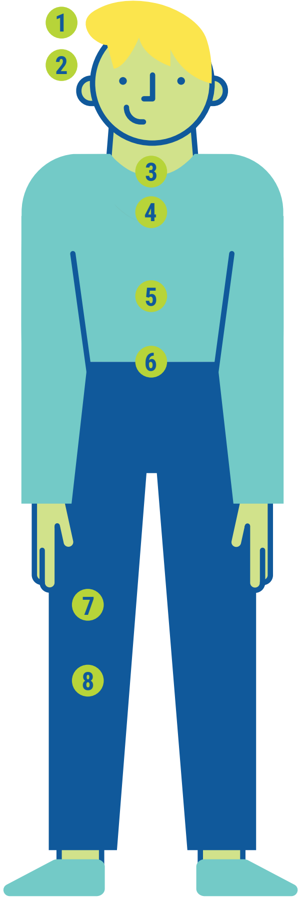
Cystinosis overview

You have probably been hearing the word "cystinosis" for several years now. As you may already know, cystinosis is an ultra-rare condition that is usually diagnosed in childhood. But what is cystinosis *really*?

WHAT IS CYSTINOSIS?

Cystinosis is a rare genetic condition. A genetic condition means that it is passed down through families.

Cystinosis leads to a buildup of cystine (a natural protein in your body) in a part of your body's cells called lysosomes. As cystine builds up, it forms crystals that cause the cells to die. Eventually, these dying cells can cause damage to organs, including your kidneys and liver.



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), 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 leads 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 will need to start **dialysis** or have a **kidney transplant** to live.

CYSTINOSIS AND YOUR KIDNEYS

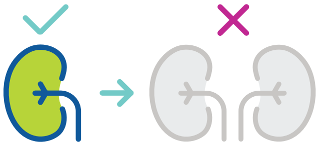
Cystinosis leads to permanent kidney damage and **kidney failure**. Kidney failure means the kidneys are unable to remove waste from your blood. The extra waste builds up in your body and makes you sick. When the kidneys lose the ability to remove waste from the blood, there are two treatment options:

1 Dialysis

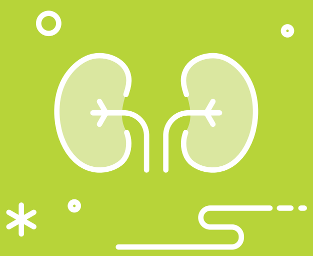
Start **dialysis**, a treatment that connects your body to a machine that cleans your blood

2 Kidney Transplant

Have a **kidney transplant**, a surgery that replaces your sick kidneys with a healthy kidney from someone else's body



With cystinosis, it is highly likely that you will need more than one kidney transplant during your life. You may get a kidney transplant when your kidneys are close to failure, as a prevention measure before you need to start dialysis. It is also possible you may start dialysis while you wait for a kidney transplant. Kidney transplant surgery is considered safe and is usually very successful. However, as with any surgery, there could be some risks involved.



***Kidney transplant** is considered the best option for people facing kidney failure because it can increase your chances of living a longer, healthier life. Dialysis is not a permanent solution to kidney failure because it only helps your kidneys do one of the kidneys' many jobs.*

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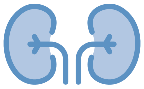

Preparing for kidney transplant

The kidneys are two bean-shaped organs, about the size of a fist. They are vital organs, meaning everyone needs at least one healthy kidney to survive. The kidneys have many functions, but their most important role is removing waste from the body by filtering your blood and eliminating waste through the urine.

Cystinosis can damage the kidneys over time, leading to kidney failure. Once the kidneys have failed, you must start dialysis or have a kidney transplant to live.

KIDNEY TRANSPLANT

Remember, a kidney transplant treats kidney failure. ***It does not cure cystinosis****. After getting a transplant, it is important to keep taking medicine for cystinosis to protect your new kidney.*

A kidney transplant is a surgery in which a healthy kidney is removed from a person's body and given to someone whose kidneys are not working. Transplants can come from **deceased donors** (who have recently passed away) or **living donors**. No matter where the new kidney comes from, doctors test it to make sure it is safe and healthy for your body.

