

Atransferrinemia is an extremely rare disorder. Approximately 10 cases in 8 families have been reported in the medical literature. Because atransferrinemia may go unrecognized or misdiagnosed, determining its true frequency in the general population is difficult.

Atransferrinemia affects males and females in equal numbers. Atransferrinemia was first described in the medical literature in 1961. A diagnosis of atransferrinemia is made based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests. Laboratory tests can reveal low or undetectable levels of transferrin in the blood.