

Understanding Cystinosis

A guide to supporting
your student with a
rare genetic disease



You can make a difference

for your student with cystinosis

You likely have never heard of cystinosis. It's a rare genetic disorder affecting only about 500 to 600 people in the United States.

One of your students is living with cystinosis. The condition can be managed, but both the disease and the treatment come with symptoms and side effects that can make children with cystinosis stand out in ways they might not want to. Students with cystinosis may also be absent more often than others because of illness and frequent medical appointments.

This guide is intended to provide you with a brief overview of cystinosis and treatments for it, as well as information on how the condition can impact students' lives at school. We hope that it will help you support your student with cystinosis and educate other students and staff about the disease.

To learn more about cystinosis, visit CystinosisUnited.com.

Cystinosis 101

What is cystinosis?

Cystinosis is a rare, genetic disease that results in a buildup of cystine, an amino acid, inside cells. This buildup leads to the formation of crystals, causing damage to nearly every cell and organ in the body.

The condition is progressive. Signs of damage usually start in the kidneys and eyes but then develop differently in each person.

A student with cystinosis may have:

- Small stature
- Sensitivity to light
- Muscle wasting
- Frequent thirst
- Fatigue
- Frequent need to urinate
- Trouble swallowing
- Diabetes

How is cystinosis treated?

Cystinosis is treated with medicines called cystine-depleting therapies (CDTs), which work to lower the levels of cystine in the cells and may limit damage to the body. People with cystinosis may also need other medicines or supplements, including eye drops, to manage specific symptoms.

As kidney function declines, people with cystinosis may require dialysis or even a kidney transplant. Dialysis can be fatiguing, typically takes hours for each visit, and may be done up to 3 times a week.

A student with cystinosis may experience these side effects from use of CDTs:

- Bad breath
- Sulfur-like body odor
- Nausea, vomiting, diarrhea, or abdominal pain
- Headache

How cystinosis affects life at school

Children with cystinosis want the same experiences as any other child at school. They want to learn, play at recess, and make friends. But sometimes the challenges of living with cystinosis can get in the way.

Accommodations to support a student with cystinosis

A student with cystinosis has to deal with some challenges and issues that other students do not.



Medicines—People with cystinosis must take medicine every day on a precise schedule. The school nurse may need to assist the student with taking medicines at scheduled times.



Bad breath or body odor—CDTs may cause these. Allowing a student to have mints, chew gum, or apply essential oils during class can help.



Dehydration—Cystinosis causes dehydration, and students experience near-constant thirst. Allow the student to carry a water bottle and to go to the restroom as frequently as needed. Have the student keep a spare set of clothes available, perhaps in the school nurse's office, in case of accidents.



Sensitivity to light—Because the eyes can be affected by cystine crystal formation, the student may need to sit away from direct sunlight, wear sunglasses in class, or regularly use eye drops that help remove the cystine crystals. Outdoor activities may need to be limited or avoided because of sun exposure.



Sensitivity to heat—An impaired ability to sweat is another symptom of cystinosis and may make a student especially sensitive to heat. Activity may need to be limited, especially in warm rooms or outdoors.



Low energy levels—Cystinosis can also cause low energy levels. Allowing the student to snack throughout the day can help.



Nausea or fatigue—These symptoms may interfere with the student's ability to get work done on time or take tests as scheduled. The student may need more time to take quizzes or tests and may need to take breaks to rest, perhaps in the school nurse's office.



Gastronomy tube (G-tube)—Some students may need to be fed or given medicines through a G-tube. School personnel may need special instruction to support students with G-tubes.

Absences

Cystinosis may cause a student to miss school. Reasons may include:

- Doctor appointments
- Feeling sick
- Hospitalizations
- Interrupted sleep due to medicine schedules
- Dialysis
- Kidney transplant

You can help by working with the student's parents or caregivers to plan for expected absences, especially those that may be long term.

A student with cystinosis will benefit from a missed-school plan to assist in keeping up with work. It is important that the student, parents, and teachers agree about what classmates will be told about the student's absence.

Speaking about cystinosis

Helping peers understand cystinosis can make a big difference for your student. The student, the student's parents, or the school nurse may want to speak to teachers and classmates at the start of the school year to explain cystinosis to them.

A video and downloadable guide that can help get you started can be found at CystinosisUnited.com/what-is-cystinosis.

Appointing an advocate

It can be helpful to have a single point person who knows the student well to be the student's advocate at school. This person should be available to answer questions and tackle problems. A counselor, nurse, or teacher are good options.

Bullying

Some symptoms of cystinosis and side effects of treatment may make a student a target of bullying. The student with cystinosis may feel more confident if plans are in place to address other students' reactions. Educating classmates about the challenges that come with cystinosis may also help them develop empathy toward the student.

