NCL-Stiftung
Für eine Zukunft ohne Kinderdemenz

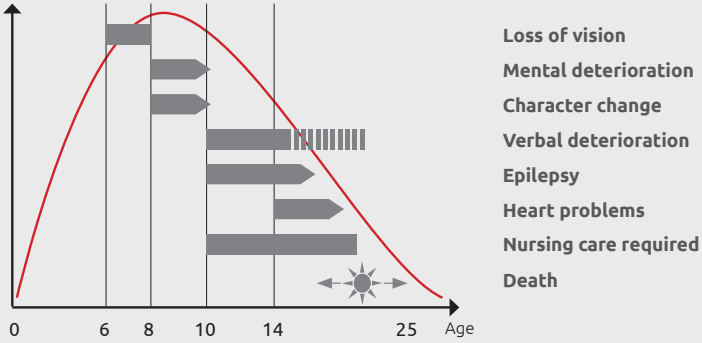
Children and adolescents with progressive retinopathy could have NCL!

Photo by Janko Ferlič on Unsplash

Factsheet for ophthalmologists and orthoptists

What is NCL?

The Childhood Dementia NCL (Neuronal Ceroid Lipofuscinosis) comprises a group of genetic storage disorders which usually present in the first decade of life. Children go blind, develop epilepsy and, as neural cells die, experience progressive loss of their mental and motor abilities. To date NCL is incurable and results in premature death, usually before the 30th birthday.
13 different genetic forms of NCL are known, presenting at different ages, and referred to as CLN1 – CLN14. Almost all NCL disorders are inherited autosomal-recessively.



Progress of juvenile NCL (CLN3)

The prototype for the NCL disorder is juvenile NCL (Spielmeyer-Vogt Disease, Batten Disease, genetic nomenclature "CLN3"), which occurs in school-age children. Typically, a previously healthy child will experience visual impairment around the time they start school. The rapidly progressing loss of vision is caused by retinopathy and can remain the only symptom for many years.

Ophthalmologists thus play a key role in diagnosing this disease.

Key symptoms of the juvenile NCL disease

1. Loss of vision in both eyes

2. Rapid progression

3. Failure to see centrally located objects (central scotoma)

Diagnosis and clinical course

Fundus

Early findings:

» Juvenile reflex absent
» Subtle RPE alterations in the macula

Later findings:

» Pale optic disc
» Macula with clear RPE alterations and atrophy
» Rarefied vessels
» Peripheral bone spicules

Optical coherence tomography (OCT)

» Loss of the outer retinal layers with perifoveal commencement and centrifugal progression

Electroretinogram (ERG)

Early findings:

» Reduced or extinguished scotopic and photopic b-wave amplitudes

Later findings:

» Extinguished ERG

CAVE:

» No anterior segment pathology
» No nystagmus or ocular motor disturbance

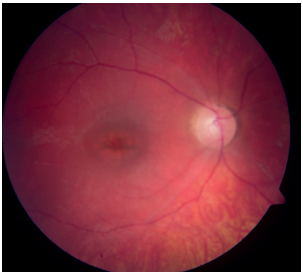


Fig. 1: Fundus of an 8-year old patient with juvenile NCL and macular RPE alteration (so-called bull's eye maculopathy).
Source: Dr. Simon Dulz, University Medical Centre Hamburg-Eppendorf

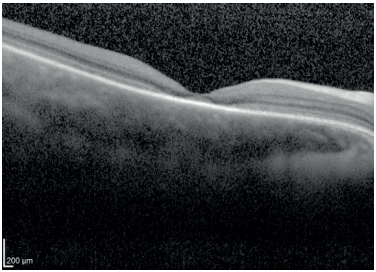


Fig. 2: Macular atrophy caused by juvenile NCL with loss of the outer retinal layers.
Source: Dr. Simon Dulz, University Medical Centre Hamburg-Eppendorf

Differential diagnosis for juvenile NCL

Stargardt Disease
» Gradual deterioration of visual acuity
» Very little change in the ERG

Cone Dystrophy
» Normal scotopic ERG response to stimuli
» Slower progression of the disease

Retinopathia pigmentosa
» Disease commences with decreased night vision and reduced scotopic ERG
» Initially without macular involvement

Advice on NCL-related issues

The following centres offer advice in German-speaking countries:

University Medical Centre Hamburg-Eppendorf,
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Macular panels for diagnosis (e.g.):
MGZ – Medizinisch Genetisches Zentrum, Munich; Ophthalmic request form (German) available to download.
Zentrum für Humangenetik Regensburg:
https://www.humangenetik-regensburg.de/netzhauterkrankungen.html

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Funded by the Spethmann Stiftung and the Stiftung ASD

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