**Supplemental materials**

**Supplemental Table 1. Summary statistics for maternal and neonatal 25-hydroxyvitamin D models by affection status and ancestry and for the potential confounding factors used in the multivariate models.**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **summary statistics in GWAS#** | | | | |  | **summary statistics in GWAS+follow-up#** | |
|  | **dataset** |  | **N** | **ASD cases** | **controls** |  | **ASD cases** | **ID cases** |
|  | mothers |  | 768 | 379 | 389 |  | 507 | 173 |
|  | neonates |  | 748 | 377 | 371 |  | 507 | 179 |
| **trait** | **dataset** | **range** | **all** | **ASD cases** | **controls** |  | **ASD cases** | **ID cases** |
| vitamin D [nmol/L] | mothers | [9.8-259.0] | 91.0(35.8) | 90.5(33.3) | 91.5(38.1) |  | 89.8(32.0) | 88.0(35.7) |
|  | neonates | [9.4-309.4] | 90.1(39.8) | 90.6(38.1) | 89.6(41.5) |  | 90.0(39.3) | 91.5(41.4) |
|  | $CAU mothers | [31.5-259.0] | 105.8(38.1) | 101.0(34.8) | 111.0(40.9) |  | 100.0(32.6) | 105.8(44.4) |
|  | $HIS mothers | [21.8-206.4] | 82.9(31.1) | 83.4(30.1) | 82.5(32.0) |  | 83.0(29.5) | 87.1(31.5) |
|  | $EA mothers | [14.3-177.3] | 81.1(30.0) | 86.7(30.6) | 73.9(27.9) |  | 85.1(29.7) | 50.0(13.2) |
|  | $AA mothers | [9.8-259.0] | 75.3(29.3) | 65.1(17.8) | 84.7(35.1) |  | 95.3(45.5) | - |
|  | $SA mothers | [9.8-187.3] | 82.9(41.6) | 95.3(45.5) | 67.9(32.6) |  | 95.3(45.5) | - |
|  | $OTH mothers | [43.5-198.6] | 108.2(38.8) | 90.5(33.6) | 124.2(37.4) |  | 99.7(30.6) | 73.2(32.9) |
| **covariates tested^** | **multivariate model** | **range/categories** | **all** | **ASD cases** | **controls** |  | **ASD cases** | **ID cases** |
| offspring status | neonates | case/control | **-** | 377 | 371 |  | 507 | 179 |
| offspring sex | neonates | male/female | 82% M | 82% M | 82% M |  | 82% M | 58% M |
| parity | both | [0-5] | 0.9(1.0) | 0.7(0.9) | 1.03(1.04) |  | 0.75(0.92) | 1.22(1.26) |
| neonatal birth weight (g) | mothers | [1,085-5,897] | 3,434(558) | 3,462(547) | 3,407(568) |  | 3,420(565) | 3,139(701) |
| maternal weight (lbs) | mothers | [84-332] | 151.5(35.1) | 152.7(36.2) | 150.4(34.0) |  | 151.8(37.2) | 158.1(37.2) |
| neonatal birth year (yrs) | both | 2000 | 135(18%) | 64(17%) | 71(19%) |  | 93(18%) | 44(25%) |
|  | 2001 | 197(26%) | 98(26%) | 99(26%) |  | 134(27%) | 52(29%) |
|  | 2002 | 319(43%) | 164(44%) | 155(42%) |  | 208(41%) | 65(36%) |
|  | 2003 | 97(13%) | 51(13%) | 46(12%) |  | 72(14%) | 18(10%) |
| maternal blood draw season | mothers | dec-feb | 222(29%) | 115(30%) | 107(28%) |  | 146(29%) | 45(26%) |
|  |  | mar-may | 193(25%) | 95(25%) | 98(25%) |  | 124(24%) | 32(18%) |
|  |  | jun-aug | 143(19%) | 68(18%) | 75(19%) |  | 105(21%) | 52(31%) |
|  |  | sep-nov | 210(27%) | 101(27%) | 109(28%) |  | 132(26%) | 44(25%) |
| neonatal birth  season | neonates | dec-feb | 134(18%) | 67(18%) | 67(18%) |  | 102(20%) | 51(28%) |
|  |  | mar-may | 226(30%) | 117(31%) | 109(30%) |  | 153(30%) | 50(28%) |
|  |  | jun-aug | 216(29%) | 111(29%) | 105(28%) |  | 139(28%) | 38(21%) |
|  |  | sep-nov | 172(23%) | 82(22%) | 90(24%) |  | 113(22%) | 40(23%) |

