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## I-2

### IDENTIFICATION

Causal gene(s)	RFC1
Repeat size or mutation	AAGGGn expansion + C.1267C>T
Cell type	Fibroblast

### DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	55
Age at collection (years)	61

### SOURCE & PUBLICATIONS

Originating lab / institution	Massachusetts General Hospital and Harvard Medical School
Links to publications or public resources	Truncating Variants in RFC1 in Cerebellar Ataxia, Neuropathy, and Vestibular Areflexia Syndrome - PubMed <a href="https://pubmed.ncbi.nlm.nih.gov/36289003/">https://pubmed.ncbi.nlm.nih.gov/36289003/</a>