Gosse Bouma

LILOG Prototype I Grammar Documentation: Semantics

Bericht des European Child & Adolescent Psychiatry

Kurzfassung

Niemann–Pick disease, a neurovisceral lysosomal lipid storage disorder, is a rare disorder that is unknown to many clinicians. The disease, that often has its onset during childhood or adolescence, shows a polymorphic clinical picture, including psychiatric symptoms. Because of its infrequence, Niemann–Pick disease is diagnosed with an average delay of 6 years. This report presents a case of an adolescent male whose symptoms had led to various hospitalisations and psychiatric diagnoses. When he presented with psychotic symptoms in our department, thorough diagnosis revealed Niemann–Pick disease type C1 as the underlying disease.