



## Patient Advocate-Driven Advancement of Clinical Recommendations in Ultra-Rare Disease: The CLN1 Batten Disease Experience

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### Abstract:

With rare diseases, an individual patient may be the only case treating physicians will see in their careers. There is often limited information available on how best to manage the clinical care of such patients due to a lack of published guidelines. As a result, clinicians and families may be left caring for patients without a point of reference or insight from what has and has not worked well for others. Management consensus based on both clinical and family experience has great value to inform clinicians and families in the future.

CLN1 disease (neuronal ceroid lipofuscinosis type 1), a type of Batten disease or neuronal ceroid lipofuscinosis (NCL), is an ultra-rare pediatric neurodegenerative disorder. To date, this ultra-rare disease has lacked a published clinical consensus likely due to low prevalence, disease course variances, lack of existing care networks, and other factors. Taylor's Tale, a CLN1 disease patient advocacy group, created a partnership between families and clinicians to build a professional, uniquely informed CLN1 disease management consensus. This project can serve as a model for other rare disease organizations.

### Objective:

Design and execute a patient-advocacy group driven initiative that partners families' perspectives with clinicians experienced in CLN1 disease in order to identify management strategies.