Recombinant Parathyroid Hormone (1-34) replacement treatment of Hypoparathyroidism in the alfacalcidol-resistant patient with severe Autoimmune Polyendocrinopathy Syndrome type 1

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Nothing to disclose

BACKGROUND: Hypoparathyroidism (HPT) is present in 80% of patients with Autoimmune Polyendocrinopathy Syndrome type 1 (APS-1) – rare monogenic complex disease characterized also by adrenal failure, chronic candidiasis and a spectrum of other autoimmune disorders, including enteropathy and malabsorbtion. Active vitamin D and calcium are currently used for HPT treatment to maintain normal serum calcium levels.

OBJECTIVE: To describe a severe case of APS-1 with HPT and a role of parathyroid hormone therapy in controlling normal calcium level.

CLINICAL CASE:

A patient was diagnosed with APS -1 at the age of 10 and was followed up for next 17 years in our department.
Two heterozygous mutations, R257Xand W78R, were found in *AIRE* gene.

Table 1. Development of clinical components

Clinical components	AGE of manifestation (yrs)	Treatment
Chronic candidiasis	4	Antifungal drugs
Adrenal insufficiency	10	Hydrocortisone Florineff
Hypoparathyroidism	10	Alfacalcidol
Malabsoption	20	Digestive enzymes
Alopecia areata	20	
Ovarian failure	21	Femoston
Enamel hypoplasia	-	
Pigmented retinopathy	26	

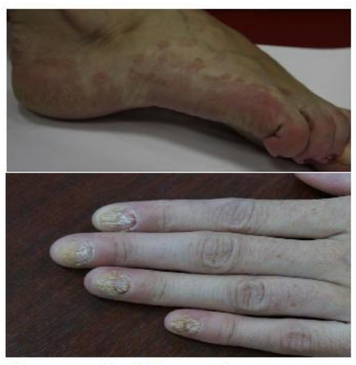

Figure 1. Chronic candidiasis of nails and skin

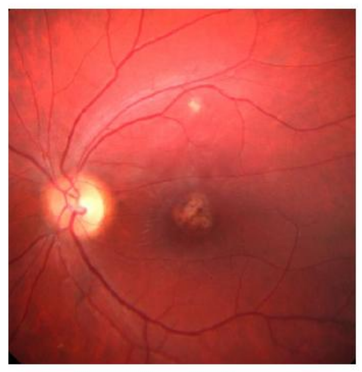

Figure 2. Pigmented retinopathy

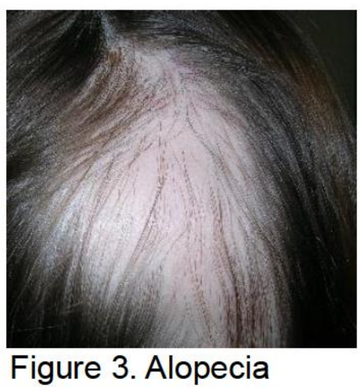

Figure 3. Alopecia

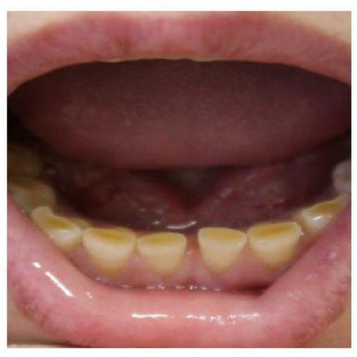

Figure 4. Enamel hypoplasia

Figure 5. The patient at the age of 26 years. Works as a dance teacher.

Course of the disease and PTH1-34 treatment:

Severe malabsorbtion was observed since 20 yrs and has led to clinically significant hypocalcemia. Progressive increasing of alfacalcidol doses up to 20 mcg/day did not normalize calcium level. Gastroscopic study showed an atrophy of the mucosa of the stomach and small intestine. The patient had cholelithiasis and cataracta. Parathyroid Hormone (PTH 1-34) treatment was initiated and normocalcemia was achieved in two weeks on the dose 40 µg per day (twice a day) without vitamin D and calcium supplementation. Calcium level remained within normal range on PTH1-34 treatment without alfacalcidol for the next 5 months but the dose increased to 80 µg per day (twice a day). Urinary calcium excretion was measured on PTH therapy at 8 and 12 months and was in normal range (2.8 mmol/day). Later on the decrease of calcium level was noticed when it was measured just before the injection. Continuous PTH 1–34 delivery by insulin pump was initiated with positive effect.