# for quantitative traits, the mean(standard deviation) are shown; for qualitative traits, N is specified for each category

$ 35% Caucasian (CAU), 42% Hispanics (HIS), 15% East Asian (EA) 3% African American (AA), 3% South Asian (SA) and 3% other (OTH) individuals

^ the subset of genotyped individuals with non-missing vitamin D or specific covariate measurement has been considered in each summary statistic estimation.

**Supplemental Table 2. Meta-analysis of the top hits in Table 1 including homogeneous groups from – three populations (Caucasian, Hispanic and East Asian ancestry)**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Maternal rs4149056-*SLCO1B*1 effects on maternal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.12** | **761** | **-0.58** | | **0.12** | | | **2.7x10-7** |
| East Asian | 0.12 | 112 | -0.69 | | 0.34 | | | 0.04521 |
| Hispanic | 0.12 | 303 | -0.17 | | 0.21 | | | 0.4145 |
| Caucasian | 0.17 | 243 | -0.83 | | 0.20 | | | 4.6x10-5 |
| **meta-analysis** |  |  | **---** | |  | | | **5.4x10-5** |
| **Maternal rs11528045-*HTR7* effects on maternal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.20** | **763** | **0.53** | | **0.11** | | | **5.4x10-7** |
| East Asian | 0.32 | 112 | 0.31 | | 0.27 | | | 0.25 |
| Hispanic | 0.22 | 304 | 0.62 | | 0.16 | | | 1.4x10-4 |
| Caucasian | 0.10 | 243 | 0.63 | | 0.26 | | | 0.016 |
| **meta-analysis** |  |  | **+++** | |  | | | **8.1x10-6** |
| **Maternal rs6486205-*CYP2R1* effects on maternal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.35** | **764** | **0.4** | | **0.09** | | | **1.9x10-6** |
| East Asian | 0.38 | 112 | 0.18 | | 0.24 | | | 0.45 |
| Hispanic | 0.35 | 303 | 0.54 | | 0.13 | | | 7.1x10-5 |
| Caucasian | 0.36 | 245 | 0.51 | | 0.15 | | | 9.9x10-4 |
| **meta-analysis** |  |  | **+++** | |  | | | **6.6x10-7** |
| **Fetal rs4588-*GC* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.26** | **688** | **-0.62** | | **0.11** | | | **3.1x10-8** |
| East Asian | 0.24 | 85 | -0.90 | | 0.36 | | | 0.013 |
| Hispanic | 0.23 | 219 | -0.45 | | 0.19 | | | 0.016 |
| Caucasian | 0.32 | 177 | -0.43 | | 0.22 | | | *0.060* |
| **meta-analysis** |  |  | **---** | |  | | | **2.4x10-4** |
| **Fetal rs11690195-*MMADHC-RND3* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.42** | **692** | **-0.47** | | **0.10** | | | **2.3x10-6** |
| East Asian | 0.42 | 87 | -1.09 | | 0.32 | | | 1.1x10-3 |
| Hispanic | 0.40 | 221 | -0.16 | | 0.16 | | | 0.32 |
| Caucasian | 0.45 | 178 | -0.49 | | 0.21 | | | 0.02 |
| **meta-analysis** |  |  | **---** | |  | | | **1.3x10-3** |
| **Maternal rs9527875-*DIAPH3-PCDH17* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.30** | **692** | **0.59** | | **0.11** | | | **6.9x10-8** |
| East Asian | 0.19 | 87 | 1.24 | | 0.37 | | | 1.4x10-3 |
| Hispanic | 0.32 | 218 | 0.44 | | 0.17 | | | 8.6x10-3 |
| Caucasian | 0.30 | 175 | 0.60 | | 0.22 | | | 7.8x10-3 |
| **meta-analysis** |  |  | **+++** | |  | | | **7.8x10-6** |
| **Maternal rs72650824-*CXCL8-CXCL6* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.07** | **693** | **1.53** | | **0.3** | | | **6.7x10-7** |
| East Asian | 0.00 | 87 | NA | | NA | | | NA |
| Hispanic | 0.02 | 219 | 0.80 | | 0.54 | | | 0.14 |
| Caucasian | 0.04 | 175 | 1.12 | | 0.60 | | | *0.06* |
| **meta-analysis** |  |  | **?++** | |  | | | **7.8x10-6** |
