Visual failure, macular dystrophy, abnormal ERG; No other abnormal neurology.

Vacuolated lymphocytes

MRI studies to exclude Finnish variant LINCL and biochemistry for other metabolic retinopathies.

CLN3 common mutation

homozygote

heterozygote

CLN3 no del present

Further CLN3 mutation testing

Review blood film, EM of skin or rectal biopsy.

FPP/CVB/RL

EM of skin or rectal biopsy.

GRODs

PPT assay. (Mutation testing CLN1)

CLN3 common mutation

Further CLN3 mutation testing

Review blood film, EM of skin or rectal biopsy.

Classic juvenile NCL

Atypical juvenile NCL

Other NCL

Not NCL

JNCL/GROD

Classic JNCL

Atypical JNCL

Not NCL

Other storage disorders

Other atypical NCL

Other typical NCL: CLN5, 6, 7, 8 etc.

Not NCL

Algorithm A