This Q & A article is from BDSRA’s “Ask An Expert” Facebook Chat in the BDSRA Closed Facebook group for parents and caregivers.

\*Please note: the information contained in this material is intended to provide

basic information to Batten families and caregivers. It is not intended to be, nor is it, medical advice for individual children. Parents and caregivers should consult the patient’s physician prior to changing medication, medical treatment

or daily activities.

“Ask a Pediatric Neurologist”

Featured Expert:

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Q: Can a child with the CLN1 type of Batten disease not experience seizures?

A: Most children with CLN1 will have seizures at some point, although there can be some individuals who are exceptions to this.

Q: I was wondering my geneticist counselor said that my son has juvenile battens but she also said that he is cln2 tpp1 so is that juvenile or later infantile BC my neurologist said last infantile

A: There was a time where we talked about type of Batten Disease based on an individual's age at symptom onset. We have since learned that the genotype (or enzyme problem) is also very important. Classification is now primarily based on genotype (eg. cln1, cln2, cln3, etc). It may be helpful to have further discussion about your son's diagnosis with one of the Centers of Excellence. The BDSRA could help facilitate this. BDSRA can help connect doctors in the community with Batten experts, just so they can make sure they are accurate in the diagnosis of the child.

Q: My child has JNCL. She is currently at CHOP and has been for over a month now. In March she was hospitalized with urosepsis and was hospitalized for almost a month. She was discharged only to have to be readmitted 5 days later for shock. Her blood pressure drops really low. She is doing better now, but is this something that our kids are susceptible to, the low blood pressure, shock, etc.? I'm scared that once she is discharged again, how will I know if she is in shock again? We almost lost her that night.

A: I am so sorry to know that she is still in the hospital. Our thoughts are with you. In general, low blood pressure is not something that we specifically know to be a problem related to JNCL. However, we do know that children, who have neurological disorders, especially when mobility is limited, are susceptible to problems with serious infections, which sometimes can result in low blood pressure - shock. Shock can be a sign of serious infection. Things to watch for are a person's alertness and responsiveness, skin coloring (being very pale), hands or feet becoming cool, heart rate being very low or very high. When these signs all come around the same time, it is an important time to be in contact with your regular doctors.

Q: Is there any new information on CLN5 and how can I connect with other researchers or people studying this particular type?

A: There aren't any CLN5 human trials now. But, Dr. David Palmer in New Zealand has incredible data on gene therapy in sheep--a necessary large animal model that appears to normalize them. With your permission, I would be glad to connect you.

Q: Is it unusual for at the age of 13 to still have his eyesight since the symptoms began ten years ago?

A: In regards to eyesight, as well as other symptoms, there is a fair degree of variability in symptom onset and course in patients with CLN1 and CLN2. Symptoms still have variability for patients with CLN3, but some symptoms, like vision loss, are fairly consistent in age when they start for CLN3 patients.

Q: My son, like many children with Battens, was initially diagnosed with a seizure disorder and our neurologist was not going to be satisfied until we found the right combo of meds to make him completely seizure-free. With the Battens diagnosis, is this still possible, or will our children continue to have seizures and should we be satisfied that our son's seizures are controlled to one a month?

A: How to define seizure control is unique for each affected individual and their family. When seizures have been difficult to manage, there is sometimes a tradeoff between side effects and number of seizures. Parents, working with their physicians are often just the right individuals to define that balance. With a Batten Diagnosis, although seizure freedom may not be achievable for many, there may still be an ideal to achieve for each child. Some families with their doctors target 'big seizures' that take a toll on their loved one's alertness or ability to engage day to day. Others focus on seizures that impact safety - such as seizures that cause drop attacks or falls.

Q: My son has been diagnosed with CLN10. I was just wondering what we could expect. He is 20 yrs. old now. He has gradually gone downhill but has really started to progress in the last 6-8 months. He is experiencing seizures, speech problems, is losing muscle tone, and just overall weakness. He has gone from eating some by mouth, to being completely tube fed within the last 3months.

A: I am sorry to hear the difficult time your son is having. We do sometimes see that patients with different forms of Batten disease will have a very sudden decline. In general, our experience is that some will continue to have rapid progression of symptoms, where some will enter a period of stability for a while, without new complications. It has been a challenge to predict who will follow which course. At a time where patients have had a sudden change in symptoms, it can be helpful to involve a palliative care team. These teams are often discussed at end-of-life times, but more and more often, they are, and should be, involved in the care of patients with complex medication needs. They can provide helpful insight and assistance with symptom management, early on.

Q: Just one more thing on this, when we brought her back in this last time, they couldn't find any sign of infection. Took lots of blood work, blood/urine cultures, X-rays, scans, etc., and nothing stood out. This is why it's so scary. I could potentially be running her back to the hospital a lot.

A: When patients have symptoms that can be serious, and that can happen over and over, it can be helpful to develop an action plan. This is something we do for patients with asthma, for patients with seizures, and patients with other kinds of problems. Once a recurring symptom is identified, the action plan helps parents, other caregivers, and teachers/schools to know a) what to look for, and b) what to do when each symptom happens - when to do some watchful waiting, when to start a treatment at home, and when to head to the hospital.

Q: My daughter is diagnosed by the NIH with CLN6. There are no trials that I currently know of. Do you know of any maybe?

A: I am not aware of currently ongoing clinical trials for CLN6 in the United States. We do, however recognize that the state of clinical trials for Batten Diseases is evolving. For the most up-to-date information about clinical trials in the United States, [www.clinicaltrials.gov](http://www.clinicaltrials.gov/) is a very good resource. Every trial conducted in the US must register on this site, before enrolling patients.

Q: My daughter is almost 5 with symptoms starting at 2 weeks of age. She was diagnosed with GRODS and fingerprint inclusions. However every known gene has been tested and we still are unable to find the affected gene. Do you have suggestions on our next step?

A: For patients with NCL-like diseases in general, we continue to discover 'new NCLs' over time. Working with your doctors to revisit possible causes and newly discovered genes can be helpful.

Q: My daughter is 20 and has JNCL. I recently heard that Biotin could possibly slow degeneration of the myelin sheath. I give it to my daughter as a hair supplement but was told for brain health it should be a much larger amount. I was wondering about your thoughts on this.

A: I am not aware of direct study of biotin for JNCL. There are uses for biotin in neurological disorders affecting children. This is typically in the context of biotin deficiencies and related disorders. In addition, there was a recent small study of biotin for patients with chronic progressive multiple sclerosis. Both of these are different kinds of diseases than the NCLs.

Q: Do you know of a neurologist in the Orange County New York area that you would recommend for Batten disease. I am also 50 miles north of the city. I haven't been able to find a neurologist that I feel comfortable with in the year that I've been here.

 A: For physician referrals, the BDSRA is knowledgeable about physicians around the nation. I would be happy to work with the BDSRA regarding a physician referral in the Orange County/NYC area.

Q: My daughter is almost 7 and was diagnosed a few days ago at NIH. Vision loss is her only symptom but the past few days she complains of skin sensitivity and feeling like bugs are crawling on her. Prior to the diagnosis I was brushing her off. Is this possibly related to her JNCL?

 A: I am glad that the NIH was able to help to find your daughter's diagnosis. For patients with JNCL, hallucinations can be a symptom. There are many causes of skin sensitivity and abnormal feelings. If this persists, it sounds like these would be good symptoms to discuss with your local neurologist.