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INVESTIGATE PATIENTS WITH KEARNS-SAYRE SYNDROME AND THEIR MOTHERS THOROUGHLY

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Letter to the Editor

With interest we read the article by Sabella-Jimenez et al. about two patients (patient-1, patient-2) with Pearson syndrome (PS) that evolved into chronic progressive external ophthalmoplegia (CPEO plus), and lastly Kearns-Sayre syndrome (KSS) with progression of the disease [1]. KSS in patient-1 was due to the single mtDNA deletion NC_012920.1:m.8286_14416del [1]. KSS in patient-2 was due to a single mtDNA duplication of 7.9kb [1]. Patient-1 deceased at age 10y and patient-2 at age 7y [1]. We have the following comments and concerns.

Patient-1 was described with muscle weakness of the upper limbs and reduced tendon reflexes but normal electromyography (EMG) [1]. We should know if the EMG was carried out with needles and it should be discussed why there is a discrepancy between the clinical exam and the EMG findings. Is it conceivable that the clinical findings were due to neuropathy? Were nerve conduction studies carried out? Which were the results?

Patient-1 was described as having had ptosis on the right side [1]. However, figure 1 indicates that the patient had bilateral ptosis with right-sided predominance [1]. Was the picture taken some time after the first clinical neurologic exam?

The ECG in patient-1 showed premature ventricular contractions [1]. We should be informed if the patient had Wolf-Parkinson White (WPW)-syndrome or Lown-Ganong-Levine (LGL) syndrome, or another conduction defect, and if she underwent ablation or another treatment.

Since patients with CPEO plus, as patient-1, may develop hypertrophic cardiomyopathy [2], we should know the results of echocardiography and eventually coronary angiography.

Patient-2 was reported with symptomatic hypocalcemia and focal seizures. We should know if these seizures were triggered by hypocalcemia or if other triggers could be identified. Since seizures were associated with hemiparesis, we should know if the patient had developed a stroke-like episode and if the MRI at that time was indicative of a stroke-like lesion.

It should be discussed why patient-2 had cerebellar ataxia but normal MRI of the brain. Recently, involvement of the spinal cord in KSS has been reported [3,4]. We should know if patient-2 had involvement of the spinal cord and if ataxia in this patient was attributable to spinal cord involvement.

Though MIDs due to single mtDNA deletions are usually sporadic, they may be maternally transmitted in 4% of the cases [5]. We should know if the mothers of the two presented patients were prospectively investigated for subclinical abnormalities or mild clinical manifestations of the disease and if they underwent genetic testing as well. We also should know the causes of decease in both patients.

Overall, this interesting report about two patients with KSS has some limitations, which should be addressed before drawing final conclusions. Concerning patient-1 it should be clarified if she had bilateral ptosis, if she had myopathy of limb muscles, which type of pre-excitation was diagnosed, and if there was hypertrophic cardiomyopathy. Concerning patient-2, it should be discussed if seizures were due to hypocalcemia or a stroke-like episode and if ataxia was due to spinal cord involvement in the light of normal cerebral MRI. It should be clarified if the mtDNA rearrangements were sporadic or maternally inherited.

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