Cell Lines SPG7 Publication

Patient 2

IDENTIFICATION

Causal gene(s)	SPG7
Repeat size or mutation	c.415C > T (exon 4) and c.941T>A (exon 7)
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male
Age at collection (years)	52

SOURCE & PUBLICATIONS

Originating lab / institution	University of Sydney
Links to publications or public resources	Pharmacological rescue of mitochondrial and neuronal defects in SPG7 hereditary spastic paraplegia patient neurons using high throughput assays - PubMed https://pubmed.ncbi.nlm.nih.gov/37766787/