Can I pass Gitelman or Bartter Syndrome on to my children?

This depends on the genes of your partner.

If your partner is not a carrier, then none of the children you have together will be affected, but they will all be carriers.

If your partner is a carrier (this is extremely unlikely), then for each child there is a 1 in 2 (50%) chance that they will have the condition. If they do not have the condition they will be a carrier.

If your partner also has the condition (this is extremely unlikely), then all the children you have together will have the condition.

If your partner has the condition, they may already know about it, but if they are a carrier they probably will not. The chances of your partner being a carrier are less than 1 in 100, unless you are related by blood (cousins), in which case it is higher. Because these chances are so low, most people with the condition have healthy children who are carriers of the gene.

DNA testing in families

If you are worried your partner may be a carrier, you may want to ask for advice about a DNA test. This is a simple blood test, which looks at the genes that cause the condition. Some people find this information useful when planning a family.

Other family members related to you by blood (including brothers, sisters, parents, children, aunts, uncles, cousins) might also want their DNA examined, so we recommend that you talk to them about it, and suggest they seek professional advice. This can be through their doctor, a renal clinic, or a geneticist.

Where can I get more information?

- www.rarerenal.org
- http://www.facebook.com/groups/Fighting GitelmansSyndrome/
- www.barttersite.org
- http://omim.org/entry/607364
- http://omim.org/entry/26380

Cambridge Renal Genetics and Tubular Disorders Clinic:
Professor Fiona Karet 01223 762617
Sister Caroline Robinson 01223-348745

UCL Centre for Nephrology
Professor Robert Unwin 020 7472 6499
Professor Robert Kleta 020 7317 7554

Great Ormond Street Hospital
Dr Detlef Bockenhauer 020 7405 9200

St. Helier Hospital
Dr Hugh Gallagher 020 8296 3464

Southmead Hospital, Bristol
Dr Charlie Tomson 0117 323 5224

Queen Alexandra Hospital, Portsmouth
Dr G Venkat Raman 023 9228 6000

Freeman Hospital, Newcastle
Dr John Sayer 0191 2336161

Birmingham Children's Hospital
Dr Sally-Anne Hulton 0121 333 9225

Document history

Authors	A Puttick, F Karet, Hypokalaemic Alkaloses Rare Disease Group
Department	Medical Genetics, Addenbrooke's Hospital
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Gitelman Syndrome and Type 3 Bartter Syndrome

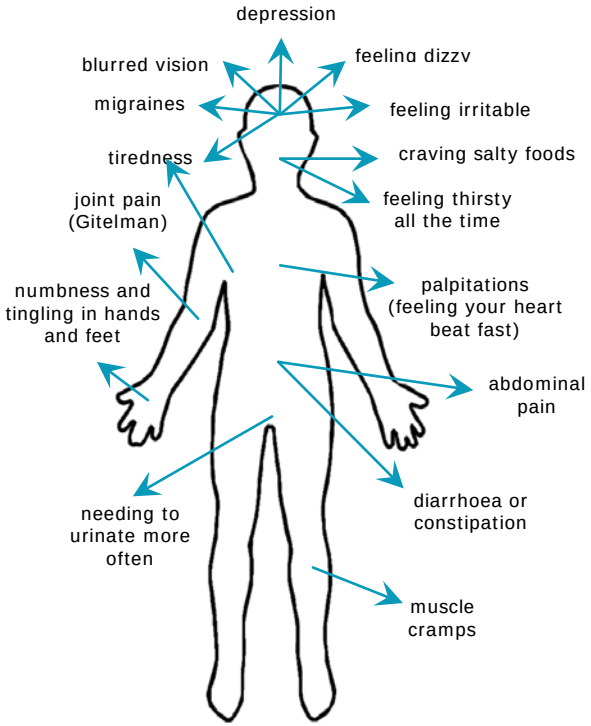
Information for patients and families

What are Gitelman Syndrome and Type 3 Bartter Syndrome?

These are rare, inherited disorders of the kidneys that cause salts, including sodium, potassium and magnesium, to be lost from the body in the urine.

What are the symptoms?

Some people will notice lots of these symptoms, and others will notice very few.



What treatments are there?

Diet - A dietician can help you choose foods with lots of salt, potassium and magnesium in them. Dietary information can be found at:- http://www.rarerenal.org/diseases/hypokalaemic-alkalosis/dietary-needs/

Supplements - Most people will need either potassium (K) or magnesium (Mg) supplements (or both) every day, to help replace what is lost in the urine.

Unfortunately, these supplements are sometimes needed in high doses and can cause unpleasant side effects, including diarrhoea, and can be difficult to digest. We find that the following types cause the fewest problems:

Potassium: SlowK and Kay-Cee-L Liquid
Magnesium: Mg lactate (MagTab SR), Mg glycerophosphate and Mg aspartate.

Other medicines - these might include:
Spironolactone (or eplerenone)

- Amiloride
- ACE Inhibitors (ending with 'pril', for example: lisinopril, enalapril, ramipril)
- Angiotensin Receptor Blockers (ending with 'sartan', for example: losartan)
- Indomethacin can be helpful for children while they are growing

These medicines work by helping your kidneys hold on to the potassium and magnesium you need.

I don't have many symptoms, so why do I need to take these medicines?

Although you may not have noticed symptoms, you have had the condition for a long time, and balancing salt and acid levels with treatment may give you more energy. Your kidneys will keep losing important salts, and without treatment, potassium and magnesium levels in the blood could fall very low. This could cause longer-term problems, including with your heart rhythm.

What else can I expect?

You will need to stay on treatment for the rest of your life and have hospital appointments and regular blood tests. The amount of supplements and medicines needed may change over time.
Although the kidneys are affected, other kidney problems do not usually occur and most of the kidneys' functions remain normal, so you are unlikely to need dialysis or a kidney transplant.

Is there a cure?

Unfortunately we are unable to offer you a cure. However, with treatment, we aim to prevent symptoms and we expect most patients to live a normal life.

How are Gitelman and Bartter Syndromes passed on?

Each condition is caused by changes in one gene, which is important for moving salt around in the kidneys:

- In Gitelman this gene is called SLC12A3 (which encodes a salt transporter)
- In type 3 Bartter this gene is called CLCNKB (which encodes a chloride channel)

Everyone has 2 copies of the gene involved:

- Healthy people have two normal copies
- Carriers have one copy that works normally and one that doesn't. Carriers are usually perfectly healthy because the normal copy can still do its job
- In patients with the condition, neither copy works normally. The gene can't do its job properly and so the kidneys lose salt
- Men and women are affected equally
- When both parents are carriers, each child could be healthy, a healthy carrier, or affected: