

for you to have a specialist consultation, which will allow plenty of time for any questions you may have.

We will review your blood results and medications to ensure you continue to receive the appropriate treatment. We also aim to provide support for you and your family, and inform you of any other support that is available, such as charities and support groups.

The frequency of these appointments may change and will be discussed with you in the clinic.

What other names are used for Gitelman's Syndrome?

- Familial hypokalemia-hypomagnesemia

Where can I find more information?

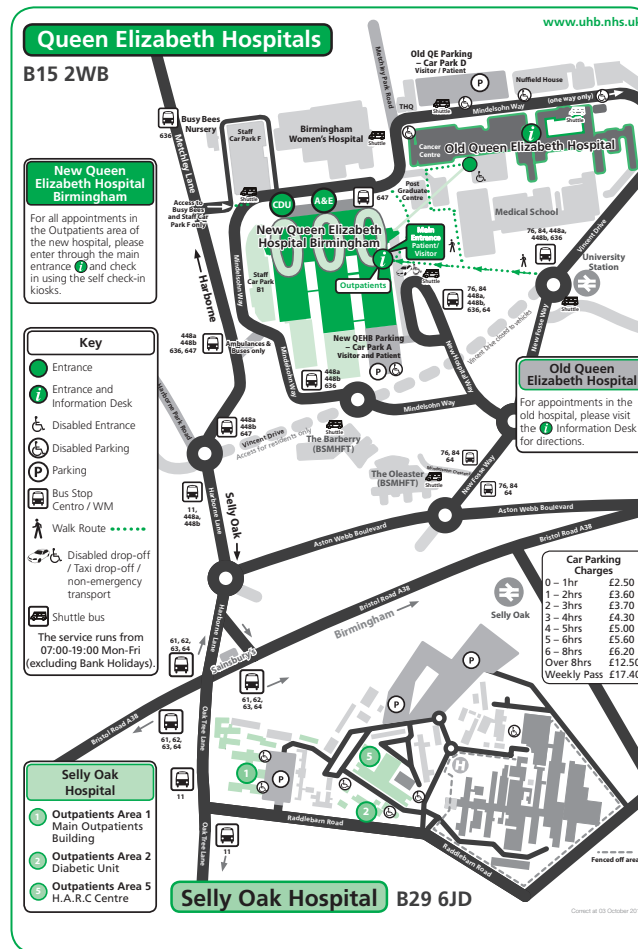
- www.barttersite.org
- www.raredisease.org.uk
- www.geneticalliance.org.uk

Useful contacts

Steven Wise
(Renal Metabolic Clinical Nurse Specialist)
07810654864

Renal.metabolic@uhb.nhs.uk

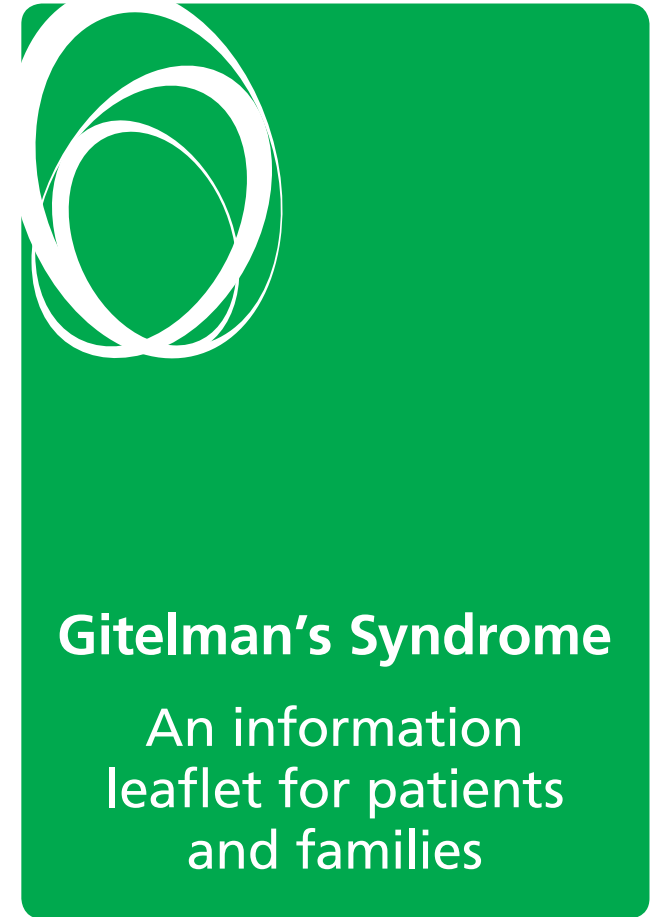
You can use the above contact details if you require further information or have any questions regarding Gitelman's Syndrome.



