



DOCK8 Deficiency

A Guide for Adolescents

What Is DOCK8 Deficiency?



DOCK8 deficiency is a rare immune disorder named after the mutated gene responsible for the disease.

People with DOCK8 deficiency have lower-than-normal amounts and function of immune cells. As a result, people with DOCK8 deficiency often have frequent respiratory infections, ear and sinus infections, lung infections, allergies, asthma, eczema, and warts and other viral skin infections.

How to Use This Guide

Like most people newly diagnosed with DOCK8 deficiency, you probably have a lot of questions and thoughts running through your head. This guide will help you to better understand what it means to have DOCK8 deficiency and provide some helpful tips and encouragement for managing this condition.

Nobody wants to have DOCK8 deficiency, but having the right kind of help and information can make a big difference in how you deal with it.

You don't need to read this guide from beginning to end or in any specific order. Different parts of this guide will be helpful to you at different times. Read the sections that are important to you for now. Later, you can always go directly to other parts of the guide as needed.

This guide should not take the place of talking to your doctor or health care providers. If you are confused or worried about your health, you should always discuss this with your medical team. They are always there to support you.

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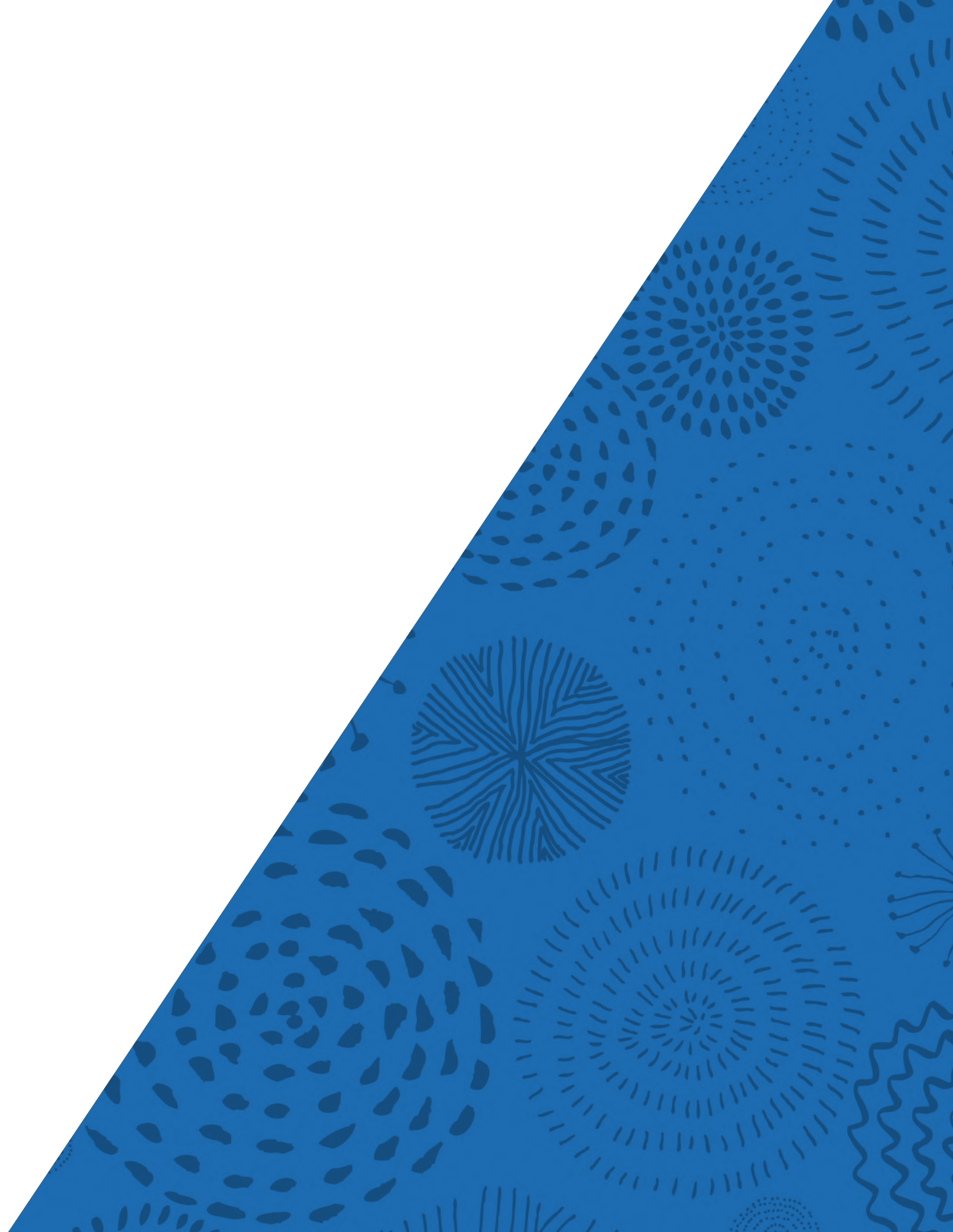
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Diagnosis



So You Have DOCK8 Deficiency –

Now What?

Every person reacts to a diagnosis of DOCK8 deficiency in their own way, but it is never easy. Whatever you are feeling right now is okay. You might feel scared, worried, confused, angry, or numb. You may have questions like, “Why me?”

There is no right or wrong way to deal with a diagnosis of DOCK8 deficiency. It is okay if it takes a little bit of time for you to come to terms with your diagnosis.

Now that you have a diagnosis, there will be many discussions of treatment options, and many decisions will need to be made. You may start to hear words that you may not understand. Just remember you are not alone. There is a lot of support available to help you get through this.



