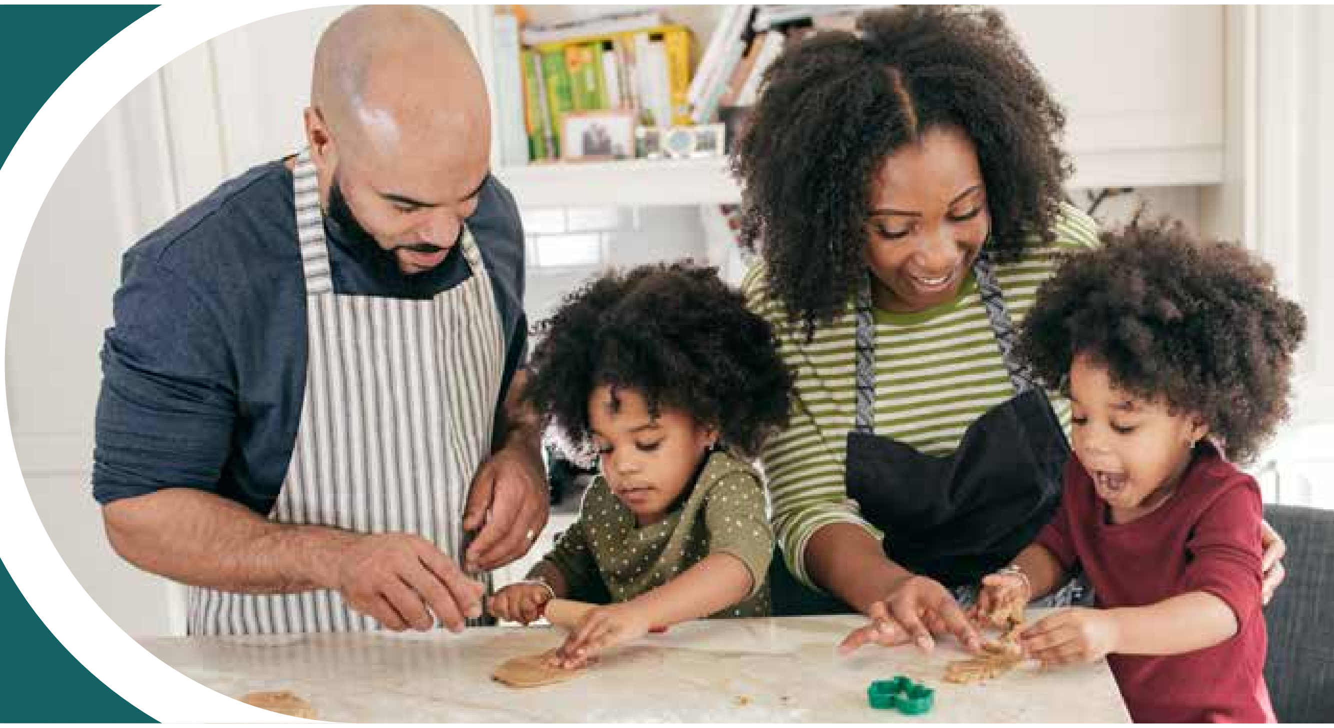
Information and support for people affected by cTTP



This resource is for people diagnosed with congenital thrombotic thrombocytopenic purpura (cTTP). Family and friends may also find it helpful. It tells you more about cTTP and helps you find information and support to better understand your diagnosis and guide your next steps.

A diagnosis of cTTP can be overwhelming. This resource has information on managing daily life, questions to ask your doctor or nurse, and activities that can help you cope with any emotional impact of your diagnosis. You can use the sections that feel most relevant to you, when you need them.

What is cTTP?

Congenital thrombotic thrombocytopenic purpura (cTTP) is an ultra-rare condition affecting how blood clots form in blood vessels throughout the body.

- **Congenital** means that cTTP is a form of TTP present from birth.
- **Thrombotic** means that blood clots form.
- **Thrombocytopenia** means that there is a low number of platelets, which help with clotting.
- **Purpura** means there are purple spots or bruises on the skin.

