Cell Lines FXTAS Publication

FMR1 conditional KO clone F17

IDENTIFICATION

Causal gene(s)	FMR1
Repeat size or mutation	hESC conditional FMR1 knockout
Cell type	hESC

DONOR INFORMATION

Donor gender	Male

SOURCE & PUBLICATIONS

Originating lab / institution	Stanford University School of Medicine
Links to publications or public resources	The fragile X mutation impairs homeostatic plasticity in human neurons by blocking synaptic retinoic acid signaling - PubMed https://pubmed.ncbi.nlm.nih.gov/30068571/ Cell-type-specific profiling of human cellular models of fragile X syndrome reveal PI3K-dependent defects in translation and neurogenesis - PubMed https://pubmed.ncbi.nlm.nih.gov/33852833/