine. Dopamine is a brain chemical that serves as a neurotransmitter and is deficient in children with tyrosine hydroxylase deficiency. Treatment options for severe tyrosine hydroxylase deficiency have been less effective. Tyrosine hydroxylase deficiency occurs due to disruptions or changes (mutations) of the TH gene. The TH gene mutation is inherited as an autosomal recessive trait. The exact incidence of tyrosine hydroxylase deficiency in the general population is unknown. Fewer than 50 cases have been reported in the medical literature. Researchers believe that the disorder is often misdiagnosed or goes undiagnosed, making it difficult to determine its true frequency in the general population. Tyrosine hydroxylase deficiency and guanosine triphosphate cyclohydrolase I deficiency (autosomal dominant Segawa syndrome) account for approximately 5-10 percent of all cases of primary dystonia in childhood. Tyrosine hydroxylase deficiency was first described in the medical literature in the 1970s.