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Mini Case Study

Gitelman's Syndrome Associated with Chondrocalcinosis. Description of a Clinical Case in an Emergency Department

Isabella Di Zio[1]* and Rodolfo Cisternino[2]

[1]*Medicina Interna, Ospedale San Massimo, Penne, Pescara, Italy*

[2]*Ospedale di comunità, SS. Ospedale Immacolata, Guardiagrele, Chieti, Italy*

***Corresponding Author:** Isabella Di Zio, Medicina Interna, Ospedale San Massimo, Penne, Pescara, Italy.

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Abstract

Gitelman syndrome (GS), is a hereditary tubulopathy (prevalence between 1.2 and 25 casi/1.000.000), characterized by hypomagnesemia, hypokalemia, metabolic alkalosis,and hypocalciuria iperreninemico hyperaldosteronism. On the clinical symptoms may have a large procession consisting of cramps, myalgia, muscle weakness, to episodes of carpal spasm-breech, tetany, and rhabdomyolysis [1-3].

The mutation responsible for the syndrome, an autosomal recessive in, interested in the NCCT gene for the protein, a sodium-chloride cotransporter in the distal convoluted tubule [1].

Several contributions intheliterature underline theassociation of SG withchondrocalcinosis, a rheumatological disorder characterized by episodes of acute orchronic synovitis, cartilaginous secondary deposit of crystals of calcium pyrophosphatedihydrate [1,4-6].

We describe a case of suspected SG associated with chondrocalcinosis occurred in our Emergency Department.

Keywords: *Hypomagnesemia; Hypokalemia; Gitelman Syndrome; Chondrocalcinosis.*

Case Presentation

Male patient, aged 41, reports prelipotymic episode associated with vomiting, asthenia, diffuse myalgias, muscle cramps, tendentially constipated. Vital Signs: BP: 120/80; Sat: 100% in the air; CF: 100/m '(R) CT: 36°C; capillar blood sugar: 134 mg/dl.

Triage code: yellow. On entering the surgery the patient presented alert, oriented in the three parameters, just visit the cooperative, markedly asthenic. The clinical objectivity highlights a significant reduction in muscle strength AAAA, the muscles are aching spontaneously and on palpation. There are no defects in strength or sensitivity to the limbs, no defects or coordination of movement or of equilibrium. Reflexes present. No signs of meningeal irritation. Objectivity cardiorespiratory and abdominal are within limits.

In history chondrocalcinosis from about 10 years for which the patient is followed by an Rheumatic Center. The patient also reported that because of pain related to his disease takes steroids and NSAIDs intramuscularly and orally, continuously for several months. Denied additional diseases, or drug abuse or drugs.

The patient is undergoing venipuncture, EKG, CT head and BGA.EKG: sinus rhythm tended tachycardia. Electrical axis balanced. Alterations of ventricular repolarization. 0-40'' long QT.

Brain CT: No alterations in the density of the brain parenchyma.

BGA: Metabolic alkalosis.

Severe hypokalaemia and Hypomagnesemia on blood tests.

The patient is then hospitalized for intensive observation, monitored from an electrocardiographic point of view and a total of 60 mEq of IV K and 1 gram of magnesium sulfate is infused. The following checks are carried out after 7 hours, at the electrolyte normalization control tests.

EKG: Sinus rhythm. Balanced electrical axis. Changes in ventricular repolarization. No QT alterations.

BGA: Partial correction of metabolic alkalosis.

The patient presents an improvement of the picture both from the laboratory, ECGraphic, blood gas analytical and clinical point of view with reduction of myalgia and muscle weakness. The patient is discharged with the prescription of a home potassium replenishment therapy and transferred to the reference rheumatological center to perform genetic testing in the clinical suspicion of Gitelman Syndrome associated with chondrocalcinosis.

