Cell Lines SPG7 Publication

CU19008

IDENTIFICATION

Causal gene(s)	SPG7
Repeat size or mutation	c.1715C>T
Cell type	Fibroblast

DONOR INFORMATION

Age at collection (years)	3

SOURCE & PUBLICATIONS

Originating lab / institution	University of Milan
Links to publications or public resources	Whole-Exome Sequencing Study of Fibroblasts Derived From Patients With Cerebellar Ataxia Referred to Investigate CoQ10 Deficiency - PubMed https://pubmed.ncbi.nlm.nih.gov/37090936/