

# NCL-Foundation

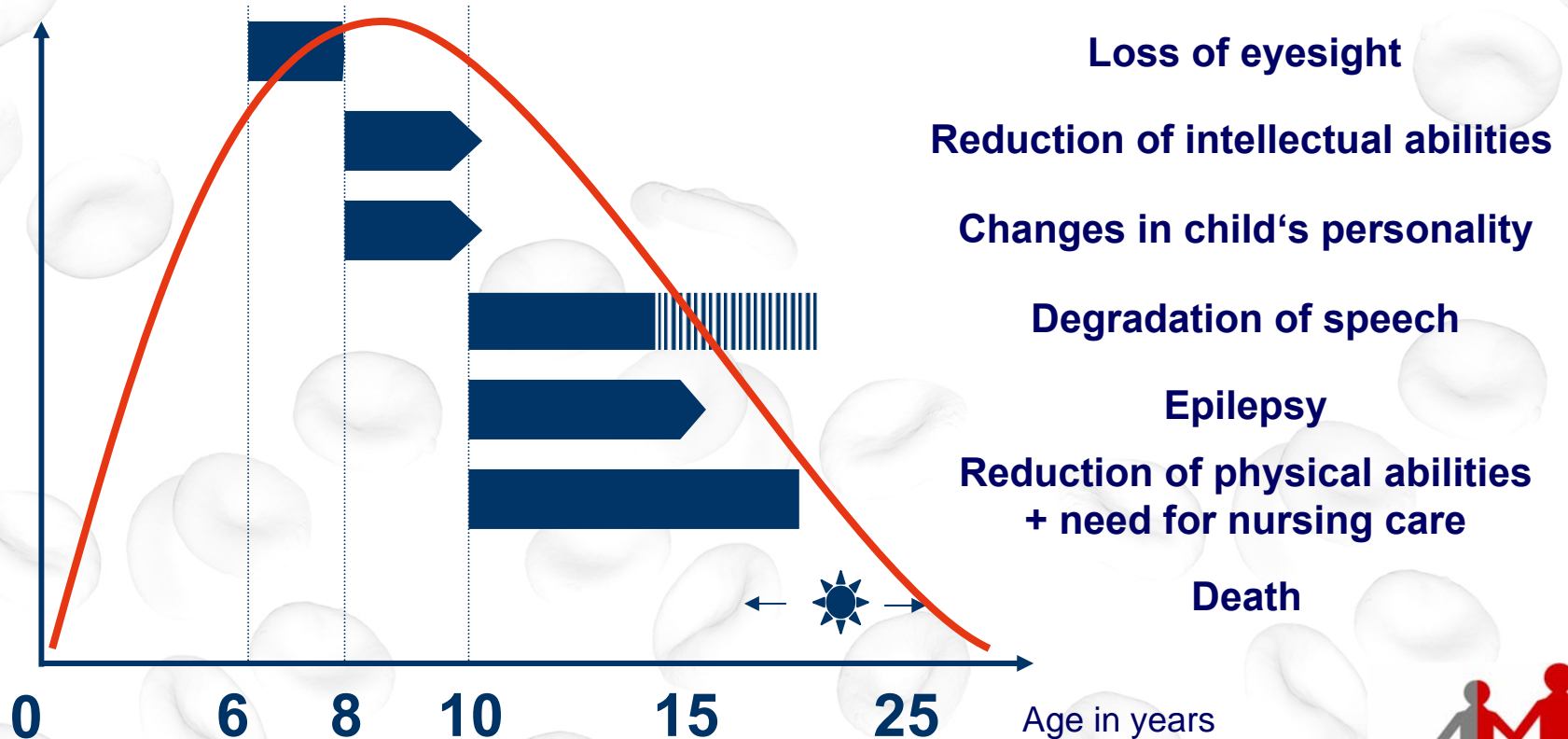
- **Address:** Holstenwall 10, 20355 Hamburg
- **Email:** frank.stehr@ncl-stiftung.de
- **Internet:** [www.ncl-stiftung.de](http://www.ncl-stiftung.de)