| **Maternal rs205761 *MKLN1* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | **SE** | | ***P*** | | |
| **whole dataset** | **0.48** | **692** | **0.50** | **0.10** | | **7.6.10-7** | | |
| East Asian | 0.30 | 87 | 0.20 | 0.33 | | | 0.56 | |
| Hispanic | 0.48 | 219 | 0.54 | 0.15 | | 5.1x10-4 | | |
| Caucasian | 0.45 | 175 | 0.59 | 0.23 | | 9.2x10-3 | | |
| **meta-analysis** |  |  | **+++** |  | | **2.8x10-5** | | |
| **Maternal rs2541497 *NUBP1* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | **SE** | | ***P*** | | |
| **whole dataset** | **0.42** | **689** | **0.53** | **0.11** | | **1.4x10-6** | | |
| East Asian | 0.24 | 87 | 0.40 | 0.38 | | | 0.30 | |
| Hispanic | 0.32 | 217 | 0.28 | 0.19 | | 0.15 | | |
| Caucasian | 0.39 | 174 | 0.18 | 0.21 | | 0.38 | | |
| **meta-analysis** |  |  | **+++** |  | | **0.07** | | |
| **Maternal rs1550598 *DOK6* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | **SE** | | ***P*** | | |
| **whole dataset** | **0.38** | **691** | **0.53** | **0.11** | | **2.6x10-6** | | |
| East Asian | 0.12 | 87 | 0.53 | 0.55 | | | 0.34 | |
| Hispanic | 0.42 | 218 | 0.36 | 0.17 | | 0.04 | | |
| Caucasian | 0.44 | 174 | 0.39 | 0.22 | | 0.08 | | |
| **meta-analysis** |  |  | **+++** |  | | **5.3x10-3** | | |
| **Maternal rs7837124 *c8orf34* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | **SE** | | ***P*** | | |
| **whole dataset** | **0.02** | **691** | **1.61** | **0.34** | | **2.7x10-6** | | |
| East Asian | - | 87 | - | - | | | - | |
| Hispanic | 0.02 | 219 | 1.87 | 0.65 | | 4.2x10-3 | | |
| Caucasian | 0.03 | 175 | 0.59 | 0.58 | | 0.31 | | |
| **meta-analysis** |  |  | **? ++** |  | | **9.0x10-3** | | |
| **Maternal rs490379 *PKN2* effects on neonatal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | **SE** | | ***P*** | | |
| **whole dataset** | **0.11** | **691** | **0.75** | **0.16** | | **4.1x10-6** | | |
| East Asian | 0.06 | 87 | 1.46 | 0.65 | | | 0.03 | |
| Hispanic | 0.13 | 218 | 0.70 | 0.23 | | 2.3x10-3 | | |
| Caucasian | 0.12 | 175 | 0.74 | 0.31 | | 0.02 | | |
| **meta-analysis** |  |  | **+++** |  | | **2.5x10-5** | | |
| **Fetal rs17666424-*ACKR1* effects on maternal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.13** | **699** | **0.59** | | **0.13** | | | **2.9x10-6** |
| East Asian | 0.04 | 88 | -0.34 | | 0.70 | | | 0.62 |
| Hispanic | 0.16 | 226 | 0.65 | | 0.21 | | | 1.8x10-3 |
| Caucasian | 0.19 | 178 | 0.54 | | 0.22 | | | 0.02 |
| **meta-analysis** |  |  | **-++** | |  | | | **2.6x10-4** |
| **Fetal rs2336664-*STIMATE* effects on maternal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.44** | **698** | **0.42** | | **0.09** | | | **2.0x10-6** |
| East Asian | 0.33 | 88 | 0.13 | | 0.27 | | | 0.62 |
| Hispanic | 0.47 | 226 | 0.47 | | 0.16 | | | 3.3x10-3 |
| Caucasian | 0.40 | 178 | 0.53 | | 0.18 | | | 2.9x10-3 |
| **meta-analysis** |  |  | **+++** | |  | | | **8.1x10-5** |
| **Fetal rs7015627-*MED30* effects on maternal vitamin D** | | | | | | | | |
| **group** | **MAF** | **N** | **beta** | | **SE** | | | ***P*** |
| **whole dataset** | **0.12** | **695** | **0.66** | | **0.14** | | | **1.9x10-6** |
| East Asian | 0.15 | 88 | 0.66 | | 0.34 | | | *0.06* |
| Hispanic | 0.17 | 223 | 0.84 | | 0.20 | | | 2.8x10-5 |
| Caucasian | 0.06 | 177 | 0.30 | | 0.40 | | | 0.46 |
| **meta-analysis** |  |  | **+++** | |  | | | **9.2x10-6** |

