CONDITION

Guenther porphyria

Guenther porphyria is a rare inherited condition that causes extreme light sensitivity and damage to red blood cells.

WHAT IT IS
Rare inherited porphyria affecting skin and red blood cells

BODY SYSTEM
Skin, blood, eyes, bones and teeth

MAIN SYMPTOMS
Light-sensitive skin, blistering, red urine, anemia

WHAT CAUSES IT
Inherited deficiency of a porphyrin-making enzyme

HOW IT IS DIAGNOSED
Porphyrin testing and genetic testing

MAIN TREATMENT
Light protection, blood care and sometimes stem cell transplant

CAN IT BE CURED?
Sometimes - stem cell transplant can cure

IS IT AN EMERGENCY?
Sometimes - fainting, breathing trouble or spreading wound infection

LONG-TERM OUTLOOK
Lifelong photosensitivity; severity and complications differ widely

Overview

Guenther porphyria is a rare inherited condition that affects how the body makes heme. Heme is an important part of hemoglobin, the protein in red blood cells that carries oxygen.

The medical name is congenital erythropoietic porphyria , often shortened to CEP . It is also called Gunther disease or Günther disease.

In CEP, the body has too little activity from an enzyme called uroporphyrinogen III synthase. Enzymes are proteins that help chemical reactions happen. When this enzyme does not work properly, chemicals called type I porphyrins build up.

Porphyrins react with visible light. When they collect in the skin, exposure to sunlight and some artificial light can cause burning, blistering and tissue damage. Porphyrins also collect in red

blood cells, making the cells fragile and shortening their lifespan. This can cause hemolytic anemia , meaning anemia caused by red blood cells breaking down too soon.

Symptoms often begin in infancy or childhood, but milder forms may not be recognized until later. Severity differs greatly between people. CEP does not usually cause the sudden abdominal, nerve or mental health attacks associated with acute porphyrias.

Symptoms

Red, pink or dark urine may be the first sign. It can be noticed in a baby's diaper. The color comes from porphyrins rather than bleeding, although other causes of red urine still need to be ruled out.

The skin is extremely sensitive to light, especially on the hands, face and other exposed areas. Reactions can happen without the usual warmth or pain of sunburn.

Possible skin symptoms include:

Repeated injury can damage cartilage, fingers, ears, the nose and facial features. Modern light protection and treatment may reduce this damage, but established scarring may be permanent.

Porphyrins can collect in the teeth, causing reddish-brown discoloration called erythrodontia . The teeth may glow pink or red under ultraviolet light.

Damage to red blood cells can cause anemia. Symptoms may include tiredness, weakness, pale skin, shortness of breath, dizziness or a fast heartbeat. The spleen, an organ that filters blood, may become enlarged because it removes damaged blood cells. This can sometimes worsen anemia or lower platelet levels.

Eye problems can develop when the eyelids, the clear front surface of the eye or nearby tissues are repeatedly injured. Symptoms may include eye pain, redness, sensitivity to light, excessive tearing or changes in vision.

In the most severe cases, symptoms begin before birth and may cause severe anemia or fluid buildup in the fetus. Other people have much milder symptoms that appear later in life.

- Burning, redness, swelling and blisters after light exposure
- Fragile skin, slow-healing wounds and repeated infections
- Scarring or changes in skin color
- Increased hair growth, especially on the face and hands

Causes and risk factors

CEP is usually caused by harmful changes in both copies of the UROS gene. This gene provides instructions for making uroporphyrinogen III synthase.

The condition is generally inherited in an autosomal recessive pattern. This means a person receives one altered gene copy from each parent. Parents who each carry one altered copy usually do not have symptoms.

When both parents are carriers, each pregnancy has:

These chances apply separately to each pregnancy. They do not predict how severe the condition would be.

Rarely, a CEP-like condition can be linked to a change in another gene involved in red blood cell development. Genetic testing helps clarify the cause.

