TTD is present at birth. Males and females are affected in equal numbers. The estimated incidence is about 1 in 1,000,000 newborns in the United States and Europe. Over 100 patients have been reported worldwide. TTD has been reported in all ethnic groups. An initial evaluation for TTD involves a diagnostic work-up, including obtaining a detailed history of the patient's prenatal and neonatal history. A detailed physical exam is performed to assess clinical features such as hair abnormalities, short stature, small chin, ichthyosis, intellectual impairment or developmental delay, and bone and teeth anomalies. Evaluation by a developmental pediatrician or neurologist may determine whether there is any developmental delay or intellectual impairment. MRI imaging of the brain to identify abnormal patterns of myelination is often performed. Laboratory testing for immune function, blood count and iron levels can also be performed. TTD is typically diagnosed by polarized light microscopy of hair shafts, revealing a tiger-tail pattern, and at times by measurement of reduced sulfur content in patient's hair. The classical tiger-tail pattern alone usually is enough to diagnose TTD. However, there are other conditions with similar hair shaft abnormalities and often genetic testing is ordered to confirm the diagnosis. However, there are a few patients with features of TTD who will not have mutations in the known genes. They may have mutations in yet to be identified TTD associated genes.