Do you feel like you’ve had a hard time focusing on anything since you were diagnosed? There are a few things that might help:

- Ways to treat DOCK8 deficiency are being studied extensively, which is helping many people with DOCK8 deficiency survive longer.
- While no one will feel exactly like you do, there are other people out there who share your feelings and experiences. There is even information online about how to connect to others and learn more about DOCK8 deficiency; see www.dock8.org.
- Having DOCK8 deficiency is not your fault. We know that DOCK8 deficiency is genetic, and nothing that you did, said, or thought caused you to have DOCK8 deficiency.
- Having the right information can help you better understand what your body is going through and what your treatment will be. If you are wondering about anything, please ask someone on your medical team. They can answer as many questions as you have.
- Remember there is no right or wrong way to feel. Each person handles this differently, and your feelings will change over time. Hiding your feelings is often not helpful, so try to be open about them as much as you can.

How do they know that I have DOCK8 deficiency?

Signs of DOCK8 deficiency include frequent ear and sinus infections, lung infections, allergies, asthma, eczema, and skin infections like shingles, molluscum, and warts. A type of white blood cells called lymphocytes do not work as well and are often low, a condition called lymphopenia, affecting the B and T lymphocytes, and low levels of antibodies to fight infections are also common. Because some signs of DOCK8 deficiency can look like other common illnesses, the diagnosis is made by genetic testing of the *DOCK8* gene. Only certain laboratories and research groups are able to perform this test.

How did I get DOCK8 deficiency?

DOCK8 deficiency is inherited. When each parent carries one copy of the abnormal DOCK8 gene and the child inherits both copies, then the child has DOCK8 deficiency and is not able to produce a functional DOCK8 protein. Of note, parents who are only carriers of the *DOCK8* gene often don't know they are carriers, because carriers do not have symptoms.

Can DOCK8 deficiency be managed?

Stem cell transplants, also called hematopoietic stem cell transplants, can cure many genetic immunodeficiency diseases. A stem cell transplant is the treatment of choice for DOCK8 deficiency and resolves most of a patient's clinical symptoms. Families must carefully consider the risks and benefits before pursuing stem cell transplant or other treatment options.

While a person with DOCK8 deficiency is being evaluated to see whether a stem cell transplant is appropriate, there are many supportive care therapies that are used based on a person's clinical condition. These therapies may include medications and other strategies for managing specific infections, allergies, and asthma.

Doctors may recommend antimicrobial drugs (like antibiotics and antivirals) to prevent infections. Doctors may also consider using blood immune globulin replacement therapy to strengthen the immune system. In some patients, a drug called interferon alpha has been used to control serious viral infections, such as widespread warts or herpes. These therapies will not cure DOCK8 but are used to prevent and treat complications of DOCK8 deficiency.

Treating the symptoms is a priority. Multidisciplinary care is preferred, with input from a team of immunologists, dermatologists, infectious disease specialists, and ear, nose, and throat specialists. Imaging using CT (computed tomography) scans of the chest and sinus are performed as necessary.

A curative treatment by hematopoietic cell transplantation is currently the standard of care for DOCK8 deficiency. Stem cell transplants are used to treat DOCK8 deficiency by preventing further infections and the development of cancer by replacing the impaired immune system with a healthy immune system. Although this treatment is still being researched, it is being performed in several areas of the world.

FAQs on Diagnosis

Getting the Right Information

Not knowing or understanding what is going on can be stressful. You have a right to know what is happening in your body, and having the right information is an important way to help yourself. It can also give you some control during a time when you cannot control very much.

Some people like to know everything, while others may want to know less. Whatever you choose is okay, and what you choose may change over time. But you should always ask questions.



Some questions you might want to ask are:

- What is DOCK8 deficiency, in language that I can understand?
- What part of my body does it affect?
- What will it do to me?
- What tests am I going to have to do?
- What kind of treatment will I have?
- Will the treatment be painful?
- Does the treatment have side effects?
- How long does the treatment take?
- Will I lose my hair?
- Will the treatment change how I look, feel, or act?
- How will we know if the treatment is working?
- What happens if I don't have treatment?
- What happens if the treatment doesn't work?
- Has anyone like me been treated before?
- Is there anything special I should be doing?
- When you talk to my parents, will you include me?
- What will happen after treatment?
- Will my DOCK8 deficiency come back?
- What about school and the rest of my life?
- Will my siblings be affected too?

A space to take notes or write down any other questions you may have for your health care team:

What Does It All Mean?



Sometimes when you are getting a lot of new information, it can be overwhelming and hard to understand. You will have lots of appointments with different members of the medical team, who might use a lot of medical terms when talking to you.

You may need to remind them to speak in plain English, not medical language — and that is okay!

Here are some tips on how to get the right information:

- Write your questions down in advance.
- Write the answers down.
- Ask people to repeat things as many times as you need to understand it.
- Ask the doctor to draw you a picture or use a model if it will help you.

Tips

Good Googling

- Make sure the site you are using is accurate and reliable.
- Check all information with your parents and health care providers.
- Keep in mind that many sites will tell you about the positives of a treatment but will often not talk about the negative side effects.
- Use information on the web as a starting point for discussions with your parents and health care team.



The Immune System

The system in your body that helps fight off infections is called the immune system. The immune system is made up of a network of cells, tissues, and organs that work together to protect the body.

White blood cells, also called leukocytes, are part of this immune system. Leukocytes are found in lots of places, including your spleen, an organ in your belly that filters blood and helps fight infections. Leukocytes also can be found in bone marrow, which is a thick, spongy jelly inside your bones.

