



# Algorithm to Identify Children with Down Syndrome

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# Goal of Study

- Our goal is to develop and validate an algorithm that accurately identifies individuals with Down syndrome using administrative data.

# Administrative Claims Data is an Efficient Approach Studying Down Syndrome

- People with Down syndrome can be identified using International Classification of Diseases (ICD) diagnosis codes
- It allows longitudinal follow up to study health conditions over time

# Using ICD Codes Alone in Case Identification can be Unreliable

- ICD codes are used for billing purposes and not designed for research
- Thus, they are subject to biases that limit their validity in identifying Down syndrome
- ICD codes that are more likely to be reimbursed by payors are more likely to be documented compared to those that are not.
- Physicians often use ICD codes that are associated with the comorbidity that they are treating rather than the underlying genetic process.

# Birth Certificates in Tennessee

- Down syndrome is noted on birth certificates in Tennessee
- Prior to 2004, there was just one check box on the birth certificate to indicate whether or not the child had Down syndrome. This was based on genetic testing or clinical suspicion.
- Starting in 2004, the birth certificates had 2 check boxes; one indicated “karyotype confirmed” Down syndrome and the other indicated “karyotype pending” Down syndrome

# Objective

- The objective of this study was to develop an algorithm using birth certificates and administrative claims data (ICD codes) to correctly identify individuals with Down syndrome, and then to validate the algorithm using manual chart review. If an accurate algorithm could be identified, it could then be used to identify individuals with Down syndrome for future research studies.

# Methods

- We used data from the Tennessee Medicaid Program (TennCare) and identified children who were born between 2000-2017 who were continuously enrolled in TennCare during their first year of life. Their record was linked to their birth certificate.
- We identified children as “Suspected of having Down syndrome” by having Down syndrome marked on their birth certificate OR having at least one ICD code for Down syndrome in their Medicaid claims data anytime during their first 6 years of life
- We then narrowed this group down to those patients who ever received care at Vanderbilt University Medical Center

# Algorithm

Children were defined as having Down syndrome if they met one of the following criteria:

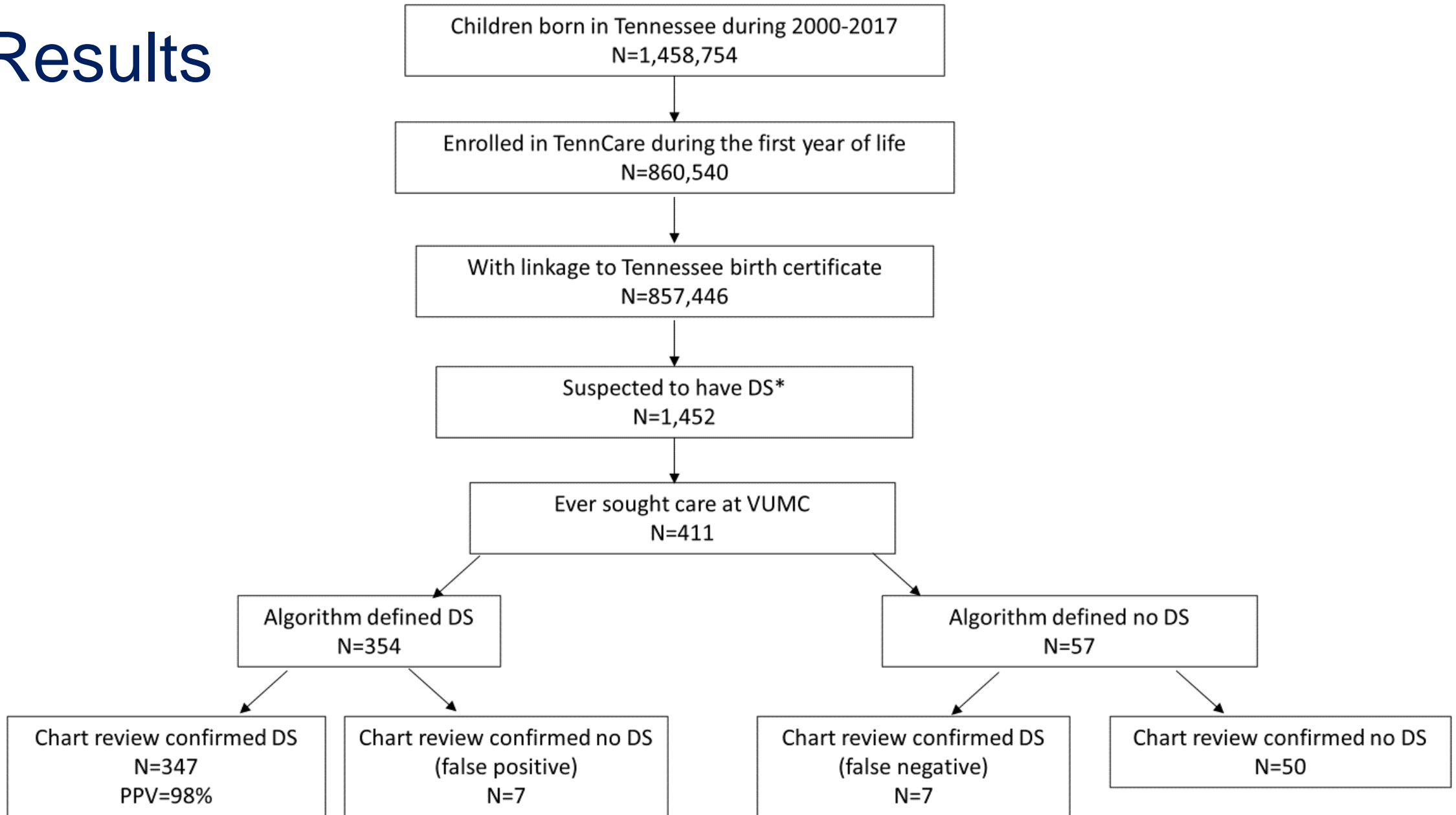
1. Having birth certificate indication for “karyotype confirmed” Down syndrome
2. Having birth certificate indication for “karyotype pending” Down syndrome or just Down syndrome if test type was not specified (i.e., prior to 2004) *and* having at least two healthcare encounters for Down syndrome during the first six years of life
3. Having at least three healthcare encounters for Down syndrome during the first six years of life, with the first and last encounter separated by at least 30 days



