DISEASE:
Action myoclonus-renal failure syndrome

NAME:	Action myoclonus-renal failure syndrome
DESCRIPTION:	A rare epilepsy syndrome characterized by progressive myoclonus epilepsy in association with primary glomerular disease. Patients present with neurologic symptoms (including tremor, action myoclonus, tonic-clonic seizures, later ataxia and dysarthria) that may precede, occur simultaneously or be followed by renal manifestations including proteinuria that progresses to nephrotic syndrome and end-stage renal disease. In some patients, sensorimotor peripheral neuropathy, sensorineural hearing loss and dilated cardiomyopathy are associated symptoms.
ORPHACODE:	163696
SYNONYMS:	AMRF EPM4 Myoclonus-nephropathy syndrome Progressive myoclonic epilepsy type 4 Progressive myoclonus epilepsy type 4
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SCARB2
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