



## POSTER PRESENTATION

### MECHANISMS

#### PP-M-01

#### PRIMARY MITOCHONDRIAL DISEASES AND ENDOCRINOPATHIES

<https://doi.org/10.15605/jafes.037.AFES.92>

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#### OBJECTIVES

Mitochondria are organelles in the cell involved in energy production by generating adenosine triphosphate (ATP). Primary mitochondrial diseases lead to the impairment of the mitochondrial respiratory chain and results in defective energy production. This review presents the endocrine disorders associated with primary mitochondrial diseases and describes the major genotypes and phenotypes in which they occur.

#### METHODOLOGY

A systematic search of literature was conducted using the search terms mitochondria, primary mitochondrial diseases, mutations, endocrine disorders, genotype and phenotype.

#### RESULTS

Primary mitochondrial diseases are due to defects in the mitochondrial or nuclear DNA. The defects can be inherited or sporadic (somatic mutations). The prevalence of primary mitochondrial diseases is at least 1 in 5,000. The most common mitochondrial DNA defect is the m.3243A > G mutation which is associated with several clinical syndromes including mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) and maternally inherited diabetes and deafness (MIDD). Diabetes mellitus (both type 1 and type 2) is the most common endocrine manifestation of mitochondrial DNA mutations mainly because of its association with the MELAS and MIDD phenotypes and the m.3243A > G mutation. Additional mitochondrial DNA mutations associated with diabetes mellitus include the m.14709T > C, m.8296A > G, and m.14577T > C mutations. Other endocrine disorders, although rarer, have also been reported in association with primary mitochondrial diseases. They include growth hormone deficiency (e.g., in MELAS with m.3243A > G mutation), hypothyroidism, hyperthyroidism, hypoparathyroidism, adrenal deficiency, and hypogonadism in both sexes.

#### CONCLUSION

Primary mitochondrial diseases, mainly due to mitochondrial DNA mutations, are important contributors to various endocrine disorders. The decrease in ATP production and/or increase in oxidative stress causes a failure in the synthesis and/or secretion of hormones, leading to multiple hormonal deficiencies. Through autoimmunity, several endocrinopathies can also occur. The most common endocrine dysfunction observed in primary mitochondrial diseases is diabetes mellitus.

