

The severity of GAMT varies from patient to patient. Global developmental delays affect all individuals with this disorder and may be the first sign, appearing before other symptoms. The majority of individuals with GAMT deficiency have intellectual disabilities, seizure disorders, muscle weakness, behavior disorders, and movement disorders. People with GAMT may have weak muscle tone and delayed development of motor skills such as sitting or walking. Severely affected patients may lose previously acquired skills such as the ability to support their head or to sit unsupported. The prevalence of GAMT deficiency has been estimated to be from 1 out of 2,640,000 to 1 out of 550,000 patients being diagnosed to a conflicting report of 1 out of 115,000 patients being diagnosed. As of 2015, there have only been 110 individuals with GAMT deficiency diagnosed worldwide. GAMT deficiency patients are frequently misdiagnosed with cerebral palsy as infants and toddlers. Children are often misdiagnosed with autism or global developmental delays.