CONDITION

Transient familial hyperbilirubinemia

Transient familial hyperbilirubinemia is a rare newborn condition in which a familial factor temporarily slows the liver's removal of bilirubin.

WHAT IT IS
Temporary familial difficulty clearing bilirubin in newborns

BODY SYSTEM
Liver and blood

MAIN SYMPTOMS
Yellow skin, yellow eyes, sleepiness, poor feeding

WHAT CAUSES IT
Familial maternal factor slowing bilirubin processing

HOW IT IS DIAGNOSED
Fractionated bilirubin and tests excluding other newborn causes

MAIN TREATMENT
Phototherapy, with exchange transfusion if bilirubin becomes dangerously high

CAN IT BE CURED?
Yes - resolves after the newborn period

IS IT AN EMERGENCY?
Sometimes - marked sleepiness, poor feeding, or abnormal movements

LONG-TERM OUTLOOK
Usually full recovery when severe bilirubin toxicity is prevented

Overview

Transient familial hyperbilirubinemia is a rare condition affecting newborn babies. It causes bilirubin to build up in the blood during the first days of life. "Transient" means temporary, while "familial" means that the condition can occur more than once within a family.

Bilirubin is a yellow substance made when old red blood cells are broken down. A newborn's liver must change bilirubin into a form that can leave the body in stool and urine. In this condition, a factor associated with the mother temporarily slows that liver process.

The bilirubin involved is usually unconjugated bilirubin . This is bilirubin that has not yet been processed by the liver. Its buildup causes jaundice, which makes the skin and whites of the eyes look yellow.

Many newborns develop mild jaundice because their livers are still maturing. Transient familial hyperbilirubinemia is different because the bilirubin can rise quickly or reach a much higher level. Prompt measurement and treatment matter because very high unconjugated bilirubin can enter the brain and cause lasting injury.

The condition usually settles as the baby's liver becomes better able to process bilirubin. It does not normally cause an ongoing liver disorder.

Symptoms

Jaundice is the main sign. It may begin during the first few days after birth and become more noticeable as the bilirubin level rises.

Possible signs include:

Skin color alone cannot show how high the bilirubin is. Yellowing may also be harder to recognize in babies with darker skin. Looking at the whites of the eyes and gums can help, but a bilirubin measurement is needed to assess the level accurately.

A baby with a very high bilirubin level may become unusually difficult to wake, floppy, stiff, or irritable. A high-pitched cry, backward arching, abnormal movements, or seizures can be signs that bilirubin is affecting the brain. These symptoms need emergency assessment.

Dark urine or very pale, chalk-colored stool is not typical of this condition. These signs can point to a problem with bile flow or the liver and need prompt medical evaluation.

- Yellow coloring of the face, chest, abdomen, arms, or legs
- Yellowing of the whites of the eyes
- Sleepiness or difficulty waking for feeds
- Poor feeding or weaker sucking

Causes and risk factors

The exact biological cause is not fully understood. The condition is thought to involve a substance produced by the mother that temporarily interferes with the newborn liver enzyme responsible for processing bilirubin. This enzyme is called UGT1A1.

The familial pattern suggests that inherited factors may influence the production or effect of this substance. However, this is not the same as a clearly defined single-gene condition with a predictable inheritance pattern.

A previous baby in the family with severe unexplained newborn jaundice raises concern that another baby could be affected. The likelihood of recurrence is not known precisely.

Premature birth, bruising, blood-group incompatibility, infection, dehydration, and certain red blood cell conditions can also raise bilirubin. These do not cause transient familial hyperbilirubinemia, but they may make jaundice more severe or provide another explanation for it.

This condition is also different from breast milk jaundice. Breast milk jaundice is more common, usually appears later, and can continue for several weeks in an otherwise well baby. Feeding method alone does not establish which type of jaundice a baby has.

Diagnosis

There is no single widely available test that confirms transient familial hyperbilirubinemia. The diagnosis is based on the pattern of jaundice, the bilirubin results, the family history, and the exclusion of other causes.

A screening device placed on the skin may estimate the bilirubin level. If the result is high, rising, or close to a treatment threshold, a blood sample is used to measure it more accurately.

The blood test separates bilirubin into unconjugated and conjugated, or "direct," forms. This condition mainly causes an increase in unconjugated bilirubin. A raised direct bilirubin level suggests a different liver or bile-flow problem.

Other tests may include:

Doctors interpret the bilirubin according to the baby's age in hours, gestational age, and other risk factors. The same bilirubin result may require different action in babies of different ages or levels of maturity.

Genetic testing may be considered when jaundice is unusually severe, lasts longer than expected, or suggests an inherited condition such as Crigler-Najjar syndrome. It does not usually confirm transient familial hyperbilirubinemia itself.

- Blood groups for the baby and birth parent
- A direct antiglobulin, or Coombs, test for immune destruction of red blood cells
- A blood count and immature red blood cell count
- A blood smear to examine the shape of red blood cells
- Testing for glucose-6-phosphate dehydrogenase deficiency when appropriate
- Tests for infection, liver disease, or inherited bilirubin disorders when indicated

Treatment and management

Treatment aims to lower bilirubin before it can affect the brain. The care team bases decisions on repeated bilirubin measurements and the baby's individual risk factors rather than on visible yellowing alone.

Phototherapy is the main treatment. The baby is placed under a special blue light, usually wearing eye protection. The light changes bilirubin into forms that can leave the body without first being processed by the liver. This is medical phototherapy, not ordinary sunlight.

