**Table S1 The information of *UGT1A1* polymorphisms**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **dbSNP\_ID** | **Allele** | **chr** | **Chr. Position（****GRCh37）** | **Ref mRNA** | **SNP Property** | **Functional Change** | **MAF** a |
| rs3755319 | A/C | 2 | 234667582 | NM\_000463.2 | 5'-flanking | / | 0.30 |
| rs887829 | C/T | 2 | 234668570 | NM\_000463.2 | 5'-flanking | / | 0.13 |
| rs4148323 | G/A | 2 | 234669144 | NM\_000463.2 | nonsynon\_exon1 | p.Gly71Arg | 0.21 |
| rs6742078 | G/T | 2 | 234672639 | NM\_000463.2 | intron1 | / | 0.13 |
| rs6717546 | G/A | 2 | 234682119 | NM\_000463.2 | 3'-flanking | / | 0.26 |

a : Selected SNPs with minor allele frequency (MAF) greater than 5% was found in the HapMap-HCB [Han Chinese in Beijing, China] records from Hap Map database.

**Table S2 Hardy-Weinberg equilibrium evaluation of *UGT1A1* polymorphisms**

|  |  |  |  |
| --- | --- | --- | --- |
| **dbSNP\_ID** | **Group** | **Genotype count（frequency %）** | **HWE *P*** |
| rs3755319 |  | A/A | A/C | C/C | 0.28 |
|  | Controls | 123 (45.56) | 112 (41.48) | 35 (12.96) |  |
|  | Cases | 152 (42.58) | 161 (45.10) | 44 (12.32) |  |
| rs887829 |  | C/C | C/T | T/T | 0.14 |
|  | Controls | 214 (79.26) | 50 (18.52) | 6 (2.22) |  |
|  | Cases | 286 (80.11) | 63 (17.65) | 8 (2.24) |  |
| rs4148323 |  | G/G | G/A | A/A | 0.18 |
|  | Controls | 193 (71.48) | 67 (24.81) | 10 (3.70) |  |
|  | Cases | 263 (73.67) | 88 (24.65) | 6 (1.68) |  |
| rs6742078 |  | G/G | G/T | T/T | 0.24 |
|  | Controls | 212 (78.52) | 52 (19.26) | 6 (2.22) |  |
|  | Cases | 284 (79.55) | 64 (17.93) | 9 (2.52) |  |
| rs6717546a |  | G/G | G/A | A/A | 0.35 |
|  | Controls | 117 (43.49) | 115 (42.75) | 37 (13.75) |  |
|  | Cases | 143 (40.63) | 159 (45.17) | 50 (14.20) |  |

a : Five cases and one control failed to be genotyped.

