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Characterization of Comorbidities in Patients with a Dual Diagnosis of Down Syndrome and Autism Spectrum Disorder Using Cerner Health Facts

Michael Slogic
Children's Mercy Hospital, mjslogic@cmh.edu

Earl F. Glynn
Children's Mercy Hospital, efglynn@cmh.edu

Cy Nadler
Children's Mercy Hospital, cnadler@cmh.edu

Meredith Dreyer
Children's Mercy Hospital, mldreyer@cmh.edu

Sarah T. Edwards
Children's Mercy Hospital, sedwards1@cmh.edu

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Michael J Slogic, MD; Earl Glynn, MS; Sarah Edwards, DO; Cy Nadler, PhD; Meredith Dreyer Gillette, PhD; Hung-Wen Yeh, PhD

Disclosures

- None

Background

- Up to 15% of patients with Down syndrome (DS) will be diagnosed with autism spectrum disorder (ASD)[1]
- Limited information on comorbidities in patients with this dual diagnosis
- Comorbidities affect the cognitive and behavioral profile[2-4]
- Medical databases provide the opportunity to isolate relatively large sample sizes for rare diagnoses

Aim of the Study

- To characterize the medical and psychological comorbidities from an organ system-based perspective in patients with a dual diagnosis
- To compare the prevalence of these comorbidities to patients with ASD or DS alone
- Utilize Cerner Health Facts, a large medical data warehouse, to facilitate this comparison

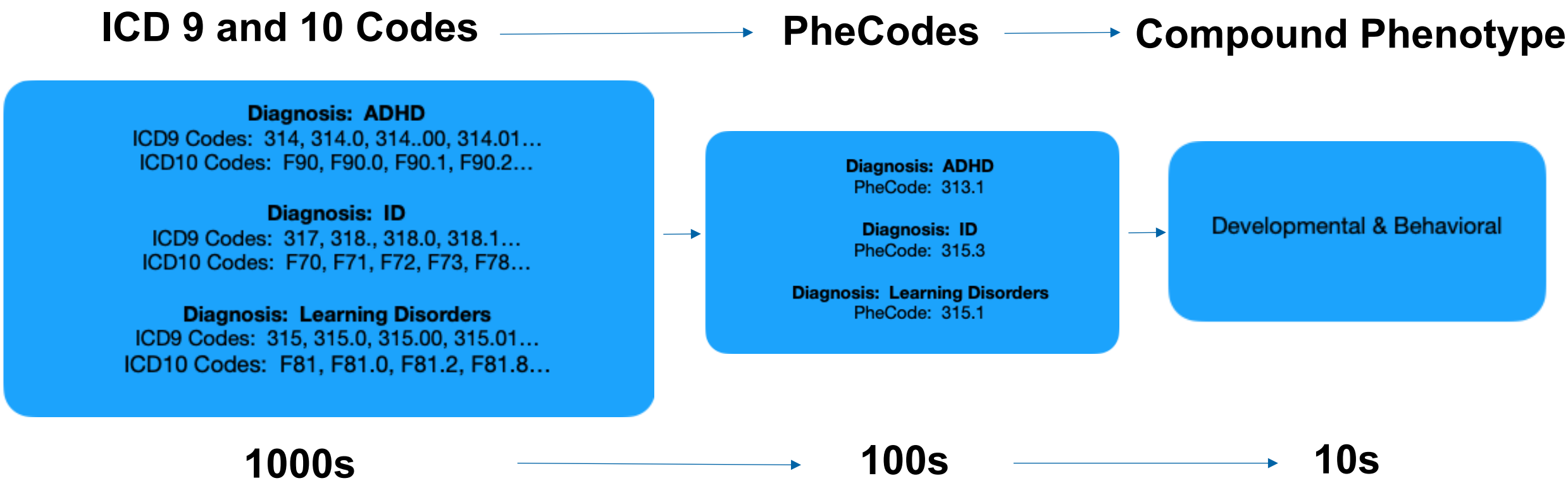
Methods: Cerner Health Facts

- Medical Data Warehouse[5]
  - Over 68 million de-identified patients
  - Over 500 million encounters
  - 100 Health Systems
  - Over 600 medical facilities
- Composed of ICD9/ICD10 diagnoses

Methods

- Inclusion criteria
  - Birth to <19 years
  - Diagnosis of DS, ASD, or dual diagnosis
  - All ICD-9/10 diagnostic codes for those with diagnoses above were extracted
- ICD-9/10 codes for ASD and DS were removed for comparisons[6-8]

Methods[6-8]