Family history is the main risk factor. CEP is not caused by diet, stress, personal hygiene or something the parents did during pregnancy. It is not contagious.

- A 1 in 4 chance that the child will have CEP
- A 1 in 2 chance that the child will be an unaffected carrier
- A 1 in 4 chance that the child will inherit neither altered copy

Diagnosis

Diagnosis starts with the pattern of symptoms, such as severe light sensitivity, blistering, anemia and red urine.

Laboratory testing measures porphyrins in urine, blood and stool. CEP usually produces a characteristic excess of type I porphyrins, particularly uroporphyrin I and coproporphyrin I. These results help distinguish it from other porphyrias and from other causes of light-sensitive skin.

Blood tests may check for anemia, red blood cell breakdown, bilirubin levels and changes in platelets or white blood cells. A blood film allows the cells to be examined under a microscope.

Genetic testing can identify changes in the UROS gene and confirm the diagnosis. It can also support carrier testing for adult relatives and testing during a future pregnancy when the family's gene changes are known.

Additional assessment may include examinations of the skin, eyes, teeth, liver and spleen. The care team may also check vitamin D and iron levels, particularly when light avoidance or

repeated transfusions are part of care.

Treatment and management

Treatment is planned around symptom severity, age and existing complications. Care often involves a porphyria or metabolic specialist working with blood, skin, eye and dental teams.

Strict protection from visible light is central to management. Ordinary sunscreen may not provide enough protection because porphyrins react to visible light as well as ultraviolet light.

Useful measures may include:

These measures can be demanding. A specialist service can help assess practical changes without requiring complete isolation from everyday life.

Anemia may need monitoring, folate support or red blood cell transfusions. Regular transfusions can reduce the bone marrow's production of porphyrin-filled red blood cells as well as treating anemia. Repeated transfusions can cause excess iron to build up, so iron levels and organs affected by iron may need monitoring. Treatment can remove excess iron when necessary.

Wounds and skin infections require early treatment. Eye care may include lubrication, light protection and treatment of eyelid or corneal damage. Dental care is important, but tooth discoloration itself is not harmful.

Removal of the spleen has been used in selected people with severe blood cell destruction. It does not correct the underlying enzyme problem and has important infection and blood-clot risks, so it is not routine treatment.

A hematopoietic stem cell transplant , also called a bone marrow transplant, can replace the blood-forming cells that produce excess porphyrins. It can cure the underlying blood-forming defect and prevent further light-related injury. However, transplantation has serious risks and does not reverse all established scarring or tissue loss. It is considered mainly for severe CEP after specialist assessment.

Gene-based treatments are being studied but are not established routine care.

- Protective clothing, gloves, hats and face coverings
- Light-filtering materials on windows at home, school, work and vehicles
- Checking indoor lights and medical equipment for unsafe wavelengths
- Planning outdoor activity around lower-light conditions
- Prompt covering and care of any blisters or wounds

Complications

Repeated light injury can lead to chronic wounds, infection, scarring and loss of skin, cartilage or parts of the fingers. Limited movement may develop when scarring affects joints or hands.

Chronic red blood cell breakdown may cause severe anemia, gallstones and enlargement of the spleen. Blood transfusions can lead to iron overload, which may damage the liver, heart or hormone-producing organs if not managed.

Eye complications can include scarring of the cornea, eyelid damage and reduced vision. Dental changes and facial scarring may affect eating, speech or self-confidence.

Avoiding sunlight can result in low vitamin D, which matters for bone strength. Restrictions around light may also affect education, work, relationships and mental health. Psychological and practical support can be an important part of care.

Prevention

CEP itself cannot usually be prevented because it is inherited.

Once the condition is present, careful light protection can help prevent blisters, wounds and further tissue damage. Monitoring and treating anemia can reduce complications from poor oxygen delivery and red blood cell breakdown.

If CEP runs in your family, genetic counselling can explain carrier testing and reproductive options. This provides information and support; it does not direct you toward a particular choice.