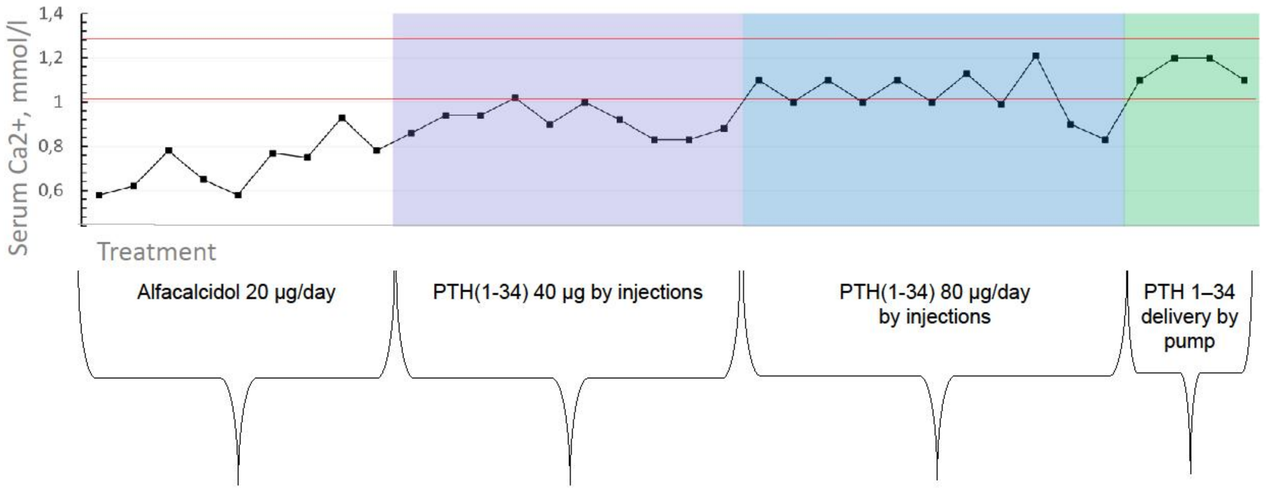

Figure 6. The level of serum ionized calcium

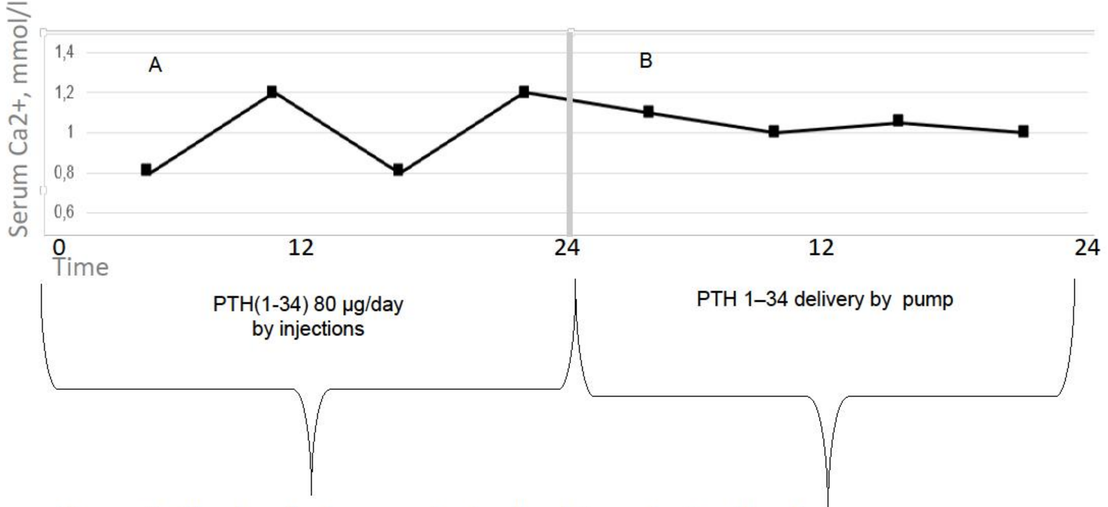

Figure 7. The level of serum ionized calcium during the day: PTH1-34 injections (A) and PTH1-34 by insulin pump (B)

CONCLUSION: Hypocalcemia in severe APS-1 cases could be difficult to control with vitamin D metabolites. PTH replacement therapy of HPT could be a treatment of choice in case of severe malabsorbtion

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