PHYSICIAN DISCUSSION GUIDE

Talking Through a Sialidosis Diagnosis

A printable companion for appointments — built with families, geneticists, and neurologists to help you ask focused questions and leave every visit with a clear next step.

How to use this guide

Bring this sheet to appointments with your neurologist, geneticist, ophthalmologist, or primary care physician. Check off questions as they're answered, and use the blank lines to note the response. There is no obligation to ask every question in one visit — use what's relevant to where your family is in the diagnostic journey.

Quick facts to confirm with your care team

Sialidosis is an ultra-rare, inherited lysosomal storage disorder caused by mutations in the **NEU1** gene, which reduces activity of the enzyme neuraminidase (sialidase).

Inheritance is **autosomal recessive** — both biological parents typically carry one copy of a NEU1 variant without symptoms themselves.

Two broad clinical types are recognized: **Type I** (later-onset, primarily neurologic/ocular) and **Type II** (earlier-onset, systemic, more severe).

Questions about diagnosis

[ ] What specific test confirmed (or will confirm) this diagnosis — enzyme activity assay, NEU1 gene sequencing, or both?

[ ] Was a cherry-red macular spot identified on dilated ophthalmologic exam? Which type is this consistent with?

[ ] Should we pursue confirmatory genetic testing to identify the specific NEU1 variant(s)?

[ ] Is this classified as Type I (cherry-red spot myoclonus syndrome) or Type II (including congenital, infantile, or juvenile-onset forms)?

[ ] Should other family members, including siblings and future pregnancies, be offered carrier testing or genetic counseling?

Questions about symptoms and monitoring

[ ] What symptoms should we expect to monitor — myoclonus (sudden muscle jerks), seizures, changes in vision, or changes in coordination and gait?

[ ] How often should vision, hearing, and neurological function be reassessed?

[ ] Which specialists should be part of the ongoing care team (neurology, ophthalmology, genetics, physical therapy, audiology)?

[ ] Are there early signs that should prompt an urgent call rather than waiting for the next scheduled visit?

Questions about treatment, trials, and support

[ ] What is currently being done to manage symptoms (e.g., anti-myoclonic or anti-seizure medication, physical or occupational therapy, low-vision support)?

[ ] Are there active natural history studies or clinical trials our family may be eligible for?

[ ] Can you refer us to a metabolic or lysosomal disease specialist, if we aren't already seeing one?

[ ] Where can we find vetted educational material and peer support for Sialidosis specifically, rather than general rare-disease resources?

[ ] Is our family eligible for patient registries that track disease progression and connect families to research opportunities?

Type I vs. Type II at a glance

	Type I	Type II
Typical onset	Late childhood through early adulthood	Birth through early childhood
Core features	Progressive myoclonus, seizures, cherry-red macular spot, gradually declining visual acuity; cognition often preserved	Cherry-red spot, coarser facial features, hepatosplenomegaly, skeletal changes (dysostosis multiplex), possible developmental/intellectual involvement
Common name	Cherry-red spot myoclonus syndrome	Includes congenital, infantile, and juvenile-onset forms
Typical pace	Slower progression	Faster progression, wider systemic involvement

This table is a general orientation, not a diagnostic tool. Presentation varies between individuals even within the same type — your care team's clinical and genetic findings take precedence.

Notes from today's visit

Date: _____ Clinician: _____

Next appointment / follow-up test: _____

Tree For Cure Foundation funds Sialidosis research and provides free educational resources for newly diagnosed families, caregivers, and clinicians. This guide does not replace medical advice — always follow the direction of your care team. Need support finding a specialist or connecting with other families? Visit the Resources section of treeforcurefoundation.org or email info@treeforcurefoundation.org.