Cell Lines CANVAS Publication

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 https://pubmed.ncbi.nlm.nih.gov/36289003/