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

The first step to getting a kidney transplant is working with your parents/caregivers to find a transplant center and schedule a transplant evaluation. Every transplant center requires different tests for a transplant evaluation, but all will include:

- Tests of physical health (e.g., blood tests)
- Psychosocial exam to make sure you are mentally and emotionally ready for transplant
- Conversations with your family about finances and health insurance to ensure that you can afford the surgery

If the transplant team decides you are ready for transplant, the next step will be for your transplant team to help you find a kidney match from a living or deceased donor.

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.

- Getting a deceased donor transplant requires being on a waitlist, which can be several months or years.
- As soon as a deceased donor kidney match becomes available for you, you will get a call to come to the hospital for surgery right away, usually in a matter of a few hours.
- To increase your chances of getting a transplant as soon as possible, you can get waitlisted at multiple transplant centers. If you are listed in multiple areas, you must be able to reach the 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 need to go through medical testing to ensure they are healthy and a good match.
- If you can find a kidney donor match, you can schedule the date of the transplant surgery.
- Living donor kidney transplants last an average of 15 years but can last for a longer or shorter amount of time.

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BENEFITS OF A TRANSPLANT

There are many things you may experience that you can look forward to after receiving a new kidney including:

- Being able to eat some of the things you may have not been able to eat before
- Eventually having to spend less time visiting the doctor
- Experiencing a growth spurt if you are still growing
- Having more energy to do the things you love
- Ending dialysis

Did you know? Transplants last longer if you take care of your health and take all your medicine; including your cystinosis medicine!

PREPARING FOR KIDNEY TRANSPLANT: YOU'VE GOT THIS!

Getting a kidney transplant is exciting, but it can also be a little scary. It is okay to feel nervous! Asking questions is one great way to feel prepared for the transplant surgery. You can use our question tool or write down your questions in a notebook to ask your transplant team. Examples of the type of questions you can ask include:

1. **Can I have medicine if I feel pain?**
2. **Can I still play sports after the surgery?**
3. **How long does the surgery last?**
4. **How soon can I go home?**
5. **Will I be fully asleep during the surgery?**
6. **Will my diet change after the surgery?**

PREPARING FOR SURGERY

It is okay to be a little nervous about the surgery. There are ways you can prepare for it, for example:

- Talk to someone who has gone through a transplant if you feel nervous or anxious. Your transplant team may be able to recommend someone you can talk to.
- Consider reaching out to the Cystinosis Research Network (Cystinosis.org) to connect with peers who have gone through something similar.
- Talk to your transplant team about how long your surgery will take and how long you will need to be in the hospital for recovery.
- Always let your care team know if you have questions or need anything before or after the surgery.

Things to bring to surgery:

- *Book/video game*
- *Laptop/tablet/phone*
- *Pillow/cozy blanket*
- ____________________
- ____________________
- ____________________
- ____________________
- ____________________

CONSIDER SCHOOLING OPTIONS

After the transplant surgery, you will need time to recover at home and temporarily avoid crowded places with lots of germs, like school.

- Talk to your parents about what schooling options are available and decide which option you prefer.
- You may be offered the options of home schooling, online learning, home visits by a teacher, or virtual classrooms.
- Let your school and teachers know that you will be absent for about 6 weeks as you recover from the surgery, and that avoiding infection will be a top priority.

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Life after kidney transplant

Receiving a new kidney is a major game-changer. Many people say they feel better quickly after surgery, if their new kidney starts working right away. **Although you may feel great, you still must take medicine to protect the health of your new kidney and to continue feeling good.**

IMMUNO-SUPPRESSANTS

Immunosuppressants, also called **anti-rejection medicines**, protect your new kidney from getting attacked by your immune system. Your immune system protects your body from outside invaders (such as germs and bacteria), and it may treat your new kidney as an outside invader, too. It is important to take immunosuppressant medicines **exactly as prescribed** to stop your immune system from attacking the new kidney.

