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SCA14-2

IDENTIFICATION

Causal gene(s)	RKCG
Repeat size or mutation	H101Q (exon 4)
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Age at collection (years)	71

SOURCE & PUBLICATIONS

Originating lab / institution	University of Oxford
Links to publications or public resources	Neurodegeneration in SCA14 is associated with increased PKCu03b3 kinase activity, mislocalization and aggregation - PubMed https://pubmed.ncbi.nlm.nih.gov/30249303/