CONDITION

Familial jaundice

Familial jaundice is a group of inherited conditions that change how the body processes bilirubin.

WHAT IT IS
Inherited conditions that alter bilirubin handling

BODY SYSTEM
Liver and digestive system

MAIN SYMPTOMS
Yellow eyes, yellow skin, intermittent episodes, often no other symptoms

WHAT CAUSES IT
Inherited changes in bilirubin processing

HOW IT IS DIAGNOSED
Bilirubin blood tests, liver tests, sometimes genetic testing

MAIN TREATMENT
Monitoring; specialist treatment for severe forms

CAN IT BE CURED?
Sometimes - liver transplant corrects severe forms

IS IT AN EMERGENCY?
Sometimes - newborn jaundice or neurological symptoms need urgent care

LONG-TERM OUTLOOK
Usually normal; severe infant forms need lifelong specialist care

Overview

Familial jaundice is a group of inherited conditions affecting bilirubin, a yellow substance produced when old red blood cells break down. The liver normally processes bilirubin and passes it into bile so it can leave the body in stool.

When bilirubin builds up, the whites of the eyes and sometimes the skin turn yellow. This is jaundice.

The main inherited forms are:

Unconjugated bilirubin has not yet been processed by the liver. Conjugated bilirubin has been processed but is not transported or removed normally.

Gilbert, Dubin-Johnson, and Rotor syndromes usually do not damage the liver or shorten life.

Crigler-Najjar syndrome can be serious, particularly in newborns and infants.

Many conditions other than inherited disorders cause jaundice. New or unexplained jaundice always needs medical assessment.

- Gilbert syndrome , a common, usually harmless cause of occasional rises in unconjugated bilirubin
- Crigler-Najjar syndrome , a rare condition in which unconjugated bilirubin can rise to dangerous levels
- Dubin-Johnson syndrome , which causes conjugated bilirubin to build up
- Rotor syndrome , another inherited cause of raised conjugated bilirubin

Symptoms

Yellowing of the whites of the eyes is often the first sign. Skin yellowing may be harder to see in darker skin tones but may be visible on the palms or inside the mouth.

Gilbert syndrome often causes no symptoms other than mild, occasional yellowing. Episodes may become more noticeable during illness, dehydration, fasting, missed meals, strenuous exercise, stress, or lack of sleep.

Dubin-Johnson and Rotor syndromes can cause persistent or intermittent jaundice. Urine may look darker because conjugated bilirubin can pass into it, but most people otherwise feel well.

Crigler-Najjar syndrome usually appears in infancy and can cause much more severe jaundice. Very high unconjugated bilirubin may enter the brain and damage nerve cells. Warning signs in a baby include poor feeding, unusual sleepiness, a high-pitched cry, stiffness, arching of the body, or seizures.

Pale or clay-coloured stool, persistent itching, fever, significant abdominal pain, or unexplained weight loss are not typical of benign familial jaundice and need assessment for other liver or bile duct problems.

Causes and risk factors

Familial jaundice is caused by inherited changes in genes that process or transport bilirubin.

Gilbert and Crigler-Najjar syndromes involve the UGT1A1 gene. This gene helps the liver make an enzyme that converts unconjugated bilirubin into conjugated bilirubin. Enzyme activity is reduced in Gilbert syndrome and severely reduced or absent in Crigler-Najjar syndrome.

Dubin-Johnson syndrome usually involves the ABCC2 gene, which helps move conjugated bilirubin from liver cells into bile. Rotor syndrome involves the SLCO1B1 and SLCO1B3 genes, which help liver cells handle bilirubin-related substances.

Most of these conditions are inherited in an autosomal recessive pattern, meaning a person usually inherits an altered gene copy from each parent. Gilbert syndrome has more complex inheritance, and not everyone with a relevant gene change develops noticeable jaundice.

A close relative with a confirmed inherited bilirubin condition increases the likelihood of having it. Lifestyle does not cause familial jaundice.

Diagnosis

Diagnosis starts with blood tests. A total bilirubin test measures the overall amount, while a fractionated test separates conjugated from unconjugated bilirubin.

Other tests may rule out more common or urgent causes:

Gilbert syndrome is often diagnosed when repeated tests show mild unconjugated bilirubin elevation while other liver tests and blood counts remain normal. Genetic testing is not usually necessary, but it may help if the pattern is unclear or a medicine affected by UGT1A1 activity is being considered.

Conjugated bilirubin elevation needs careful assessment because hepatitis, blocked bile ducts, medicines, and other liver conditions can also cause it. Specialised urine tests can help distinguish Dubin-Johnson from Rotor syndrome. Genetic testing may confirm the diagnosis.

Crigler-Najjar syndrome is investigated urgently, especially in a jaundiced newborn. Doctors consider bilirubin levels, treatment response, family history, and genetic test results.

Family history alone does not confirm familial jaundice.

- Liver enzyme tests
- A full blood count
- Tests for increased red blood cell breakdown, or haemolysis
- Urine tests
- Ultrasound or other imaging if a liver or bile duct problem is possible

Treatment and management

Gilbert syndrome usually needs no treatment and does not normally damage the liver. Regular

meals, adequate fluids and sleep, and avoiding known triggers may reduce visible episodes.

