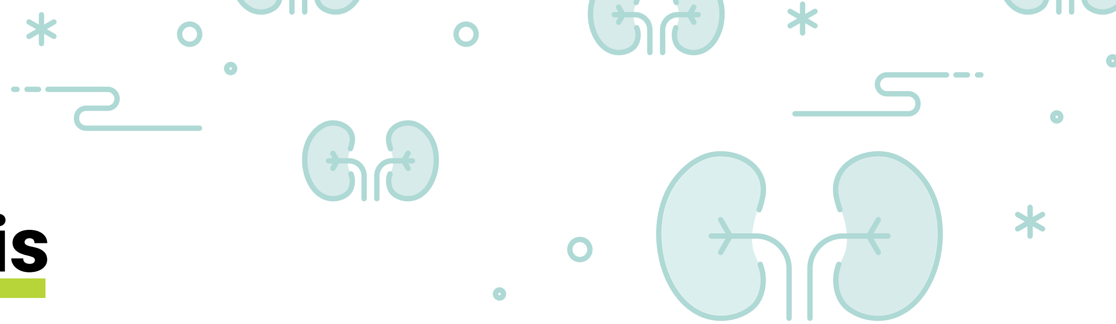
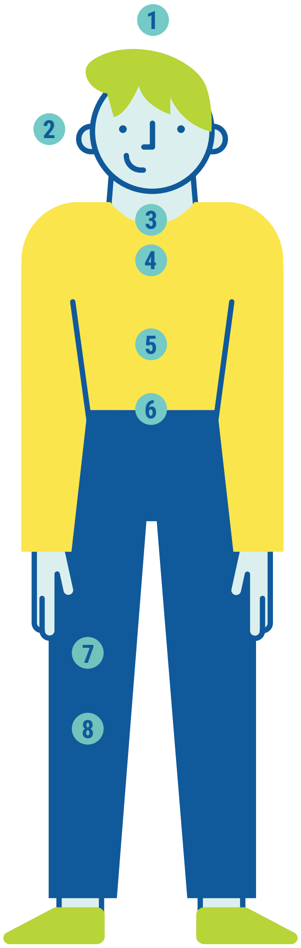


Cystinosis overview

WHAT IS CYSTINOSIS?

Cystinosis is a rare **genetic** condition that leads to a buildup of cystine (a naturally occurring protein) within the lysosomes of the body's cells. Individuals with cystinosis have a non-working transporter that doesn't allow for the cystine to escape from the lysosome. The build up of cystine crystallizes and causes cell death, which leads to organ damage. Cystinosis is an autosomal recessive genetic condition caused by **mutations** or changes in a specific gene called CTNS.

We all have two copies of the CTNS gene but individuals with cystinosis have two copies of the CTNS gene that are not working properly due to mutation(s) in them. Cystinosis is passed down through families.
If two parents happen to be carriers of a gene mutation in one of their CTNS genes, they will have a 25% chance of having a child with cystinosis, a 50% chance of having a child who is a carrier for cystinosis (not affected) and a 25% chance of having a child who is neither affected nor a carrier for cystinosis.



Having too much cystine can damage different parts of the body. Talk to your child's doctor to learn more about the ways cystinosis may be affecting different parts of their body.

① **BRAIN**
Visual or learning issues

② **EYES**
Sensitivity to light (photophobia) or blindness

③ **THYROID**
Poorly working thyroid (hypothyroidism), leading to slowed growth

④ **THROAT**
Trouble swallowing

⑤ **PANCREAS**
Diabetes

⑥ **KIDNEYS**
Fanconi syndrome (a type of kidney disease)

⑦ **MUSCLES**
Muscle weakness and decreased muscle mass (myopathy)

⑧ **BONES**
Softening or weakening of bones (rickets)

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WHAT IS CYSTINOSIS? (CONTINUED)

Over time, cystinosis can lead to permanent **kidney damage** and **kidney failure**. Each person is different, and some people with cystinosis will develop kidney disease as a child or adolescent while others may not until early adulthood. As kidney disease gets worse and the kidneys lose the ability to function, your child will need to start **dialysis** or have a **kidney transplant** to live.