**Supplemental Table 3. GWAS top SNPs or their proxies (*r2*>0.8) showing evidence for replication or functional evidence in ENSEMBL, GTEx and ENCODE databases.**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **GWAS top SNP or proxy (*r2*>0.8)** | **MAF** | **variant type** | **annotation** | ***top eQTL*- gene target - top tissue** | **reference** |
| **rs4149056** | 0.12 | top SNP missense | replicated | - | Jiang et al 2018 |
| **rs7896173** | 0.24 | intergenic (rs11528045 proxy) | ccRE | - | *EH37E0186003* |
| **rs6486205** | 0.35 | top SNP  intronic | - | - | - |
| **rs4588** | 0.26 | top SNP  missense | replicated | - | Jiang et al 2018 |
| **rs4588** | 0.26 | top SNP  missense | ccRE | *GC* | *EH37E1236527* |
| **rs11690195** | 0.42 | top SNP  intergenic | - | - | - |
| **rs9527875** | 0.30 | top SNP  intergenic | - | - | - |
| **rs72650824** | 0.07 | top SNP  intergenic | TF binding site prediction | *rs17202249* in moderate LD - predicted TF binding site | ENSEMBL |
| **rs205761** | 0.48 | top SNP  intronic | - | - | - |
| **rs2541497** | 0.42 | top SNP  intronic | - | - | - |
| **rs1550598** | 0.38 | top SNP  intronic | - | - | - |
| **rs7837124** | 0.02 | top SNP  intronic | - | - | - |
| **rs490379** | 0.11 | top SNP  intronic | - | - | - |
| **rs72713603** | 0.10 | intergenic (rs17666424 proxy) | ccRE | *FCER1A* | *EH37E0117796* |
| **rs2336664** | 0.44 | top SNP intronic | - | - | - |
| **rs2710314** | 0.41 | intronic (rs2336664 proxy) | ccRE | *SFMBT1-TMEM110* | *EH37E0659240* |
| **rs116366455** | 0.42 | top SNP  intergenic | promoter | *MED30* | *EH37E0974013* |

*Abbreviations:* eQTL: expression quantitative trait loci; ccRE*:* candidate *cis*-regulatory element

**Supplemental Table 4. Maternal and neonatal 25-hydroxyvitamin D multivariate models for potential confounding factors.**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  |  | **maternal vitamin D** |  | **neonatal vitamin D** |
| **covariate** |  | ***P*** |  | ***P*** |
| parity(nulliparous up to 5) |  | 0.07 |  | 0.05 |
| birth season\* |  | NS |  | 6.9x10-5 |
| birth year\*$ |  | 3x10-5 |  | 0.01 |
| PC 1-10 |  | <10-6 |  | <10-6 |
| birth weight |  | 0.05 |  | NS |
| blood draw season |  | 1.8x10-7 |  | NS |
| maternal weight |  | 2.7x10-7 |  | NS |
| offspring sex\* |  | NS |  | NS |
| offspring ASD outcome\* |  | NS |  | NS |

\* birth year, birth season, offspring sex and outcome were included to account for matched case-control sampling.