Your lymphatic system is home to these germ-fighting cells, too. You've encountered your lymphatic system if you've ever had swollen "glands" on the sides of your neck, like when you have a sore throat. Although we call them "glands," they are actually lymph nodes, and they contain clusters of immune system cells. Normally, lymph nodes are small and round

and you don't notice them. But when they're swollen, it means your immune system is working hard.

Lymph nodes work like filters to remove germs that could make you sick. Lymph nodes and the tiny channels that connect them to each other contain lymph, a clear fluid with leukocytes in it.

You can help your immune systems by washing your hands regularly to prevent infections, eating nutritious foods, getting plenty of exercise, and getting enough sleep.

Treatment



Ear, Sinus, and Lung Infections

Most individuals experience these infections during childhood, but these infections often occur more frequently than expected or are chronic in people with DOCK8 deficiency. Pneumonias are often recurrent and may lead to bronchiectasis, a disease in which lung airways are enlarged and can result in a chronic cough. Prophylactic antibiotics and intravenous or subcutaneous immunoglobulin are sometimes used to help prevent these infections.

Allergies and Asthma

Food and environmental allergies are prevalent. Many individuals need to use inhalers or nebulizers to control asthma. Some may use an epinephrine autoinjector (e.g., EpiPen).

Eczema

The degree of eczema is variable, ranging from largely absent to severe and difficult to control. Eczema is treated with creams, steroid ointments, and sometimes wet wraps.

Recurrent Viral Infections

As children age, an increased number of viral infections of the skin are seen. Warts, molluscum, zoster (shingles), and herpes infections can be problematic.

Squamous Cell Carcinomas/Lymphomas

These cancers are seen at a higher frequency in children and adults with DOCK8 deficiency. Lymphomas are usually treated with chemotherapy. Squamous cell carcinomas may be treated with surgery if small (such as a small spot on the skin) or with combination of chemotherapy and/or radiation.

CBC Abnormalities

Complete blood counts (CBCs) may show low levels of lymphocytes, particularly as children get older. The antibody level of immunoglobulin M (IgM) levels may be low, and immunoglobulin E (IgE) levels are usually high. Antibody responses to prior vaccines may be absent, and some individuals are given intravenous immune globulin (IVIG) because of this. Lab tests are drawn as needed to monitor these abnormalities.

Stroke/Central Nervous System Events

Sometimes there can be inflammation of the blood vessels with DOCK8 deficiency. This can be caused by viruses (such as the virus that causes chickenpox or shingles), but sometimes it is not clear why the blood vessels are inflamed. If this happens in the brain's blood vessels, this can lead to stroke or neurologic changes. Sometimes there have been almost no symptoms of this. Sometimes other blood vessels, such as the aorta, can be involved. The medical team can evaluate for problems with the blood vessels through special MRI or CT scans.

Liver Disease

There is a higher incidence of liver disease with DOCK8 deficiency. This often affects the bile ducts and is usually caused by a parasite called *Cryptosporidium* that can be in the water and infect the intestines. There are some medicines that help a little, but a stem cell transplant usually is needed to clear this infection in patients with DOCK8 deficiency.

Common Symptoms and Treatment of DOCK8 Deficiency



Blood Testing/ Other Tests/ Procedures You May Undergo

Chest x-ray

A procedure done to check your lungs for pneumonia.

CT scan

A procedure done of the head, neck, sinus, chest, abdomen, and/or pelvis to look for signs of infections and sometimes to look for changes in blood vessels.

MRI scan

MRI and MRA (special MRI to look at blood vessels) are done of the brain and sometimes of the liver.

Dental visit

It is important to have healthy teeth when undergoing a transplant so you may undergo a teeth cleaning and checkup.

EKG (electrocardiogram)

A procedure done to look at your heart rhythm.

Sample “cultures”

Tests done from throat, urine, stool, and nose samples to check for infections.

Blood draws

Blood will be drawn to check your blood cell counts and run other tests, including genetic testing.

HLA typing

Human leukocyte antigen (HLA) typing is a way to tell how closely the tissues of one person match the tissues of another person. It is important in bone marrow and stem cell transplants to know how closely the transplant recipient matches the donor.

Bone marrow aspiration and biopsy:

A small surgical procedure where a small amount of bone marrow is collected from your hip in order to assess disease stage or the success of treatment.

Central line

A tube that is surgically placed in your chest and connects to a large vein so that you can have IV meds, blood transfusions, blood draws, and other procedures painlessly without having to be poked with needles several times a day.

Chemotherapy

About 1 week before getting your transplant, you will get chemotherapy to make space for the new bone marrow in your body.

Pulmonary function test

A procedure done to see how well your lungs work to make sure that your body can handle the medication you will receive.

Radiation

You may receive radiation therapy in addition to chemotherapy to help prepare your body for transplant. Powerful x-rays will be aimed at specific parts of your body to help get rid of bad cells and prepare your body for the new bone marrow you will receive. Radiation therapy is a lot like getting an x-ray.



What Is a Stem Cell Transplant?

- A stem cell transplant is a procedure in which healthy bone marrow cells (stem cells) are transplanted into a person who has diseased or damaged marrow.
- The healthy stem cells are often donated by someone whose marrow type closely matches your own. If family members are not a match, your doctors will look for an unrelated donor.
- When new cells are available for you, you receive them through an IV. They will travel through your bloodstream to the space in the center of your bones. There they will make their new home and begin produce healthy new blood cells.
- Usually, patients getting a stem cell transplant spend a minimum of one month in the hospital. How long you stay depends on how quickly your body and bone marrow recover from the transplant.

Side Effects and Risks of Transplant



Many of the problems that occur shortly after transplant come from having your bone marrow wiped out by medicines (chemotherapy) or radiation just before the transplant. Others may be side effects of the treatments themselves. It is important to know that your infections may temporarily get worse than they were before transplant and cause more complications until the new immune system recovers.

