Cell Lines CANVAS Publication

CANVAS 2C Heterozygous

IDENTIFICATION

Causal gene(s)	RFC1
Repeat size or mutation	~0/1300 AAGGG
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Female

SOURCE & PUBLICATIONS

Originating lab / institution	Michigan Medicine; pub: University of Michigan
Links to publications or public resources	AAGGG repeat expansions trigger RFC1-independent synaptic dysregulation in human CANVAS neurons - PubMed https://pubmed.ncbi.nlm.nih.gov/39231235/