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Progressive myoclonus epilepsy without renal failure in a Chinese family with a novel mutation in *SCARB2* gene and literature review

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Statement of the Problem: To describe the clinical and genetic features of a Chinese progressive myoclonus epilepsy (PME) patient with *SCARB2* mutation without renal impairment and review 27 *SCARB2*-related PME patients from 11 countries.

Method: The patient was a 27-year-old man with progressive action myoclonus, ataxia, epilepsy, dysarthria and absence of cognitive deterioration. Renal functional test was normal. Electroencephalography showed progressively slowed background activity and sporadic generalized spike-and-wave discharges. Electromyography showed slowed motor and sensory nerve conduction velocities and distal motor latency delay accompanied by normal Compound Motor Action Potential (CMAP) and amplitudes of Sensory Nerve Action Potential (SNAP). The amplitude of cortical components of Brainstem Auditory-Evoked Potential (BAEP) was normal with slightly prolonged latencies. Generalized atrophy, ventricle enlargement and white matter degeneration was observed in brain magnetic resonance imaging. Open muscle biopsy and genetic analysis were performed. 200 healthy individuals were set for control. qPCR, western blotting and immunofluorescence were carried out to evaluate the fate of the *SCARB2* mRNA and lysosomal-membrane type 2 (LIMP2) protein level.

Findings: One homozygous mutation in *SCARB2* gene (c.1187+5G>T) was identified in the patient. Each of his parents carried a heterozygous variant. This mutation was not detected in healthy controls and predicted to be disease causing by prediction tools. qPCR revealed a significantly lower level of *SCARB2* mRNA in peripheral blood cell of the proband compared with his parents and healthy control. Muscle biopsy showed mild variation in fiber size. Western blotting and immunofluorescence detected an extremely weak signal of LIMP2 protein from skeletal muscle of the proband.

Conclusion: In this study, we identified a *SCARB2*-related PME patient with normal renal function and a novel homozygous splicing mutation. *SCARB2* gene should be analyzed in patients with progressive action myoclonus, epilepsy, peripheral neuropathy, without cognitive deterioration or renal failure.

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