

Nonalcoholic Fatty Liver Disease (NAFLD)

A **fatty liver** or **steatotic liver** is one that has too much fat built up. A fat content over 5% of the liver's weight is considered excessive and can lead to liver inflammation and damage. Here, we focus on fatty liver diseases that can happen in people who do not drink alcohol.

In **nonalcoholic fatty liver disease (NAFLD)** or metabolic dysfunction-associated steatotic liver disease (MASLD), the liver contains excess fat but no damage or scarring. This can be seen in overweight or obese patients who drink no or very little alcohol.

Nonalcoholic steatohepatitis (NASH) or metabolic-associated steatohepatitis (MASH) is a serious form of NAFLD. The excess fat has caused liver inflammation (hepatitis), tissue damage, and scarring (**fibrosis**). This condition can worsen into serious and permanent scarring as well as a hardening of the liver (**cirrhosis**), which can lead to fatal conditions such as liver failure and liver cancer.

Causes

- Overweight / Obesity
- Type 2 Diabetes
- High cholesterol / fat levels
- High blood pressure
- Side effects of certain medications
- Hypothyroidism (low thyroid hormones)
- Other hormonal conditions
- Other causes

Symptoms

Often, NAFLD has no symptoms, but some people experience **fatigue** and **pain** in the upper right area of the abdomen.

Symptoms of NASH and cirrhosis include (but are not limited to):

- Itchy skin (long-lasting)
- Swelling of the abdomen and legs due to fluid buildup
- Shortness of breath
- Weight loss
- Prominent blood vessels beneath the skin
- Tiredness or weakness
- Yellowing of the skin and eyes (jaundice)

Treatment

- **Weight loss** – starting with diet and exercise. If necessary, weight-loss surgery and/or medications may be considered.
- Individuals who have cirrhosis may need a **liver transplant**.

References / Resources

Clinical practice recommendations

- [Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome](#) – 2023
- [Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome](#) – 2023

Websites of medical institutions

- [Steatotic \(Fatty\) Liver Disease](#) – Cleveland Clinic
- [Nonalcoholic fatty liver disease](#) – Mayo Clinic
- [Nonalcoholic Fatty Liver Disease](#) – Johns Hopkins Medicine

Fatty liver and 22q11.2DS

Nonalcoholic fatty liver disease happens in a minority of adults with 22q11.2DS. This condition tends to happen in patients who are overweight or obese, and individuals with 22q11.2 are susceptible to becoming obese starting in their adolescence. It is therefore important to maintain a healthy weight to avoid obesity and its associated issues like fatty liver.

Diagnosis of fatty liver

Fatty liver often does not lead to obvious symptoms. It is often diagnosed when blood tests (which may be ordered for a different reason) indicate a liver problem.

Fatty liver may also be detected through abdominal ultrasound scanning. The [Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome](#) recommends abdominal ultrasound at the initial assessment after 22q11.2DS diagnosis and to check for conditions including fatty liver.

Newer imaging techniques can measure the stiffness of the liver, which is related to scarring.

If advanced liver disease is suspected, the doctor may use a needle to take a tiny piece of the liver (biopsy) and check for damage.