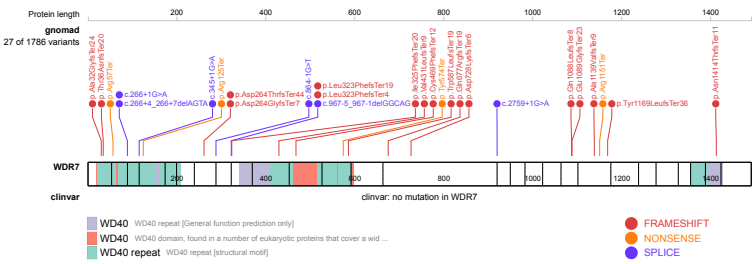
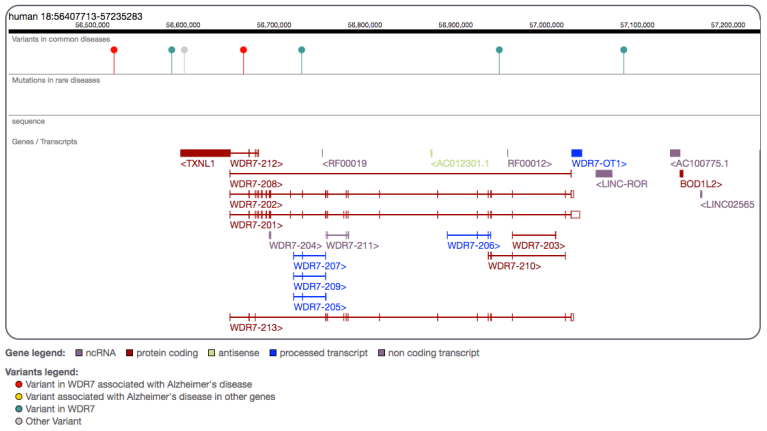
Investigating the potential of WDR7 as an ALS gene

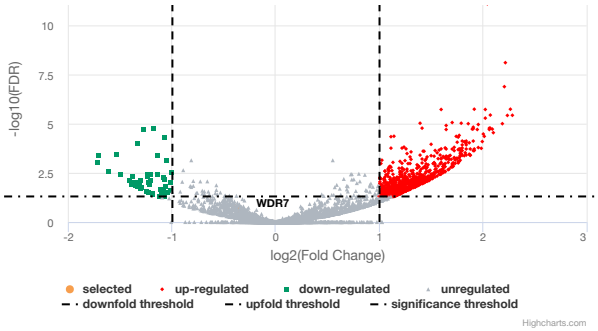
WDR7 is a member of the WDR family, interacts with Rab3A GDP/GTP exchange protein, and participates in calcium-dependent neurotransmitter release. A poster presentation at ASHG 2018 (M. Course, U. of Wash.) described discovery of a novel 69bp repeat region in an intron within the WDR7 locus that in an expanded state segregated with ALS diagnosis in one family. Here we investigate the potential of WDR7 as an ALS gene using applications available on Bigdata.biogen.com

A survey of >60K exomes reveals many loss of function variants in WDR7.

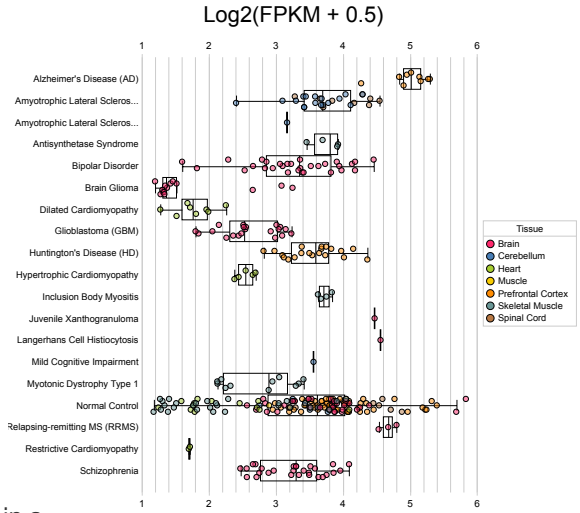




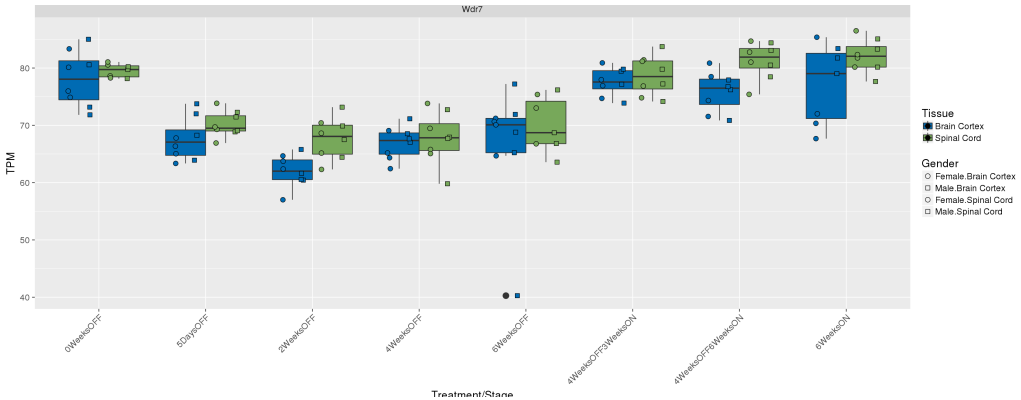
Variants at the 5' end of the WDR7 locus are sub-associated with AD and other traits.



WDR7 is not differentially expressed in ALS motor neurons.



Wdr7 is modestly reduced during disease progression in a TDP43 murine neurodegenerative model.



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and all contributors