# Statistical Analysis

- Among individuals who the algorithm defined as having Down syndrome, we calculated the Positive Predictive Value (PPV) with manual chart review confirmed Down syndrome as the gold standard.
- All analysis were performed using R software version 4.3.1.

# Results



# Results

<b>Maternal characteristics</b>	Children with suspected Down syndrome (N=411)
<b>Age at delivery</b>	31 (23, 38)
<b>Education (n=410)</b>	
Some high school or less	108 (26.3%)
High school graduate	133 (32.4%)
At least some college education	169 (41.2%)
<b>Residence</b>	
Urban	134 (32.6%)
Suburban	136 (33.1%)
Rural	141 (34.3%)
<b>Married</b>	241 (58.6%)
<b>Smoking during pregnancy (n=409)</b>	72 (17.6%)
<b>Prenatal care started at first trimester (n=381)</b>	264 (69.3%)
<b>Parity (n=405)</b>	
Primiparous	123 (30.4%)
2	116 (28.6%)
3+	166 (40.9%)
<b>Delivery method</b>	
Vaginal/assisted	217 (52.8%)
Cesarean section	194 (47.2%)

# Results

<b>Infant characteristics</b>	<b>Children with suspected Down syndrome (N=411)</b>
<b>Male sex</b>	224 (54.5%)
<b>Non-Hispanic White</b>	209 (50.9%)
<b>Gestational age in weeks</b>	38 (36, 39)
<b>Birth weight in grams</b>	2920 (2495, 3280)
<b>Small-for-gestational-age at 10<sup>th</sup> percentile (n=410)</b>	59 (14.4%)
<b>Singleton birth</b>	404 (98.3%)
<b>One or more older siblings (n=406)</b>	283 (69.7%)
<b>Congenital heart disease</b>	343 (83.5%)
<b>Birth year</b>	
2000-2004	74 (18.0%)
2005-2009	118 (28.7%)
2010-2017	219 (53.3%)

# Results:

Criterion 1 (karyotype confirmed Down syndrome)	Criterion 2 (karyotype pending DS or just DS <i>and</i> $\geq 2$ ICD diagnosis for Down syndrome)	Criterion 3 ( $\geq 3$ ICD diagnosis for Down syndrome)	Study population - children with suspected Down syndrome (N=411)	Children with chart review confirmed Down syndrome (N=354)	Children with chart review confirmed no Down syndrome (N=57)	PPV
X			1	1	0	100.0%
X		X	34	34	0	100.0%
	X		3	3	0	100.0%
	X	X	63	63	0	100.0%
		X	253	246	7*	97.2%
			57	7^	50	NA

\*False positives.

^ False negatives

# Discussion and conclusions

- Using birth certificate data and ICD diagnosis data, we developed an algorithm to accurately determine children with Down syndrome.
  - Of the 411 children who were suspected to have DS and sought care at an academic medical center, our algorithm defined 86.1% as having Down syndrome.
  - The algorithm is accurate in differentiating suspected Down syndrome children with and without Down syndrome, with 98.0% and 87.3% accuracy, respectively, in determining those truly with Down syndrome and those truly without Down syndrome.
- ICD coding alone can be an efficient and valid approach in Down syndrome case identification.
  - 350 (98.9%) children met ICD coding only criterion
  - Of the 253 children who exclusively met ICD coding only criterion, 97.2% had Down syndrome confirmed by chart review

# Discussion and conclusions

- Birth certificate as a source in Down syndrome case identification was less sensitive but accurate
  - 24.8% (n=102) had Down syndrome coded on the birth certificate
  - 101 out of 102 had their Down syndrome identified by algorithm and confirmed by chart review