Your transplant team can help you cope with side effects.

Some can be prevented, and most can be treated to help you feel better. This is not a complete list, and you should tell your doctor or transplant team about any problems you have or changes you notice.

Nausea, Vomiting, and Diarrhea

These are common when undergoing the preparative regimen but can often be controlled with medications.

Skin Changes

A rash is a change in the skin that affects its color, appearance, texture, or smooth surface. Rashes can be caused by many factors, including drug reactions; allergies; bacterial, fungal, or viral infections; chemotherapy; or graft-versus-host disease (see below). Your skin can also have areas of discoloration (some areas of your skin may get very dark [hyperpigmented] or light [hypopigmented]), and you may continue to see changes in the skin for up to a year after transplant.

Nail Changes

Similar to the changes in the skin, you may also see discoloration or darkening of the nails. This is very common and typically will get better with time.

Hair Loss

Hair loss is an expected side effect from chemotherapy treatment. Scarves, wigs, or hats can be used until your hair grows back.

Mucositis

Undergoing chemotherapy can result in mouth and throat sores called mucositis. Mucositis can make it painful to eat or drink but can be treated with medications. This will usually improve when the new immune system starts to work.

Bleeding and Transfusions

After transplant, your risk for bleeding increases, because the conditioning treatment weakens your body's ability to make platelets. Platelets are the blood cells that help blood to clot. Platelet counts are low for at least several weeks after a transplant, during which you might notice easy bruising and bleeding, such as nosebleeds and bleeding gums. If your platelet count drops below a certain level, a platelet transfusion may be needed. It also takes time for your bone marrow to start making red blood cells, and you might need red blood cell transfusions from time to time as you recover.

Bladder Irritation (cystitis)

In the first few weeks after transplant, some patients may develop irritation of the bladder or have pain or muscle spasms when they are trying to urinate. This is called cystitis and is usually due to complications of a viral infection (BK virus) and chemotherapy. In most patients, the symptoms are mild and go away quickly, but in others, this can be more severe, and you may see blood when you urinate. If this happens, your doctors will talk to you about how to best manage this.

Infection

The first 6 weeks after transplant is a critical time when you can easily get serious **infections**. Bacterial infections are common during this time, but viral infections that were controlled by your immune system can become active again, and fungal infections can also occur.

Infections that cause only mild symptoms in people with normal immune systems can be quite dangerous for you. This is because right



after the transplant, you don't have many white blood cells that are working well, and they are the primary immune cells that fight off infections. You may be given antibiotics to try to prevent infections until your blood counts reach a certain level.

After engraftment, the risk of infection is lower, but it still can happen. It can take up to a year or longer after transplant for the immune system to work as well as it should. It can take even longer for patients with graft-versus-host disease (see below).

Post-transplant, you will be asked to avoid several things that pose an increased risk for infection. Since flowers and plants can carry bacteria and fungi, they're not allowed in your room. For the same reason, you may be told not to eat certain fresh fruits and vegetables and to make sure all your food is well-cooked and handled very carefully by you and family members. You may also be told to avoid contact with soil (no gardening), feces (stool, both human and animal), aquariums, reptiles, and some other pets. Your medical team will give you specific instructions.

Graft-Versus-Host Disease (GVHD)

Sometimes when a person receives bone marrow or stem cells from another person, the new cells react very strongly to your original cells. When this happens, you can show symptoms on your skin or in your gut, liver, or eyes.

GVHD does not mean the transplant is not working. These symptoms can be short- or long-term side effects, depending on how strong the reaction is. There are medicines to prevent GVHD from happening and others that can help if it does happen. Sometimes, the effects can be very severe and even life-threatening.

Acute Versus Chronic GVHD

Acute GVHD usually happens within the first 100 days after a transplant, though it is typically seen by day 60. The first signs are usually a rash, burning, and redness of the skin on the palms and soles. If acute GVHD does occur, it is most often mild, mainly affecting the skin. But sometimes it can be more serious or even life-threatening. Your doctors will give you

some treatments to try and prevent the onset of GVHD, but if it does occur, mild cases can often be treated with a steroid drug applied to the skin (topically) as an ointment, cream, or lotion or with other skin treatments. More serious cases of GVHD might need to be treated with a steroid drug taken as a pill or injected into a vein. If steroids aren't effective, other drugs that affect the immune system can be used. You can also start to see acute GVHD develop after you stop immunosuppressive medication, and you will be closely monitored.

Chronic GVHD can start anywhere from about 100 days to several years after the stem cell transplant. This may present differently than acute GVHD. Chronic GVHD is treated with medicines that suppress the immune system, much like those used for acute GVHD.

Isolation

If you get an infection that can be spread to others, you will be placed in isolation. When you are in isolation, you'll be in a private room, and staff and visitors may be required to wear gloves, gowns, and masks.

Neutropenia

Neutropenia occurs when your white blood count has low levels of neutrophils, which are the cells that fight bacteria and infections. Neutropenia usually occurs in the first 2 weeks after transplant and can make you very susceptible to infections.

Other Problems

The process of going through transplant affects every part of your body. This is a short list of possible complications, but your team will discuss with you the other risks that may affect you or other parts of your body (like the liver, lungs, or kidneys).

Although the risks of transplant can be severe and maybe even cause life-threatening complications, the goal is to safely get you through transplant so that you can be cured of DOCK8 deficiency.

Graft Failure

Graft failure occurs when the body does not accept the new stem cells (the graft). The stem cells that were given do not go into the bone marrow and multiply like they should.