**Table S3 Association between maternal *UGT1A1* polymorphisms and the risks of specific CHD subtypes**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Genotype** | **Septal defects**  | **aOR** a **(95%CI)** | **Conotruncal heart defects** | **aOR** a **(95%CI)** | **Right-sided obstructive malformations** | **aOR** a **(95%CI)** | **Left-sided obstructive malformations** | **aOR** a **(95%CI)** | **Anomalous pulmonary venous return** | **aOR** a **(95%CI)** |
| **rs3755319** |  |  |  |  |  |  |  |  |  |  |
| AA | 103 | Ref | 72 | Ref | 45 | Ref | 27 | Ref | 35 | Ref |
| A/C-C/C | 132 | 1.06 (0.72, 1.55) | 88 | 1.05 (0.68, 1.61) | 69 | 1.24 (0.77, 1.99) | 45 | 1.35 (0.75, 2.41) | 29 | 0.64 (0.35, 1.18) |
| **rs887829**  |  |  |  |  |  |  |  |  |  |  |
| CC | 193 | Ref | 127 | Ref | 88 | Ref | 61 | Ref | 51 | Ref |
| C/T-T/T | 42 | 0.87 (0.53, 1.42) | 33 | 0.99 (0.58, 1.67) | 26 | 1.02 (0.58, 1.81) | 11 | 0.61 (0.27, 1.37) | 13 | 0.86 (0.40, 1.86) |
| **rs4148323** |  |  |  |  |  |  |  |  |  |  |
| GG | 176 | Ref | 118 | Ref | 75 | Ref | 53 | Ref | 48 | Ref |
| G/A-A/A | 59 | 1.04 (0.68, 1.60) | 42 | 1.16 (0.72, 1.88) | 39 | 1.58 (0.95, 2.63) | 19 | 1.15 (0.61, 2.18) | 16 | 1.05 (0.53, 2.10) |
| **rs6742078** |  |  |  |  |  |  |  |  |  |  |
| GG | 192 | Ref | 125 | Ref | 88 | Ref | 62 | Ref | 51 | Ref |
| G/T-T/T | 43 | 0.83 (0.51, 1.36) | 35 | 0.99 (0.59, 1.67) | 26 | 0.94 (0.53,1.67) | 10 | 0.50 (0.21, 1.16) | 13 | 0.76 (0.35, 1.66) |
| **rs6717546** b  |  |  |  |  |  |  |  |  |  |  |
| GG | 95 | Ref | 68 | Ref | 42 | Ref | 32 | Ref | 28 | Ref |
| G/A-A/A | 137 | 0.88 (0.60, 1.27) | 91 | 0.88 (0.58, 1.35) | 71 | 1.18 (0.74, 1.87) | 40 | 0.80 (0.45, 1.43) | 36 | 0.85 (0.46, 1.57) |

aOR =adjusted odds ratio, CI=confidence interval

a : Adjusted for maternal age, gestational week, housing renovation, factory or landfill nearby, cooking at home, parental smoking or ETS exposure, maternal alcohol consumption, folic acid supplements.

b : Five cases and one control failed to be genotyped for *UGT1A1* rs6717546.

**Table S4** **The interaction between maternal PAHs exposure and UGT1A1 rs4148323 influencing specific CHD subtypes based on additive model**

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **rs4148323** | **PAHs exposure** | **Septal defects**  | **aOR** a **(95%CI)** | **Conotruncal heart defects** | **aOR** a **(95%CI)** | **Right-sided obstructive malformations** | **aOR** a **(95%CI)** | **Left-sided obstructive malformations** | **aOR** a **(95%CI)** | **Anomalous pulmonary venous return** | **aOR** a **(95%CI)** |
| GG | low | 17 (7.23) | Ref | 11 (6.88) | Ref | 6 (5.26) | Ref | 7 (9.72) | Ref | 4 (6.25) | Ref |
|  | high | 159 (67.66) | 2.17 (1.11, 4.21) | 107 (66.88) | 2.21 (1.04, 4.70)  | 69 (60.53) | 2.55 (1.00, 6.49)  | 46 (63.89) | 1.58 (0.61, 4.09) | 44 (68.75) | 2.53 (0.80, 8.05) |
| G/A-A/A | low | 10 (4.26) | 1.07 (0.39,2.98) | 5 (3.13) | 1.00 (0.29, 3.46) | 5 (4.39) | 1.65 (0.43, 6.29) | 0 (0.00) | - | 1 (1.56) | 0.62 (0.61, 6.30) |
|  | high | 49 (20.85) | 2.39 (1.13,5.04) | 37 (23.63) | 2.78 (1.20, 6.45) | 34 (29.82) | 4.29 (1.58,11.64) | 19 (26.39) | 2.33 (0.82, 6.63) | 15 (23.44) | 2.99 (0.84, 10.63) |

aOR =adjusted odds ratio, CI=confidence interval

a : Adjusted for maternal age, gestational week, housing renovation, factory or landfill nearby, cooking at home, parental smoking or ETS exposure, maternal alcohol consumption, folic acid supplements.