

**Supplementary information**

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**Genomics, convergent neuroscience and progress in understanding autism spectrum disorder**

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| Gene     | FDR        | Relative risk | Labelled in Fig. 1d |
|----------|------------|---------------|---------------------|
| CHD8     | 0          | 66.441        | Yes                 |
| SCN2A    | 0          | 124.201       | Yes                 |
| SYNGAP1  | 0          | 82.536        | Yes                 |
| ADNP     | 8.52E-15   | 120.051       | Yes                 |
| FOXP1    | 1.77E-12   | 143.854       | Yes                 |
| POGZ     | 1.09E-10   | 46.41         | Yes                 |
| ARID1B   | 2.58E-10   | 51.783        | Yes                 |
| SUV420H1 | 5.48E-10   | 107.618       | Yes                 |
| DYRK1A   | 8.22E-10   | 85.431        | Yes                 |
| SLC6A1   | 1.92E-09   | 118.961       | Yes                 |
| GRIN2B   | 2.04E-08   | 42.031        | Yes                 |
| PTEN     | 5.26E-08   | 188.814       | Yes                 |
| SHANK3   | 1.72E-07   | 58.42         | Yes                 |
| MED13L   | 1.84E-06   | 26.361        | No                  |
| GIGYF1   | 3.51E-06   | 31.638        | No                  |
| CHD2     | 5.47E-06   | 30.286        | No                  |
| ANKRD11  | 9.55E-06   | 17.107        | No                  |
| ANK2     | 1.43E-05   | 25.049        | No                  |
| ASH1L    | 2.04E-05   | 23.949        | No                  |
| TLK2     | 2.76E-05   | 69.144        | No                  |
| CTNNB1   | 3.98E-05   | 42.403        | No                  |
| DEAF1    | 6.92E-05   | 83.815        | No                  |
| KDM6B    | 0.00010125 | 26.601        | No                  |
| DSCAM    | 0.00013466 | 19.563        | No                  |
| SETD5    | 0.00018404 | 31.636        | No                  |
| KCNQ3    | 0.00023649 | 48.359        | No                  |
| SRPR     | 0.00028816 | 64.745        | No                  |
| KDM5B    | 0.00034543 | 56.596        | No                  |
| WAC      | 0.00040864 | 57.925        | No                  |
| SHANK2   | 0.00054203 | 14.113        | No                  |
| NRXN1    | 0.00067145 | 27.027        | No                  |
| TBL1XR1  | 0.00087038 | 81.571        | No                  |
| DNMT3A   | 0.00110973 | 61.856        | No                  |
| MYT1L    | 0.0013663  | 32.829        | No                  |
| BCL11A   | 0.00162693 | 32.619        | No                  |
| RORB     | 0.00199239 | 75.748        | No                  |
| RAI1     | 0.00234804 | 15.727        | No                  |
| DYNC1H1  | 0.002727   | 10.535        | No                  |
| DPYSL2   | 0.00324328 | 49.032        | No                  |
| AP2S1    | 0.00388129 | 112.634       | Yes                 |
| KMT2C    | 0.00448884 | 20.93         | No                  |
| PAX5     | 0.00510723 | 50.641        | No                  |
| MKX      | 0.00593604 | 31.938        | No                  |
| GABRB3   | 0.00674915 | 23.363        | No                  |
| SIN3A    | 0.0075843  | 25.736        | No                  |
| MBD5     | 0.00845793 | 18.528        | No                  |
| MAP1A    | 0.00930941 | 0             | No                  |

Supplementary Table 1 | **Effect sizes of the 47 high-confidence ASD genes (FDR < 0.01) in Figure 1d.** Column guide: 'Gene', gene symbol; 'FDR', false discovery rate from Satterstrom et al. 2020; 'Relative risk', the ratio of the frequency of a given variant in cases versus unaffected controls, where the frequency in cases is the total number of de novo damaging variants observed in a given gene, and the frequency in controls is based on estimates of the number of mutations expected per generation; 'Labelled in Fig. 1d', indicates whether the gene is labelled in Fig. 1d or not.

## Reference

1. Satterstrom, F. K. *et al.* Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell* **180**, 568–584 (2020).