1. Tasks and objectives of the NCL Group Germany e.V.

- Providing support for the optimal care and fostering as well as for the medical care of the affected children.
- Facilitate contacts between affected parents, helping them, and giving advice to the family members.
- Providing a time-out for children and parents through "guest accommodation" for the affected children.
- Organize meetings and conferences for parents, children, doctors, and assistants.
- Promoting scientific research in the field of NCL diseases.
- Raise and further the awareness of the disease with authorities.

2. The work of the association

The association has currently about 400 members. It is led by an 8-member honorary executive committee. Four regional groups (north, west, east, south) were formed for better care of the families and ease of staying in contact.
Close contact is held also with NCL self-help groups of in other countries.

3. Promotion of the Association

The association is a not-for-profit organisation. It is financed through donations, membership fees, subsidies from health insurance funds according to § 20c SGB V and through fines collection fund of the judicial authority of the City of Hamburg.

4. The disease pattern of the NCL

The NCL diseases - also known as CLN (after the affected genes) or English Batten Disease - occur in different forms and at different ages. Altogether about 320 cases of illness are known in Germany. However, the estimated number of undiagnosed cases is probably much higher because of the low degree of awareness.
Only since a few years there has been an improvement in the international efforts to promote scientific research aiming to uncover the causes of the disease and to develop a therapy. An international NCL congress is held every two years.
In Prof. Dr. A. Kohlschütter, the association has a recognized scientific adviser as its patron, who has numerous international contacts with other NCL researchers and physicians.

5. The causes

While the mechanism of the disease is always similar in different variants, the cause is to be found in different defective genes. Over the past few years, researchers were able to localise all of these genes. Heredity, with the exception of one adult form, is always recessive.
The defective genome causes accumulation of a grease-like material in all body cells (therefore NCLs belong to accumulation diseases) because a protein that is necessary for the normal metabolism is missing. This process is particularly critical in the sensitive nerve cells, which gradually die off. So far, there is no known way to prevent the brain degeneration caused by this.
Researchers internationally are working together to make known approaches to treating genetic defects (including enzyme replacement, gene and stem cell therapy) also usable for NCL.
The clinical (and prenatal) diagnosis of these diseases is based on electron microscopy of tissue, measurement of protein concentrations and analysis of the known CLN genes in blood samples.
An early diagnosis is important to interpret the changed behaviour of the child correctly and to cater to its needs.
Healthy siblings can seek advice at the Humangenetisches Institut of the University of Hamburg, since they, like their parents, could also be a carrier of the disease.

6. The course of the disease and its impact on the family

All the different forms of NCL have the following traits in common: progressive blindness, mental breakdown, epilepsy, and movement disorders. The children gradually lose their ability to speak. Due to their simultaneously declining motoric skills, they also have difficulties to communicate in other ways. Memory and orientation loss, as well as complete absence of day-night rhythm are further stages of the disease. The children are gradually declining physically, start needing a wheelchair and become incontinent very early on. Since the swallowing reflex is also affected at a later stage, the children must then be fed via stomach tube.
Due to their missing ability to communicate, it is unclear how much the NCL children understand their situation. The hearing is not affected by the degradation, so the children can still perceive and react to their environment.

They understand more than is commonly assumed and are very sensitive to everything that happens around them.

The children generally live and are cared for at home. The parents thus experience their unstoppable degradation, right up to the point when they accompany them in their last hours.
Since the first symptoms are very unspecific and the disease is still unknown to many physicians, it can take years before the correct diagnosis is made. A period in which, in addition to the child's growing difficulties, the family is often confronted with allegations from society. Family problems are incorrectly assumed to be the cause of the developmental delay. During this period, more potentially affected siblings are born because the parents are not aware of this genetic defect.
Prior to the possibility of genetic diagnosis, the NCL variants were distinguished only by the age of the onset of the disease.
In Germany the juvenile NCL (usually CLN3) and the late-infantile form (usually CLN2) occur most frequently.

Late-infantile NCL (orig. Jansky-Bielschowsky disease)

The disease begins at the age of two to four years. The first signs are medically hardly controllable epilepsy and an initial delay in development: words learned are forgotten again, instead of running, the child will relapse to crawling. Since at this age, development of the brain has not yet completed, an "up and down" can be observed, when already lost skills are regained again for a short period of time. The onset of blindness often remains unobserved, because the children cannot yet express themselves. Most children die between 10 and 13 years of age.

Juvenile NCL (originally Spielmeyer-Vogt's disease)

During the first years the children develop normally and are seemingly healthy. In the first year of school, they are usually diagnosed with a beginning vision disorder, which leads to complete blindness within two to four years.
As they are due to the loss of their verbal skills unable to express their desires or feelings properly, they often become aggressive or depressed.
As the disease progresses, epileptic seizures with all known symptoms appear first sporadically, later multiple times a day. In most cases the affected person will die between the ages of 25 to 30 years.

7. Ways to help

Due to the absence of treatments of the root-causes, the primary and most important task of all specialists - medical, educational and others - is to deal empathetically with the children and their problems. These specialists also need the courage to use unconventional methods if they want to be a real help to the families.

These days, there is no known therapy or drug against the disease itself. The only way to help is to alleviate the symptoms, treat the epileptic attacks with anticonvulsants and try - with great patience and understanding - to accompany the children and their families.

This is important because parents of NCL-affected children, a rare disease not well-known in medical circles, get little understanding from authorities or other organizations: Any measure or help that is self-evident for other fatal diseases has typically to be fought for.

8. Regular events

The following events are offered annually to the members:

- Three-day convention for all members and children.
- Regular meetings in the regional groups.
- Weekend for healthy siblings (if there is demand).
- Mothers weekend.
- Fathers weekend.
- Two guest accommodations for affected children (ten days each).

9. Exchange of experience and information

Both at the annual convention and during the regional group meetings, parents have the opportunity to exchange their experiences personally. This is important and gives strength and courage to continue the daily chores.

Mutual assistance is also important applying new legislation and for dealing with the authorities.

The quarterly "Members-Info" and our homepage www.ncl-deutschland.de serve as a permanent connector and information source

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We welcome any support! Whether as a donation or permanently through membership:

For donations of 50,- Euro or above, a confirmation of donation can be given. The membership fee (40 Euros per person per year, 60 Euros for families) is also tax deductible.

Donation account:
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Neuronale-Ceroid-Lipofuszinose

NCL

Gruppe Deutschland e. V.

Zum Wohle an NCL erkrankter Kinder

The Neuronal Ceroid Lipofuscinoses (NCL) are a group of currently incurable, recessive, neurodegenerative diseases in childhood.

The NCL Group Deutschland e.V.
represents the interests of the affected
children and their families.

www.ncl-deutschland.de

We are member of the following umbrella organizations:
Der PARITÄTISCHE, Achse, BAG Selbsthilfe,
Kindernetzwerk und Eurordis