

## CLINICAL IMAGE

# Colonic pseudo-obstruction caused by MELAS syndrome

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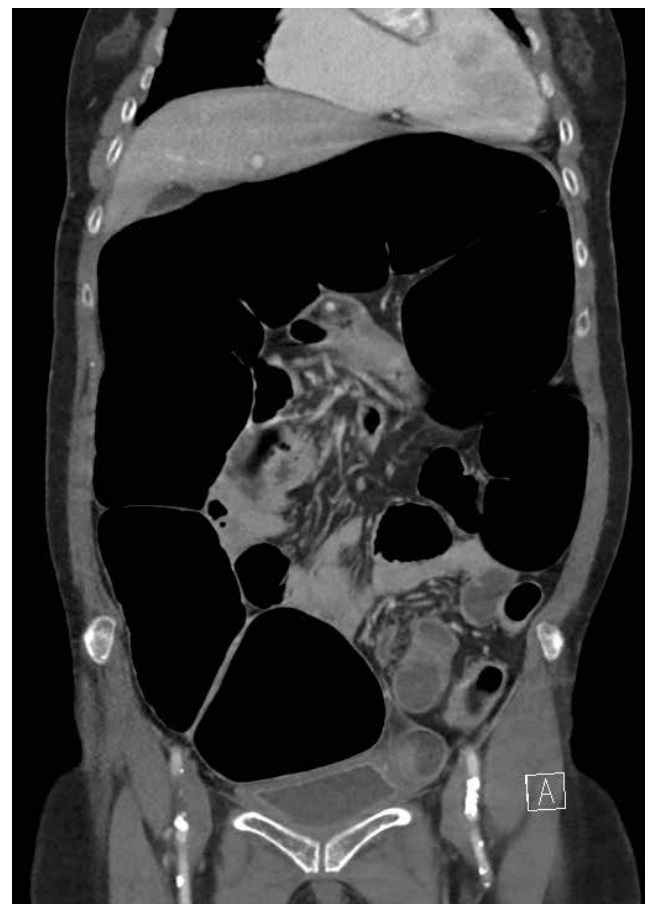
**Keywords** - paralytic ileus, intestinal pseudo-obstruction, MELAS syndrome, sepsis

## Case

A 60-year-old woman presented to the emergency department with nonketotic hyperglycaemia. She was known with the mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS) syndrome since the age of 51. Her mother and grandmother also suffered from this syndrome. She was admitted to the department of internal medicine because of the risk to develop a lactic acidosis during the hyperglycaemia. During hospital admission, she developed respiratory insufficiency due to sepsis and concomitant abdominal distention. She was admitted to the intensive care unit (ICU). Computed tomography of the abdomen revealed pathological dilatation of the entire colon with a maximum cecum diameter of 10 cm (*figure 1*). No signs of mechanical obstruction were



**Figure 1.** Computed tomography (coronal section) of the abdomen showing pathological dilatation of the entire colon



**Figure 2.** Computed tomography (transversal section) of the abdomen showing pathological dilatation of the colon without signs of mechanical obstruction

seen (*figure 2*). X-ray of the chest did not show pneumonia and urine culture was negative. The paralytic ileus with secondary bacterial translocation was considered to be the most likely cause of the sepsis. She was treated with ceftriaxone, metronidazole,

an osmotic laxative, enemas and infusions with glucose 10% to support aerobic metabolism, with initial good clinical response and discharge to the ward.

In the following days, the constipation persisted, for which she received supportive therapy with an osmotic laxative, a stimulant laxative, the serotonin agonist prucalopride and enemas. After initially receiving nutrition through a gastric feeding tube, oral diet was introduced. However, one week after ICU discharge, she ended up in an in-hospital-cardiac arrest due to hypoxaemia after aspiration of gastric content. This aspiration was probably a consequence of the persistent ileus. Although she had return of spontaneous circulation, her neurological prognosis was extremely unfavourable. Supportive treatment was discontinued and she died two days after being readmitted to the ICU.

### Background

MELAS is a maternally inherited syndrome characterised by mitochondrial dysfunction, resulting in insufficient energy for normal metabolism. Reported prevalence varies between 1:13.000 and 1:424 and 25% of the patients present with gastrointestinal symptoms such as nausea, vomiting and anorexia. The most common genetic mutation is a m.3243A>G mutation in the mitochondrial DNA. This mutation was also detected in our patient. Classically, MELAS presents with stroke-like episodes, seizures, (cardio)myopathy and lactic acidosis. Heteroplasmy, meaning that each patient has a unique distribution of normal and aberrant mitochondria, plays a

role in timing of onset and expression of symptoms. In our patient, the syndrome was expressed by premature dementia after two stroke-like episodes and a cerebrovascular accident, cardiomyopathy, hearing impairment, diabetes mellitus and constipation. More recently, she had also developed changing consciousness, possibly due to absences. A rare manifestation of the MELAS syndrome is intestinal pseudo-obstruction. In our opinion, it is likely that our patient developed the paralytic ileus in the context of the MELAS syndrome. In the last weeks to months, progression of several MELAS manifestations was observed accompanied by chronic constipation. In case reports various laxative agents, acetylcholinesterase inhibitors and parenteral feeding are suggested as effective treatment of paralytic ileus in patients with MELAS syndrome. In addition, optimal glycaemic control is essential to prevent and manage lactic acidosis in patients with MELAS syndrome.

In conclusion, the common diagnosis colonic pseudo-obstruction can be caused by a rare disease, such as the MELAS syndrome in this case.

### Acknowledgement

We would like to thank Dr. Monraats (Department of Radiology, Academic Medical Center, Amsterdam, the Netherlands) for assessing the computed tomography.

### Disclosures

The author declares no conflict of interest. No funding or financial support was received.