Dubin-Johnson and Rotor syndromes also usually need no bilirubin-lowering treatment. Follow-up may confirm that the bilirubin pattern remains stable and that another liver problem has not developed.

Crigler-Najjar syndrome requires specialist care. Treatment may include intensive blue-light phototherapy, which changes bilirubin into forms the body can remove without normal liver processing. Treatment may be needed for long periods and can become less effective as a child grows.

Some forms respond to medicine that increases remaining UGT1A1 enzyme activity. An experienced clinical team must select and monitor this treatment.

A dangerous bilirubin rise requires urgent hospital care. Treatment may include exchange transfusion, in which some of a baby's blood is replaced to lower bilirubin quickly. Liver transplantation may be considered for severe Crigler-Najjar syndrome.

These conditions can affect how the body handles certain medicines. Before starting a new prescription, tell the prescriber about a confirmed diagnosis, especially Gilbert or Crigler-Najjar syndrome. This does not mean most medicines are unsafe.

Complications

Gilbert syndrome does not progress to liver failure. Some people may have a higher risk of pigment gallstones or side effects from certain medicines, but serious complications are uncommon.

Dubin-Johnson and Rotor syndromes generally do not cause scarring, cirrhosis, or loss of liver function.

The main complication of severe unconjugated hyperbilirubinaemia is bilirubin-related brain injury. Lasting neurological damage is called kernicterus and may cause movement problems, hearing loss, difficulty looking upward, or developmental disability. This risk is associated mainly with severe newborn jaundice and Crigler-Najjar syndrome, not typical Gilbert syndrome.

Someone with benign familial jaundice can still develop an unrelated liver, blood, or bile duct condition. A major change from the usual pattern needs assessment.

Prevention

Familial jaundice cannot be prevented because the gene changes are inherited.

People with Gilbert syndrome may reduce visible episodes by avoiding dehydration, long periods without food, and severe sleep loss. These measures manage triggers but do not cure the condition.

If a severe inherited bilirubin disorder runs in your family, genetic counselling can explain inheritance and testing options before or during pregnancy. Testing is a personal choice.

Prompt testing and treatment of significant newborn jaundice can help prevent brain injury, whatever its cause.

Living with it and what to expect

Most people with Gilbert syndrome live normally without ongoing treatment. Bilirubin levels can rise during illness or after missed meals and then return to their usual level. The condition does not normally worsen with age.

Dubin-Johnson and Rotor syndromes are also usually lifelong but stable. Keeping a record of the diagnosis and typical test results can help avoid repeated investigations.

Routine follow-up is not always needed for a stable, confirmed benign condition. It may still be appropriate after diagnosis, during pregnancy, before certain medicines, or if symptoms change.

Crigler-Najjar syndrome may require daily treatment, regular bilirubin tests, specialist appointments, and plans for illnesses that could raise bilirubin. Early recognition and continued treatment reduce the risk of permanent complications. Liver transplantation may change the long-term course for selected people.

When to get medical help

Arrange a medical assessment if you develop jaundice for the first time, even if relatives have Gilbert syndrome.

Seek prompt medical care if jaundice occurs with:

A jaundiced newborn should be assessed promptly. Treatment depends on the baby's age, bilirubin level, gestational age, and other health factors.

Get emergency help if a jaundiced baby is difficult to wake, feeds poorly, has a high-pitched cry, becomes unusually floppy or stiff, arches their body, or has a seizure.

Adults and older children need emergency help for jaundice with confusion, extreme

drowsiness, fainting, severe abdominal pain, uncontrolled bleeding, or vomiting blood.

- Dark urine and unusually pale stool
- Persistent itching
- Fever, repeated vomiting, or abdominal pain
- Unexplained weight loss or marked weakness
- A rapid increase in yellowing
- A clear change from your usual familial jaundice pattern

Common questions

Is familial jaundice the same as Gilbert syndrome?

Not always. Gilbert syndrome is the most common inherited cause of mild unconjugated bilirubin elevation, but familial jaundice also includes Crigler-Najjar, Dubin-Johnson, and Rotor syndromes.

Can familial jaundice damage my liver?

Gilbert, Dubin-Johnson, and Rotor syndromes do not usually damage the liver. Crigler-Najjar syndrome mainly threatens the brain through very high unconjugated bilirubin.

Why do my eyes look more yellow when I am ill or stressed?

In Gilbert syndrome, illness, dehydration, missed meals, heavy exercise, stress, and poor sleep can temporarily increase bilirubin. New or much stronger jaundice still needs assessment.

Can I pass familial jaundice to my children?

Possibly. The chance depends on the condition and the other parent's genes. Genetic counselling can explain your family's inheritance pattern.

Do I need to avoid alcohol or special foods?

No special diet removes familial jaundice. Regular meals and hydration may reduce Gilbert syndrome episodes. Alcohol can affect the liver for unrelated reasons, so general lower-risk guidance still applies.

Can jaundice come back after it has faded?

Yes. Bilirubin can rise and fall. Persistent, worsening, or different symptoms should not automatically be attributed to a known familial condition.

Sources

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