TIPS ON SUPPORT

Cystinosis can be tough on children, but there are ways you can support them and help them cope. Here are a few tips:

1 Listen

Take notice of the verbal and nonverbal cues from your child. Talk to them about how they are feeling and let them freely express their emotions.

2 Educate

Talk to your child about their health in terms that they can understand. Involve them during doctor visits. Let your child ask questions and let them know what will be happening ahead of time before any procedures, big and small.

3 Normalize

Help your child feel comfortable with cystinosis. Help your child accept this part of their life by having frequent, open talks about cystinosis.

4 Reassure

Focus on the positives. Remind your child of what they can accomplish and what they can do well.

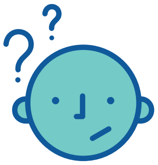
5 Empower

Help your child feel as much in control of their life as possible. Let them make their own decisions. Small choices, like which arm to have blood drawn from or if they want juice or water with their medicine, can go a long way.

Understanding what your child is going through

The diagnosis of kidney disease due to cystinosis can be overwhelming and even unexpected. As a parent, it is natural to feel a wide range of emotions as you adjust to this news and what it will mean for your child and your family.

Your child may also feel a combination of emotions such as:


Confused


Overwhelmed


Scared


Angry


Sad


Frustrated

YOUNG CHILDREN

Behavior changes in younger children can include:

- Acting younger than they are (e.g., a 7-year-old suddenly sucking their thumb or wanting to sleep with you in your bed).
- Showing how they feel by misbehaving more than usual.
- Refusing to take medicine or resisting doctor visits.

Children who are too young to understand what their diagnosis of cystinosis-related kidney disease means may react differently than older children.

YOUNG CHILDREN (CONTINUED)

Some things you can do as a parent are:

- Explain what is happening and what to expect in a way your child can understand.
- Bring familiar items such as your child's favorite toy, book, or blanket to the hospital. Child Life programs are also available in certain children's hospitals to provide a more welcoming environment.
- Acknowledge their emotions (e.g., "It seems like you are feeling very sad, do you want to talk about it?").
- Support positive behaviors (e.g., "You are so brave for coming to the doctor like a big kid!").

PRETEENS AND TEENS

No matter what emotion your child is feeling or expressing, you can let them know that any emotion is okay. Let your child know that they are welcome to talk openly about how they feel.

Reactions from preteen and teenage children can include:

- Asking a lot of questions about how cystinosis affects the kidneys.
- Withdrawing and not wanting to talk at all.
- Expressing feeling very isolated. This might be because they don't know anyone else with their condition and they feel like "the only one."

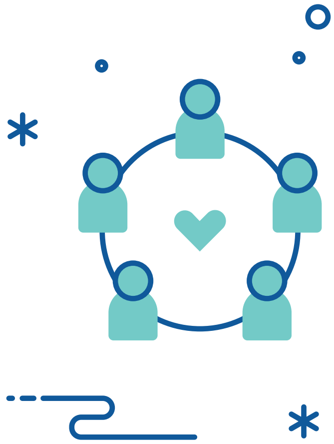
Some things you can do as a parent are:

- Let your child know you are there for them when they're ready to talk.
- Check with your child's medical team to see if there are any patient support groups in the area or activities with other pediatric patients.
    - *You can also connect with the Cystinosis Research Network (Cystinosis.org) to attend their conferences. You and your child will have the opportunity to meet others who are going through a similar experience.*
- Teach your teen to advocate, or speak up, for themselves. Being comfortable talking about their disease will make it easier for them to take charge and advocate for themselves in difficult situations.

Caregiving for parents

As a parent, you act selflessly and always put your child first. But every parent needs time and space to refuel. To provide your child with the best care possible, one of the most important things you can do is **take care of yourself**.

CAREGIVING UNIT



Caregiving is challenging, and you may feel that it is your responsibility alone to care for your child. Isolating yourself so you can be "on-call" 24/7 can lead to stress, burnout, and bitterness. This is why building a support network or "caregiving unit" is extremely important when raising a child with a chronic disease like cystinosis.

