Cell Lines AOA1 Publication

H201R-iPSC

IDENTIFICATION

Causal gene(s)	APTX
Repeat size or mutation	c.602A>G (heterozygous); CRISPR/Cas9 modified from control
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male

SOURCE & PUBLICATIONS

Originating lab / institution	Southern Medical University
Links to publications or public resources	Induced pluripotent stem cells carrying novel APTX mutations presented defective neural differentiation with the accumulation of DNA single-strand breaks - PubMed https://pubmed.ncbi.nlm.nih.gov/41136416/