CLN2 disease

Intracerebroventricular enzyme replacement therapy (ICV-ERT) for CLN2 disease represents the first approved treatment for neuronal ceroid lipofuscinosis (NCL) diseases. It is the first treatment where a recombinant lysosomal enzyme, cerliponase alfa, is administered into the lateral cerebral ventricles to reach the central nervous system, the organ affected in CLN2 disease. If untreated, CLN2 children show first symptoms such as epilepsy and language developmental delay at 2-4 years followed by rapid loss of motor and language function, vision loss, and early death. Treatment with cerliponase alfa has shown to slow the rapid neurologic decline. However, the mode of administration by 4 hour-long intracerebroventricular infusions every 14 days represents a potentially greater risk of infection compared to intravenous enzyme replacement therapies. The Hamburg NCL Specialty Clinic was the first site worldwide to perform intracerebroventricular enzyme replacement therapy in children with CLN2 disease. In order to ensure maximum patient safety, we analysed data from our center from more than 3000 intracerebroventricular enzyme replacement therapies in 48 patients over 6 years with regard to the occurrence of device-related adverse events and device infections. Since starting intracerebroventricular enzyme replacement therapy, we have also developed and continuously improved the “Hamburg Best Practice Guidelines for ICV-Enzyme Replacement Therapy (ERT) in CLN2 Disease.” Results from this study showed low rates for device-related adverse events and infections with 0.27% and 0.33%, respectively. Therefore, following our internal procedural guidelines has shown to improve standardization and patient safety of intracerebroventricular enzyme replacement therapy for CLN2 disease [1)]

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Schwering C, Kammler G, Wibbeler E, Christner M, Knobloch JK, Nickel M, Denecke J, Baehr M, Schulz A. Development of the “Hamburg Best Practice Guidelines for ICV-Enzyme Replacement therapy (ERT) in CLN2 Disease” Based on 6 Years Treatment Experience in 48 Patients. J Child Neurol. 2021 Feb 5:883073821989154. doi: 10.1177/0883073821989154. Epub ahead of print. PMID: 33543660.