Glossary

Autosomal recessive disease
A pattern of inheritance where 2 mutated genes (one from each parent) are passed to their child, which then causes a certain disease.

Batten disease
A group of rare, inherited, neurodegenerative disorders that mostly begin in childhood. It is named for Dr. Batten, who discovered the disease. CLN2 disease is a common form of Batten disease.

Cerebrospinal fluid (CSF)
Body fluid surrounding the brain and spine.

Cerliponase alfa
A drug form of human TPP1, the enzyme that is absent, missing, or not working properly in patients with CLN2 disease. It is the generic name for Brineura® (cerliponase alfa), which is approved to slow loss of ability to walk or crawl (ambulation) in symptomatic pediatric patients 3 years of age and older with late-infantile CLN2 disease.

CLN2 disease
A rare autosomal recessive disorder where lysosomes in the brain don't have enough of the TPP1 enzyme to break down materials in cells.

CLN2 Clinical Rating Scale
This scale was designed specifically for CLN2 disease and evaluates motor function. It can also help doctors determine how well treatment with Brineura is working.

Enzyme
Proteins that cause reactions to occur in cells.

Enzyme replacement therapy (ERT)
A treatment that helps replace missing or improperly working enzymes. In the case of CLN2 disease, treatment with ERT helps replace the TPP1 enzyme.

Extension study
After the main clinical study has ended, patients may be able to continue treatment by enrolling in this kind of study.

Hypersensitivity
A set of reactions produced by the immune system, including fever, vomiting, irritability, and severe or serious allergic reactions (called anaphylaxis).

Infusion
A way in which drugs are delivered into the veins, or ventricles.

Intraventricular access device
A device that is surgically implanted below the scalp and allows Brineura to be delivered through intraventricular infusion.

Intraventricular infusion
A method in which a drug is infused directly into a ventricle in the brain. This is how Brineura is delivered in children with CLN2 disease.

Late-infantile
An age range that generally includes toddlers to very young children. This age range is used to describe when signs of a disease typically begin to appear.

Lysosome
Lysosomes are found within cells, and contain enzymes. One of their primary purposes is to recycle particles using enzymes.

Lysosomal storage disorder (LSD)
An inherited disease that is characterized by an abnormal build-up of various materials in lysosomes in the body's cells as a result of a reduced amount of a particular enzyme. Nearly 50 various types of LSD have been identified to date.

Magnetic resonance imaging (MRI)
A noninvasive technique that uses a magnetic field and radio waves to produce computerized images of the organs and tissues within the body.

Motor function
The ability to walk or crawl, which is what the CLN2 Clinical Rating Scale measures.

Neurodegenerative
Having a decline or change in brain function.

Neuronal ceroid lipofuscinoses (NCLs)
Neuronal ceroid lipofuscinoses, or NCLs, are a group of rare, genetic, neurodegenerative disorders.

Tripeptidyl peptidase 1 (TPP1) enzyme
An enzyme that breaks down material in cells. A reduced amount of this enzyme is what causes CLN2 disease.

Ventricle
A hollow cavity in the brain that contains CSF.