Discussion

SG is a rare hereditary tubulopathy that usually begins in adulthood and is inherited in an autosomal recessive manner. Clinical suspicion arises as a result of the association of the clinical picture with changes in laboratory tests (hypomagnesaemia, hypokalemia and hypocalciuria), however the diagnosis of certainty makes use of genetic testing [1,3].

SG is caused by the mutation of a gene (SCL 12A3, located on chromosome 16p13) that encodes the NCCT protein, a sodium-potassium cotransporter, expressed on the apical membrane of the distal convoluted tubule cells [9]. This protein is responsible for the sodium and potassium reabsorption deficit and the consequent electrolyte imbalance. To date, more than 140 gene mutations have been described in subjects with SG [1,3].

Chondrocalcinosis in SG is secondary to hypomagnesemia. The magnesium deficiency, in fact, reduces the activity of pyrophosphatases, favoring the precipitation of calcium pyrophosphate crystals at the cartilage level [1,5,7,8].

Treatment of SG consists in correcting the electrolyte deficiency and the consequences that can derive from it. Sometimes oral therapy is not sufficient, and periodic intravenous administration of magnesium and potassium is necessary. The use of antialdosteronic diuretics may be useful [1,10].

Conclusion

Despite the rarity of the disease, and while the definitive diagnosis remains strictly specialist relevance, it is clear how important diagnostic-therapeutic support in the Emergency Department was in this clinical case. The objectives we set ourselves were a general diagnostic framework and stabilization of the patient's clinical picture. What was important to us Emergency Physicians was basically the treatment of severe hypokalaemia which had already begun to give electrocardiographic and blood gas analytical changes and the improvement of the patient's general clinical condition.

The beating heart of this whole process remains Brief Observation and Emergency Medicine, fundamental supports to be able to follow the patient's clinical evolution, his response to the therapy practiced and the confirmation of the diagnostic hypothesis. Thanks to the OBI

in particular, the role of the Emergency Doctor turns from the simple role of "stabilizer" of the patient's clinical condition to a leading role in the multidisciplinary approach of the diagnostic chain.

Bibliography

1. Gutierrez M., *et al*. "Gitelman's syndrome associated with chondrocalcinosis: description of two cases". *Reumatismo* 62.1 (2010): 60-64.

2. Knoers NV and Levtchenko EN. "Gitelman syndrome". *Orphanet Journal of Rare Diseases* 30.3 (2008): 22.

3. Hisakawa N., *et al*. "A case of Gitelman's syndrome with chondrocalcinosis". *Endocrine Journal* 45 (1998): 261-267.

4. Resnick D., *et al*. "Clinical, radiographic, and pathologic abnormalities in calcium pyrophosphate dihydrate deposition disease (CPPD): pseudogout". *Radiology* 122 (1977): 1-15.

5. Cobeta-Garcia JC., *et al*. "Chondrocalcinosis and Gitelman's syndrome. A new association?" *Annals of the Rheumatic Diseases* 57 (1998): 748-749.

6. Domínguez JC., *et al*. "Chondrocalcinosis and Hypomagnesemia: Clinical and Radiological Progression". *Reumatología Clínica* 4.1 (2008): 37-39.

7. Ea HK., *et al*. "Chondrocalcinosis secondary to hypomagnesemia in Gitelman's syndrome". *The Journal of Rheumatology* 32 (2005): 1840-1842.

8. Garcia Nieto V., *et al*. "Chondrocalcinosis and hypomagnesaemia in patient with a new mutation in the gene of thiazide-sensitive Na-Cl cotrasporter". *Nefrologia* 23 (2003): 504-509.

9. Colussi G., *et al*. "Correction of hypokalemia with antialdosterone therapy in Gitelman's syndrome". *The American Journal of Nephrology* 14 (1994): 127-135.

10. Bettinelli A., *et al*. "Long-term follow-up of a patient with Gitelman's syndrome". *Pediatric Nephrology* 7 (1994): 67-68.

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