**Development and validation of a diagnostic algorithm for Down syndrome using birth certificates and International Classification of Diseases codes**

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**Table S1**: Children who were suspected of having DS by an indication for DS on the birth certificate and/or with at least one ICD diagnosis for DS, born 2000-2017, enrolled in TennCare during infancy, and ever/never sought care at VUMC.

|  |  |  |  |
| --- | --- | --- | --- |
|  | Ever sought care at VUMC | | P value |
| Yes (N=411) | No (N=1041) |  |
| Qualified for suspected DS |  |  |  |
| Having at least one ICD code specific for DS | 411 (100%) | 1041 (100%) | 1 |
| DS coded on birth certificate | 102 (24.8%) | 162 (15.6%) | <.001 |
| Algorithm defined as having DS | 354 (86.1%) | 726 (69.7%) | <.001^ |
| Criterion\* 1 only | 1 (0.2%) | 4 (0.4%) |  |
| Criterion 1 & 3 only | 34 (8.3%) | 38 (3.7%) |  |
| Criterion 2 only | 3 (0.7%) | 1 (0.1%) |  |
| Criterion 2 & 3 only | 63 (15.3%) | 113 (10.9%) |  |
| Criterion 3 only | 253 (61.6%) | 570 (54.8%) |  |
| Algorithm defined as *not* having DS | 57 (13.9%) | 315 (30.3%) |  |

\*Criterion 1: Karyotype confirmed DS; Criterion 2: Karyotype pending DS + 2 ICD coded visits; Criterion 3: 3 ICD coded visits with first and last visits separately by at least 30 days apart.

^P value comparing algorithm defined DS and not DS by ever/never sought care at VUMC

**Table S2**: Maternal and infant characteristics of children with suspected DS, born 2000-2017, enrolled in TennCare during infancy, and ever/never sought care at VUMC.

|  |  |  |  |
| --- | --- | --- | --- |
|  | Ever sought care at VUMC | | P value |
| Yes (411) | No (1041) |  |
| **Maternal characteristics** |  |  |  |
| Age at delivery (n=1450) | 31 (23, 38) | 27 (22, 36) | <.001 |
| Education (n=1446) |  |  | 0.007 |
| Some high school or less | 108 (26.3%) | 305 (29.4%) |  |
| High school graduate | 133 (32.4%) | 394 (38.0%) |  |
| At least some college education | 169 (41.2%) | 337 (32.5%) |  |
| Residence (n=1451) |  |  | <.001 |
| Urban | 134 (32.6%) | 279 (26.8%) |  |
| Suburban | 136 (33.1%) | 249 (23.9%) |  |
| Rural | 141 (34.3%) | 512 (49.2%) |  |
| Married (n=1451) | 241 (58.6%) | 456 (43.8%) | <.001 |
| Maternal smoking during pregnancy (n=1446) | 72 (17.6%) | 236 (22.8%) | 0.03 |
| Prenatal care started at first trimester (n=1341) | 264 (69.3%) | 679 (70.7%) | 0.60 |
| Parity (n=1441) |  |  | 0.61 |
| Primiparous | 123 (30.4%) | 341 (32.9%) |  |
| 2 | 116 (28.6%) | 277 (26.7%) |  |
| 3+ | 166 (40.9%) | 418 (40.3%) |  |
| Delivery method |  |  | 0.03 |
| Vaginal/assisted | 217 (52.8%) | 617 (59.3%) |  |
| Cesarean section | 194 (47.2%) | 424 (40.7%) |  |
|  |  |  |  |
| **Infant characteristics** |  |  |  |
| Male sex | 224 (54.5%) | 586 (54.7%) | 0.80 |
| Race and Ethnicity |  |  |  |
| Non-Hispanic White | 209 (50.9%) | 430 (41.3%) | <.001 |
| Non-Hispanic Black | --\* | 186 (17.9%) |  |
| Hispanic | -- | 153 (14.7%) |  |
| Other | -- | 272 (26.1%) |  |
| Gestational age in weeks (n=1448) | 38 (36, 39) | 38 (36, 39) | 0.26 |
| Birth weight in grams | 2920 (2495, 3280) | 2892 (2410, 3260) | 0.15 |
| Small-for-gestational-age at 10th percentile (n=1446) | 59 (14.4%) | 151 (14.6%) | 0.93 |
| Singleton birth | 404 (98.3%) | 1014 (97.4%) | 0.31 |
| One or more older siblings (n=1441) | 283 (69.7%) | 694 (67.1%) | 0.33 |
| Congenital heart disease | 343 (83.5%) | 689 (66.2%) | <.001 |
| Birth year |  |  |  |
| 2000-2004 | 74 (18.0%) | 295 (28.3%) | <.001 |
| 2005-2009 | 118 (28.7%) | 301 (28.9%) |  |
| 2010-2017 | 219 (53.3%) | 445 (42.7%) |  |

\*Cells were suppressed for n<11