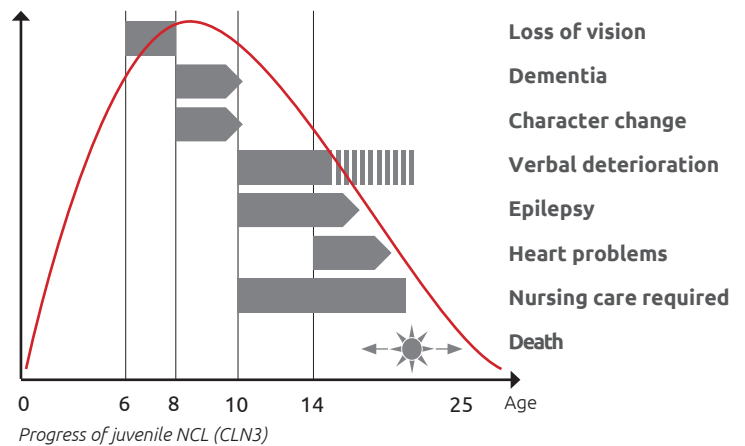


# Epilepsy, developmental regression, vision loss – it could be NCL!

## What is NCL?

Neuronal Ceroid Lipofuscinosis is a genetic lysosomal storage disorder. Genetic defects in one of the 13 CLN genes which are currently known result in lipopigments becoming locked in the lysosomes in almost all body tissues. The nerve cells in the brain's grey matter are, however, subject to the most damage. The disease is generally caused by autosomal recessive inheritance. NCL disorders are the most common group of neurodegenerative diseases among children and adolescents, with sufferers experiencing deterioration of cognitive and motor abilities, epilepsy, diffuse retinal degeneration and reduced life expectancy.



## When could NCL be the cause of illness?

If the following are present in **combination**:



**Loss of visual acuity (unclear retinopathy)**



**Delayed speech development**



**Developmental regression (incl. dementia)**



**Epilepsy**

An NCL disorder should be considered as a cause if at least two of the symptoms are present in combination. Initially sufferers' development is usually completely in line with their age, until the disease breaks out, causing their first seizures or their psycho-motoric development to stagnate.

In the case of the CLN3 form the first symptom is often a progressively worsening visual impairment at school age, caused by retinopathy.

## How can NCL be diagnosed?

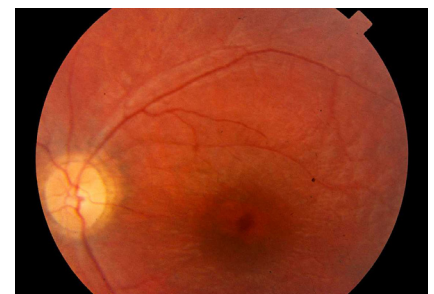
Depending on the age at which NCL manifests and the sufferer's clinical picture, the cause can be narrowed down to a specific form of the disorder, using molecular-genetic criteria to obtain the diagnosis.



In the case of unclear loss of visual acuity and suspected retinopathy, a thorough examination by an ophthalmologist will be required (such as an electroretinogram or optical coherence tomography) to identify changes to the eyes which are typical for NCL.