Graft failure can lead to serious bleeding and/or infection. Graft failure is suspected in patients whose counts do not start going up within 3 to 4 weeks of a bone marrow or peripheral blood transplant or within 7 weeks of a cord blood transplant. Although it can be very upsetting to have this happen, these people can get treated with a second dose of stem cells if the cells are available.

Late Effects

The type of problems that can happen after a transplant depend on many factors, such as the type of transplant done, the pre-transplant chemo or radiation treatment used, the patient's overall health, the patient's age when the transplant was done, the length and degree of immune system suppression, and whether chronic GVHD is present and how bad it is. The problems can be caused by the conditioning treatment or by other drugs used during transplant. Possible long-term risks of transplant include:

- Organ damage
- Relapse (if cancer was present pre-transplant)
- Secondary (new) cancers
- Abnormal growth of lymph tissues
- Infertility (the inability to produce children)
- Hormone changes, such as changes in the thyroid or pituitary gland
- Cataracts (clouding of the lens of the eye, causing vision loss)

Although the risks of transplant can be severe and maybe even cause life-threatening complications, the goal is to safely get you through transplant so that you can be cured of DOCK8 deficiency.



A photograph of a man with short dark hair, smiling broadly, holding a golden retriever puppy. They are inside a car, with the window and interior visible. The background is bright and slightly blurred. A thick teal diagonal line runs from the top right towards the bottom left, separating the image from the text on the right.

Clinical Trials

Doctors, scientists, and researchers are always working on developing new treatments. This often takes many years, but an important part of this process is called a clinical trial.

Clinical trials are research studies that test new types of treatment to make sure they are possible, safe, and effective.

What are the benefits?

When you take part in a clinical trial, you will be monitored very closely. Any changes in your health will be detected and treated much more quickly as a result.

How does it work?

In order to be on a clinical trial, you will have regular tests and appointments to monitor how you are doing. If, at some point, the doctors find that the trial is not good for you because it is not working or because the side effects are too bad, they will take you off the trial. You can also withdraw from a trial whenever you want.

How do I decide whether a clinical trial is good for me?

- Talk about it with your parents.
- Ask any questions you have.
- Ask for written information or take notes so that you don't have to remember everything on your own.

Remember, no one will be upset with you if you choose not to participate.

Your Information





Who's Who?

Your Medical Team Members

Giving names to new faces...

During your treatment, you'll get to know a whole new team that will take care of you. It's not always easy to remember all the names and who does what. This chart will help you to organize all the information about who is looking after you. You may not have all these people on your team, or there may be more than one person doing the same job.

Medical Team

Profession / Specialty	What Do They Do?	Your Team Member's Name
Allergist	A physician specially trained to diagnose and treat allergies, asthma, and immunologic disorders.	
Care Coordinator	A nurse who is the main point of contact and helps patients and families communicate and deal with the different members of their medical team.	
Dermatologist	A physician specially trained to deal with diseases of skin, nails, and hair.	
Fertility Specialist	A physician specially trained to deal with fertility issues and discuss possibilities to reduce infertility risks due treatment.	
Gynecologist	A physician specially trained in dealing with women's health issues related to female reproductive organs..	
Hospital Teacher	A teacher who helps patients to continue their studies out of school. They help connect the student, the parent, and the school with the goal of remaining current with their academic program.	
Infectious Disease Specialist	A physician specially trained to diagnose and treat immunology and infectious diseases patients.	
Nurse Practitioner	A clinician with expertise in preventing, diagnosing, and treating health conditions.	
Occupational Therapist	A professional who helps individuals to be independent and improve their abilities to perform daily activities and self-care.	

Medical Team

Profession / Specialty	What Do They Do?	Your Team Member's Name
Oncologist/Transplant Physician	A physician specially trained to do a stem cell transplant who also treats patients with cancer.	
Ophthalmologist	A physician specially trained to deal with eye conditions.	
Otorhinolaryngologist (Ear, Nose, and Throat)	A physician specially trained to deal with diseases of the ear, nose, and throat.	
Pharmacist	A professional with knowledge about the drugs and medications that may be part of your treatment.	
Physiotherapist	A professional who helps with recovering physical movement, like walking, bending, and strengthening.	
Psychologist	A therapist who helps with emotional and intellectual well-being during diagnosis and treatment.	
Pulmonologist	A physician specially trained to diagnose and treat lung conditions and diseases.	
Registered Dietician	A professional who provides information to patients and their families about nutritional needs related to their disease and treatment.	
Registered Nurse	A nurse who provides regular care in the hospital or outpatient clinic.	
Social Worker	A trained professional who helps patients and their families adjust to life with disease and treatments. They can offer support and help with financial and other practical issues.	

When you are diagnosed with DOCK8 deficiency, it's likely that the hospital will be part of your life, either as a day hospital patient or as an inpatient for longer stays.

Being in the Hospital

Your first visit

The first time you come for an appointment, it's common that they ask you all sorts of questions — from what your date of birth is and your address or who lives with you to all your prior illnesses and treatments. It may seem annoying, but it's really important for the team to get to know you better — and have all these answers in your medical file for the following consults to come.

Being an inpatient

Sometimes for your treatment, you will need to stay at the hospital. It may be just for a day or two, if you just need some exams, or it can be for longer stays, when you come for chemo or a transplant.

Long stays in hospital can be really boring. Here are some tips from other young people that might help you have a better experience:

- Make your room look like your own space: You can bring some of your favorite stuff, pictures, and pillows — everything that may make you feel more comfortable.
- Invite your friends to come visit you! You can play games, watch TV, and catch up on everything that has happened since you were hospitalized. The hospital will have rules about the number of visitors, and it may also depend on how you're feeling, so it's good to check with your doctors before planning with your friends.
- You can also make a visitor book and take pictures with people who visit you. It can be fun and help you make nice memories for the future.