$Note: A positive association is observed between maternal vitamin D and birth year. Starting around the same time as this cohort, the Institute of Medicine (IOM) recommendations for vitamin D intake during pregnancy increased, as did the number of U.S. adults consuming daily vitamin D supplements [Rooney et al., JAMA 2017].

**Supplemental Table 5. Number of independent maternal and fetal SNPs for each candidate gene showing *P*<0.05 in association with maternal and neonatal 25-hydroxyvitamin D in array-genotyped EMA samples.**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  | **maternal vitamin D** | | |  | **neonatal vitamin D** | | |
| **gene** |  | **N maternal SNPs**  ***P*** ≤**0.05** |  | **N fetal SNPs  *P*** ≤**0.05** |  | **N maternal SNPs**  ***P*** ≤ **0.05** |  | **N fetal SNPs**  ***P*** ≤**0.05** |
| ***GC*** |  | 2 |  | 2 |  | 4 |  | 4 |
| ***VDR*** |  | 0 |  | 1 |  | 1 |  | 0 |
| ***CYP2R1*** |  | 2 |  | 1 |  | 1 |  | 1 |
| ***CYP24A1*** |  | 1 |  | 1 |  | 0 |  | 1 |
| ***LGMN*** |  | 0 |  | 1 |  | 2 |  | 0 |
| ***CYP27B1*** |  | 0 |  | 0 |  | 0 |  | 0 |
| ***DHCR7*** |  | 0 |  | 0 |  | 0 |  | 0 |
| ***CUBN*** |  | 3 |  | 2 |  | 3 |  | 1 |
| ***LRP2*** |  | 4 |  | 4 |  | 1 |  | 1 |

**Supplemental Table 6. Genetic association between 22 candidate SNPs and 25-hydroxyvitamin D** **in three models using the ASD/control discovery dataset (GWAS), the full ASD/ID/control dataset (including 131 reclassified ASD and 179 ID affected samples), and in the full ASD/control dataset (excluding 179 ID samples).**

