

## INFORMATION

NCL is the most common neurodegenerative disease in childhood. This fatal metabolic disease causes nerve cells of the brain to gradually die (s. fig.). The NCL-Foundation concentrates on juvenile NCL (jNCL). Here rapid loss of eyesight - the first symptom - already shows at the age of school enrolment. The disease is based on a genetic defect, which is recessively inherited. The affected gene CLN3 lies on the 16th chromosome.

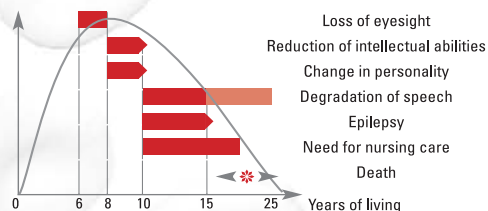


Healthy brain



NCL-affected brain

Intensified research is urgently needed because of the harshness and inevitability of the course of this disease. NCL-patients die between the age of 20 and 30 after a long way of suffering. Death occurs through respiratory paralysis



Schematic diagram of the average course of JNCL.

or serious infections. Since NCL is so rare false diagnoses are still common. That is why the disease is oftentimes diagnosed very late. In order to inform families concerned and educate relevant groups of physicians, the foundation again arranged for different projects in the year 2006:

### Physicians

- **Ophthalmologists:** Initiation of a publication in the professional journal „Klinische Monatsblätter für Augenheilkunde“ and its distribution to all eye clinics in Germany. Foundation lectures were held in the eye clinics in Bonn and Zurich.
- **Pediatricians:** Development of an NCL-leaflet and its distribution via the field services of HEXAL and ratiopharm. Furthermore, the leaflet was put to use as a congress enclosure at various events for physicians. An advanced NCL training was initiated at the Magdeburger Klinik.
- **Neurologists:** Distribution of the leaflet at the annual migrating convention with 400 attendants.
- **Human geneticists:** Initiation of an NCL-satellite symposium at the annual meeting of neuro-geneticists in Rostock.

### High-school students, College students, Postgraduates

- **Biology up-to-date:** Continuation of the school project at high-schools in Hamburg to early sensitise students to a rare disease. Aim: Integration of a rare hereditary disease into the biology classes of the senior years of secondary school.
- **Lectures:** Information lectures were held at the biochemist convention (Hamburg), as well as at the European convention of scholarship holders of the Boehringer Ingelheim Fonds.

### Others

- **Co-operation:** Active collaboration as a member of the ACHSE (Alliance of Chronic Rare Diseases), a network of patient organizations of those infected with rare diseases.
- **Publications:** Regular articles in the member journal of the NCL-self-help group, as well as an article in the member journal of Pro retina.



## RESEARCH STATUS

### Basic research

- **Immune system:** Besides the already proven autoantibody against GAD65, recently **further autoantibodies** were detected in patients. Whether they contribute to the pathogenesis needs to be verified. That is why a **(neuro)immunologist** has to be won for the NCL-research.
- **Cell-Processes:** Aside from the endocytosis and certain transport processes, the **autophagocytosis** also seems to be defective in the juvenile NCL. This phenomenon also appears in other neurodegenerative diseases.
- **Cell-Areals:** It has been newly discovered that next to the recycling center (lysosome) and the power station (mitochondrion) of the cell, also the **cell framework** (cytoskeleton) and the **nerve distributing center** (synaptosome) are affected. Electrophysiological methods need to be applied.
- **New NCL-genes/ forms of disease:** Ms. Lehesjoki (Finland) has localized the **CLN7-gene**. Mutations in this gene lead to the Turkish variant of the late-infantile form of NCL. Mr. Steinfeld (Göttingen) has shown that mutations in the **CathepsinD-gene** (CLN10) lead to a disease in humans similar to NCL.

### Clinical Research

- **Clinical scale:** In 2006 a **clinical scale** to evaluate the psychological and psychiatric problems was established.
- **Gene therapy:** This year the first **gene therapeutic study** in late-infantile NCL-children was completed (Laboratory R. Crystal, USA). A new vector in the knock-out mouse model showed promising results. The initiation for the juvenile form is a "must".
- **Immunosuppression:** In Finland the **immunosuppression therapy** has been continued with juvenile NCL patients. It still needs to be statistically evaluated.

