Cell Lines SPG7 Publication

Patient 3

IDENTIFICATION

Causal gene(s)	SPG7
Repeat size or mutation	chr16:g.89623293A > G, c.2182-2A > G (intron 16- exon 17) and c.1529C>T (exon 11)
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male
Age at collection (years)	47

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/