**National Contest**  
for **Life\_NCL**

**Imagine your child is blind.  
And this is just the beginning.**

**Schematic diagram of the average course of jNCL**



# There are different NCL-forms. All are deadly.

## Mutated genes in Neuronal Ceroid Lipofuscinosis (NCL)

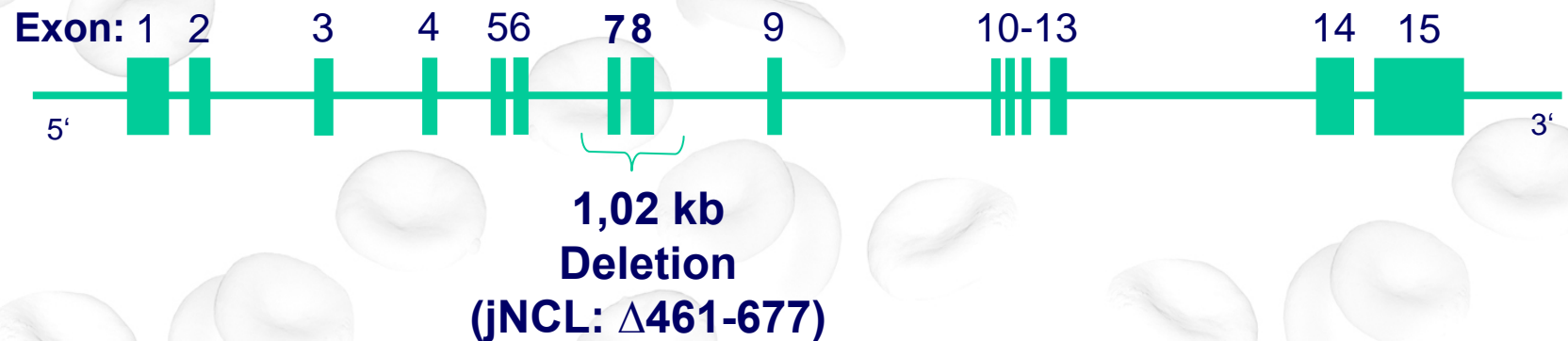
Mutated gene	Protein and function
CLN1	Palmitoyl-Protein-Thioesterase 1 (PPT1); <i>infantile NCL</i>
CLN2	Tripeptidyl Peptidase 1; <i>late-infantile NCL</i>
<b>CLN3</b>	<b>Battenin; lysosomale transmembrane protein; function unknown; <i>juvenile NCL</i></b>
CLN5	Protein with two transmembrane regions; function unknown; <i>late-infantile NCL</i>
CLN6	Transmembrane protein; function unknown; <i>late-infantile NCL</i>
CLN8	Transmembrane protein (endoplasmatic retikulum); function unknown; <i>variante NCL</i>



Enzymes or membrane proteins are missing.

# The mutated gene of the juvenile form (CLN3) is known.

## Structure of CLN3 and position of major deletion (jNCL) on chromosome 16p12.1



Protein: 438 aa (47.622 Da); highly N-glycosylated; hydrophobic; phosphorylated, lipid modification sites

**Tim Husemann is one of the affected children. His father has established the NCL-Foundation and fights against time.**

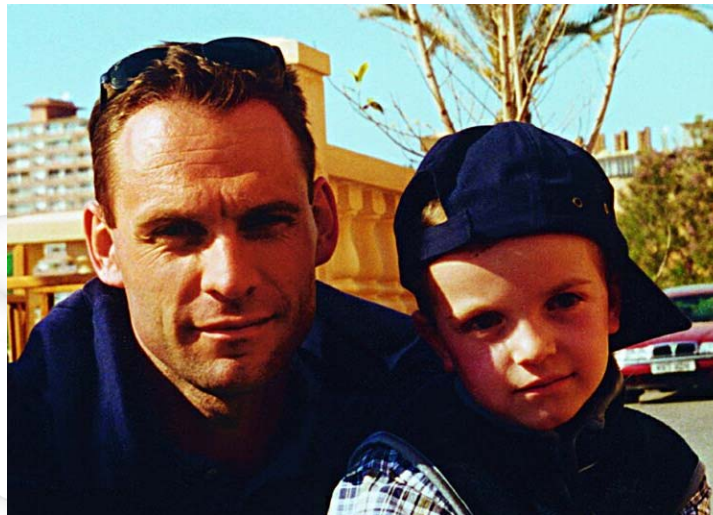
## **The disease and possibilities of treatment**

### **What is the cause of the disease?**

- NCL-children have a **gene defect**.
- A **protein** is missing.

• Build-up of **storage material**.

