Cell Lines CANVAS Publication

CANVAS 4P

IDENTIFICATION

Causal gene(s)	RFC1
Repeat size or mutation	~1080/1180 AAGGG
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male

SOURCE & PUBLICATIONS

Originating lab / institution	Laboratory of Vikram Khuruna, Harvard 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/