NCL diagnostic protocol

NCL Clinical Symptoms

Light microscopy

Vacuoles in lymphocytes
Mutation analysis CLN3

No vacuoles in lymphocytes

Enzyme analysis

PPT1, TPP1 or CTSD enzyme deficiency
Mutation analysis CLN1, CLN2 or CTSD

Not CLN1, CLN2, CTSD

Electron microscopy

Typical NCL deposits
Mutation analysis other NCL genes CLN3, CLN5, CLN6, CLN7, CLN8

Typical NCL deposits, not known NCL gene

Store research samples DNA, fibroblast cell line

Mutation analysis novel NCL genes

www.ucl.ac.uk/ncl
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