• **Cells die**. Forever.



- First the **eyes** are affected, then the **brain**.
- Gradual loss of different **body functions**. At last the respiration.

### **What can one do?**

• **Protein replacement**: Generation of protein.

• **Medication**: Use of drugs which work in comparable diseases.

• **Animal models**: Simulate course of disease.

• **Genetherapy**: Transport of functional gene into cells.

# The basic concept of the foundation calls for sharp focusing of aims and activities.

## Aims and Activities

**Employment of one scientist. His main areas of focus are**

- increase **knowledge** of NCL, to increase the possibility of an **early diagnosis**
- activities are concentrated on setting up/extending an interdisciplinary **NCL-network** of scientists
- to evaluate, develop and pursue **clinical trials** against NCL
- secure **donations and financing** to give initial impulses in research

# The NCL-Foundation wants to combine and focus knowledge of different disciplines on one question.

## Comparison of “classical” sponsoring and “NCL- research initiation”

### Classical sponsoring

### NCL-research initiation

#### Proceeding

Established: application, appraisal of the research concept, funding

New: initiative funding to build up research networks

#### Idea

Direct funding on one research idea

Indirect funding: Initiation of research to establish research networks

#### Aim

Research, extend knowledge

Knowledge management and its application

#### Area of application

By the majority on well known and wide spread diseases (e.g. cancer, Alzheimer)

On an orphan disease (NCL, especially the juvenile form)

#### Application of funds

For the institute, clinic, chair etc., which applied for the funding

For the coordinator (scientist) of the non-profit NCL-Foundation

#### Incentive system

Direct research funding, reputation

The dedicated scientist sets incentives, encourages experts to do NCL-research, to apply for funding

#### Sponsors

Industry, big foundations, the state

Private and company donations

#### Elements (e.g. Board)

Crosslinking of traditional structures

New establishment, volunteer

# Successful work of the foundation requires a lot.

## Prerequisites for the NCL-Foundation work

- A **Foundation concept**, which makes allowance of risks and restrictions.
- **Focusing** of activities.
- **Combat cause** not symptoms. In contrast to patient organization, it is not in the foreground to help to deal with this disease, but the aim is to quickly overcome the disease by research.
- **Cooperation** with patient organization „NCL Gruppe Deutschland“ (Rüther).
- **Size** of Foundation, which limits expenses of management and finances.
- **Volunteer** take-over of foundation functions by socially dedicated, cooperative and qualified persons.
- An **open organization**. Human resources must be quickly adjustable.
- **Virtual organization**. Intensive use of electronic media for communication, coordination and extension of network.
- Permanent **awareness training and public relations**.

# Organization of the NCL-Foundation.

## SCIENTIFIC ADVISORY COUNCIL (EXCERPT)

- Prof. R. Baumeister
- Prof. C. Hübner
- Prof. K. Rüther
- Prof. P. Saftig

## NCL CONTACT NETWORK (EXCERPT)

- Prof. T. Braulke
- Prof. H.-H. Goebel
- Prof. A. Jalanko
- Prof. A. Kohlschütter
- Prof. K. Sandhoff

## RESEARCH AND MARKETING

- Scientist

- IT: J. Nörskau
- Graphics: E. Koch
- Legal: T. Burkert
- Taxes: I. Steffen
- Translation: C. Jädke

- Dr. C. Borchard
- Dr. F. Husemann
- Mr. M. Hartleif

## SUPPORTING FUNCTIONS (EXCERPT)

## BOARD



The board is comprised of tax accountants and a business economist.

