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# P1028

## IDENTIFICATION

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

## DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	6
Age at collection (years)	32

## SOURCE & PUBLICATIONS

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