Since it is likely you will need a second and maybe even a third kidney transplant during your lifetime, it is important to extend the life of your transplant by taking care of it. A big part of taking care of your new kidney is taking immunosuppressive medicines exactly as instructed. **If you skip even one dose of your medicine, your body can start rejecting your new kidney**.

AFTER SURGERY

For a period of time after the transplant surgery, you will need to take extra precautions to stay healthy.

- Avoid being around pets since they can carry germs.
- Avoid being near people who are sick.
- Avoid participating in sports until the doctor tells you it is safe.
- Keep your surgery spot clean and dry to prevent infection.
- Limit physical activities, like riding a bike, jumping on a trampoline or playing sports, until the surgery area is fully healed.

RECOVERY

When you arrive home after your transplant surgery, you may feel much better, but you will need to take it easy for a while. During the first three months after surgery your immune system is much weaker than normal, meaning you can get sick very easily. For this reason, your doctor will recommend you mostly stay at home during this time and avoid crowded places like school, restaurants and public transportation.

The time during the recovery period can feel isolating, so it is important to prepare yourself mentally by finding other ways to stay busy and keep in touch with your friends or classmates. Just remember, you have had a very important surgery that will change the course of your life.

Once your recovery is complete and your doctor says you can return to your regular lifestyle, there are many activities you can look forward to enjoying. Are any of these activities on your list?

- Driving
- Exercising
- Eating more of the things you like
- Going to school/college
- Hanging out with friends
- Traveling
- Swimming
- Getting a job
- Hiking
- Playing sports

TAKING CYSTINOSIS MEDICINE

Take your medications for cystinosis as prescribed at the same time every day. **Adherence is extremely important. Even after you have had a kidney transplant, you will still need to take medicines for cystinosis.** If you stop taking your cystinosis medicines, your new kidney transplant could stop working and other symptoms of cystinosis will get worse. Although it can be challenging, you must take your medicines for cystinosis for the rest of your life and take immunosuppressant medicines for as long as your kidney transplant lasts.

Have an honest talk with your parents and health care team if you are having trouble taking all your medicine, whether it is because of the side effects, a busy schedule or simply not wanting to. Your parents and doctors may have solutions that could make things easier.

MAKE TAKING MEDICINE EASIER

Try some of these techniques to remember to take medicines when you are on the go.

- Set alarms or reminders on your phone.
- Carry your medicine in a pill case or small sack. You can even use a 'disguised' pill case like an empty and clean mint container.
- If you do not like taking medicine in front of others at school, tell your teachers and school nurse that you want to take your medicine in complete privacy.
- Identify where you can take your medicine if you are going out. Keep extra medicine in your backpack or car in case you ever forget your medicine at home.

If you stop taking your medicine, ***you might feel just fine at first, but skipping medicine can have very bad effects,*** *such as:*

- *Your new kidney could get damaged and stop working.*
- *Your cystinosis could get worse and cause long-term damage – cystinosis affects more than just your kidneys.*
- *You might end up having to spend more time at the doctor's office.*
- *You might not have the energy to do the things you like doing or are looking forward to doing, like playing sports, going to school or learning to drive.*

Remember, taking care of your health today is the first step to living your best life. Prioritize taking your medicine and managing your health so you can continue doing all the other things you like and spending time with those you love.