- Turn off your phone and create a "Do not disturb" sign for your door for those days when you don't feel like talking to anyone.
- Get to know the night nurses well. They will help you when you can't sleep.
- Try to be involved in decisions. Simple things like deciding when to sleep or take a bath or telling people what you don't like to eat can make all the difference.
- Bring your music, books, movies, or a laptop. Starting a journal or writing poems (or a novel) can be interesting as well.
- Find your release — arts and crafts, scrapbooking, puzzles, games, crosswords, or knitting.
- Make plans about things you'll do when you leave the hospital. It helps to have something to look forward to.
- Make new friends — you may find patients your age and try to meet up.
- Chat online with people dealing with similar experiences.
- Ask your friends or schoolteachers to bring things from school for you to do.
- Always ask for help when you need it.



Cultural differences

Whether you have to travel far for treatment or just around the corner, hospitals are a different world, and the people, the environment, or the language can be really different from what you are used to. You might find it difficult to talk to the doctors about symptoms and personal issues, especially when they don't have the same cultural background. Try to keep this in mind and help the health care team to understand cultural issues that might be part of how you are experiencing things during treatment. At NIH, there is a free language service available to assist you and your family during your stay if English is not your first language. Ask your care coordinator about it!

Going home from the hospital

After the transplant, you had to stay in the hospital for at least a month, with a lot of people helping you heal, taking care of your medications, and giving you special care. You have probably become used to the health care team, the hospital routine, and having people around who know and understand

your situation. It's normal to feel anxious about taking this big step. Remember: All the support will continue to be there for you when you leave the hospital.

Usually you will be requested to stay in the area around 100 days after your transplant. Have the clinic phone number in hand, in case you need it. During these first months, you'll have regular appointments at the outpatient clinic, when exams will be done to make sure recovery is going well. These appointments are also an opportunity to ask questions and get help to better adapt to this new phase of your treatment. It may also be good to arrange to continue to see the social worker or psychologist during this adaptation period to take care of the emotional part of this transition. Everyone will react in a different manner, and it may be nice to have someone to confide in who can help think about all of it with you.

Going home from the hospital can feel exciting, but sometimes it can feel a little bit scary too. And that's okay!



Notes

A space for your questions
and thoughts

Coping and Emotions



Staying Healthy



Staying healthy while living with DOCK8 deficiency can help you to better cope with treatment and help your body to recover.

Eating well and drinking liquids

A good diet is really important during treatment. Losing weight or not being well-hydrated can make it difficult for your body to deal with treatment side effects and cause you to not have enough energy to do the things you enjoy. Talk to your registered dietician to explore what foods and drinks can be good for you during treatment.

Physical activity

Even a small amount of exercise is good for your body and mind. It can help you to increase your energy, improve your sleep, manage your weight, keep your bones strong, and deal with all the different sort of emotions that may come up during treatment.

Being mentally active

It's important to keep yourself active, both physically and mentally. Try to keep up with schoolwork and do activities that challenge your mind. For example, you can play games, solve puzzles and logic challenges, read, and keep in touch with your friends.

Taking your medication

Taking meds is an important part of your health care. It can be harder to be compliant with your medications if you don't really understand why and how they can help your body fight the disease. Talk to your health care team about why you need your medications and about any other questions you might have.

Routine and schedules

Your routine will change a lot with treatment. You will have lots of doctor appointments, exams, and medications to remember. Having a weekly planner or a to-do list can help you to organize everything. It's also important to try to get back to a daily routine when possible. Maintaining your normal home life and having something to look forward to in your day can be helpful.

Neutropenic precautions

After receiving chemotherapy and radiation, your white blood cell count will be low. This means that germs that normally would not bother you can make you very sick. There are things you can do to protect yourself until they recover:

Avoid large crowds

Especially after stem cell transplant, avoiding large crowds and public spaces will be important to protect you from getting an infection and keep you healthy.

Wear a mask

A mask will protect you from other people's germs, especially colds and flu.

Wash your hands

A lot of germs are spread on hands, so washing your hands or using a waterless hand sanitizer will keep you safe by removing or killing those germs. Hand hygiene is important at the hospital and when you go home. Wash your hands when you leave or return to your room, after you use the toilet, before you eat, and before you touch your face.

http://intranet.cc.nih.gov/nursing/practicedocs/sop_pdf/SOP_Neutropenic_Patient.pdf

YOUR Emotions

Dealing with DOCK8 deficiency can bring on many different feelings, and these feelings may change over time. Some people even describe it as being on a roller coaster — having ups and downs that you have no control over.

The important thing to remember is that there is no right or wrong way to feel.

The most helpful thing you can do for yourself is share your feelings — even if this isn't what you are normally comfortable doing. Some people feel like if they ignore their feelings, they will go away, but they won't. Keeping too many feelings to yourself will often lead to meltdowns or angry outbursts — so find ways to express yourself.





Oftentimes, even naming a feeling can be hard. Here are some common feelings people experience:

Shock/Disbelief

This is often experienced when someone is first diagnosed. While some people may feel relief to know the reason for their illness, it isn't uncommon to also have trouble focusing on what you were being told. Some people cry; others feel numb or drained. These are all normal reactions to feeling shock.

Fear

No matter how tough you are, being diagnosed with DOCK8 deficiency can be scary. Sometimes that fear is based on things you know will happen, but other times that fear can be because of things you imagine might happen. Learning about things that you are unsure of can help calm these fears.

Uncertainty

While your medical team will tell you everything they can about your illness and treatment, it's impossible for anyone to give you a 100% guarantee that things will go exactly as planned. Not knowing what will happen or how to plan for the future can be overwhelming, but it is possible to live with uncertainty.