|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **ASD/control discovery dataset**  **(GWAS)** | | | | **ASD/ID/control dataset**  **(GWAS + follow-up)** | | | | **full ASD/control dataset**  **(ASD+ ID-ASD+GP)** | | | |
| **SNP** | **N** | **beta** | **SE** | ***P*** | ***P*** | **N** | **beta** | **SE** | ***P*** | **N** | **beta** | **SE** |
| **rs11528045** | 763 | 0.53 | 0.11 | 5.4x10-7 | 1.7x10-3 | 1041 | 0.29 | 0.09 | 3.7x10-5 NS | 878 | 0.42 | 0.10 |
| **rs6486205** | 764 | 0.41 | 0.09 | 1.9x10-6 | 7.3x10-6 | 1036 | 0.34 | 0.07 | 8.8x10-7 | 879 | 0.40 | 0.08 |
| **rs4149056** | 761 | -0.58 | 0.12 | 2.7x10-6 | 8.0x10-6 | 1024 | -0.50 | 0.11 | 2.5x10-6 | 871 | -0.56 | 0.12 |
| **rs830958^** | 765 | 0.45 | 0.19 | 0.016 | 7.6x10-3 | 1004 | 0.43 | 0.16 | 0.044 | 868 | 0.36 | 0.18 |
| **rs117913124** | 764 | -1.59 | 0.35 | 5.7x10-6 | 2.3x10-4 | 1009 | -1.18 | 0.32 | 9.9x10-6 \* | 868 | -1.52 | 0.34 |
| **rs17666424** | 699 | 0.59 | 0.13 | 2.9x10-6 | 5.0x10-6 | 956 | 0.49 | 0.11 | 5.3x10-6 | 813 | 0.53 | 0.12 |
| **rs2336664** | 698 | 0.42 | 0.09 | 2.0x10-6 | 3.7x10-5 | 951 | 0.33 | 0.08 | 7.4x10-5 | 805 | 0.34 | 0.08 |
| **rs7015627** | 695 | 0.67 | 0.14 | 1.9x10-6 | 2.9x10-4 | 908 | 0.44 | 0.12 | 1.9x10-6 \* | 784 | 0.62 | 0.13 |
| **rs780827** | 698 | -0.26 | 0.10 | 7.2x10-3 | 6.2x10-3 | 953 | -0.22 | 0.08 | 5.5x10-3 | 807 | -0.24 | 0.09 |
| **rs4667591** | 689 | -0.39 | 0.12 | 9.9x10-3 | 2.2x10-3 | 926 | -0.31 | 0.10 | 1.4x10-3 | 809 | -0.35 | 0.11 |
| **rs56097748** | 693 | 0.90 | 0.29 | 2.1x10-3 | 1.1x10-3 | 984 | 0.85 | 0.26 | 1.6x10-3 | 833 | 0.88 | 0.28 |
| **rs62302166** | 692 | -1.02 | 0.29 | 4.5x10-3 | 2.0x10-3 | 936 | -0.79 | 0.25 | 1.5x10-4 | 812 | -1.05 | 0.28 |
| **rs705124** | 693 | 0.47 | 0.18 | 8.1x10-3 | 0.024 | 948 | 0.35 | 0.15 | 7.9x10-3 \* | 822 | 0.45 | 0.17 |
| **rs17139719** | 692 | 0.49 | 0.19 | 9.5x10-3 | 0.063 | 963 | 0.30 | 0.16 | 0.018 \* | 821 | 0.42 | 0.18 |
| **rs11690195** | 692 | -0.48 | 0.10 | 2.3x10-6 | 1.4x10-5 | 933 | -0.40 | 0.09 | 4.1x10-6 $ | 814 | -0.45 | 0.10 |
| **rs4588** | 688 | -0.63 | 0.11 | 3.1x10-8 | 1.0x10-9 | 943 | -0.61 | 0.10 | 4.0x10-10 | 811 | -0.66 | 0.10 |
| **rs490379** | 691 | 0.75 | 0.16 | 4.1x10-6 | 2.9x10-3 | 953 | 0.41 | 0.14 | 1.6x10-5 \* | 823 | 0.64 | 0.15 |
| **rs7837124** | 691 | 1.61 | 0.34 | 2.7x10-6 | 1.1x10-6 | 993 | 1.40 | 0.29 | 2.3x10-5 | 838 | 1.38 | 0.32 |
| **rs11574143** | 690 | -0.42 | 0.15 | 5.7x10-3 | 0.018 | 957 | -0.30 | 0.13 | 2.9x10-3 \* | 827 | -0.41 | 0.14 |
| **rs9527875** | 692 | 0.60 | 0.11 | 6.9x10-8 | 1.8x10-5 | 991 | 0.41 | 0.09 | 1.1x10-5 | 837 | 0.46 | 0.10 |
| **rs2541497** | 689 | 0.53 | 0.11 | 1.4x10-6 | 3.3x10-3 | 994 | 0.34 | 0.09 | 3.7x10-5 NS | 836 | 0.42 | 0.10 |
| **rs17202249^** | 692 | 0.85 | 0.20 | 2.6x10-5 | 0.034 | 952 | 0.37 | 0.17 | 1.8x10-3 | 819 | 0.58 | 0.18 |

*Abbreviations: GP: general population (controls)*

^ Since the SNPs rs2544385 and rs72650824 were not available in Taq-Man assay, two proxies, rs830958 and rs17202249, were genotyped for the follow-up analyses.

