Cell Lines AOA1 Publication

FD104

IDENTIFICATION

Causal gene(s)	APTX
Repeat size or mutation	pW279X/pW279X
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Links to publications or public resources	The ataxia-oculomotor apraxia 1 gene product has a role distinct from ATM and interacts with the DNA strand break repair proteins XRCC1 and XRCC4 - PubMed https://pubmed.ncbi.nlm.nih.gov/15380105/ Synergistic decrease of DNA single-strand break repair rates in mouse neural cells lacking both Tdp1 and aprataxin - PubMed https://pubmed.ncbi.nlm.nih.gov/19303373/