Cell Lines AOA1 Publication

P1

IDENTIFICATION

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

SOURCE & PUBLICATIONS

Originating lab / institution	Columbia University College of Physicians and Surgeons
Links to publications or public resources	Coenzyme Q deficiency and cerebellar ataxia associated with an aprataxin mutation - PubMed https://pubmed.ncbi.nlm.nih.gov/15699391/ Lack of aprataxin impairs mitochondrial functions via downregulation of the APE1/NRF1/NRF2 pathway - PubMed https://pubmed.ncbi.nlm.nih.gov/25976310/ Familial cerebellar ataxia with muscle coenzyme Q10 deficiency - PubMed https://pubmed.ncbi.nlm.nih.gov/11294920/