Living with it and what to expect

CEP is lifelong unless the underlying blood-forming problem is successfully treated with a stem cell transplant. Its course is not the same for everyone.

People with severe early symptoms may need extensive light protection, regular transfusions and help managing skin, eye or mobility problems. Milder forms may be controlled with light avoidance and periodic monitoring. Symptoms can also become more noticeable later if anemia worsens or light exposure increases.

Damage from past blistering, such as scars or altered fingers and facial features, may not improve even when porphyrin production is reduced. Effective treatment can still help prevent new injury.

Regular follow-up commonly focuses on blood counts, iron levels, spleen size, skin wounds, eye health, dental care and vitamin D. People receiving transfusions or who have had a transplant need additional long-term monitoring.

Practical planning can make school, work and travel more manageable. Adjustments may include safe lighting, filtered windows, flexible schedules and protective clothing. Patient organizations and rare-condition services may help with equipment, education and connecting with others who have porphyria.

When to get medical help

Arrange routine medical advice if you notice new blistering, worsening tiredness, increasing shortness of breath, changes in vision or wounds that are not healing. New red or dark urine also needs assessment if CEP has not already been confirmed, because urine color has several possible causes.

Contact your specialist team promptly for:

Seek emergency help for difficulty breathing at rest, chest pain, fainting, confusion or severe weakness. These can be signs of serious anemia or another urgent problem.

Emergency assessment is also appropriate for rapidly spreading skin redness, severe swelling, fever with marked illness, or difficulty waking. These may indicate a serious infection. A large burn or extensive blistering may need urgent wound and fluid care.

- Increasing weakness, dizziness or a fast heartbeat
- Fever, pus or worsening redness around a wound
- New eye pain, redness or blurred vision
- More frequent blistering despite usual light protection
- Increasing abdominal fullness that may suggest an enlarged spleen

Common questions

Is Guenther porphyria contagious?

No. It is an inherited condition caused by gene changes. It cannot spread through touch, blood, food, coughing or sexual contact.

Can I go outside if I have CEP?

Many people can go outside with carefully planned light protection. The amount and type of

protection needed are personal because visible light, not just ultraviolet light, can trigger injury. Protective clothing and specialist-approved window or face filters may be more important than ordinary sunscreen.

Does CEP cause acute porphyria attacks?

Usually not. CEP is mainly an erythropoietic porphyria, meaning it starts in blood-forming cells. It causes light-sensitive skin and red blood cell breakdown rather than the sudden abdominal pain and nerve symptoms typical of acute hepatic porphyrias.

Can Guenther porphyria be cured?

A stem cell transplant can cure the blood-forming defect in some people. Because transplantation carries major risks, it is generally considered for severe cases. It may prevent future damage but cannot reliably reverse established scars or tissue loss.

Will my children inherit it?

A person with the usual autosomal recessive form passes one altered UROS gene copy to every child. A child would generally develop CEP only if the other biological parent also passes on a harmful UROS change. Genetic testing and counselling can clarify the chance for a particular family.

Why do the urine and teeth turn red?

Type I porphyrins have a red color and can collect in urine and developing teeth. This causes red or dark urine and reddish-brown teeth. The color is a feature of porphyrin buildup rather than proof of blood in the urine.

Sources

- GeneReviews®. "Congenital Erythropoietic Porphyria." NCBI Bookshelf, University of Washington, Seattle
- National Organization for Rare Disorders. "Congenital Erythropoietic Porphyria." Rare Disease Database
- National Institutes of Health Genetic and Rare Diseases Information Center. "Congenital erythropoietic porphyria"
- Orphanet. "Congenital erythropoietic porphyria." ORPHA:79277
- International Porphyria Network. "Congenital erythropoietic porphyria (CEP)." Patient information

- British Porphyria Association. "Congenital erythropoietic porphyria (CEP)." Patient information