

Renal Genetics and Tubular Disorders Clinic



Information about Gitelman's Syndrome

What is Gitelman's Syndrome?

- A rare inherited disorder (of the renal tubules of the kidney).
- It causes the kidney to waste potassium, magnesium, sodium and chloride into the urine, instead of reabsorbing it back into the bloodstream.
- Levels of calcium in the urine are low although calcium levels in the blood are normal. Because calcium is retained this protects against getting osteoporosis.
- It does not cause the kidneys to function abnormally in other respects.
- It does not cause kidney failure.
- An affected person has two altered genes, one from each parent.
- Brothers and sisters have a 25% chance of also having Gitelman's syndrome and 50% chance of being a carrier.
- Parents are not themselves affected but are carriers.

Symptoms of Gitelman's Syndrome

The symptoms associated with Gitelman's syndrome are due to the loss of potassium, magnesium and sodium from the body, but the combination of symptoms vary from patient to patient.

- Some patients have no symptoms and the syndrome is only discovered when blood is taken for some other reason.
- Fatigue and tiredness.
- Excessive thirst.
- Salt craving.

- Muscle cramps including leg cramps and muscle spasms.
- Joint pain or stiffness.
- Numbness of hands and feet.
- Heart palpitations.
- Dizziness.
- Blurred vision.
- Increased need to pass urine because the urine is diluted.

Treatment and prognosis

- To replace potassium and magnesium with supplements.
- The medications amiloride, spironolactone and triamterene can be helpful as they help the kidney to retain potassium.
- Regular blood tests to monitor levels of potassium, magnesium and sodium.
- The syndrome has only been recognised since the 1960's, so long term effects are unknown.
- So far the only complications are related to low potassium and magnesium levels.
- It can overlap with Bartter's, a related condition.

Following your clinic visit we may ask you to perform 24-hour urine collections to accurately measure various substances in your urine. Please ask for instructions if you are unsure how to do this.

Where can I get further information?

- The Clinic Sister is Caroline Robinson. You can contact her by:
 - phone: 01223 348745
 - e-mail: caroline.robinson@addenbrookes.nhs.uk
 - <http://barttersite.org/gitelmans>
 - www.ncbi.nlm.nih.gov/omim

- Addenbrookes Kidney Patient Association (AKPA) contact details available from the Renal Genetics Clinic.
- As part of this clinic we are developing patient information about some disorders of the kidney. We would like to include patients' views and experiences in this. If you would like to be involved as a patient representative, please let us know in clinic or on the contact number above.

Renal Genetics and Tubular Disorders Clinic:

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Consultants

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We are currently working towards a smoke-free site. Smoking is only permitted in the designated smoking areas.

For advice and support in quitting, contact your GP or the free NHS Stop Smoking helpline on 0800 169 0 169

Help with this leaflet:



If you would like this information in another language, large print or audio format, please ask the department to contact Patient Information: 01223 216032 or patient.information@addenbrookes.nhs.uk



Document history

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