Anyone you trust can be a part of the caregiving unit, including:

- Spouse
- Siblings
- In-laws, cousins or other family members
- Friends
- Neighbors
- Coworkers
- Teachers
- Religious community

People in your caregiving unit should understand your child's condition and be able to step in if you need them. For example: if you are your child's primary caregiver and you are planning to donate your kidney to your child, you will need a second or third backup caregiver to step in while both you and your child recover.

Potential caregivers might not always come to you, so you may need to reach out first to gather members for your caregiving team. You may be surprised by who is willing and able to step up to help, if you ask.

If you need to step away from caregiving for a bit but do not have the funds to arrange for a qualified caregiver to take your place, you can apply for financial assistance. The National Organization for Rare Disorders (NORD) offers a grant called the Rare Caregiver Respite Program to help caregivers take time off without compromising the care of their loved one.

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CAREGIVING UNIT (CONTINUED)

The more trusted individuals who are in your caregiving unit, the better. That way if one person is busy, someone else can step up.

Finding additional caregivers and asking them to help you as needed will benefit everyone.

- You will get more time to complete simple tasks such as showering, napping, and running errands.
- You will be able to give your other children or family members more of your time.
- Your child with cystinosis will have someone else who can answer their questions or help them do everyday tasks, like driving them to or from school.
- People who want to help you in some way, but do not know how, will have a chance to do something helpful.

Practical suggestions for managing your caregiving unit include:

- Create a group text using tools like GroupMe or WhatsApp for regular updates on your child's overall health and lifestyle (e.g. latest lab results, medicine changes, favorite TV shows, school schedule, etc.).
- Use online survey tools, such as Doodle or When2Meet, to track and see everyone's availability each week or month.
- Use the Find My Friends app in case of an emergency to see who is nearby and can help complete an errand, stop by the house, or take your child to an appointment.

PASSING ON THE RESPONSIBILITY

As your teenager moves closer to adulthood and independence, this is a good time to slowly let them take over the responsibility of caring for themselves.

- Involve your teen in every discussion related to their health (e.g. treatment choices, medicine decisions, kidney transplant preparation, etc.).
- Teach your teen how to schedule their own doctor's appointments and arrange medicine pick-up with the pharmacy.
- Ensure your teen knows the names of all their medicines and what each pill looks like outside of the bottle.
- Trust your teen to take their medicine on time, as directed.
- Encourage your teen to track their treatment by writing down progress, side effects, and questions for the doctor in a notebook.
- Help your teen develop healthy coping skills such as participating in support groups, openly discussing their illness, and recognizing when to ask for help.

Preparing for your child's kidney transplant

Cystinosis affects several organs such as the kidneys, which can lead to permanent kidney damage and kidney failure.

Kidney transplant is considered the best treatment option for kidney failure because it can increase your child's chances of living a longer, healthier life. Even children as old as 1 or 2 years of age may be able to get a kidney transplant depending on the transplant center. However, as with any surgery, there are some risks involved.

CONSIDERING TRANSPLANT

When someone receives a kidney transplant as a child, teen, or young adult, it is highly likely that they will need more than one kidney transplant during their life. Dialysis is the only other treatment option for kidney failure, but it causes stress on the body and is not considered a life-long solution.

Getting a kidney transplant will take time and effort on the part of your child, your family, and your child's care team. In some cases of cystinosis, you may have years before you need to seriously consider kidney transplant for your child. This can give you the necessary time to research the transplant process and identify a transplant center that works best for you and your family.

PREPARING FOR TRANSPLANT

When your child's medical exams begin showing signs of decreasing kidney function, you should start preparing your child for a kidney transplant.

1 Find a transplant center and schedule a transplant evaluation.

- Many children's hospitals in the United States have a transplant center.
- Some transplant centers require a referral from a doctor, but others do not. Check to see what the requirements are at the center you are interested in.
- Your child must have a full evaluation of physical health, mental health, and finances by a transplant team to help them decide if your child is ready for the kidney transplant.
- If the transplant team decides your child is ready, the next step will be for your transplant team to help you find a kidney match from a living or deceased donor.

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PREPARING FOR TRANSPLANT (CONTINUED)

Whether your child receives a living donation or a deceased donation, taking cystinosis medicine and other prescribed medicine will help the new kidney last longer.

2 Consider the types of kidney transplants.

A **deceased donor transplant** comes from someone you may or may not have known who has just passed away but had a healthy kidney.

