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Gitelman's Syndrome: Can It Be a Cause of Rhabdomyolysis?

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Abstract

Gitelman syndrome is an autosomal recessive disorder characterised by hypokalaemia, metabolic alkalosis, hypomagnesaemia, hypocalciuria and hypertension [1].

We report the case of a female patient presenting with significant rhabdomyolysis and motor deficiency associated to Gitelman's syndrome.

Observation

A 37-year-old female patient, with no previous personal or family history for hereditary or autoimmune diseases, presented with a proximal and distal muscle weakness with generalised oedema and rhabdomyolysis (100 X normal) following an influenza-like episode. The patient was hospitalized in an intensive care unit.

Initial laboratory tests showed a low blood sodium level of 115 with hyperkalaemia of 6 without renal failure. The infectious disease investigations were all negative. Given the severity of the clinical presentation and the suspicion of an inflammatory myopathy the patient was treated with corticosteroids and plasma exchange. The muscle enzymes were back to normal but a persistent hypokalaemia appeared and remained asymptomatic and refractory to correction.

Gitelman's syndrome was retained on the basis of blood and urine ionograms and metabolic alkalosis.

Anti-nuclear antibodies and muscle antibodies were negative, a muscle biopsy revealed no abnormalities. As corticosteroid tapering was initiated, she presented swallowing problems with abnormal oesophageal mannometry and dysphonia, ENT examination was normal. After 3 months of corticosteroids, we observed a rise in muscle enzymes levels despite potassium supplementation and normal blood and urine ionograms.

Discussion

Gitelman syndrome was first described by Gitelman et al and is now a known inherited tubulopathy [1]. The physiopathology of this syndrome is an abnormality in the renal tubular transport mechanism, caused by mutations in the gene coding for the thiazide-sensitive Na-Cl transporter in the distal tubule [2]. Bartter's syndrome is the main differential diagnosis of Gitelman's syndrome. The calcium-creatinine ratio of 24-hour urine can differentiate between the two entities [3]. The prognosis for Gittelman syndrome is generally good. A potassium and magnesium supplementation, or even treatment with spironolactone if blood pressure permits is indicated in all patients [4]. To the best of our knowledge, its association with inflammatory myopathy has never been described in the literature. What argues in favour of this association is the improvement of myolysis with corticosteroids and relapse following the tapering and elevated muscle enzymes despite normal potassium levels. But Gittelman syndrome associated to rhabdomyolysis can also be a complication of an infectious disease.

Conclusion

Gitelman's syndrome should be considered in the differential diagnosis of hypokalaemic rhabdomyolysis, although it is a rare condition. An even rarer association seems possible in our patient and we are awaiting the results of a second muscle biopsy in a referral center.

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