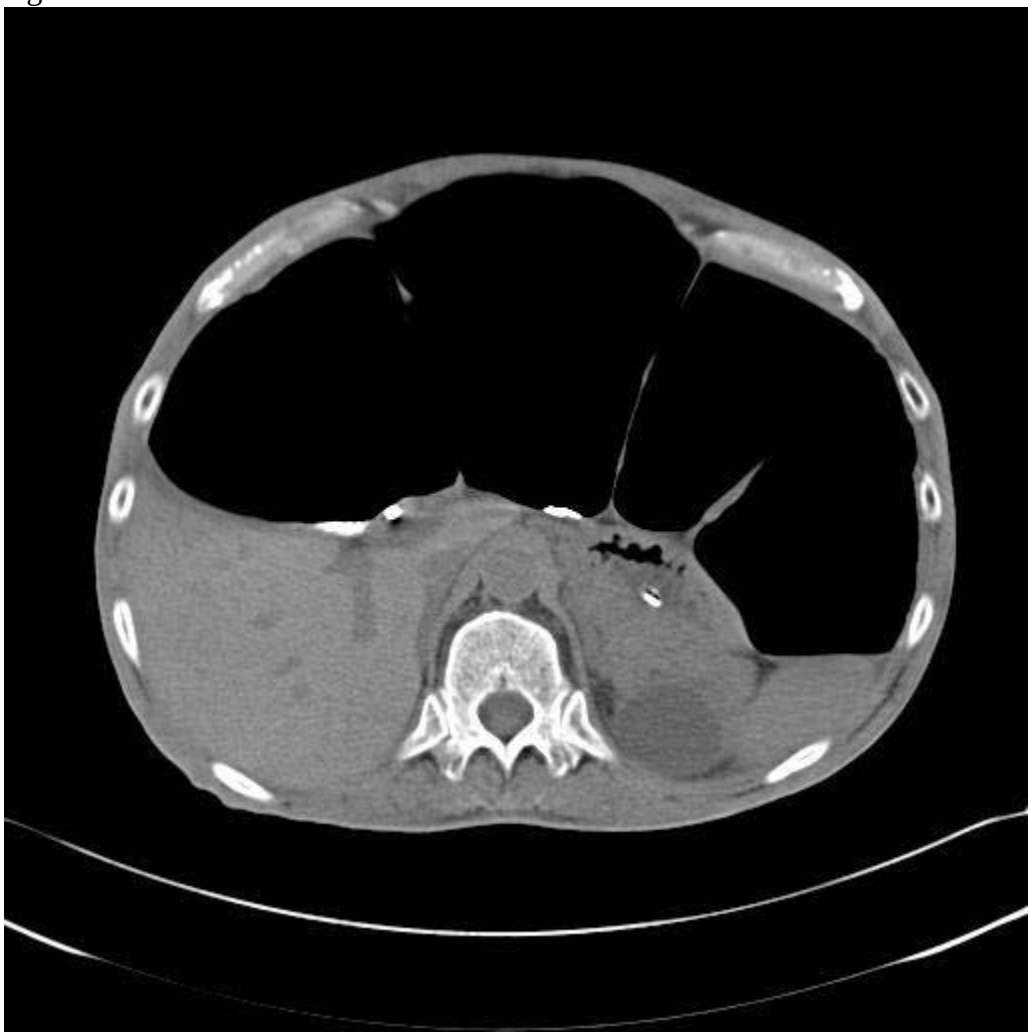
Question 9 – Week of February 6

A 64 year old Caucasian male with history of deafness, diabetes and cardiac dysrhythmia presented with 3 month history of abdominal distention. A non-contrast CT of the abdomen is shown below (figure 1). He was on TPN for nutritional support. Tests for secondary causes of intestinal dysmotility including connective tissue disorders, amyloidosis, spinal lesions and paraneoplastic dysmotility were negative. Colonic motility testing revealed megacolon with an absent contractile responses to a meal and to intravenous neostigmine. His other past medical history was significant for stroke at the age of 40 with no residual deficit. His mother also had a stroke and deafness at 40. Genetic tests revealed substitution of A with G at position 3243 in mitochondrial DNA. The most likely diagnosis is

A. MNGIE
B. MELAS
C. LHON
D. NARP

Figure 1:



Answer: B

The patient has typical features seen in mitochondrial encephalopathy, lactic acidosis and stroke like episodes (MELAS) associated with A3243G mutation in mitochondrial DNA. MNGIE is associated with nuclear DNA mutation in thymidine kinase gene. LHON (lebers hereditary optic neuropathy) and NARP (Neuropathy, Ataxia and Retinitis Pigmentosa) are also mitochondrial disease but gastrointestinal disturbance and strokes are not hallmarks in these conditions.

Reference:

Betts J, et al, Neurology. 2008 Apr 8;70(15):1290-2.