

MCT8-specific thyroid hormone cell transporter deficiency (THCT deficiency) is an inherited disorder that is characterized by severe intellectual disability, an impaired ability to speak, diminished muscle tone (hypotonia), and/or movement abnormalities. THCT deficiency is a rare inherited disorder that manifests in males only. One hundred and forty-four males from 56 separate families (kindreds) have been identified, two thirds of whom have been reported in the medical literature. The frequency of THCT deficiency in the general population with intellectual disability is not known.