ICD 9 and 10 Codes
PheCodes
Compound Phenotype
Diagnosis: ADHD
ICD9 Codes: 314, 314.0, 314..00, 314.01…
ICD10 Codes: F90, F90.0, F90.1, F90.2…
Diagnosis: ID
ICD9 Codes: 317, 318., 318.0, 318.1…
ICD10 Codes: F70, F71, F72, F73, F78…
Diagnosis: Learning Disorders
ICD9 Codes: 315, 315.0, 315.00, 315.01…
ICD10 Codes: F81, F81.0, F81.2, F81.8…
Diagnosis: ADHD
PheCode: 313.1
Diagnosis: ID
PheCode: 315.3
Diagnosis: Learning Disorders
PheCode: 315.1
Developmental & Behavioral
1000s
100s
10s

Statistical Comparison

- Prevalence and prevalence ratios for 32 compound phenotypes for DS, ASD, and dual diagnosis
- Effect sizes and p-values calculated comparing all three cohorts
- P-values, and odds ratios for each comparison (Dual vs DS, Dual vs ASD, DS vs ASD)
  - Logistic regression models adjusted for several factors (age, duration in months across encounters, sex, race, urban/rural setting, census region, and teaching facility)
- Focus on moderate (0.5) or greater effect sizes given large sample size and multiple comparisons