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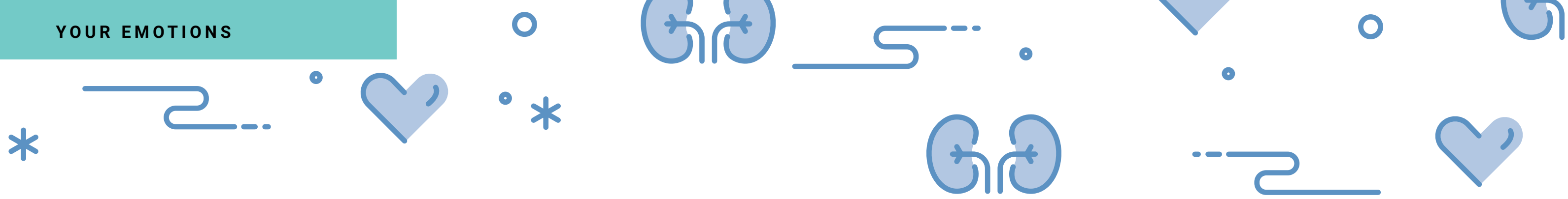
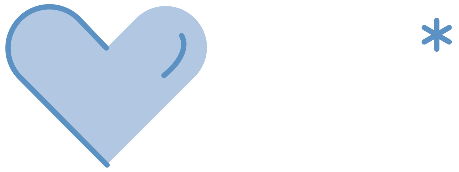
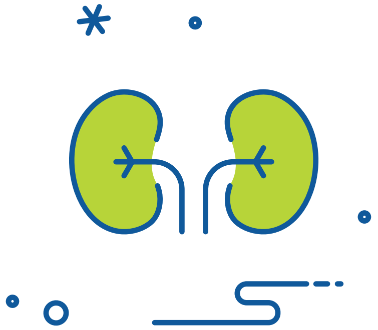


Emotional health after transplant



Getting a transplant is usually an exciting event, but because it is also a major life change, it is normal to feel a mix of emotions afterward. If you feel anxious, depressed, scared, stressed or unhappy, **please know that you are not alone**. Many people who receive transplants experience these feelings at first, for many different reasons.



After transplant some people experience:

- Changes in mood, which can sometimes be a side effect of the immunosuppressant medicine you are taking
- Feelings of stress or anxiety about your new lifestyle
- Guilt about getting a kidney from a living or deceased donor
- Family members who have emotional changes as they adjust to your new lifestyle

You do not have to deal with these feelings alone. Getting a kidney transplant is a **major** life change, and it is **okay** to feel stressed and anxious about events that change your life.

You have been through a lot after living with cystinosis for so many years. Sometimes it is not possible to deal with everything alone. Reach out to your family and friends when you need to talk.

There may be times when you do not want to talk to someone you know or would rather talk to someone new. Tell your parents or social worker that you would like to be connected to a counselor. Also, let your transplant team know about your emotional changes so they can help support you and adjust your medicines if needed.

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Your transplant center may host local support groups in your area. You can also join other trusted support groups to talk to people who have been in your shoes:

- NEPHKIDS
  Cyber-support group
- AMERICAN ASSOCIATION OF KIDNEY PATIENTS
  AAKP's Pediatric Kidney Pals – Summer camps
- CYSTINOSIS RESEARCH NETWORK
  Cystinosis Research Network Facebook – Open Page
  Cystinosis Research Network Facebook – Closed Group
  Cystinosis Research Network Twitter
  Cystinosis Research Network Instagram

If you believe you are experiencing depression, anxiety or having thoughts of self-harm, call:

NATIONAL SUICIDE PREVENTION LIFELINE:

1-800-273-8255

or

NATIONAL ALLIANCE ON MENTAL ILLNESS:

1-800-950-6264
Text NAMI to 741741

Remember, you are not alone!

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Living your best life with cystinosis

Everyone deserves to live their best life, including you! As a young person, you have your whole life ahead of you, so here are some ways to make the most of it:

Advocate for yourself — help others learn more about cystinosis and what they can do to help.

Find a hobby – start doing something you enjoy and find interesting, like photography, painting, cooking, biking, writing, dancing or playing an instrument.

Help others – give back to your community and those in need by volunteering.

Make new friends – surround yourself with supportive people who make you happy.

Stay active – find a sport or outdoor activity you enjoy and your doctor approves of.

Practice positivity – a positive attitude is key to success; try meditation, mindfulness or positive affirmations.

Set goals – whether you want to go to college or start working after high school graduation, set high goals for yourself.

Share your story – inspire and motivate others by sharing your journey.

