A patient guide to Gitelman Syndrome

Delivering the best in care

UHB is a no smoking Trust

To see all of our current patient information leaflets please visit www.uhb.nhs.uk/patient-information-leaflets.htm
Introduction

One of the main functions of the kidney is to control the level of magnesium, sodium, potassium and chloride in your body. Gitelman syndrome is a rare inherited disorder that causes the kidney to waste magnesium, sodium, potassium and chloride into your urine, instead of reabsorbing it back into your blood stream. It is estimated that Gitelman syndrome occurs 1 in 40,000 people and can affect both males and females of all ethnic backgrounds.

How Gitelman syndrome affects the kidneys

1. The kidney contains millions of tiny filters called nephrons that make urine and help control the level of salts and minerals within our body. A normal kidney will filter everything, except for blood and protein, into the tubule (small tube) of the nephron. The tubules then take back everything that the body needs and disposes of the things it does not.

2. The part of the filtering unit that should reabsorb sodium does not work in Gitelman syndrome. This results in sodium (salt) being wasted into the urine. This is why you may hear Gitelman syndrome called a ‘salt wasting’ condition.
Water absorbed from body

The high level of salt in the fluid passing through the nephron attracts more water from the body which causes you to pass more dilute (weaker) urine. Taking more water from the body can result in low blood pressure and will increase your thirst.

Excessive urination

Sodium

Potassium

This part of your kidney then works hard to try and take back some salt so that the levels of sodium in your blood do not become too low. However, to take back sodium the nephron has to swap it for potassium. This results in more potassium being wasted in the urine and low levels of potassium in the blood.
What symptoms may I experience?
The symptoms associated with Gitelman syndrome are usually caused by the levels of potassium and magnesium being too low. If you are regularly experiencing the following symptoms you should contact your health care professional to discuss your treatment:

- Tiredness and fatigue
- Muscle cramping
- Heart palpitations
- Joint pain
- Tingling sensation, particularly in the face and hands
- Muscle twitching

How is it diagnosed?
Gitelman Syndrome usually presents itself in adolescence or early adulthood. Diagnosis can be made through physical examination, and blood and urine tests.

Blood tests will show low levels of potassium and magnesium. The urine tests will show low levels of calcium in the urine.

Treatment for Gitelman Syndrome
The aim of treatment is to keep the levels of potassium, magnesium, sodium and chloride at a safe level. This can be done through the prescribing of supplements. The amount of supplements required are different for each patient, it is not unusual to be on a large dose so please do not be alarmed. Some of the medications you may come across are:

- **Slow–K (potassium chloride)**: A potassium chloride supplement that comes in tablets.
- **Kay–Cee–L (potassium chloride)**: A potassium chloride supplement that comes in a syrup formation. This medication is suitable for people who can not tolerate tablets.
• **Magnesium supplements**: These vary in efficiency and side effects. It may take a little while to find a magnesium tablet and dose that works for you and minimises side effects. Tablet types include:
  > Magnesium lactate
  > Magnesium citrate
  > Magnesium glycorophosphate
  > Magnesium oxide

• You may be prescribed additional medications such as potassium sparing diuretics, ACE inhibitors, and angiotensin receptor blockers. These medications help the body hold on to potassium, however they should be taken with caution as they can cause low blood pressure.

**Monitoring**

Blood level targets are lower for patients with Gitelman syndrome. The target for potassium levels is to be around 3.0mmols/L and the target for magnesium is to be greater than 0.6mmols/L.

Potassium levels between 2.5–3.0mmols/L are quite common and may require an increase in medication.

Contact your health care professional if you have potassium levels below <2.5mmols/L. This may require more immediate action.

**Blood tests**

You should have regular blood tests to check your levels are within the safe limits and to check whether you are taking the right amount of medication. Common blood tests you might hear mentioned are as follows:

• **Potassium**: Potassium is essential to regulate your heart beat. If your potassium levels fall too low it may increase the risk of an abnormal heartbeat.

• **Sodium**: Sodium plays a major role in keeping your bodies cells functioning normally. Sodium also helps regulate the amount of water in your body.
• **Bicarbonate**: Bicarbonate keeps the level of acid in your body at the right level.

• **Magnesium**: Magnesium is necessary for your muscles and nerves to function correctly. It also plays a role in keeping bones healthy.

**Investigations**

**24 hour tape**

If you are regularly experiencing palpitations your health care professional may request a 24 hour tape. This is a test carried out to monitor your heart during your daily activities over a 24 hour period. You will not be required to stay in hospital for this test.

**X–ray**

X–rays produce an image of solid tissue in your body, such as bone. This test may help identify the cause of joint pain.

**Diet**

You may notice that you naturally crave more savoury and salty foods. This is normal and we encourage you to have a high sodium salt diet. Patients with Gitelman syndrome are spared the risk factors associated with high salt intake.

There are certain types of food that are high in potassium. You should try to incorporate these foods into your diet to help maintain your potassium levels:

• Vegetables (such as potatoes, mushroom, spinach, tomato and pulses)

• Fruit (such as dried fruit, bananas, grapes, oranges, strawberries)

• Savoury snacks (such as potato snack and nuts)

• Drinks, such as coffee, hot chocolate, fruit juices, and cocoa

**Top tip**: Boiling vegetables can lower the potassium content of food. Try steaming or microwaving vegetables to retain the potassium.
You should avoid liquorice, as this causes the body to waste more potassium in the urine.

**How will Gitelman syndrome affect me?**

With treatment and monitoring the general outlook for patients with Gitelman syndrome is good. You may experience fatigue which at times may affect your daily activities.

Some patients may experience some joint pain. The symptoms can often be mistaken for gout (a type of arthritis). The pain is actually caused by a long standing magnesium shortage and can be resolved through anti-inflammatorily medication. Speak to your health care professional if you are experiencing any joint pain.

During episodes of diarrhoea and vomiting the levels of potassium and magnesium in your blood will fall even lower. During this time you are at an increased risk of dangerously low potassium and magnesium levels, it may be necessary to temporarily increase your medication during this time.

The low levels of potassium, and increased urine output, can make your blood less acidic than it should be (you may hear this referred to as alkalosis). This can contribute towards the symptoms associated with Gitelman syndrome. The acid levels in your blood will return to normal with appropriate treatment.

Patients should be aware of activities that may increase electrolyte depletion (reduction of minerals in your body). Patients should ensure to keep well hydrated when taking part in sports activities.

**Tips for clinic**

There is a specialist clinic for patients with Gitelman syndrome where you will be seen by a doctor or specialist nurse knowledgeable in Gitelman syndrome. Below are a few tips you can follow to make the most of your appointment:

- Write down any questions you may have because it can be easy to forget when you enter the busy clinic environment.
- Write down any symptoms you may have experienced since your...
last clinic appointment. Make sure you include when the symptoms started and what makes them worse or better.

- Never be afraid to double check and ask for things to be explained if you do not understand
- Ask for any words you do not understand to be written down and explained
- Write down a summary of any plans that have been made or any tests that have been booked
- Ask when your next clinic appointment will be

**Genetics**

Gitelman syndrome is inherited in an autosomal recessive pattern. This means that a person must inherit two changed copies of the same gene in order to have Gitelman. If a person inherits one changed gene and one normal gene, then that person will be a healthy carrier. If both parents are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child.

Each child of parents who both carry the Gitelman gene therefore has a 25% chance of inheriting a changed gene from both parents and being affected by Gitelman syndrome.

There is a 50% chance that the child will inherit just one copy of the Gitelman gene. This means they will be healthy carriers like their parents.

There is a 25% chance that the child will inherit both normal copies of the gene and will therefore not have Gitelman syndrome or be a carrier of Gitelman syndrome.

The chance remains the same in every pregnancy and is the same for boys and girls.
Patients with Gitelman syndrome are advised to discuss any plans for pregnancy with the specialist team.

**MyHealth@QEHB**

MyHealth@QEHB is a secure internet based programme that allows you to view blood results, clinic letters, and much more. Speak to your health care professional if you would like to find out more.

**Help with health care costs**

Information can be found in the leaflet HC11, “Are you entitled to help with health costs?” available from the cashiers office in the Queen Elizabeth Hospital, Birmingham.

Or visit: www.nhs.uk/healthcosts
Other names for Gitelman syndrome
You may hear Gitelman syndrome referred to slightly differently. Other names include:
• Familial hypokalaemia–hypomagnesaemia
• Hypokalaemic alkalosis
• Salt wasting tubulopathy

Further information
Further information about Gitelman syndrome is available at:
www.rarerenal.org
www.barttersite.org
www.gitelmansyndrome.co.uk

Useful Contacts
Renal Metabolic Disease Nurse Specialist
Tel: 07810 654 864
Office: 0121 727 2518

Renal Outpatients
Nurses: 0121 371 5635 or 0121 371 5633
Appointments: 0121 371 4447 or 0121 371 4446
The Trust provides free monthly health talks on a variety of medical conditions and treatments. For more information visit www.uhb.nhs.uk/health-talks.htm