### CAVE incorrect diagnoses:

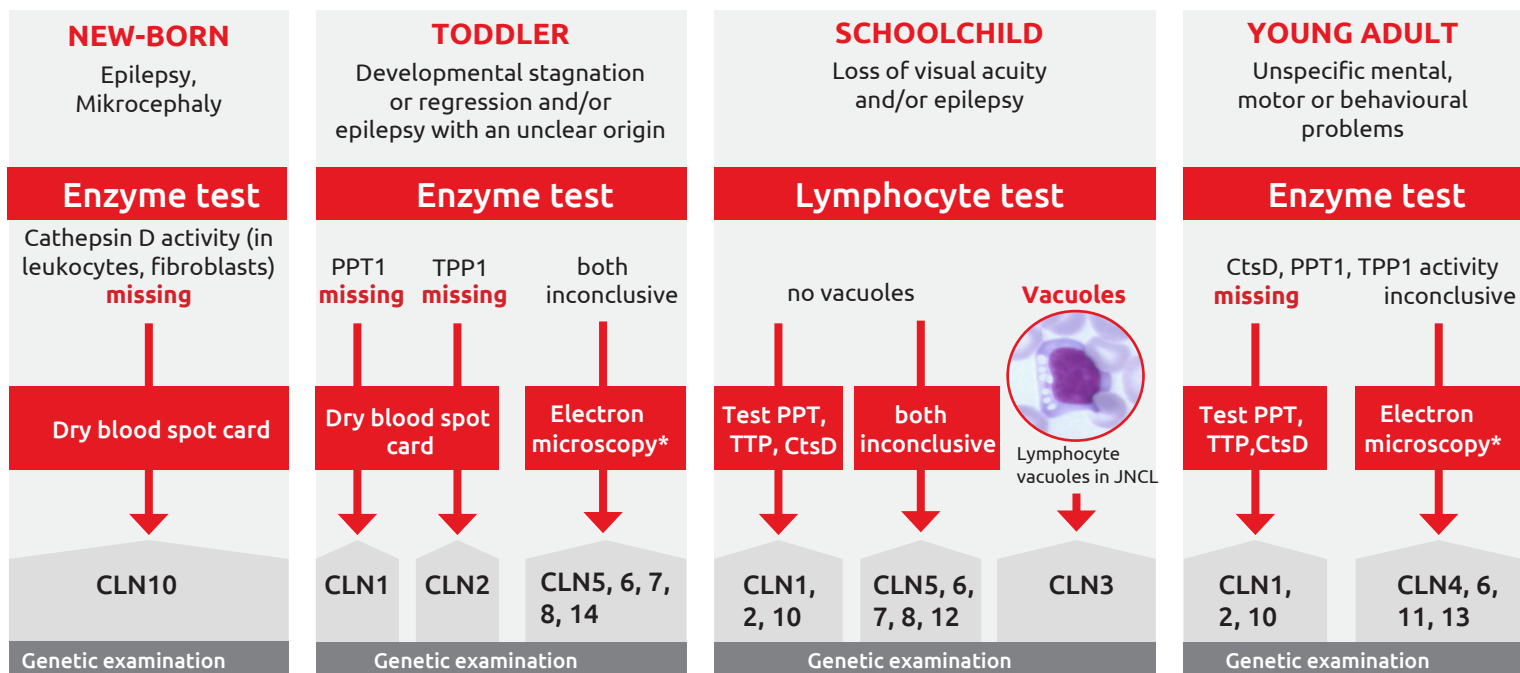
- Epilepsy with an unclear origin in combination with delayed speech development
- Retinopathia pigmentosa



*Juvenile NCL fundus with so-called bull's eye maculopathy.*

# Suspected NCL!

## How can NCL be diagnosed?



\*Alternatively panel diagnosis of all genes

## What are the treatment options for NCL?

### Palliative therapies

- Control of symptoms for epilepsy, disturbed sleep, movement disorder, behavioural problems, swallowing difficulties (PEG gastronomy), breathing/secretion problems (aspirators)
- Movement therapies – physiotherapy, speech therapy, ergotherapy, equine-assisted therapy
- Adequate provision of aids

### Causal therapies

- Development of causal therapeutic approaches is difficult as the pathomechanism of some sub-forms of NCL is not yet known
- Since July 2017, an approved intravitreal enzyme replacement therapy has been available to CLN2 patients
- Experimental therapy: gene therapy for some sub-forms of NCL (in preparation)

### Recommendation for palliative medication:

- Epilepsy: **Valproate** and **Lamotrigine**
- Spasticity: **Baclofen** and **Tizanidine**
- Myoclonus: **Pregabalin**, **Piracetam** and **Zonisamide**

### Not recommended:

- **Phenytoin**, **Vigabatrin** and **Carbamazepine**

## Advice on NCL-related issues

**University Medical Centre  
Hamburg-Eppendorf**  
Clinic of Paediatrics  
Dr. med. Angela Schulz  
+49 (0) 40 74 102 0440  
an.schulz@uke.de

**University Children's Hospital Zurich  
– Eleonorenstiftung**  
Department of Paediatric Neurology  
Prof. Dr. Dr. med. Robert Steinfeld  
+41 (0) 44 266 7330  
robert.steinfeld@kispi.uzh.ch

**NCL-Gruppe Deutschland e.V.**  
Juliane Sasse  
+49 (0)30 25044916 (AB)  
Juliane.Sasse@ncl-info.de

**NCL-Stiftung**  
Dr. Frank Stehr  
+49 (0) 40 69 666 7419  
frank.stehr@ncl-stiftung.de

Certified NCL online further training (2  
CME) at [Medlearning.de](http://Medlearning.de)

With the support of

**Rotary**  
Club Ahrensburg-Schloß

Authors:  
Dr. Angela Schulz and Dr. Eva Wibbeler

NCL-Stiftung  
Holstenwall 10  
20355 Hamburg

Tel.: +49 (0)40 6966674-0  
E-Mail: [contact@ncl-stiftung.de](mailto:contact@ncl-stiftung.de)  
[www.ncl-stiftung.de/english/](http://www.ncl-stiftung.de/english/)