**A genome-wide association study of obstructive heart defects among participants in the National Birth Defects Prevention Study**

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**Supplementary Table 1. Type of heart defect for each included case infant.**

|  |  |  |
| --- | --- | --- |
|  | **Discovery**† **(N = 666)** | **Replication**† **(N = 471)** |
| Right obstructive heart defects |  |  |
| Pulmonary atresia | 45 (6.76) | 19 (4.03) |
| Tricuspid atresia | 30 (4.50) | 11 (2.34) |
| Pulmonary valve stenosis | 286 (42.94) | 183 (38.85) |
| Tetrology of Fallot | 9 (1.35) | 2 (0.42) |
| Ebstein Malformation | 8 (1.20) | 2 (0.42) |
| Left obstructive heart defects |  |  |
| Coarctation of the aorta | 216 (32.43) | 172 (36.52) |
| Aortic stenosis | 89 (13.36) | 77 (16.35) |
| Hypoplastic left heart syndrome | 59 (8.86) | 37 (7.86) |
| Interrupted Aortic Arch | 0 (0) | 6 (1.27) |
| Missing phenotype data‡ | 3 (0.45) | 4 (0.85) |

† Data are presented as N (% of Discovery or Replication infants with this defect). Some infants were affected by more than one obstructive heart defect.

‡ All are coded for heart defects, but the specific obstructive heart defect code is not listed in available National Birth Defects Prevention Study clinical dataset

**Supplementary Figure 1. Manhattan and QQ plots for discovery TDT.** The red line in the Manhattan plot represents the genome-wide significance threshold (P < 5x10-8), and the blue line represents a suggestive threshold (P < 1x10-6).





**Supplementary Figure 2. Manhattan and QQ plots for replication TDT.** The red line in the Manhattan plot represents the genome-wide significance threshold (P < 5x10-8), and the blue line represents a suggestive threshold (P < 1x10-6).



**Supplementary Figure 3. Manhattan and QQ plots for combined TDT.** The red line in the Manhattan plot represents the genome-wide significance threshold (P < 5x10-8), and the blue line represents a suggestive threshold (P < 1x10-6).





**Supplementary Figure 4. Manhattan and QQ plots for discovery mother case-control GWAS.** The red line in the Manhattan plot represents the suggestive significance threshold (P < 1x10-6), and the blue line represents a relaxed threshold (P < 1x10-5).





**Supplementary Figure 5. Manhattan and QQ plots for replication mother case-control GWAS.** The red line in the Manhattan plot represents the suggestive significance threshold (P < 1x10-6), and the blue line represents a relaxed threshold (P < 1x10-5).





**Supplementary Figure 6. Manhattan and QQ plots for the mother case-control meta-analysis.** The red line in the Manhattan plot represents the suggestive significance threshold (P < 1x10-6), and the blue line represents a relaxed threshold (P < 1x10-5).





**Supplementary Figure 7. Manhattan and QQ plots for discovery infant case-control GWAS.** The red line in the Manhattan plot represents the suggestive significance threshold (P < 1x10-6), and the blue line represents a relaxed threshold (P < 1x10-5).





**Supplementary Figure 8. Manhattan and QQ plots for replication infant case-control GWAS.** The red line in the Manhattan plot represents the suggestive significance threshold (P < 1x10-6), and the blue line represents a relaxed threshold (P < 1x10-5).





**Supplementary Figure 9. Manhattan and QQ plots for infant case-control meta-analysis.** The red line in the Manhattan plot represents the suggestive significance threshold (P < 1x10-6), and the blue line represents a relaxed threshold (P < 1x10-5).



