Autoimmune polyendocrinopathy type 1 (APS1), or APECED syndrome, is a rare autosomal recessive genetic disease. Autoimmune hypoparathyroidism is usually the first clinical manifestation and the most prevalent. Other manifestations of the disease are classically chronic mucocutaneous candidiasis and adrenal insufficiency.

**Treatment** will depend on the affected organ and could be challenging throughout life, especially because of development of combination of different auto-immune disorders.

**APECED syndrome**

**Malabsorption syndrome** secondary to exocrine pancreatic insufficiency was strongly suspected. Indeed, malabsorption and other gastrointestinal disorders occur in about 25 percent of patients [1]. A treatment with Creon Forte® was initiated and permitted a resolution of the digestive symptoms (steatorrhea and abdominal pain). The inflammatory syndrome had also resolved under treatment for exocrine pancreatic insufficiency. On the other hand, within a month following the initiation of the treatment with Creon®, the patient developed a severe hypercalcemia (corrected serum calcium 3.29 mmol/L) despite the reduction of the oral dose of calcium (2 g/day) and calcitriol (1.5 µg/day). Calcitriol and oral calcium intake was then interrupted until normalization of serum calcium. Thereafter, oral calcium supplements and calcitriol were reintroduced at initial regimen (1.5 g/day and 0.75 µg/day respectively) that maintained the serum calcium between 2.14 and 2.33 mmol/L.

**Case report**

A 22-year-old woman was admitted to the emergency department for severe acute hypocalcemia. She was diagnosed for hypoparathyroidism at the age of 4 after seizure caused by hypocalcemia. At 14-year-old, she developed an ovarian failure. At that time, APS1 was confirmed by molecular analysis: homozygous mutation in exon 6 of AIRE gene [c.798del(p.Gly267ValfsX111)]. Puberty was induced by estradiol transdermal patch, later replaced by a pill of Désol 30°. She had also a history of vitiligo and asthma treated by leukotriene receptor antagonist.

Serum calcium was stable over time under oral calcium (1.5 g/day) and calcitriol (0.75 µg/day). However, she developed nephrolithiasis three years before admission. Despite persistent adherence to the treatment, she developed severe acute hypocalcemia (corrected serum calcium 1.3 mmol/L). In addition to hypocalcemia related-tetany, she complained of abdominal pain and diarrhea. Acute treatment was initiated with intravenous administration of calcium gluconate. Persisting hypocalcemia required increased oral doses of calcium (4 g/day) and calcitriol (2 µg/day).

1.25-dihydroxy-vitamin D raised slightly despite the doubling of calcitriol dose [from 28.5 to 50.5 pg/ml (N: 22-111)].

**Biology** revealed inflammatory syndrome. Repeated microbiological samples remained negative. Digestive disturbances were investigated by the following tests: measure of steatocrit and fecal calprotectin, triolein breath test, gastroscopy, ileocolonoscopy, abdominal magnetic resonance imaging and enterolysis.

Results were compatible with the presence of a steatorrhea with a steaticrit value of 82% although triolein breath test was normal. Fecal calprotectin was 54 µg per g of stool. Duodenal biopsies showed non-specific mild duodenitis and ileal and colonic mucosa were normal. Abdominal imaging were also normal.

**Conclusion**

High dose of calcium and calcitriol use is limited by the risk of hypercalcemia and by side effects as nephrolithiasis. This case illustrates the necessity to test new therapies like subcutaneous recombinant PTH that could be a good alternative for patients with hypoparathyroidism associated with malabsorption.

**Reference**

1. J Clin Endocrinol Metab. 1998; 83 (4): 1049

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