Cell Lines AOA1 Biobank

JUCTCi002-A

IDENTIFICATION

Causal gene(s)	APTX
Repeat size or mutation	c.837G > A (p.Trp279Ter) (homozygous)
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Female
Ethnicity	Jordanian
Age at collection (years)	27

SOURCE & PUBLICATIONS

Originating lab / institution	The University of Jordan
Links to publications or public resources	Generation and characterization of induced pluripotent stem cell (iPSC) line (JUCTCi002-A) from a patient with ataxia with oculomotor apraxia type 1 (AOA1) harboring a homozygous mutation in the APTX gene - PubMed https://pubmed.ncbi.nlm.nih.gov/32769066/