Yes

No

Vacuolated lymphocytes

MRI for cerebellar atrophy or other changes. Photosensitive EEG at 1-2Hz and/or giant VEP at about 8 years.

Yes

No

CLN5 mutation.

EM of skin or rectal biopsy, EM lymphocytes.

One or more of:
1. EM of skin, rectal biopsy or lymphocytes shows GRODS,
2. PPT deficiency,
3. CLN1 mutation.

Yes

No

EM of skin or rectal biopsy, EM lymphocytes, if not already done.

See Algorithm A

Finnish origin

Onset < 18 months. Other INCL features: microcephaly, slowing down of development.

Yes

No

Previously normal development to 8 months. Age 1-7 years, two or more of: seizures, ataxia, regression, myoclonus, visual failure.

One or more of:
1. EM of skin, rectal biopsy or lymphocytes shows GRODS,
2. PPT deficiency,
3. CLN1 mutation.

Yes

No

CLN5 mutation.

Finnish variant LINCL

Other NCL- CLN2, CLN6, CLN7, CLN8

Not NCL

INCL

Other NCL

Not NCL

Other storage disorders

Other atypical NCL

Not NCL

Vacuolated lymphocytes

Photosensitive EEG at 1-2Hz, and giant VEP at age <8 years.

Yes

No

Repeat electrophysiology. EM of skin, rectal biopsy or lymphocytes. MRI brain.

See Algorithm A

Algorithm B