LRRC7 – leucine rich repeat containing 7

“Predicted to be involved in several processes, including establishment or maintenance of epithelial cell apical/basal polarity; positive regulation of neuron projection development; and receptor clustering. Located in several cellular components, including centrosome; cytosol; and nucleoplasm”

“Biased expression in brain (RPKM 2.3)”

Lead SNP Chr1:69,692,599: rs12729671

- Intronic variant
- Correlated w/ expression of LRRC7 in 5 tissues (see “rs1272967_GTEx.csv”, this file has expression data from GTEx for this SNP, only kept entries with pval < 0.01)
- 57 SNV’s in a +- 50kb window that are high LD (R2 > 0.90) and correlated with expression of LRRC7 (see “rs1272967_LDExpress_90R2_50kb.csv”)

NCKAP5 – NCK associated protein 5

“Predicted to be involved in microtubule bundle formation and microtubule depolymerization. Predicted to be active in microtubule plus-end.”

“Broad expression in lung (RPKM 2.7), kidney (RPKM 0.7) and 20 other tissues”

Lead SNP Chr2:133,004,221: rs1437897

- Intronic Variant
- Correlated w/ expression of CTH and GPR39 in 2 tissues (see “rs1437897.csv”, GTEx expression data for this SNP, only kept entries with pval<0.01)
- LDExpress returned no variants in LD in GTEx

ATXN3—ataxin3

“Machado-Joseph disease, also known as spinocerebellar ataxia-3, is an autosomal dominant neurologic disorder. The protein encoded by this gene contains (CAG)n repeats in the coding region, and the expansion of these repeats from the normal 12-44 to 52-86 is one cause of Machado-Joseph disease.:

“Ubiquitous expression in skin (RPKM 4.6), testis (RPKM 3.0) and 25 other tissues”

BMP6 - bone morphogenetic protein 6

“This gene encodes a secreted ligand of the TGF-beta (transforming growth factor-beta) superfamily of proteins. Ligands of this family bind various TGF-beta receptors leading to recruitment and activation of SMAD family transcription factors that regulate gene expression. The encoded preproprotein is proteolytically processed to generate each subunit of the disulfide-linked homodimer. This protein regulates a wide range of biological processes including iron homeostasis, fat and bone development, and ovulation. Differential expression of this gene may be associated with progression of breast and prostate cancer. Mutations in this gene may be associated with iron overload in human patients.”

“Ubiquitous expression in placenta (RPKM 15.8), lung (RPKM 11.0) and 24 other tissues”