*\* empirical P<0.05; $ empirical P<0.1; NS empirical P>0.1.* Empirical *P* estimated for a subset of 10 SNPs with 10-fold decrease in magnitude in 'GWAS + follow up' dataset compared to 'GWAS' dataset, showing milder decrease in magnitude in 'ASD + ID-ASD + GP' dataset. Empirical *P* assessed via association analysis of 100 sets of 131 randomly sampled ID subjects (replacing 131 ID-ASD subjects) combined with array-typed ASD subjects and controls.

**Supplemental Table 7. Association between 22 SNPs and ASD or ID outcome using logistic regression adjusted for offspring sex and 10 inferred PCs.**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **ASD vs GP**  **maternal SNP** | | **ASD vs GP**  **fetal SNP** | | **ID vs GP**  **maternal SNP** | | **ID vs GP**  **fetal SNP** | |
| **SNP** | **OR** | ***P*** | **OR** | ***P*** | **OR** | ***P*** | **OR** | ***P*** |
| **rs11528045** | 1.00 | 0.97 | 1.19 | 0.19 | 0.81 | 0.27 | 1.09 | 0.65 |
| **rs6486205** | 0.97 | 0.76 | 1.08 | 0.50 | 1.32 | 0.07 | *1.42* | *0.04* |
| **rs4149056** | 1.08 | 0.64 | 0.95 | 0.73 | 0.76 | 0.28 | 1.02 | 0.92 |
| **rs830958** | 1.03 | 0.90 | 1.12 | 0.59 | *2.21* | *9.4x10-3* | 1.58 | 0.14 |
| **rs117913124** | 0.85 | 0.71 | 1.03 | 0.93 | 1.12 | 0.85 | NA | NA |
| **rs17666424** | 0.95 | 0.76 | *0.74* | *0.03* | 1.08 | 0.71 | 0.85 | 0.39 |
| **rs2336664** | 0.93 | 0.52 | 0.85 | 0.11 | 1.02 | 0.92 | 0.88 | 0.42 |
| **rs7015627** | 1.08 | 0.61 | 0.97 | 0.85 | 0.96 | 0.87 | 0.86 | 0.51 |
| **rs780827** | 0.99 | 0.92 | *1.30* | *0.02* | 1.03 | 0.85 | 1.18 | 0.32 |
| **rs4667591** | 0.90 | 0.37 | 0.84 | 0.14 | 1.15 | 0.43 | 1.26 | 0.18 |
| **rs56097748** | 0.77 | 0.36 | 0.72 | 0.25 | 1.71 | 0.14 | 1.03 | 0.94 |
| **rs62302166** | 0.63 | 0.10 | 0.72 | 0.26 | 1.01 | 0.97 | 1.15 | 0.73 |
| **rs705124** | 1.22 | 0.24 | 0.96 | 0.80 | 0.92 | 0.75 | 1.03 | 0.90 |
| **rs17139719** | 1.08 | 0.66 | 1.11 | 0.58 | 0.70 | 0.18 | 0.94 | 0.83 |
| **rs11690195** | 1.17 | 0.13 | 1.07 | 0.53 | 1.07 | 0.68 | 0.83 | 0.24 |
| **rs4588** | 0.92 | 0.49 | 0.93 | 0.50 | 0.74 | 0.08 | 0.73 | 0.09 |
| **rs490379** | *0.66* | *7.3x10-3* | 0.79 | 0.15 | 0.72 | 0.17 | 1.30 | 0.24 |
| **rs7837124** | 1.65 | 0.18 | 1.25 | 0.51 | 2.28 | 0.08 | 1.17 | 0.76 |
| **rs11574143** | 1.16 | 0.33 | 0.92 | 0.57 | *1.54* | *0.05* | 1.12 | 0.58 |
| **rs9527875** | 0.84 | 0.11 | 0.92 | 0.47 | 0.97 | 0.85 | 0.89 | 0.47 |
| **rs2541497** | 1.13 | 0.26 | 1.20 | 0.10 | 0.87 | 0.37 | 0.98 | 0.89 |
| **rs17202249** | 1.17 | 0.44 | *1.45* | *0.08* | 1.01 | 0.98 | *1.73* | *0.05* |

*Abbreviations: GP: general population (controls)*

NID=179 ; NASD=507 and NGP=379