### Care Research

- In May 2006 the first international conference concentrating on the educational area of juvenile NCL took place in Sweden. Besides doctors and researchers, also educators and therapists were in attendance. Innovative programmes concerning handling of the disease, as the usage of **voice computers**, were presented.



## RESEARCH SUPPORT

The Foundation follows a co-operative research support based on networking concerning researchers and foundations / sponsors. The scientific **NCL-network** is being expanded by personal contacts and on-site visits, as well as the national NCL-Congress. Furthermore, speeches at international meetings (e.g. Sweden), poster presentations (Alzheimer-Congress, Berlin) and the initiation of special issues of professional journals concentrating on NCL help the cause.

The focus of the **5th NCL-Congress** lay on the discussion of different strategies for the development of a gene therapy of juvenile NCL.

### Provisional results of two research projects that are being supported:

- Prof. Dr. K. Rütger (Analysis of the retina degeneration in CLN3-mice): After 14 months a measurable difference was found between wild type and knock-in-mice.
- Dipl. S. Pohl (Finding the unknown function of CLN3): The protein concerned most likely takes over transport functions.

## FUNDRAISING

In the fundraising area the following events were organized:

### 1/06: Dental gold event

In co-operation with Heraeus Kulzer, dentist Dr. Jens Hauptmann from Frankfurt initiated a dental gold drive. The professional journal „zm“ reported on this event. We are looking for more help and innovative ideas.

### 5/06: Foundation Portrait

A portrait of the NCL-Foundation was released by the Schümann publishing house in the Hamburg handbook issue no. 12.

### 6/06: Benefit Golf Tournament Adendorf

The Golf-Club Adendorf organized a charity-tournament in favour of the NCL-Foundation. Prominent participants were Jan-Josef Liefers, Caroline Beil and Martin Wilhelmi.

### 8/06: American Barbecue



The staff of the Marriott Hotel Hamburg voluntarily organized a summer party in favour of the NCL-Foundation.

### 9/06: 3. Charity-Dinner „Cooking for Kids“

The dinner took place in the E.ON guest house in Hamburg. Our special thanks go to Tim Mälzer (NCL-ambassador), patrons Dr. Dreyer / Dr. Blümel (Haspa), Ms. Seeger (E.ON), Ms. Huth (festpunkt) and Mr. Winkler (Winklers Catering).



### 11/06: Wiesbaden Gala Ball of real estate

Under the patronage of Mayor Roth and initiated by Anja Strumpf Events 1.000 guests supported the foundation.

### New issue of the foundation brochure

For the new issue of the foundation brochure Mrs. Eva-Luise Köhler, the wife of the German Federal President, wrote the greeting.

*We would like to thank our sponsors Aldiana, Avis, Bild-Metro-press, Die Bahn, E-plus, HanseNet, Robinson, Round Table, Rotary, Springer Biobackwerk and Wempe, as well as all others who donated, sponsored and volunteered.*

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## NCL-ACCOUNT FOR DONATIONS

Account number 10 59 22 30 30  
Bank code 200 505 50  
Bankverbindung Hamburger Sparkasse

For all donations exceeding 100 € you will receive a receipt for tax purposes. We therefore ask you to state your name and address in the purpose field.

In order to help the daily handling of the disease, those affected have organized a self-help group.

### Die NCL-Gruppe Deutschland e.V.

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# 5<sup>th</sup> Annual Report 2006

of the charitable NCL-Foundation

The NCL-Foundation was founded in 2002 and aims at developing a therapy of the deadly NCL disease (Neuronal Ceroid Lipofuscinosis / Batten disease) by 2020 the latest.

The Foundation concentrates on four areas:



1. Much **education** is necessary – meaning those concerned and doctors. That is the only way an early diagnosis of NCL is possible.



2. Knowing the disease and keeping up with the latest developments are basic requirements for particular **research**.



3. By push financing and third-party funds consulting, **patient oriented research** is being supported. Raising the quality of life for those concerned is the minimum intention.



4. Research costs money. With specific **PR** the Foundation tries to sensitize the public for rare diseases and earn donations.

