

Bartter and Gitelman syndrome: what is it?

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A word on the kidneys

Key task: clean our blood. This includes:

- Removing waste products
- Ensuring the right balance of salts in the blood
- Ensuring the right balance of acid in the blood

A curious way of cleaning...

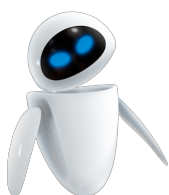
- In the first instance, the kidney filters the blood, forming a primary urine that contains everything but big proteins that do not pass the filter
- In analogy: imagine removing the garbage, by carrying everything outside except those items too big to fit through the door.....
- A large amount is filtered this way: ~180 litres per day

Fixing the cleaning

- We do not want to lose 180 litres of fluid from our blood
- After filtration: most of the fluid and the solvents in there are “reabsorbed”, put back into the blood.
- Done by a whole army of specialised transporters and channels.
- Analogy: imagine an army of robots bringing back all your household items (except the garbage)

And now to: Bartter&Gitelman

- In these disorders, one of these robots is broken.
- Specifically: these robots are needed to bring salt back into the blood



A word on salt

- Salt is a very important part of our blood
- It helps keep the water in the blood vessels
- With not enough salt, there is not enough fluid in our blood



Salt must be rescued

- Other robots take over bringing back the salt
- But: they are not as good: the loose other salts in the process: potassium, chloride and acid



So, this is Bartter & Gitelman

- One of the transporter (robots) needed to bring back salt is broken
- Other transporters take over the task, but loose potassium, chloride and acid in the process (lost in the urine)
- In some forms, calcium is also lost in the urine
- Patients have low potassium, low chloride and high bicarbonate values in their blood. Some deposit calcium in their kidneys (nephrocalcinosis)

Associated complications

- Children can have failure-to-thrive. Some need feeding tubes to supply enough nutrition, fluid and salt
- Some patient complain of fatigue, cramps and aches, probably related to the electrolyte (salt) abnormalities

Special forms

- Some transporters are more important than others. Thus, severity of symptoms can vary
- In some forms the transporters are not only important in the kidneys, but also in the ear and/ or brain.
- These patients can also have deafness and some can have seizures or movement problems
- These complications occur ONLY in those patients where these special transporters are broken