Queen Elizabeth Hospital Birmingham
Mindelsohn Way
Edgbaston, Birmingham, B15 2WB
Telephone: 0121 627 2000

(Old) Queen Elizabeth Hospital
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Queen Elizabeth Hospital Birmingham **NHS**
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www.uhb.nhs.uk/patient-information-leaflets.htm

What is Gitelman's Syndrome?

One of the main functions of the kidney is to control the level of magnesium, sodium, potassium and chloride in your body. Gitelman's 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's Syndrome occurs in one in 40,000 people and can affect both males and females of all ethnic backgrounds.

What are the symptoms?

The symptoms associated with Gitelman's Syndrome occur due to the low levels of magnesium, potassium, sodium and chloride within your body. The symptoms of Gitelman's Syndrome include:

- Tiredness and fatigue
- Muscle cramping
- Salt cravings
- Excessive thirst
- Excessive urination
- Heart palpitations

How is it diagnosed?

Gitelman's Syndrome is not usually diagnosed until adolescence or early adulthood. Diagnosis is based on physical examination, symptoms and the results of blood and urine tests. Often Gitelman's is only diagnosed when adolescents or adults see their doctor for other reasons and the

results of routine blood tests show low levels of potassium within the blood.

What is the treatment?

Treatment of Gitelman's Syndrome is focused on keeping the level of potassium, sodium, magnesium and chloride within your blood at a normal level. This can be done through magnesium and potassium supplements, and a diet rich in potassium and sodium.

Additional medicines such as Amiloride, Spirinolactone and Triamterene may help reduce the loss of potassium.

What effect will it have on my health?

Gitelman's Syndrome can make you feel fatigued which may affect you in some of your daily activities. However, in general, you can expect normal growth and intellectual development. The chance of developing osteoporosis (fragile bones) is severely reduced as Gitelman's syndrome causes your body to hold on to calcium and progression into renal failure is extremely rare.

What about my family?

Gitelman's 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's. If a person inherits one changed gene and one normal gene, then that person will be a healthy carrier.

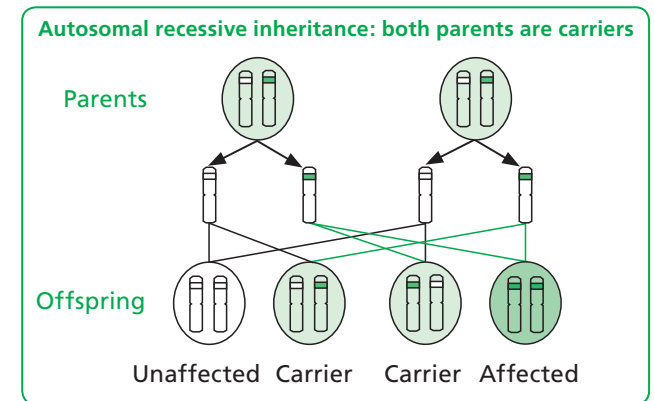
If both parents are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child.

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

There is a 50% chance that the child will inherit just one copy of the Gitelman's 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's Syndrome or be a carrier of Gitelman's Syndrome.

The chance remains the same in every pregnancy and is the same for boys and girls.



What happens now?

You will be seen in the Renal Metabolic Clinic. We aim to provide an opportunity