Kearns-Sayre syndrome requires cardiac MRI and an ICD

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In a recent article by Krishna et al. published in the Indian Pacing Electrophysiol journal a 16yo Indian male with Kearns-Sayre syndrome (KSS) due to a single large-scale mtDNA deletion was presented. He manifested phenotypically in the skeletal muscles as easy fatigability, ptosis, external ophthalmoplegia, and lactic acidosis, in the eyes as pigmentary retinopathy, in the heart as AV-block III requiring a pacemaker at age 14y, in the endocrine system as short stature, and in the gastrointestinal tract as recurrent vomiting. The patient died suddenly at age 16y without undergoing autopsy. We have the following comments and concerns.

Sudden cardiac death (SCD) has been repeatedly reported in KSS. The most common cardiac manifestations in KSS are conduction disease with atrio-ventricular block, or bradycardia-related polymorphic ventricular tachycardia, which are attributed to progressive myocardial scarring. This is why cardiac MRI is indicated in these patients not to miss the point at which a pacemaker should be upgraded to an implantable cardioverter defibrillator (ICD). Did the reported patient undergo cardiac MRI since KSS was diagnosed and were any abnormalities detected, which might have been missed on transthoracic echocardiography?

Additionally, it would be interesting to know if the patient had undergone electrophysiological investigations prior to pacemaker implantation or afterwards. Was there any evidence for vulnerability of the cardiac conduction system in addition to the AV-block, such that ventricular arrhythmias could be triggered upon electrical stimulation? Ventricular arrhythmias have been repeatedly reported as a cardiac manifestation of KSS and may be responsible for the increased risk of SCD in these patients.

Furthermore, we should be informed if the pacemaker was intact and pacing when the patient underwent resuscitation or was there complete electrical silence on arrival of the parents or the ambulance? Was there electrical activity of the pacemaker on arrival at the hospital?

KSS patients may also develop prolongation of the QT-interval, which could be a trigger of ventricular arrhythmias and SCD. Was there ever an indication for QT-prolongation? In this context it should be communicated which drugs the patient was taking at the time of his death. Was he regularly taking any QT-prolonging compounds?

Since KSS may be associated with left ventricular hypertrabeculation / noncompaction (LVHT) it would be interesting to know if echocardiography was revised for this myocardial abnormality, which is frequently missed if the imaging of the apex is insufficient or if echocardiographers are not aware of this pathology. Since LVHT is associated with an increased risk of developing ventricular arrhythmias, LVHT could be another source of SCD. It is also conceivable that the patient died from the global type of Takotsubo syndrome (TTS), which could be triggered by acute stress due to external or internal stimuli. Which links were found on the computer the patient was using before decease?

Overall, this interesting case suggests that KSS is not only associated with conduction defects but also with ventricular arrhythmias implying that these patients should receive rather an ICD than a pacemaker at diagnosis. Furthermore, KSS patients should undergo cardiac MRI or electrophysiological stimulation to assess the risk of conduction defects and arrhythmias upon the presence or absence of myocardial scars or inducible arrhythmias.

References

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