Blood clotting is an important response to an injury or cut to help stop bleeding. However, in cTTP, blood clots form when they are not supposed to, which can make it difficult for blood to flow to the rest of the body. This can cause people to feel weak and tired, and can lead to more serious health problems, such as a seizure or stroke. It can also mean that cells that help with clotting (platelets) reduce in number, meaning that people with cTTP may bruise easily or may not be able to stop bleeding after an injury.

Symptoms of cTTP

Symptoms are typically caused by blood clots and a low level of platelets. cTTP can be diagnosed in early childhood or experienced for the first time in adulthood or in pregnancy. Symptoms may include:

- feeling weak or tired
- a high temperature (fever)
- feeling confused or having speech changes
- shortness of breath or a fast heart rate
- having a lower amount of urine
- bruises or blotches on the skin
- headache
- bleeding (for example, from the nose or gums)
- the skin or whites of the eyes appearing yellow (jaundice)
- abdominal pain

You can print this page to share information with people who may not be familiar with cTTP.

Symptoms can come on suddenly and last for days or weeks. You may find it difficult to know whether your symptoms are related to cTTP or something else, like the flu. Common illnesses, like an infection, can also make any cTTP symptoms that you manage daily, such as tiredness, feel worse.

If you would like some support or reassurance, you can speak to your doctor or nurse.

Acute TTP events

Although there is currently no cure for cTTP, it can be managed with treatment. However, as cTTP is a long-term condition, there may be times when symptoms become worse and need medical attention – this is known as an acute TTP event.

While acute TTP events can have no apparent cause, there are some things that may increase their risk. For example, having an infection or other inflammatory condition, surgery, trauma, or excess alcohol.

Symptoms of an acute TTP event can affect everyone differently, but may lead to serious health problems if they are left untreated.

If you are concerned about your symptoms, feel like your symptoms have changed, or that they have become worse, you should feel able to seek medical attention. Your doctor or nurse can tell you more about acute TTP events, when and how to seek medical attention, and how to take precautions to live well with cTTP.

If you visit a healthcare professional who is not your usual cTTP doctor or nurse, it may help to:

- tell the doctor or nurse that you have cTTP, your history with cTTP, and that you are being treated
- tell them about any treatments you are taking and where your cTTP specialist team is located, if you have one.

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What causes cTTP?

A protein called ADAMTS13 helps blood to flow normally and clot when it needs to. It does this by acting like a pair of scissors to cut up another protein called Von Willebrand factor (VWF) into smaller pieces. This stops VWF from catching too many platelet cells, which means they cannot form blood clots.

People with cTTP are born with a gene that affects how ADAMTS13 is made and functions, meaning that it does not work properly. If ADAMTS13 does not work properly, VWF is not cut and there is more risk of blood clots forming.

The faulty *ADAMTS13* gene is passed down from a parent (inherited). To learn more, visit the **Genetic and Rare Diseases Information Center**.

You can watch a video explaining more about ADAMTS13 and cTTP.

Getting information and support

A diagnosis of cTTP can be overwhelming. It may feel like a relief to be diagnosed after having tests or treatment for your symptoms over a long period of time. You may also feel uncertain or anxious about what this means for your daily life. Finding information and support to better understand cTTP can help ease some of these feelings.

It is important to remember that you are not alone and there is support available. If you wish, you can speak to your doctor or nurse about any questions or worries you have. Talking to family and friends can help them understand how you are feeling and how best they can support you. We have information about talking to other people about cTTP on page 6.

Managing cTTP

Your doctor may give you medicines to manage any symptoms. There are also different treatments for cTTP, which all aim to get more ADAMTS13 into your blood. You may already have had some treatment before, or during, your diagnosis.

Preventative (prophylactic) treatment is given regularly to prevent any future acute TTP events and to stop symptoms from becoming worse.

On-demand (acute) treatment is given at the hospital during an acute TTP event until your symptoms are managed.

