Cell Lines AOA1 Publication

EAOH2

IDENTIFICATION

Causal gene(s)	APTX
Repeat size or mutation	689insT (homozygous)
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Female
Age at collection (years)	31

SOURCE & PUBLICATIONS

Originating lab / institution	Nara Medical University
Links to publications or public resources	DNA single-strand break repair is impaired in aprataxin-related ataxia - PubMed https://pubmed.ncbi.nlm.nih.gov/17315206/