Cell Lines AOA1 Publication

A1015

IDENTIFICATION

Causal gene(s)	APTX
Repeat size or mutation	pW279X/pQ181X
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	3
Age at collection (years)	27

SOURCE & PUBLICATIONS

Originating lab / institution	Istituto Neurologico Carlo Besta
Links to publications or public resources	Ataxia with oculomotor apraxia type1 (AOA1): novel and recurrent aprataxin mutations, coenzyme Q10 analyses, and clinical findings in Italian patients - PubMed https://pubmed.ncbi.nlm.nih.gov/21465257/ Lack of aprataxin impairs mitochondrial functions via downregulation of the APE1/NRF1/NRF2 pathway - PubMed