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Letter to the Editor

Heart Disease in Kearns-Sayre Syndrome and its Therapeutic Management

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Letter to the Editor

We read with great interest the article by Krupka *et al*. about a 46-year-old man with Kearns-Sayre syndrome (KSS) who clinically presented with AV block requiring pacemaker implantation, as well as dilated cardiomyopathy requiring cardiac resynchronisation therapy (CRT) and implantable cardioverter defibrillator (ICD) implantation and heart failure therapy, hypothyroidism, myopathy, pulmonary hypertension, pigmentary retinopathy, renal insufficiency, unresponsive pupils, and dysphagia [1]. Initially, the patient was considered for heart transplantation (HTX), but after reassessment, this decision was reversed [1]. After discharge, the patient died from complications of SARS-CoV-2 infection [1]. The report is noteworthy, but several points should be discussed.

The first point is that we disagree with the diagnosis of KSS. The diagnosis of KSS requires that the disease begins before the age of 20, that there is progressive external ophthalmoplegia, that there is pigmentary retinopathy, and that there is heart block, cerebellar symptoms, or elevated protein levels in the cerebrospinal fluid [2]. Since the patient did not have ophthalmoplegia and, according to the case description, the disease began at the age of 25, the diagnostic criteria are not met and the diagnosis should be revised.

The second point is that the diagnosis of KSS was not genetically substantiated [1]. Did the patient have a single mtDNA deletion or an mtDNA point mutation? Knowledge of the underlying genetic defect is crucial not only for assessing the course and outcome of the disease, but also for genetic counseling. Given that KSS is caused by single mtDNA deletions in most cases and is inherited in 4% of cases [3], we should also know whether the suspected deletion was inherited or occurred de novo.

The third point is that the patient received a pacemaker at the age of 25 due to drop attacks caused by a heart block, but there is evidence that cardiac involvement in KSS includes not only AV blocks but also malignant ventricular arrhythmias [4]. Was a long-term ECG recording performed on the patient prior to pacemaker implantation, and why did the patient not receive an ICD at the age of 25?

The fourth point is that the patient was diagnosed with dysphagia, which is rather unusual in KSS. Was the dysphagia due to involvement of the central nervous system, impairment of the striated pharyngeal muscles, involvement of the smooth gastrointestinal muscles, or depression? Knowing the cause is crucial, as it can determine the course of the disease and have implications for treatment, which depends on the underlying cause. There is also no explanation as to why the patient's pupils did not respond to light [1]. Was this due to amaurosis, impairment of autonomic innervation, iris disease, or intraocular infection?

The fifth point is that the current medications the patient was taking regularly were not specified [1]. Was the patient taking medications that contributed to the development of heart failure? Was the patient taking antiepileptic drugs (ASM) regularly? KSS can rarely manifest as seizures [5].

Finally, it should be explained why the patient did not take his heart failure medication regularly: was this due to refusal or cognitive impairment?

In summary, the diagnosis of KSS should only be made if the diagnostic criteria are met and if genetic testing reveals a single mtDNA deletion or a causative mtDNA mutation. Before implanting a pacemaker in these patients, it should be clarified whether or not there are indications for an ICD.

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References
1. Krupka D, Rakoczy K, Chełmoński A, Zakliczyński M, Przybylski R, Sokolski M. Crucial Role of Early Detection in Managing Heart Failure in Kearns-Sayre Syndrome: A Case Report. Am J Case Rep, Aug 18, 2025; 26:e947439. Doi: 10.12659/AJCR.947439
2. Shemesh A, Margolin E. Kearns-Sayre Syndrome, Jul 17, 2023. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing, Jan 2025. PMID: 29493966.
3. Poulton J, Finsterer J, Yu-Wai-Man P. Genetic Counselling for Maternally Inherited Mitochondrial Disorders. Mol Diagn Ther, Aug 2017; 21(4):419-429. Doi: 10.1007/s40291-017-0279-7
4. Wiseman K, Gor D, Udongwo N, Alshami A, Upadhaya V, Daniels SJ, *et al*. Ventricular arrhythmias in Kearns-Sayre syndrome: A cohort study using the National Inpatient Sample database 2016-2019. Pacing Clin Electrophysiol, Dec 2022; 45(12):1357-1363. Doi: 10.1111/pace.14607
5. Allen RJ, Di Mauro S, Coulter DL, Papadimitriou A, Rothenberg SP. Kearns-Sayre syndrome with reduced plasma and cerebrospinal fluid folate. Ann Neurol, Jun 1983; 13(6):679-682. Doi: 10.1002/ana.410130620