



Introducing BDFA Funded Researcher Dr Susanne Lezius



Dr Susanne Lezius is a bi-mathematician at the Institute for Medical Biometrics and Epidemiology at the University Medical Center Hamburg-Eppendorf. Her collaboration with the DEM-CHILD database team began in 2014, analysing data on the course of Batten disease and defining clinical endpoints for clinical trials. Susanne brings a wealth of experience to her new expanded role for the NCL

International Registry, made possible by the joint funding initiative of the BDFA, BDSRA, Noah's Hope and Hope for Bridget.

"We are deeply grateful for the generous financial support from the funding partners which will allow Dr Lezius to continue her work with us on the International NCL registry"

Dr Angela Schultz
MD, Children's Medical Centre, Hamburg-Eppendorf

Why do we need registries?

Having accurate and up to date information is vital for the successful clinical trials and to better understand what is happening, in the disease. This can lead to more effective care and treatments and help with improved methods of diagnosis.

How can you help?

The letter opposite details how UK patients can participate in the NCL registry. To take part or for more information please contact Dr Ruth Williams, the lead clinician for the UK.



Dear Parents/Guardians,

We are writing to you to let you know about an International Registry and Database for children diagnosed with Neuronal Ceroid Lipofuscinoses (NCL), also known as Batten disease.

NCL are a rare group of progressive diseases that mainly affect the brain and cause symptoms such as epilepsy, movement disorders, dementia and blindness. In general, symptoms worsen with time but the age at which symptoms start and the speed at which the disease progresses is very variable. The diagnosis is usually made by examination of the patient, MRI brain scan and blood tests. The diagnosis is then confirmed by genetic analysis.

At the moment we cannot predict how the disease will progress in any one individual. It may depend on many different factors, including the person's genetic makeup, their environment and lifestyle.

An International Batten Disease Registry has already been established and we hope that the information we collect from UK families will contribute to this international project and increase our understanding of how the disease progresses and why the progression is so variable between different people. Currently we do not have good treatments for these diseases, and we also hope that the information we collect from children and families worldwide will in time help us develop and test treatments for these diseases.

We would like to include as many people with NCL in the database as possible. The more information we are able to collect from different people with the disease, the greater our understanding of the disease will become.

We are looking to collect information from your child's medical records as well as the results of any tests that your child may have had, and we may also ask your permission for researchers to use existing samples of biological material (i.e. blood, skin cells, other cells) that may have been taken during the period of confirming the diagnosis. We will not ask your child to undergo any extra tests or procedures for the purpose of this study.

If you would be willing to help us with this study, please contact me on **0207 188 3998** or ruth.williams@gstt.nhs.uk for further information.

Yours sincerely,

Dr Ruth E Williams,
Consultant Paediatric Neurologist,
GMC 3057036