Bartter Syndrome

Bartter syndrome is an inherited kidney disease, which significantly decreases their absorptive capacity resulting in excessive loss of minerals and fluid into the urine.

Causes

The syndrome is caused by a genetic mutation that is inherited. There are several types of the mutation resulting in variable severity of the manifestations. The disease usually begins to manifest in early or late childhood.

Symptoms

The disease affects the renal tubules, which serve for absorption of ions and fluids from the newly formed urine. The disorder of absorption leads to excessive loss of some minerals in the urine (e.g. potassium, sodium, calcium and chlorine) and the fluid loss. The disease typically manifests with excessive urination, large fluid loss and excessive thirst. In situations with insufficient fluid intake, this can cause rapid dehydration with low blood pressure, and dizziness. Potassium losses usually cause hypokalemia and its symptoms (muscle weakness, heart rhythm disorders, etc.).

Some forms of the disease are associated with increased loss of calcium into the urine. Increased concentration of calcium in urine may form urinary stones and hypocalcemia in blood may cause muscle spasms.

Diagnosis

The diagnosis can be suspected in a patient with positive familial history of Bartter syndrome, the above mentioned symptoms and signs of ion deficiency in the blood tests (hypokalemia, hypocalcemia), while there is abundance of those ions in urine (found by urinalysis in a laboratory. The disease and its specific subtype can be confirmed by special genetic testing.

Treatment

We can not cure the disease and we treat the affected only symptomatically. It is necessary to ensure adequate intake of fluids and minerals to cope their losses. In some cases, when the situation progresses into chronic renal failure, dialysis may be indicated.