DISEASE:
CAMOS syndrome

NAME:	CAMOS syndrome
DESCRIPTION:	A disorder that is characterised by the association of a non-progressive congenital ataxia, severe intellectual deficit, optic atrophy and structural anomalies of the skin vessels. It has been described in five children from a large consanguineous Lebanese family. Short stature and microcephaly were also reported. Transmission is autosomal recessive.
ORPHACODE:	83472
SYNONYMS:	Cerebellar ataxia-intellectual disability-optic atrophy-skin abnormalities syndrome SCAR5
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ZNF592 WDR73
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- WD repeat domain 73
- zinc finger protein 592

Related Gene Panels

- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG