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INTRODUCTION

- Mitochondrial diseases usually occur by mutations of mitochondrial or nuclear DNA.
- Mitochondrial cytopathy is a disease affecting many systems including endocrine system.
- We present a case diagnosed as mitochondrial myopathy previously accompanying multiple endocrinological pathologies.

CASE

- Fifty two years old female patient admitted in another center with a complaint of drooping of upper eyelids 25 years ago.
- Acetylcholine receptor antibodies were negative and her EMG was compatible with progressive external ophthalmoplegia.
- She was diagnosed as mitochondrial myopathy by left biceps muscle biopsy.
- Coenzym Q 10 and L-carnitine treatments were started. She admitted to our center with numbness and spasm in her hands. Serum Calcium was 6.1 mg/dl, Phosphorus was 6.9 mg/dl, Magnesium was 1.5 mg/dl and parathyroid hormone level was 8.9 pg/mL.
- There were common symmetric calcifications at basal ganglions, bilateral cerebellar hemispheres, thalamus and periventricular white matter in cranial CT. EEG was normal. Creatine kinase was 507 U/L, LDH was 435 U/L and plasma lactate level was 2,4 mmol/L. Calcium carbonate and calcitriol were administered to the patient with diagnosis of hypoparathyroidism.
- Thyroid autoantibodies were positive. Thyroid hormon levels were normal. Thyroid ultrasound was compatible with chronic thyroiditis. In eye examination bilateral ptosis and cataract were detected. Retina examination was normal. Her audiometry was normal. She was diagnosed as type 2 DM.



CONCLUSION

- Mitochondrial myopathies can be together with various endocrinological problems.
- Our patient had adult onset pure mitochondrial cytopathy and primary hypoparathyroidism, chronic autoimmune thyroiditis and type 2 DM were accompanying endocrinological pathologies.
- Associations with hypoparathyroidism, autoimmune thyroiditis, DM and growth hormone deficiency have been reported in patients with Kearns Sayre Syndrome in the literature. Also MELAS (mitochondrial myopathy, encephalopathy, lactic acidosis, stroke) and adrenal insufficiency's association was reported.
- Because of probability of affecting multisystem, cases with mitochondrial myopathies should be evaluated for various endocrinological pathologies.

