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Renal Dysfunction is a Common Phenotypic Feature of Kearns-Sayre Syndrome

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Letter to the Editor

In a recent article, Choe et al. reported about a 7yo female with Kearns Sayre syndrome (KSS) who presented with a Bartter-like phenotype [1]. We have the following comments and concerns.

Bartter-like or Bartter syndrome is not an unusual phenotypic manifestation of KSS and has been previously reported in patients with KSS (Table 1) [2]. Bartter syndrome is due to mutations in the *SLC12A1* (type 1), *KCNJ1* (type 2), *ClCNKb* (type 3), *BSND* (type 4), or the *CSAR* gene (type 5) respectively and characterised by a hyper-reninemic hyperaldosteronism, hyponatriemia, hypokaliemia, hypo-osmolarity, arterial hypotension despite hyper-reninemic hyperaldosteronism, and alkalosis. Were mutations in these genes excluded as differential diagnoses of the renal abnormality? Other manifestations of renal disease that have been repeatedly found in KSS are Toni-Debre-Fanconi syndrome, renal tubular acidosis, and renal insufficiency (Table 1) [3].

Renal abnormality	NOP	Age	Sex	Reference
Renal insufficiency	1	10	m	[4]
	4	6-21	1f, 3m	[5]
	1	na	na	[6]
Renal tubular acidosis	1	7	m	[7]
Bartter-like syndrome	1	14	m	[2]
	1	7	f	[1]
	1	10	m	[8]
Toni-Debre-Fanconi syndrome	1	11	f	[9]
	1	10	m	[10]
	1	10	m	[11]
	1	5	m	[12]
	1	18	m	[3]
	1	8	f	[13]
	1	43	f	[14]
	1	13	f	[15]
NOP: number of patients, f: female, m: male, na: not accessible				

Table 1: Renal dysfunction in KSS.

A typical electrocardiographic manifestation of KSS is atrio-ventricular conduction block requiring pacemaker implantation. Did the patient develop AV-block during follow-up? Did he require implantation of a pacemaker or an implantable cardioverter defibrillator (ICD) because of QT-prolongation or ventricular arrhythmias?

Which was the indication for muscle biopsy? Did the patient complain about muscle symptoms or did blood tests show elevated muscle enzymes or elevated serum lactate?

Diagnosing KSS also requires elevation of protein in the cerebrospinal fluid (CSF). It would be interesting to know if there was elevated protein or lactate in the CSF?

Though single mtDNA deletions are usually sporadic in KSS, there are some cases in which the mutation is inherited. We should be informed about the family history, particularly if there were any clinical indications for a mitochondrial disorder in the parents or siblings of the patient. Was there renal dysfunction in any of the index patient's relatives? Was there consanguinity between the parents?

At age 8y the patient presented with bilateral ptosis but no ophthalmoparesis [1]. Ophthalmoparesis or ophthalmoplegia, however, is a key phenotypic feature of KSS. Did the patient develop ophthalmoparesis during further follow-up?

Overall, this interesting case requires work-up for mitochondrial disease in other family members, long-term follow-up of the index case to see if key phenotypic features of the syndrome had developed, and exclusion of differential diagnosis that could explain renal dysfunction.

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