# Compounds for Suppressing Fragile X Premutation rCGG Repreat-Mediated Toxicity

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## Technology description

**Technical Summary**

Fragile X syndrome (FXS) is the most common inheritable form of cognitive disability affecting nearly 1 in 4000 males and 1 in 6000 females. FXS usually results from the expansion of the CGG trinucleotide repeat in the 5' untranslated region (5' UTR) of the fragile X mental retardation 1 (FMR1) gene. Within the last decade, Fragile X-associated tremor/ataxia syndrome (FXTAS), a late age of onset neurodegenerative disorder, has been recognized among many FXScarriersin or beyond the fifth decade of life. Interestingly, 1 in every 800 males is a carrier and considered at high risk for developing FXTAS.

FXTAS is uncoupled from the neurodevelopmental disorder, FXS, and its most common clinical feature is a progressive action tremor with ataxia. More advanced or severe cases can show a progressive cognitive decline that ranges from memory deficits to dementia. Patients may also experience increased anxiety, mood liability, depression, muscle weakness, and numbness and/or pain in the lower extremities.

Dr. Peng Jin and his colleagues developed a dual assay screen - viability and behavioral - in the drosophila model of FXTAS and identified three compounds that rescued the FXTAS phenotype. Two of the compounds are phospholipase A2 (PLA2) inhibitors, which the researchers believe, based on the biological mechanisms of PLA2, can suppress rCGG-mediated neurodegeneration.

## Application area

Compounds for the treatment of Fragile X-associated tremor/ataxia syndrome.

## Advantages

11 known, FDA approved drugs identified that reduced rCGG-mediated lethality to a certain degree.
3 of these drugs also rescued locomotion deficits.

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