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Nephropathy in MELAS

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LETTER TO THE EDITOR

In a recent article, Rudnicki et al. [1] reported about a 37yo female with m.3243A>G-associated MELAS-syndrome manifesting as mitochondrial multiorgan disorder syndrome (MIMODS) [2] with hypertrophic cardiomyopathy, deafness, cognitive impairment, dysarthria, short stature, renal insufficiency, and diabetes [1]. The following comments are provided.

Renal involvement in mitochondrial disorders is not unusual and includes renal insufficiency, nephrotic syndrome, renal tubular acidosis, Fanconi-syndrome, Bartter-like-syndrome, renal cysts, tubulo-interstitial nephritis, nephrocalcinosis, nephrolithiasis, kidney neoplasms, or focal segmental glomerulosclerosis [3]. Renal involvement may be the dominant or non-dominant phenotypic feature [4].

The patient had diabetes and we should be informed about the severity of diabetes and if renal dysfunction could be attributed at least in part to diabetic nephropathy. Which were the HbA1c values? MELAS may also manifest as hyperlipidemia, atherosclerosis, or arterial hypertension [5]. To which degree did arteriopathy contribute to renal insufficiency? Which were the results of 24h-blood-pressure monitoring? Did the patient require statines, antihypertensives, or other drugs? Was any of these specimens nephrotoxic or mitochondrion-toxic? How did the patient tolerate the immunosuppressive medication after transplantation? Did she regularly take cofactors, vitamins, or antioxidants? Was a ketogenic diet tried?

Was the mutation inherited or spontaneous? Did other family members carrying the mutation, also present with kidney disease or was there phenotypic heterogeneity between them?

Which were the cerebral manifestions of MELAS in addition to cognitive impairment?

Overall, this interesting case deserves provision of supplementary data to determine if MELAS was hereditary/sporadic, if renal failure was multifactorial, if drugs/diet were beneficial, and if cerebral imaging / functional studies were abnormal.

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