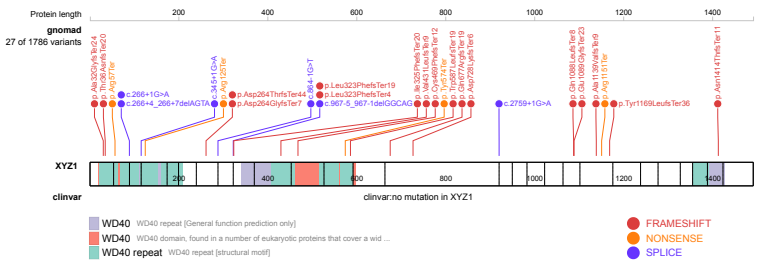
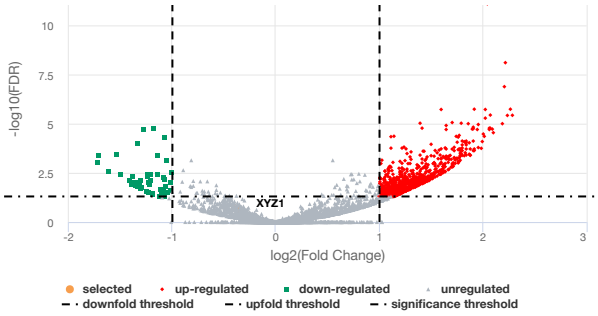
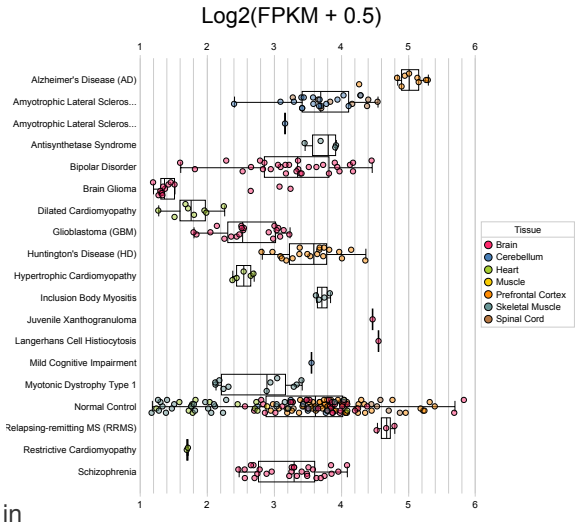
Investigating the potential of XYZ1 as a disease Z gene

XYZ1 is a member of the XYZ family, interacts with XYZ2 GDP/GTP exchange protein, and participates in calcium-dependent neurotransmitter release. A poster presentation at ASHG 2018 (M. Course, U. of Wash.) described discovery of a novel 69bp repeat region in an intron within the XYZ1 locus that in an expanded state segregated with disease Z diagnosis in one family. Here we investigate the potential of XYZ1 as a disease Z gene using applications available on http://bigdata.biogen.com.

A survey of >60K exomes reveals many loss of function variants in XYZ1.

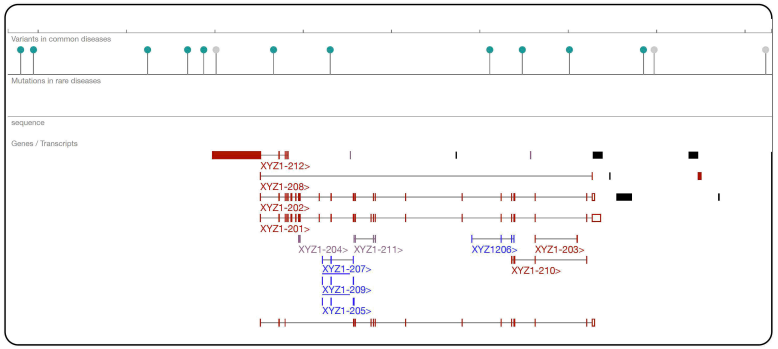




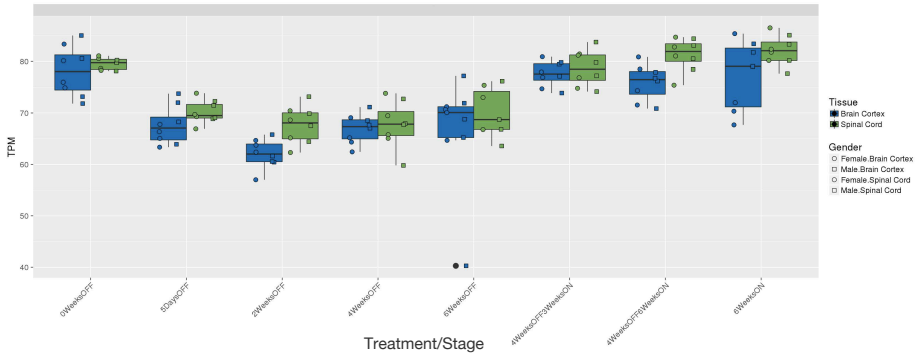


XYZ1 is not differentially expressed in disease Z motor neurons.

XYZ1 is modestly reduced during disease progression in a Z murine neurodegenerative model.



Variants at the 5' end of the XYZ1 locus are sub-associated with disease Z and other traits.



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