Cell Lines SCA27B Publication

Patient P1

IDENTIFICATION

Causal gene(s)	FGF14
Repeat size or mutation	295/304 GAA-TTC
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Originating lab / institution	McGill University
Links to publications or public resources	Somatic instability of the FGF14-SCA27B GAAu2022TTC repeat reveals a marked expansion bias in the cerebellum - PubMed https://pubmed.ncbi.nlm.nih.gov/39378335/