DISEASE:
Gerstmann-Straussler-Scheinker syndrome

NAME:	Gerstmann-Straussler-Scheinker syndrome
DESCRIPTION:	A rare inherited human prion disease characterized by adult onset of slowly progressive cerebellar ataxia, with dementia developing relatively late in the disease course (classic ataxic phenotype). Patients may present with gait disturbances and frequent falls, dysarthria, dysphagia, nystagmus, dysmetry, and eventually pancerebellar syndrome, myoclonus, spasticity, severe dementia, and mutism. The disease is invariably fatal after five years on average. Neuropathological hallmark is the presence of numerous multicentric prion protein plaques in the cerebral and cerebellar cortex.
ORPHACODE:	356
SYNONYMS:	Subacute spongiform encephalopathy, Gerstmann-Straussler type
XREF(S):	Orphanet OMIM MeSH MedDRA ICD-10
ANALYTE(S):	PRNP
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