Cell Lines AOA1 Publication

FD105

IDENTIFICATION

Causal gene(s)	APTX
Repeat size or mutation	pW279X/pW279X; sibling of FD106
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/ Aprataxin, poly-ADP ribose polymerase 1 (PARP-1) and apurinic endonuclease 1 (APE1) function together to protect the genome against oxidative damage - PubMed https://pubmed.ncbi.nlm.nih.gov/19643912/ 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/