There are many antibullying resources available. Here are a few:

- **TEEN LINE: TEENLINEonline.org**
- **StopBullying.gov**
- **Pacer's National Bullying Prevention Center: PACER.org/bullying**

504 plans and individualized education programs (IEPs)

Because of the complexity of cystinosis, it may be necessary to have a formal IEP or 504 plan in place to ensure challenges are addressed. Written documentation of accommodations made early in a student's career can be important in ensuring appropriate accommodations are made later on.

Here is a list of possible modifications or accommodations that may be considered for a 504 plan or an IEP:

- Arrange extra time to complete assignments, tests, and quizzes
- Share a copy of teacher or student notes, or use technology resources to record missed lessons
- Provide modifications in the event that students need them (for example, no stairs, modified PE schedule)
- Allow the student to show proficiency in a different way, instead of completing all requirements or assignments
- Permit water bottles or provide easy access to water for hydration
- Accommodate extra trips to the restroom
- Permit sunglasses for light sensitivity
- Permit gum or mints to counter body and breath odor
- Create an emergency medical plan with the family, school, and doctor
- Allow snacks to help the student overcome low energy levels
- Provide a quiet place for the student to rest if he or she is feeling tired
- Let the student keep an extra set of clothes with the school nurse
- Offer preferential seating near the teacher, near the front of class, or in a quiet space
- Assign a peer tutor for class work and assignments, and look into tutoring programs offered through the school
- Make at-home services and instruction available as needed



IEP and 504 plan resources:

- **Disability Rights Education & Defense Fund:** <https://dredf.org/special-education/sample-letters/>
- **KidsHealth:** <https://kidshealth.org/en/parents/iep.html>

5 ways to help your student with cystinosis

ALLOW THE STUDENT TO:

- 1** Chew gum or have breath mints
- 2** Have water and snacks in class
- 3** Take frequent bathroom breaks
- 4** Take more time to complete homework assignments and tests
- 5** Create a plan for making up work when unable to attend class

To learn more about cystinosis,
visit CystinosisUnited.com.

Accommodations planning checklist



Student name

Parent name

Parent's phone number

Schoolwide considerations

- Establish a school plan for the management of cystinosis
- Identify a student advocate at school
- Establish procedures for absences
- Discuss with the child's parents what other students will be told about cystinosis
- Become familiar with special food and drink needs
- Discuss physical abilities and limitations in physical education

For the student services staff or school administration team

- Establish a plan for the student to take medicines at school, if needed
- Review the medicine schedule with parents
- Keep an emergency supply of medicines at school
- Store an extra set of clothes for the student in case of bathroom accidents
- Identify a quiet, safe place for the student to rest if needed
- Create an emergency medical plan with parents and the student's doctor

For classroom teachers

- Provide extra time for tests and quizzes
- Provide extra time to complete assignments
- Discuss if 504 plans or IEPs are needed
- Allow the student to have gum or mints to mask breath and body odor
- Allow the student to have a water bottle to stay hydrated
- Allow snacks in the classroom to help the student overcome low energy levels
- Allow the student to wear sunglasses and/or sit away from the light as needed
- Allow the student to go to the bathroom as frequently as needed

Other notes and considerations

Parents/caregivers: Please list any special information about your child that school personnel should know.

Connect with the cystinosis community

The Cystinosis United website (CystinosisUnited.com) is a good source for more information on cystinosis. Additional information is available from multiple sources, including those below, which are not associated with Horizon Pharma.

Cystinosis Research Foundation (CRF)

CystinosisResearch.org

949-223-7610

CRF supports research that aims to improve the quality of life of patients with cystinosis, find better treatments, and ultimately find a cure.

Cystinosis Research Network (CRN)

Cystinosis.org

866-276-3669

CRN is an all-volunteer, nonprofit organization dedicated to supporting and advocating for research, providing family assistance, and educating the public and medical communities about cystinosis.

Global Genes

GlobalGenes.org

949-248-RARE (7273)

Global Genes is a nonprofit advocacy organization for patients and families fighting rare and genetic diseases, including cystinosis. Its mission is to eliminate the challenges of rare disease.

National Organization for Rare Disorders (NORD)

RareDiseases.org

203-744-0100

NORD is a patient advocacy organization dedicated to people with rare diseases and the groups that help them. NORD provides patients and families with advocacy information, assistance programs, and connections to patient organizations.

A school nurse or appointed school advocate may want to have a copy of this form.

Student name	Student date of birth
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Grade/homeroom/teacher

Doctor's name	Doctor's phone number
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Parent/guardian

Name	Relationship	Phone number
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Name	Relationship	Phone number
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Medicines list

Medicine	Dosage (amount)	Time of dose
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Medicine	Dosage (amount)	Time of dose
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Medicine	Dosage (amount)	Time of dose
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Medicine	Dosage (amount)	Time of dose
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