Anger/Irritability

Feeling angry is normal and understandable. Why did this happen? And why did it happen to you? It's very common to feel this way. Anger can be felt as rage or as irritability. Sometimes you might feel like you don't have the patience to deal with other people; remember, this is normal. It is okay to feel angry, but there are healthy ways to express your anger.

Sadness

There are many reasons to feel sad when you have DOCK8 deficiency. Sadness will often come and go, but if you notice that sadness doesn't go away and it starts to get in the way of enjoying things you like, or if you start to feel hopeless, then it is really important to talk to someone about these feelings. These may be signs of depression, and depression can be treated.

Loneliness

Having DOCK8 deficiency can make you feel really lonely, even when you are surrounded by people you love. You might feel like other people can't understand what you are going through, and you may notice that some people interact with you differently after you

are diagnosed. You may also feel lonely when spending long periods of time in the hospital, away from school and your normal activities. It may be helpful to join a support group to be connected to others with DOCK8 deficiency or similar experiences.

Withdrawal

There may be times when you feel down and want to keep to yourself, and this is okay. But if you notice that you are feeling this way most of the time and not wanting to talk to anyone, this may be because you are depressed. Make sure you are talking to someone about how you are feeling.

Frustration

It is easy to feel frustrated when you start to feel like you can't do anything for or by yourself anymore. Always being around other people and being told what to do can make you feel impatient or short-tempered. This is normal, and talking to someone can help you find ways to assert your independence and control where possible.

Guilt

Guilt can be felt for many reasons. You may feel guilt about the stress your illness has on your family. Remind yourself that having DOCK8 deficiency is not your fault.

Embarrassment

You may be embarrassed about how the DOCK8 deficiency experience has made you feel, or you may feel embarrassed by changes in your appearance that result from treatment. Other people feel embarrassed when they have to answer questions about their condition or treatment and they aren't sure how. Talking to someone about this can help you practice comebacks or responses to better prepare you for these situations.

Jealousy

You may feel jealous of others (e.g., brothers, sisters, friends) who don't have DOCK8 deficiency and don't have to go through what you are going through. It is very common and okay to feel this way.

Staying Positive

You might feel, and other people may encourage you to always stay, positive. While it is helpful to have an optimistic outlook, it is important to remember that you are not expected to feel this way all of the time. Having DOCK8 deficiency can bring on a full range of emotions. Remember that feeling sad, crying, and talking about your fears will not delay your recovery.

Coping Strategies

Coping strategies are thoughts and actions that help you deal with challenging things in your life. Coping isn't just putting up with a problem but finding a way to control it and make it feel better. Coping strategies can be really helpful in dealing with the stress and anxiety associated with your diagnosis.

Everyone deals with stress differently, so it's important to find techniques that work for you. Here are some things you can try:

Learn about your illness

Having a better understanding of your illness can help you feel more in control when you understand what is happening and know what to realistically expect. Let your medical team know how much or how little you want to know.

Ask questions

Don't be afraid of asking anyone anything. Write your questions down if no one is around when you think of them, and don't be afraid to ask the same question again if you still don't understand the answer.

Get involved

Become an active member of your care team. Find out about your tests, procedures, results, and plans for your care. Make decisions about things you can control, like when you want to be woken up and what you want to watch or listen to.

Look after yourself

Take control of your own medications and care. You will feel less dependent on others.

Talk

Expressing your feelings is very important. Find someone you trust and vent. This person can be a good friend or family member, or it can be a member of the medical team. Some people find it helpful to talk to someone who can keep their conversations private like a counselor/therapist.

Journal or blog

Writing is a way of expressing your feelings. You can choose whether to keep what you write private or to share it with others.

Laugh

Try to find ways to have fun and be silly.

Set goals

Goals help keep you focused on things you want to accomplish and help provide structure to your day. You can set short-term goals (like finishing reading a book) or long-term goals (like finishing schoolwork).

Get creative

Find ways to release your emotions through arts, crafts, music, writing, or photography.

Keep up with friends/activities

When possible, hang out or FaceTime with your friends. It helps to stay in the loop with life away from your illness.

Exercise

Exercise helps reduce stress. Whether it is walking or yoga, talk to your doctor about what exercise is okay for you.

Try something new

You will have a lot of downtime; try to use it to learn a new skill.

Get plenty of sleep

Dealing with emotions and stress is really difficult when you are tired.

Learn relaxation/meditation techniques

These can be helpful in dealing with feeling nervous, worried, or scared.

Maintain a routine

Try to maintain a normal routine even when going through treatment. Be sure not to sleep too much during the day so that you can maintain a normal sleep schedule at night.

Accept help

Needing help does not mean you are weak. Make a list of things that you need help with and give these tasks to people who offer to help.



There is no right or wrong way to grieve.

Common losses experienced:

- Missing out on going to school
- Changes in your appearance
- Losing contact with friends
- Missing out on being a kid
- Changes in how your body feels and functions
- Missing out on activities and events
- Things being canceled/postponed because of treatment
- Changes in family financial situation
- Changes in ability to plan for the future
- Loss of confidence or certainty

The way you feel about losses is called grief. Grief is a normal process that helps you adjust to loss. It is important to notice when you are experiencing grief. It can look different in different people.

Common signs of grief:

- Trouble sleeping or wanting to sleep all of the time
- Having sad dreams or nightmares
- Eating too much or too little
- Crying a lot or feeling extremely sad
- Avoiding things, people, or places that remind you of what you've lost
- Wanting to be alone or not wanting to talk to others

There is no right or wrong way to grieve. How long grief lasts is different for different people. Be patient with yourself and remember that getting support and expressing yourself are the best ways to deal with grief.