Feeding is also assessed. Regular, effective feeding supports hydration and helps the baby pass bilirubin in stool. Support may include help with positioning, attachment, expressing milk, or supplementary feeding when medically needed. Parents should not stop breastfeeding without advice from the baby's care team.

Bilirubin is checked again to see how well treatment is working. Monitoring may continue after phototherapy because the level can rise again.

If bilirubin becomes dangerously high or does not fall adequately with intensive phototherapy, an exchange transfusion may be needed. During this hospital procedure, small amounts of the baby's blood are gradually replaced with donor blood. This rapidly removes bilirubin and can also remove antibodies if immune blood-cell destruction is present.

Other treatments are used only when another cause is found. For example, treatment for infection or immune blood-group incompatibility addresses that separate problem rather than transient familial hyperbilirubinemia itself.

Complications

Most babies recover without lasting effects when high bilirubin is recognized and treated in time.

The main concern is bilirubin entering the brain. Early bilirubin-related brain dysfunction may cause marked sleepiness, poor feeding, unusual muscle tone, a high-pitched cry, or backward arching.

Severe, lasting bilirubin injury is called kernicterus . It can cause movement difficulties, hearing loss, problems with eye movement, and developmental disability. This complication is uncommon where jaundice is measured, followed closely, and treated promptly.

Phototherapy is generally well tolerated. Babies may need closer attention to temperature, hydration, skin condition, and feeding while receiving it. Exchange transfusion has more significant risks, so it is reserved for situations in which the expected benefit outweighs those

risks.

Prevention

The familial tendency behind this condition cannot currently be prevented.

Complications can often be prevented through early recognition and bilirubin monitoring. Before a future birth, tell the maternity and newborn teams if a previous baby had severe jaundice, needed intensive phototherapy, received an exchange transfusion, or developed bilirubin-related neurological problems.

This history may lead to earlier bilirubin measurement and closer follow-up after birth. Keep all arranged follow-up appointments, even if the baby appears well, because bilirubin may rise after discharge.

Adequate feeding helps reduce additional bilirubin buildup, but it does not remove the underlying familial factor. Do not use direct sunlight as a substitute for prescribed phototherapy. Sun exposure cannot be controlled accurately and can cause overheating or skin injury.

Living with it and what to expect

This is mainly a condition of the newborn period. During the first days or weeks, a baby may need repeated blood tests, hospital phototherapy, or follow-up visits soon after discharge. Feeding and wet or dirty diapers may also be monitored.

The bilirubin should fall as the temporary effect fades and the liver's processing ability matures. The exact timing is not identical for every baby. Persistent jaundice needs reassessment rather than being assumed to be part of this condition.

After the bilirubin has returned to a safe level, most babies do not need long-term liver treatment. The condition is not expected to return later in childhood or adulthood.

If a baby experienced bilirubin-related brain injury, longer-term follow-up may include hearing, movement, growth, and developmental assessments. The outlook then reflects the extent of that injury rather than an ongoing bilirubin problem.

Families may be offered additional planning before another pregnancy because another newborn could be affected. A clear record of the baby's bilirubin results, timing, treatment, and tests can help the next newborn team arrange early monitoring.

When to get medical help

Contact the newborn care team promptly if your baby looks yellow, especially if the yellow color is increasing, spreading toward the legs, or appearing during the first 24 hours after birth. A bilirubin test may be needed even when the baby otherwise seems well.

Seek urgent same-day medical care if your baby:

Call emergency services if your baby has a seizure, stops breathing, is unresponsive, becomes very floppy or stiff, has abnormal repetitive movements, or develops marked backward arching with a high-pitched cry.

Routine bilirubin rechecks are different from urgent care. Keep scheduled tests even if the yellow color appears to improve. Seek urgent help between appointments if concerning symptoms develop.

- Is much sleepier than usual or difficult to wake
- Is feeding poorly or has a weak suck
- Has fewer wet diapers than expected
- Develops dark urine or pale, chalk-colored stool
- Has worsening jaundice after discharge
- Still has jaundice beyond the follow-up period given by the care team

Common questions

Is transient familial hyperbilirubinemia the same as ordinary newborn jaundice?

No. Both cause unconjugated bilirubin to rise, but ordinary newborn jaundice is usually mild. Transient familial hyperbilirubinemia can cause a faster or more severe rise and may recur within a family.

Does this mean my baby has liver disease?

Usually not. The liver is temporarily less able to process bilirubin, but the condition does not normally cause permanent liver damage. Other liver problems must still be excluded if direct bilirubin is raised, stools are pale, or jaundice persists.

Can breastfeeding continue during treatment?

Often, yes. The priority is effective feeding and safe bilirubin levels. The care team may recommend expressed milk or temporary supplementation in some situations. Do not stop breastfeeding or change feeding plans without medical guidance.

How long does the jaundice last?

The condition usually improves during the newborn period as bilirubin processing matures. The timing depends on how high the bilirubin became, the baby's maturity, treatment response, and whether another cause is also present.

Will my baby have this condition for life?

No. The bilirubin-processing difficulty is temporary. Once it resolves, it is not expected to return later in life. Lasting problems are mainly a concern if extremely high bilirubin has already injured the brain.

Could another baby in the family have it?

Yes, recurrence is possible because the condition has a familial pattern. The exact chance cannot usually be calculated. Tell the maternity and newborn teams about the family history so bilirubin can be checked early.

Sources

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