Support causes you believe in – whether it is protecting animals, equal rights or the environment, take a stand.

Participate in advocacy – use your voice to support laws relating to cystinosis or kidney disease in general. Anyone affected by kidney disease can join the American Kidney Fund's Advocacy Network, where you can work to make a difference in the lives of others.

TALKING TO OTHERS ABOUT CYSTINOSIS

It can be frustrating or awkward having to explain cystinosis to people all the time. Remember: cystinosis does not define you as a person, and it is only one part of your life. If you feel comfortable sharing your diagnosis with others, then keep it up! If you need some time before opening up to others about your condition, then that is perfectly okay and very normal.

If you do decide to tell other people about cystinosis, here are some tips to help you have the conversation:

- Explain the basics of cystinosis. It will be easier for others to remember key points, rather than a lot of detailed information at once.
- Be honest about what you may not be able to do and tell others what you can do instead.
- Let others know that you take care of yourself by taking medication and seeing a doctor.
- Inform others that you do not want to be treated differently (if that is the case).
- If you do not want to talk about cystinosis anymore, steer the conversation in a different direction.
- You do not have to answer every question if you do not feel comfortable. Instead, you can direct others toward resources like the American Kidney Fund or Cystinosis Research Network to get more answers.

Let's take these tips and see what they would look like in a real-life situation.

Friday 5:00 PM

Hey this is my number.

Let's find a time to work on our history project soon.

Cool, sounds good

Soo...

Not to pry, just curious, saw you taking some pills a few weeks ago

Can I ask what for?

Actually, I'm not very comfortable talking about that

Oh! I'm sorry!

No, it's okay! Maybe some other time.

Does Sunday work with you to work on the project?

Yeah see you then!

Sunday 8:00 PM

I had fun playing on your Nintendo Switch after working on our project today! Thanks for having me over

I had fun too!

Come over again next weekend, I need a rematch

Lol done!

Hey so...Remember you asked about my pills? I think I'm comfortable telling you about it now

If you still want to know lol

I'm listening 👂

Basically, I was born with a rare disease called cystinosis. There is a protein called cystine that builds up in my cells. I take medicine so that it doesn't build up. Otherwise it can damage some of my organs

Wow I don't really know what to say...

Sorry you have to go through that

Are you okay?

Yeah I'm okay! I take meds and go to the doctor regularly

I can do most things anyone else can. I don't really like being treated differently

Ok got it! So how do you get cystinosis?

Umm it has to do with genetics

www.kidneyfund.org

www.cystinosis.org

Here, these websites have way more information than I do

Cool, thanks! I'll look into it

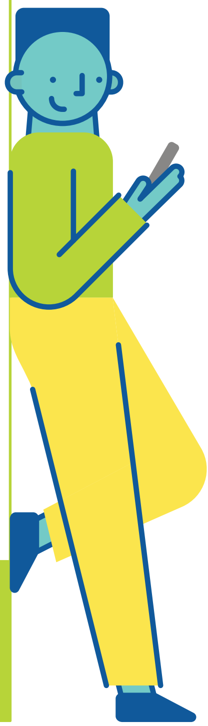
Anyways, enough about me

Do you really like pineapples on your pizza?

...Yes

I don't know if we can be friends LOL

LOL 🤦



You can practice role-playing different scenarios with someone you trust in order to feel more comfortable handling real-life situations.

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Additional resources

Cystinosis and Kidney Disease

American Kidney Fund (AKF)

www.kidneyfund.org/cystinosis

Phone: 866.300.2900

Cystinosis United

Horizon Therapeutics

www.cystinosisunited.com

Phone: 866.479.6742

Cystinosis Parent Handbook

Cystinosis Research Network (CRN)

www.cystinosis.org

Phone: 847.735.0471

Cystinosis Patient Assistance Program

National Organization for
Rare Disorders (NORD)

www.rarediseases.org

Phone: 855.201.5087

Organ Transplants: What Every Kid Needs to Know

United Network for Organ Sharing (UNOS)