## NCL-Board



**Dr. Frank Husemann:** Senior manager at Accenture. Dr. Husemann is **the founder** of the NCL-Foundation.



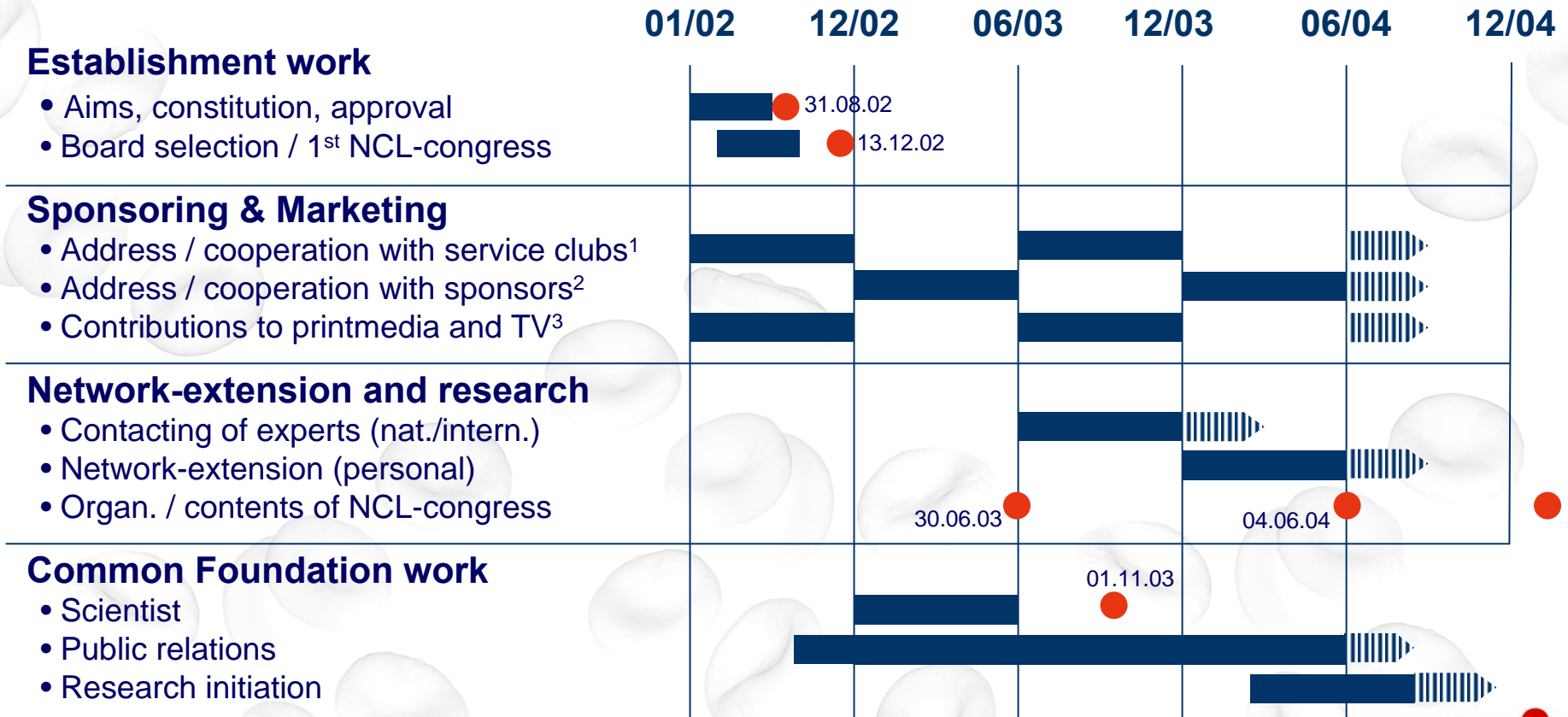
**Martin Hartleif:** Independent tax accountant working in Dortmund. Martin Hartleif is a member of **Round Table** (Dortmund).



**Dr. Christian Borchard:** Independent tax accountant working in Hamburg. Dr. Borchard is a member of a **Lions-club** (Hamburg).

# The ongoing development of the NCL-Foundation is very promising.

## Formation and planned development of the NCL-Foundation



<sup>1</sup> Round Table: national service project. Lions: approx. 5 clubs, and Leos (national service day)

<sup>2</sup> Accenture, Lucia, Lufthansa AG, Mark-E, Management-Circle, Schwarzkopf

<sup>3</sup> Article in: Welt am Sonntag (6/02; 12/03), FAZ (6/02), Hamburger Abendblatt (8/02), Ruhrnachrichten (3/02)  
 TV: SAT 1 („17.30“), RTL 2 („Die Redaktion“), PRO 7 („taff“), ZDF („Hallo Deutschland“).

# Open for discussion.

## Future directions

**Contact**

- *Patient organization*
- *Scientists*

**Exchange**

- *Knowledge (4<sup>th</sup> NCL-congress)*
- *Ideas*

**Network**

- *Database*
- *(Inter)national research group*

**Support**

- *Financial*
- *Networking*

**Combined  
action**

- *What can you do for the NCL-children?*