- Deceased donor transplants are the most common type in the United States.
- Getting a deceased donor transplant requires being on a waitlist for several months or years.
- As soon as a deceased donor kidney becomes available for your child, you will get a call to come to the hospital for surgery right away.
- To increase your child's chances of getting a kidney transplant as soon as possible, you can get listed at multiple transplant centers. However, you must be able to reach each center in a short amount of time if you are called.
  - *Check with each center to ensure that they allow patients who are listed at multiple centers.*
- Deceased kidney donor transplants last an average of 10 years but can last for a longer or shorter amount of time.

A **living donor kidney transplant** comes from a living person who offers to donate one of their kidneys.

- A kidney donor does not need to be a family member.
- The donor will have to go through medical testing to make sure they are healthy and a good match.
- If you can find a living kidney donor match for your child, you can schedule the date of the transplant surgery based on what works best for your child.
- Living donor kidney transplants last an average of 15 years but can last for a longer or shorter amount of time.

3 Make sure other parts of your child's life are in order leading up to and during the transplant surgery.

- Write a list of questions for the transplant team to answer before and after the surgery.
- Prepare your child for the surgery well in advance by telling them what to expect before and after.
- Let your child enjoy doing things that they will not be able to do immediately after surgery (e.g. go to the mall).
- Bring your child's favorite toy, blanket, or keepsake for comfort during the hospital stay.
- Try to maintain your child's usual routine as much as possible (e.g. follow the same bedtime, eating schedule, and medication schedule).

PREPARING FOR TRANSPLANT (CONTINUED)

Donating a kidney to your child

As a parent, your first thought may be that you or your spouse will donate a kidney to your child. If you want to be a living donor for your child, you will need to have a medical exam with blood tests to be sure you are healthy enough to donate a kidney. Some of the tests needed may include:

- Blood tests, urine tests, gynecological exam, colonoscopy (if over age 50), screening tests for cancer, antibody test, X-ray, electrocardiogram (EKG) to look at your heart, and other image testing like a CT scan.
- You are also required to meet with a psychologist and an Independent Living Donor Advocate to be sure you are mentally and emotionally ready to donate one of your kidneys.

Even if you wish to donate a kidney to your child, there are reasons you might be excluded as a donor. For example, taking certain medicines, excess alcohol and drug use, or not being an antibody or blood type match.

Therefore, it is important to look for other people who are willing to donate a kidney if you are not a good match. Start the evaluation process as soon as you begin thinking about a kidney transplant for your child, so that you can find out whether you are a match.

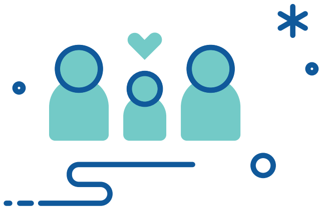
If you are told that you cannot donate a kidney to your child, try not to feel discouraged. Finding the best kidney match for your child supports the health and safety of both the donor and recipient. If you are not a match, talk to the transplant center about paired kidney exchange.

CONSIDER SCHOOLING OPTIONS

- Let your child's school and teachers know that your child will be absent for about 6 weeks as they recover from the surgery, and that avoiding infection will be a top priority.
- Talk to your child's school and teachers to see what options are available as your child recovers. This could include home schooling, online learning, home visits by a teacher, or virtual classrooms.

Once you know the available schooling options, talk to your child about what option they like best.

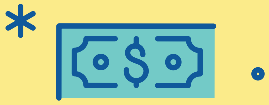
THE KIDNEY TRANSPLANT TEAM



Remember, you are not alone during the transplant process. Your child will have an entire transplant team of professionals to provide support and guidance throughout the process. The kidney transplant team will include:

- Pediatric nephrologist
- Pediatric transplant surgeon
- Social worker
- Child psychologist or psychiatrist
- Dietitian
- Nurses
- Pediatrician
- Financial coordinator

PAYING FOR A KIDNEY TRANSPLANT



Some organizations that can help you fundraise for a kidney transplant include:

Children's Organ Transplant Association:
www.cota.org

National Foundation for Transplants:
www.transplants.org

Help Hope Live:
www.helphopelive.org

If you have private insurance, call a representative before your child's transplant surgery to discuss what is and is not covered by your insurance plan. You should also talk to the transplant center's financial coordinator to ensure your financial status makes you eligible for transplant.