Grief is often associated with someone dying, but there are many things about an illness experience than can cause you to feel grief. Grief happens when you lose something or someone important to you.

Dealing With Grief

Dealing With Changes in Your Body

At some point of your treatment, you may look in the mirror and think, “Who is that person?”

Some treatment options can cause changes in your appearance, such as weight loss or weight gain, loss of hair, skin changes, discoloration of your nails, and scarring. At times, you may feel uncomfortable with your body. The good news is that your body and your feelings about your body will change over time.

It’s common for you to feel:

- **Self-conscious** of your physical changes
- **Shy** about being in public
- **Frustrated** that your body doesn’t work like it used to
- **Angry** that you have to go through this and that you don’t have control over what’s happening
- **Nervous** that you might be laughed at or stared at
- **Embarrassed** about the way you look
- **Grief** over the loss of your former self
- **Scared** that others will treat you differently
- **Worried** about feeling unattractive and not finding a partner in the future

How to cope:

- Have a makeover. Experiment with a new look, wig, or hat.
- Try to focus on other things that give you confidence, like something you are good at.
- Talk about it! People will understand.
- Treat your body with respect. Eat healthy foods, get enough sleep, and exercise if possible.
- Hang out with people who make you feel good.
- Catch up with friends individually so that you don’t have the pressure of seeing everyone at the same time.
- Remember that you are not your illness; even though your appearance may have changed, who you are has not.

Getting Support



When you're living with DOCK8 deficiency, it is important to find people who you feel comfortable talking with and getting support from.

It may feel hard to open up about your feelings, concerns, and experiences, but having someone to share them with can be really helpful.

Who can you get support from?

Some people will choose their **parents** to talk with, as they may be able to offer you the most support. Just remember that it will be important to make it clear how they can help and what you can do by yourself. ;-)

Others will rely on **brothers and sisters**. There's a special relationship and trust that may make you feel comfortable telling things to them that you would never share with anyone else.

You may have **relatives** or **teachers** that you really get along with and think would be a great source of support. That's totally fine!

Talking with your **friends** can be helpful as well. Sometimes you may have to initiate the conversation or ask for special help, and that can be good! You would be able to have a space to share your feelings, get support, and also have some fun together!

Another option is to talk to a **counselor**. A counselor will be able to listen to your thoughts and emotions in a different way than your family and friends. During your treatment, you will meet social workers and psychologists specifically trained to help people cope with living with DOCK8 deficiency, treatment, and life outside of the hospital. Don't be afraid to talk to them.

You don't need to choose just one person. Figure out what works best for you and take advantage of all possible sources of support.



Telling People

It's not always easy to talk about your disease with close family and friends.

Deciding who and what to tell can be a challenge! You might be concerned about people acting weird, or you may be feeling embarrassed talking about DOCK8 deficiency, but sometimes starting this conversation can be helpful.

Who should I tell?

There's no rule; it's up to you!

Some of your friends may notice that you are missing school days and wonder what happened. Some of them may have an idea of the reasons, but they might not know for sure how to act around you. Let them know what you're comfortable talking about and whether it's okay to ask questions. Sometimes an open conversation might seem scary, but it can clear the way and lead you to nice relationships.

If you think that your treatment may influence your schoolwork and classroom participation, telling teachers may be important to organize dates and workload. In the same way, if you work, telling your boss may be helpful as well. Together, you'll be able to make arrangements and better adjust schedules, considering your work responsibilities and treatment needs.

How can I tell people?

First of all, it's important to think about what you believe is important for people to know and be prepared to answer some questions. To be more comfortable answering questions others may ask, you can practice beforehand (by yourself or with friends, family, or someone from your health care team). Some people prefer to write a letter or an email or make a telephone call over talking in person. Some people prefer to talk in larger groups or to have one-on-one conversations. Explore your possibilities and choose what makes you more comfortable. Remember to make it clear whether it's okay if they share this information with others or if you prefer to keep it private.

What's Next



Post-Transplant, Back to “Normal”

So much happens while you are going through treatment that it is hard to think about what happens next. But eventually you will be done with treatment.

There’s nothing ordinary about returning to ordinary life after a transplant. No matter how much you’ve been looking forward to this time, it can still be weird, difficult, and scary.

So much has changed, even for the better, that it’s best to look at redefining life post- transplant. Now you will be starting a new chapter and defining a new “normal.” Although you won’t be undergoing long hospitalizations anymore, your medical follow-up care will still be part of your life.

Post-transplant, you will have to stay out of school for a minimum of 6 months, but this can go longer if you have not been weaned fully from high-dose immunosuppressive medications.



There’s nothing ordinary about returning to ordinary life after transplant. No matter how much you’ve been looking forward to this time, it can still be weird, difficult, and scary.



What Happens if Treatment Doesn’t Work

Medical advances in the study and treatment of DOCK8 deficiency have been very successful. But some individuals have treatment-related complications, such as GVHD, that persist. Although these complications can require additional treatments, most are not life-threatening.

If your transplant is not successful, you will often have the option of a second transplant.

Resources

niaid.nih.gov/diseases-conditions/dock8-deficiency

dock8.org





Notes

A space for additional
resources and info



National Institutes of Health
Turning Discovery Into Health

This guide was developed through a collaboration between the NIH Clinical Center, the National Cancer Institute and the National Institute of Allergy and Infectious Diseases.

About the National Institutes of Health (NIH): NIH, the nation's medical research agency, includes 27 Institutes and Centers and is a component of the U.S. Department of Health and Human Services. NIH is the primary federal agency conducting and supporting basic, clinical, and translational medical research, and is investigating the causes, treatments, and cures for both common and rare diseases. For more information about NIH and its programs, visit www.nih.gov.



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