Cell Lines SPG7 Publication

11/082

IDENTIFICATION

Causal gene(s)	SPG7
Repeat size or mutation	c.1529C>T (het) and p.A510V (exon 11) and c.1727C>G (het) and p.S576W (exon 13)
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Age at collection (years)	58

SOURCE & PUBLICATIONS

Originating lab / institution	University of Sydney
Links to publications or public resources	Single cell morphology distinguishes genotype and drug effect in Hereditary Spastic Paraplegia - PubMed https://pubmed.ncbi.nlm.nih.gov/34404843/