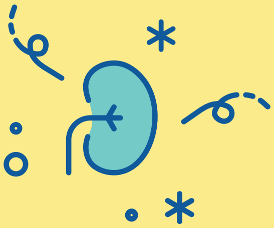
If your child has kidney failure, they may qualify for Medicare or the Children's Health Insurance Program (CHIP). If your child is on dialysis, talk to your child's social worker for more information.

Whether you have private or government insurance, there will be out-of-pocket costs. Keep in mind that your out-of-pocket costs will depend on the type of insurance you have, your geographic location, the hospital, and other factors.

If you need to take time off from work, know that the Family and Medical Leave Act might be able to protect your employment, but you may not get paid for the time off. The amount of time you need to take off will vary depending on how long it takes your child to heal in the hospital, whether you have an alternate caregiver once they return home, and how long your employer can give you leave.

It is a good idea to begin planning for transplant costs even before your child needs a transplant. Keep in mind that you will not just need funds for your child's surgery, but for your time off work, and for your recovery if you are the donor. Begin saving early, and make sure you keep a good credit score in case you need to take out a loan or use credit cards.

Helping your child manage their kidney transplant



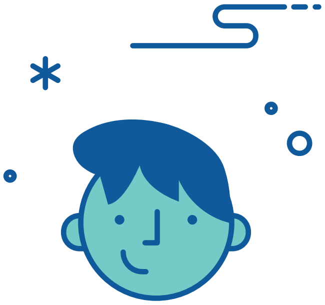
Receiving a new kidney is worthy of celebration! However, it is important to recognize that a transplant does not cure cystinosis and a new kidney comes with new responsibilities.

After the transplant surgery, your child will stay in the hospital for about a week to recover. Things that you can expect during this week include:

- Your child will need blood tests to check on the health of the new kidney, and medicines to help with pain.
- A nurse will help your child walk every day, including the day of the surgery. The nurse will also coach your child through deep breathing exercises needed for healing.
- Some children may need to go on dialysis for a short time after the transplant. This **does not** mean the kidney is bad or the transplant did not work. It may just mean the kidney needs a bit more time to start working.
- The transplant team members will talk with you, your child, and your family members about caring for your child. This includes the importance of taking their medicines exactly as instructed to keep their new kidney healthy.

During the recovery period, your doctor may recommend the following lifestyle changes:

- Limit physical activities until the surgery area is fully healed.
- Avoid participation in sports or rough play until the doctor says it is safe.
- Keep your child's surgery spot clean and dry to prevent infection.
- Keep your child away from people who are sick.
- Talk to your doctor about letting your child be around pets immediately after the surgery, since pets can carry germs.
- Help your child avoid grapefruits, pomelos, pomegranate, and drinks that contain these fruits because they can stop the immunosuppression medicines from working.



During this time, there are several things you can do to help your child have a smooth emotional transition:

- Encourage your child and their friends to stay in touch, whether through phone, social media, email, or even greeting cards. Staying connected will make returning to school easier.
- Keep reminding your child of all the things that they will be able to do once they recover.

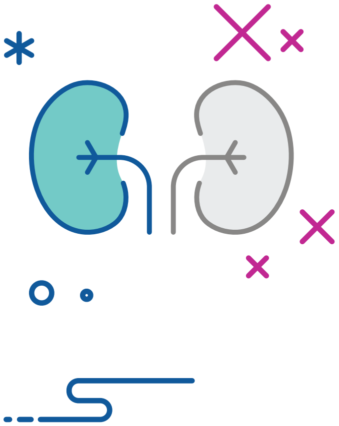
MEDICINES YOUR CHILD MUST TAKE

Immunosuppressants, also known as **anti-rejection medications**, are the medicines that your child will need to take every day for as long as they have their new kidney. Your child's immune system, which protects the body from infections, may not recognize the cells from the donated kidney as friendly. Immunosuppressant medicines slow down the immune system, so that it is less likely to attack and reject the new kidney.

Your child may need a second or even a third kidney transplant in their lifetime. In order to extend the life of each transplant, it is essential that your child takes their immunosuppressive medicines exactly as instructed. If your child does not take their immunosuppressant medicine, **or skips even one dose**, their body can start rejecting the new kidney.

Your child will still need to take medicines for cystinosis *after they have had a kidney transplant. Even with a new kidney, your child still has cystinosis. If they do not take their cystinosis medications as prescribed, their new kidney transplant could stop working. Although it can be challenging, you must ensure your child* ***always*** *takes their medicines for cystinosis* ***AND*** *immunosuppressants for their kidney transplant.*

KIDNEY TRANSPLANT REJECTION



It is natural for a parent whose child has a kidney transplant to fear transplant rejection. However, episodes of rejection are quite common, and do not always lead to complete rejection of the new kidney. **Acute rejection** will usually happen within the first 3-6 months of a kidney transplant. Many kidney transplant patients have some acute rejection episodes, which means their body shows signs that it is fighting the new kidney. Less than 1 in 20 transplant patients have an acute rejection episode that leads to complete failure of their new kidney.

Chronic rejection happens slowly over the years after a kidney transplant. Over time, your child's new kidney may stop working because their immune system will constantly fight it.

If your child has a rejection incident or episode, it **does not** mean their new kidney is not working or will be completely rejected. Changing the dosage of their immunosuppressant medicine can usually treat this problem. This is why it is so important for your child to take their immunosuppressant medicine exactly as prescribed.

Make sure your child attends all appointments with their transplant team. During these visits, the doctors will run blood tests and check your child's lab work to make sure rejection is not happening.

It is important to know the signs of rejection, so you can quickly recognize them and take action. The most common signs are:

- Fast heartbeat
- Sudden weight gain (fluid retention)
- Swelling in the body
- Urinating less or very little
- Pain or tenderness at surgery site
- Fever
- Symptoms similar to the flu

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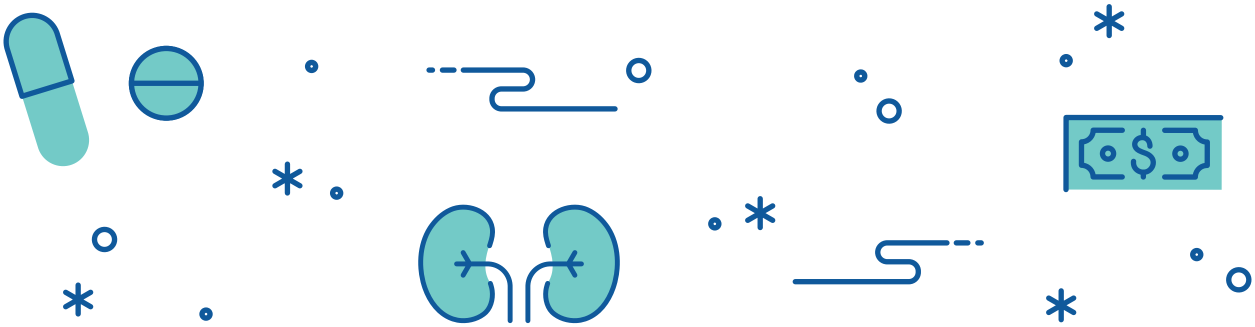
TAKING TRANSPLANT MEDICINES

Taking transplant medicines can be challenging for children and teens. As a parent, there are things you can do to make it easier. Some practical tips to help your child take their medicine properly include:

- Discuss *why* your child does not want to take their medicine, rather than trying to enforce compliance with arguments and punishments.
- Come up with solutions that will make taking medicine easier for your child.
- Explain what might happen if the medicine is not taken but try to avoid "scare tactics."
- Long-term issues might not feel as important to children and teens. Instead, focus on how the side effects of skipping medicine doses will disrupt your child's daily activities (e.g. going to school, participating in sports, learning to drive, hanging out with friends).

PAYING FOR MEDICINES

Coordinate with your doctor and insurance provider before the transplant surgery to ensure that necessary medicines will be covered by your insurance. If your child has Medicare, it will help pay for immunosuppressive drugs for a limited time after your child's transplant. Typical coverage is up to three years after the surgery.



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