# 3URJUHVVLYH P\RFORQXV HSLOHSV\ ZLWKRXW UHQDO IDLOXUH SCARB2 JHQH DQG OLWHUDWXUH UHYLHZ

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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: e 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 nerv conduction velocities and distal motor latency delay accompanied by normal Compound Motor Action Potential (CMAP) and amplitudes of Sensory Nerve Action Potential (SNAP). e 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 performe 200 healthy individuals were set for control. qPCR, western blotting and immuno uorescence were carried out to evaluate the fate of the SCARB2 mRNA and lysosomal-membrane ivvalu EMC Lor contnl ed laAEP (e)8 (10 0 3li-pan <</Lang (en-US)(t)6 /