097 SYMPTOMS OF MITOCHONDRIAL DISEASE FALLING ON DEAF EARS

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Case report Mitochondrial diseases show a broad phenotypic spectrum and are underdiagnosed. We present a 37-year-old woman referred to our neurology clinic after an incidental finding of bilateral caudate, lentiform and pulvinar calcification during workup for cochlear implant surgery. She was born at 30 weeks' gestation with her identical twin sister and was found to have progressive sensorineural hearing loss from eight years of age. She had had no seizures, ataxia, weakness, psychosis or involuntary movements. Her mother had diabetes mellitus. On examination, she was areflexic but otherwise normal. Neurophysiology showed a mild sensory neuropathy. Mitochondrial gene testing showed heteroplasmy for m.3243A>G mutation.

Discussion Our patient was seen by a number of different specialists for her deafness and the possibility of mitochondrial diseases was never explored. Pathogenic mitochondrial gene mutations are present in up to 0.5% of the general population. The m.3243A>G mutation is associated with deafness and diabetes mellitus. Neurologists see it in MELAS syndrome. Radiologically, it is associated with basal ganglia calcification. Reaching a diagnosis allows the patient and family to understand their illness and prognosis, to undergo predictive testing if wished and to undergo screening for other affected organ systems.