Your doctor and nurse will work with you to decide which treatment is best for you, and to explain what your treatment involves. You may have tests to check the level of ADAMTS13 or platelets in your blood to find out when treatment may be needed.

Speaking with your doctor or nurse

You may have questions about managing your symptoms, your treatment plan, and who you should contact for information and support. All of your questions are important. You can use the questions below as a starting point to make your own list.

Being open with your doctors and nurses about how cTTP affects you and your daily life can help them to give you the best care possible. If something is not clear, ask for it to be explained to you in a different way.

- Could anyone else in my family be affected by cTTP?
- Where can I find more information on cTTP and my treatment?
- What symptoms should I look out for?
- Who do I contact if my symptoms get worse or I think I am having an acute TTP event?
- Who can I contact if I have any questions?
- How will you know if my treatment is working?

Practical information and support

Living with a long-term condition can affect many aspects of your daily life. You may feel anxious or stressed about how cTTP impacts school, work, finances, or your social life. Or you may find it difficult to talk to other people about cTTP and how it affects you. There may be times when you need to manage cTTP alongside life changes like relocating, going to college, or deciding to travel. This section has information to help you cope in your daily life.

Leaving the hospital

After your diagnosis or recovery from an acute TTP event, you may feel tired and need time to rest.

It may be useful to ask your doctor or nurse for a contact number to use if you have any questions after leaving the hospital, such as symptoms to look out for when you are recovering at home.

Talking to people about cTTP

Living with a rare condition can sometimes feel isolating. You may feel that family, friends, colleagues, and teachers do not fully understand your experiences. Talking to people about cTTP can help lessen anxiety around continuing work or school, socializing, and asking for support.

Sharing this resource with people supporting you may help them to better understand cTTP. You could use it to help you start conversations about your experience and the type of support you may need.

You could also share it with the doctors or nurses looking after you if you are receiving care outside of your usual hospital or cTTP team.

This resource can be sent by email or message, or you can print a copy. You could also ask your cTTP doctor for a letter explaining your condition and the risk of acute TTP events.

Connecting with family and friends. If you are feeling low or anxious, it is helpful to have understanding from the people you trust.

We are all different, so it can help if you explain the type of support you need. For example, support with specific tasks, like grocery shopping, babysitting, or driving you to hospital appointments.

Talking to your employer or child's teacher. If you need time off for treatment or additional support to continue your studies, it may help to speak to your employer or your child's teacher about cTTP. For example, you could ask them about flexible hours or working from home.

Talking to people affected by cTTP

Support groups allow you to connect and share experiences with people in a similar situation to you. They can also be a valuable source of information. There are support groups for people from all around the world, and there may be such groups in your country. Family and friends may also find this support useful.

- cTTP is the inherited form of thrombotic thrombocytopenic purpura (TTP). Searching online for "TTP support" can help you connect with people who have cTTP as well as immune-mediated TTP (iTTP), and who can share similar experiences with you.
- Your doctor or nurse may be able to help you find support from other people affected by cTTP.
- You can ask your doctor or nurse about patient organizations that may provide information or more practical and financial support.

Find more information on talking about cTTP and asking for support in our resource "***Finding support and connecting with others affected by cTTP***".

Daily life with cTTP

There are things you can do to help manage daily life with cTTP, and stay as healthy and as active as possible. Consider speaking to your doctor or nurse about how cTTP affects you. They can give you more information and support to overcome specific challenges.

Follow a healthy lifestyle. Although acute TTP events can occur without reason, we know that eating a nutritious diet, getting enough sleep, and practicing good hygiene to prevent an infection may help you avoid an acute TTP event.

Do the activities you enjoy. Living with a blood disorder may mean you need to be more cautious about avoiding an injury. This can affect you if you play contact sports, or do any other activity that could put you at risk. However, you can work with your doctor or nurse to be able to continue enjoying your social life and activities that are important to you as much as possible.