www.unos.org

Phone: 888.894.6361

Pediatric Kidney Transplantation: A Guide for Patients and Families

American Society of Transplantation (AST)

www.myast.org

Phone: 856.439.9986

What Every Parent Needs to Know: A Guide When Your Child Needs a Transplant

Organ Procurement and Transplantation Network | UNOS

www.Optn.transplant.hrsa.gov

Phone: 877.464.4772

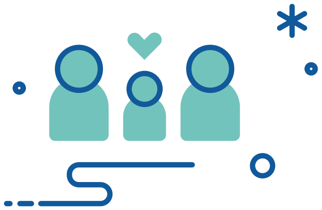
WORK WITH YOUR STUDENT’S PARENTS TO CREATE THE BEST PLAN OF ACTION.

To learn more about kidney transplant, visit the American Kidney Fund:

www.kidneyfund.org

To learn more about cystinosis, visit the Cystinosis Research Foundation:

www.cystinosisresearch.org



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Your student requires a kidney transplant:

What you can do as their educator

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. The buildup of cystine crystalizes and causes cell death, which leads to organ damage.

Over time, cystinosis can lead to permanent kidney damage and **kidney failure**. As kidney disease gets worse and the kidneys lose the ability to function, your student will need to start **dialysis** or have a **kidney transplant** to live.

A kidney transplant is considered the best treatment option for kidney failure and is life changing. However, it comes with some new changes in lifestyle.

After a transplant, it is vital to take a medicine called **immunosuppressants**. This medicine prevents the immune system from fighting the new kidney and causing a **rejection**. However, because immunosuppressants weaken the immune system, your student will be at a higher risk for **infections**.

After a transplant, your student will need to stay at home and avoid crowded places where they are more vulnerable and exposed to germs. This includes school.

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CONSIDERATIONS WHILE YOUR STUDENT RECOVERS AT HOME

School is a big part of a child's life and it is challenging for them to sit out on the experience. As an educator there are ways you can help your student have the best experience possible as they recover at home post-transplant.

Here are some things to start considering now:

- Work with your student and their parents to figure out an alternate schooling option (e.g. home schooling, online learning, home visits, or virtual classrooms).
- Discuss with the parents what other students will be told about the transplant and absence.
- Encourage other students to stay in touch (e.g. greeting cards, texts, e-mails, social media).

CONSIDERATIONS ONCE YOUR STUDENT RETURNS TO SCHOOL

Most successful transplants result in children being able to return home in 1-2 weeks. It takes about 6 weeks to fully heal and return to school, but it varies for each child. When your student returns to school, they will require additional considerations in order to protect and maintain their new kidney while continuing treatment for cystinosis.

Transitioning back to school can be difficult—academically, emotionally, and socially. As an educator, there are several ways you can help your student have a smooth transition back into the classroom, for example:

- Consider creating a 504 plan or an individualized education program (IEP) if one does not already exist for the student.
- Create a 'missed-days' plan for future sick days/doctor appointments.
- Create a plan for managing your student's medicine at school, including immuno-suppressant medication.
- Discuss how transplant affects your student's participation in physical activities and/or recess (e.g., avoid rough contact sports until doctor says it is safe).
- Discuss any new changes in diet (e.g., avoid buffet style food serving due to increased risk of germs).
- Discuss a plan for keeping your student protected from other sick students.

Other notes

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You are in charge:

Questions to ask your doctor

Getting a kidney transplant is exciting, overwhelming, and maybe even a bit scary. It is okay to feel a mix of emotions. A great way to feel prepared for the transplant surgery is by asking questions. You can take this Q&A sheet with you to the doctor and write down answers the doctor gives you.

How safe is the surgery?

How long does the surgery take?

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Will I be fully asleep during the surgery?

Can I have medicine if I feel pain?

Will my diet change after the surgery?

Can I still play sports after the surgery?

What happens if I don’t take my medicine after the surgery?

Can cystinosis harm my new kidney?

Write your own question: ____________________

Write your own question: ____________________