Cell Lines CANVAS Biobank

# MCRIi026-A

## IDENTIFICATION

| | |
|---|---|
| **Research Resource Identifier (RRID)** | CVCL_D0HV |
| **Causal gene(s)** | RFC1 |
| **Repeat size or mutation** | Disease |
| **Cell type** | hiPSC |

## DONOR INFORMATION

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|---|---|
| **Donor gender** | Female |
| **Age at collection (years)** | 61 |

## SOURCE & PUBLICATIONS

| | |
|---|---|
| **Originating lab / institution** | Murdoch Children's Research Institute, University of Melbourne |
| **Links to publications or public resources** | Generation and heterozygous repair of human iPSC lines from three individuals with cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS) carrying biallelic AAGGG expansions in RFC1 - PubMed<br>https://pubmed.ncbi.nlm.nih.gov/36805468/<br>MCRIi026-A u00b7 Cell Line u00b7 hPSCreg<br>https://hpscreg.eu/cell-line/MCRIi026-A |