Plan events or travel. Your doctor or nurse may be able to support you in planning future events or travel. They can help you take the right steps and create an action plan ahead of time in case of an emergency. You can talk to them about:

- having a test to check your ADAMTS13 or platelet levels to find out more about your risk of an acute TTP event
- how to contact a doctor if you have any symptoms when you are away from home
- which vaccinations you may need to travel abroad, and if you can have them
- which documents you need to be able to take any medicines with you
- how to arrange treatment elsewhere, if needed
- how to find travel insurance for your trip.

Planning day-to-day life

- Writing down a plan of your activities for the day can help you feel more in control and focus on what is most important to you.
- Breaking up tasks across different days can save you energy. For example, you could cook a few meals and put them in the freezer to eat on days when you do not have the energy to cook.
- Thinking ahead about some of the challenges or concerns you have can help you plan actions to overcome them. Being prepared can help you feel less stressed if a situation does arise. This is called "if/then" planning.

  "**If** I feel too tired at work, **then** I will speak to my manager about taking time off."

  "**If** I am worried about my symptoms getting worse, **then** I will contact my doctor."

  "**If** I feel overwhelmed by a busy day, **then** I will write down the tasks I need to do and ask family and friends for the help I need."

Find more information about managing treatment and any changes to daily life in our resource **"*Living with cTTP*"**.

Speaking with your doctor or nurse

- What can I do to manage my symptoms in day-to-day life?
- How can I prevent an acute TTP event?
- Are there any lifestyle changes I should make to manage my cTTP, or activities I should avoid?
- What do I need to arrange before traveling abroad?
- How can I find other people affected by cTTP to talk to?
- Are there any local support groups or organizations I can contact?
- Is there any financial support I can access?

Coping with cTTP

Whether you have been newly diagnosed with cTTP, were diagnosed some time ago, or care for someone with cTTP, it is important to take steps to look after yourself and manage any emotional impact the situation may have on you. This section has examples of ways you can take care of yourself.

Taking time to look after yourself

Managing a long-term condition can be overwhelming and may cause you to feel stressed at times. Stress affects our thoughts, emotions, and body—often without us realizing. These feelings can be more difficult to cope with if we feel tired or unwell.

Focusing on what you can control

Many causes of stress are beyond our control. Focusing on what **is** within your control can make a difference to how you cope. Rather than trying to change unhelpful thoughts, think about what you can do that would be most helpful for you. This may be taking time to rest, talking to family and friends, speaking to your doctor or nurse, or doing something you enjoy.

Making time for the things we value

When we feel overwhelmed, we can sometimes forget about the things we enjoy doing, or we cannot find the time to do them. Doing the things we value is important for managing our emotions and helping with stress. Simple activities, like going for a walk or planning to meet friends for a coffee or playdate, can help us feel more positive.

Activity: Some people find it helpful to close their eyes, take three deep breaths, and ask "What do I see myself doing that I enjoy?"

Practicing self-care

Sometimes, it can be helpful to take a few minutes to think about what you are feeling, without judging those feelings. Having compassion for what you and your family are experiencing can help you settle your emotions and take actions to look after yourself. The activity below may help you to practice self-care if there are times when you feel anxious or overwhelmed.

Meditation activity: Sit or lie down in a comfortable position and close your eyes. When you are ready, take three slow, deep breaths. Take a couple of minutes for each of the suggested steps below.

1. **Recognize.** Notice your surroundings, body, thoughts, and feelings. Say your feelings out loud or silently to yourself to acknowledge them.
2. **Allow.** Instead of trying to change them or think of your feelings as good or bad, view them as if you were watching a movie, letting them come and go just as they are. It is okay to feel the way you are feeling. You may want to say to yourself, "It is okay that I feel this way right now."
3. **Investigate.** Notice what words are going through your mind. What emotions are you feeling, and where in your body are they coming from?
4. **Nurture.** Be kind and comforting to your own experiences and feelings. Ask yourself what you could do that would help you right now, such as thinking about how you would talk to and comfort a friend if they were in a similar situation.

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