Systemic involvement (hepato) Splenomegaly Neonatal Absent in ~15% of cases Cholestasis Age of onset is variable always before neurological signs Foetal May regress with age Liver ascites/ (Hepato) hydrops **Splenomegaly** Neonatal fatal Age, years 2 3 6 10 20 30 50 60 40 Birth Infantile Early) Late Infantile Juvenile Delay in Adult motor Gait problems milestones School problems

Psychiatric problems

Ataxia, Dystonia

(Dementia)

Vertical supranuclear gaze palsy-

Ataxia

(Seizures)

(Cataplexy)

Neurological involvement

Clumsiness

Cataplexy

Speech delay

Hypotonia