

[Cell Lines](#) [SPG7](#) [Publication](#)

# 14/065

## IDENTIFICATION

|                         |   |
|-------------------------|---|
| Causal gene(s)          | SPG7  |
| Repeat size or mutation | c.1454_1462del (het) and p.Arg485_Glu487del (exon 11) and c.1529C>T (het) and p.A510V (exon 11) |
| Cell type               | Fibroblast  |

## DONOR INFORMATION

|                           |      |
|---------------------------|------|
| Donor gender              | Male |
| Age at collection (years) | 70   |

## 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<br><a href="https://pubmed.ncbi.nlm.nih.gov/34404843/">https://pubmed.ncbi.nlm.nih.gov/34404843/</a> |