



## MELAS Syndrome: An Update

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### Introduction

MELAS syndrome (mitochondrial cytopathy, encephalomyopathy, lactic acidosis and stroke-like episodes) is a rare maternally inherited multisystemic disease caused by mitochondrial dysfunction [1,2]. It is characterised by stroke like episodes, resulting in hemiparesis, hemianopia or cortical blindness. Other features include migraine-like headaches, progressive dementia, and muscle weakness, diabetes mellitus, hearing loss, cardiac disease and short stature.

MELAS typically presents in childhood after a normal early development recently it has been recognised that adult onset of symptoms can occur [1-4]. A relapsing-remitting course is most common, with stroke-like episodes leading to progressive neurological dysfunction and dementia. There is persistent lactic acidosis. Histologically, there are ragged red fibres on muscle biopsy and accumulation of abnormal mitochondria in smooth muscle.

Several transfer RNA mutations can be responsible for MELAS, however about 80% of patients with MELAS syndrome exhibit a heteroplasmic A3243 mutation [5,6]. These defects consequently affect mitochondrial energy metabolism, leading to myopathy, metabolic abnormalities and other multisystemic effects.

Gastrointestinal manifestations of MELAS syndrome are varied. Chronic diarrhoea is known to occur in 65% to 70% of patients with MELAS [7]. Intestinal pseudo-obstruction has also been documented in the literature and high calorie parenteral nutrition has been suggested as a treatment modality to resolve symptoms [7,8]. We have previously reported a patient presenting with ischaemic colitis subsequently diagnosed with MELAS [9].

Diagnosis is challenging as MELAS is a rare entity, has multisystemic manifestations and a broad phenotypic heterogeneity [10]. At first presentation, the clinical picture is often incomplete, further hindering the diagnosis. A careful review of family history as well as histological and genetic evaluation can confirm the diagnosis. If genetic studies are affirmative, this obviates the need for muscle biopsy or exhaustive metabolic evaluation. Current prognosis is unfavourable, with encephalomyopathy and cardiac involvement in MELAS associated with high morbidity and mortality.

Patients with MELAS and their families would benefit from genetic counselling to inform of the probability of transmission of the disorder to the next generation and provide social and psychological support

[11]. Siblings of MELAS patients could also choose to undergo genetic testing to determine if they are also affected. Advances in reproductive medicine such as mitochondrial replacement via pronuclear transfer or maternal spindle transfer hold promise for reducing the transmission of MELAS to subsequent generations [12].

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