# Supplemental Data

## Linkage, Association, and Gene-Expression Analyses

## Identify *CNTNAP2* as an Autism-Susceptibility Gene

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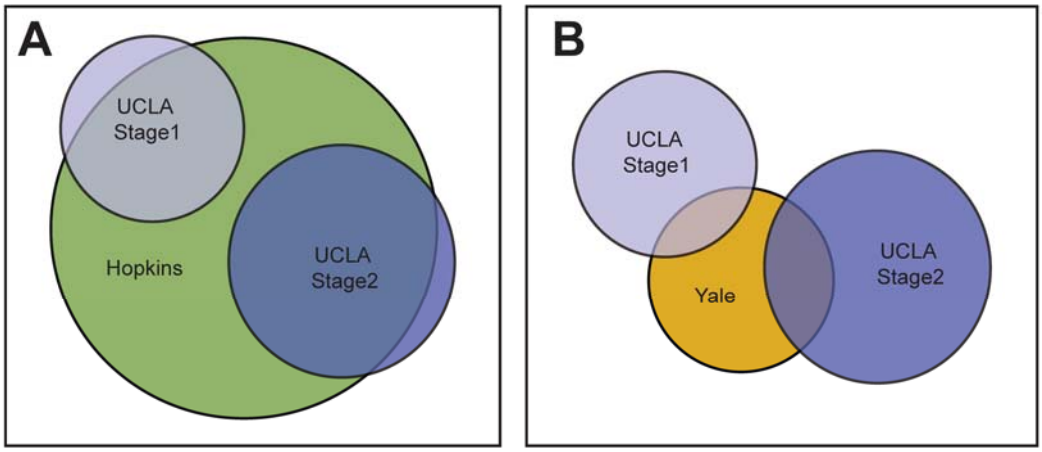
### Supplemental References

S1. Bakkaloglu, B., O'Roak, B.J., Louvi, A., Gupta, A.R., Abelson, J.F., Morgan, T.M., Chawarska, K., Klin, A., Ercan-Sencicek, A.G., Stillman, A.A., et al. (2008). Molecular cytogenetic analysis and resequencing of *Contactin Associated Protein-Like 2* in autism spectrum disorder. Am. J. Hum. Genet. *82*, this issue.

S2. Arking, D.E., Cutler, D.J., Brune, C.W., Teslovich, T.M., West, K., Ikeda, M., Rea, A., Guy, M., Lin, S., Cook, E.H., Jr., and Chakravarti, A. (2008). A common genetic variant in the neurexin superfamily member *CNTNAP2* increases familial risk of autism. Am. J. Hum. Genet. *82*, this issue.




C

| AGRE Family | Included in UCLA? | UCLA Stage | Functional Prediction |
|---|---|---|---|
| AU0358 | yes | Stage2 | Deleterious |
| AU0139 | yes | Stage1 | Deleterious |
| AU0140 | yes | Stage1 | Deleterious |
| AU0668 | yes | Stage1 | Deleterious |
| AU0284 | yes | Stage1 | Deleterious |
| AU0334 | yes | Stage1 | Deleterious |
| AU0176 | yes | Stage1 | Deleterious |
| AU0668 | yes | Stage1 | Benign |
| AU0116 | yes | Stage2 | Benign |
| AU0230 | yes | Stage1 | Benign |

**Figure S1. Sample Overlap for UCLA, Yale,[S1] and Hopkins[S2] Studies**

(A) UCLA vs. Yale

19.3% (326 / 1693) samples used in the UCLA analyses were present in the Yale cohort. Sample overlap for Stage1 and Stage2 analyses was comparable at 16.8%

(113 / 672) and 20.9% (213/1021), respectively. Only 7.1% (8 / 113) of UCLA Stage 1 families sequenced in the Yale study were identified as having a rare *CNTNAP2* coding variant. Similarly, under 1% (2 / 213) of Stage 2 families sequenced in the Yale study had rare coding variants. Together these results suggest that rare *CNTNAP2* coding variants do not in themselves explain the association with WORD.

(B) UCLA vs. Hopkins

86.9% (1472 / 1693) samples used in the UCLA analyses were present in the HOPKINS cohort. Sample overlap for Stage1 and Stage2 analyses was comparable at 83.2% (559 / 672) and 89.4% (913 / 1021), respectively.

(C) Only 7.1% (8/113) of UCLA Stage 1 families sequenced in the Yale study were identified as having a rare *CNTNAP2* coding variant. Similarly, under 1% (2/213) of Stage 2 families sequenced in the Yale study had rare coding variants. Together these results suggest that rare *CNTNAP2* coding variants do not in themselves explain the association with WORD.