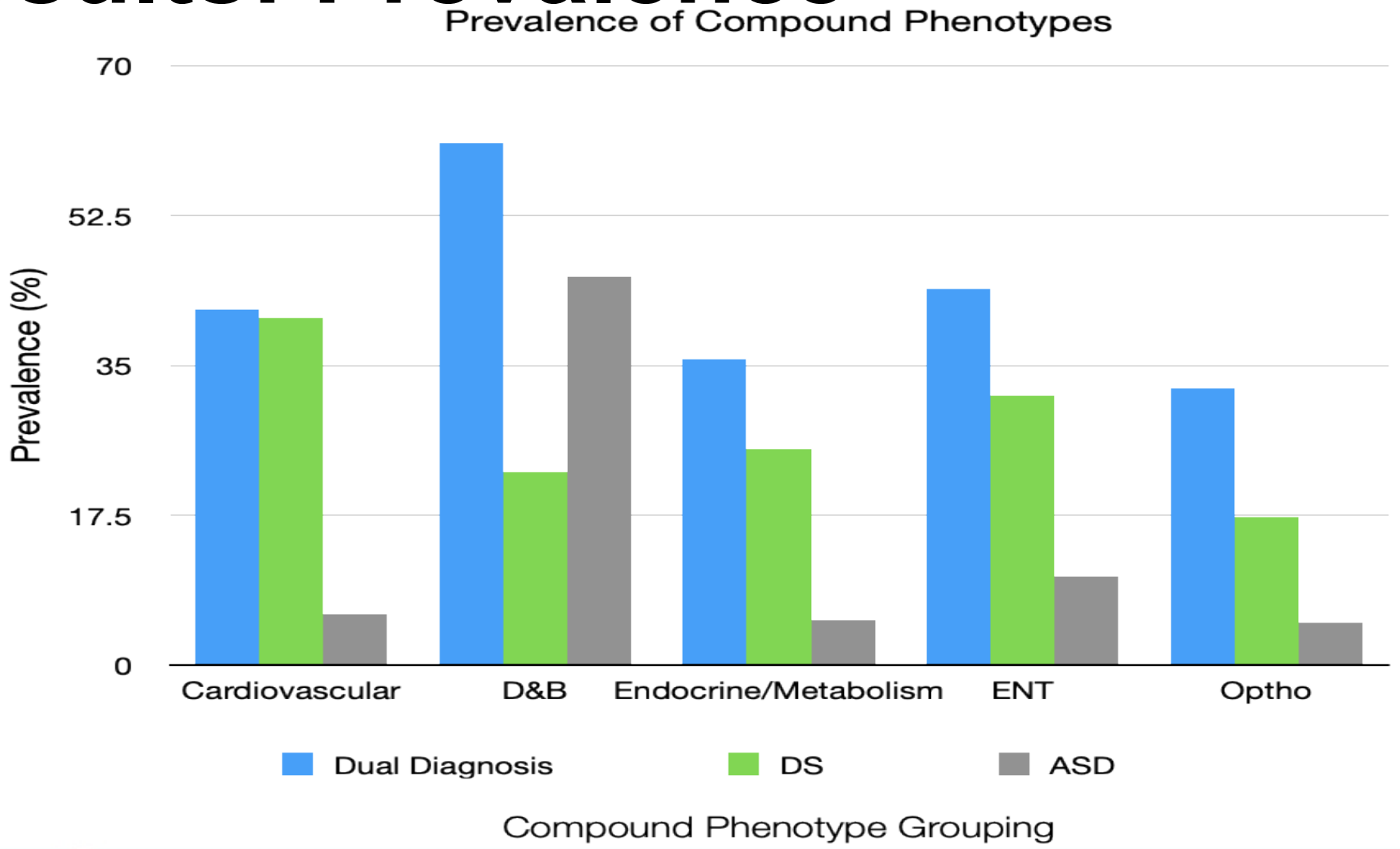
Results: Demographics

Population (%)	Dual Diagnosis (N=1,075)	DS (N=21,187)	ASD (N=97,181)
Sex			
Male	65.3	53.4	77.8
Female	34.7	46.5	22.1
Race/Ethnicity			
Caucasian	54.3	52.2	60.7
African American	11.4	12.5	14.9
Hispanic	3.2	3.9	2.4
Other	31.1	31.4	21.9

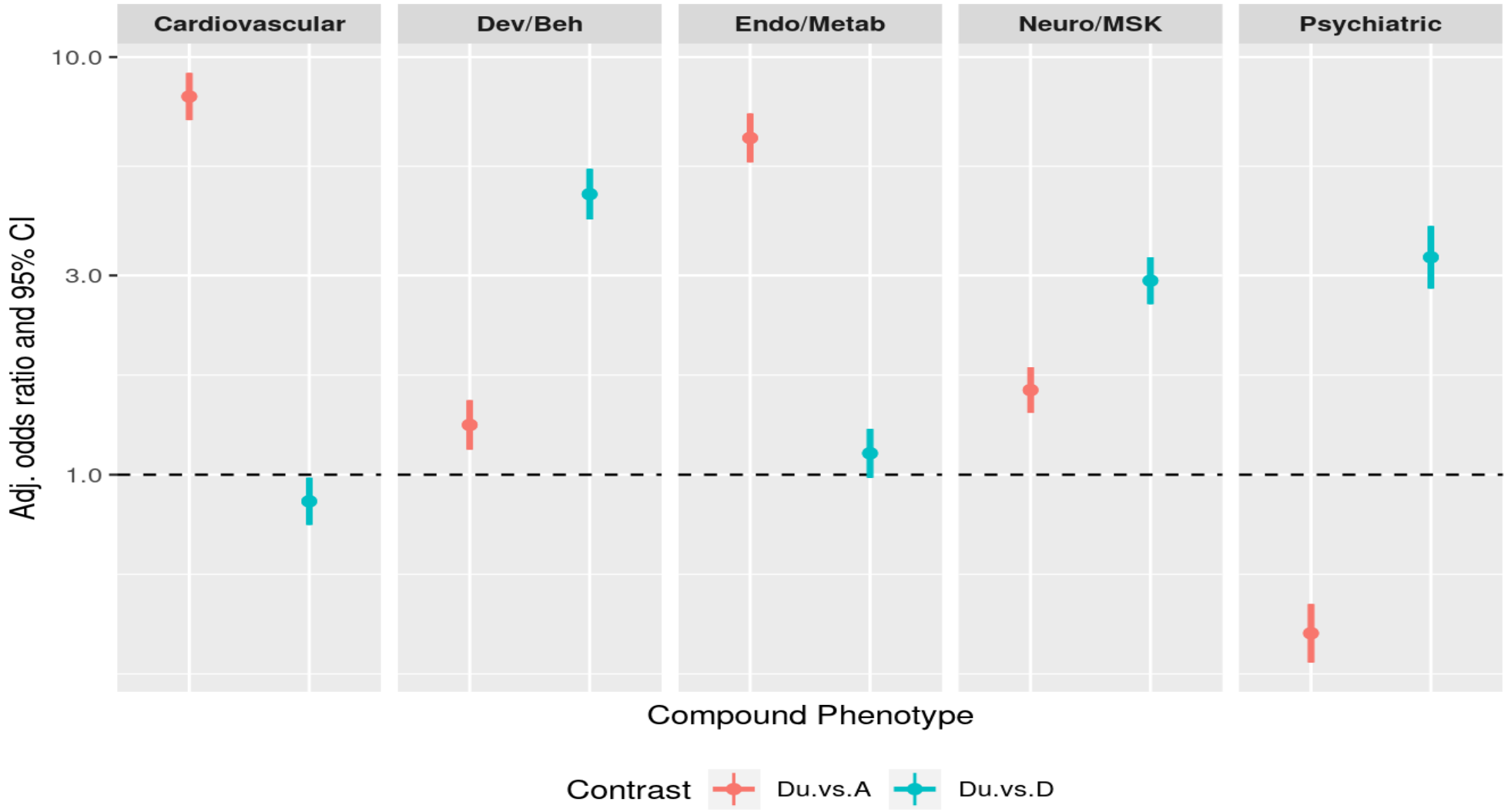
Results: Prevalence and Effect Size

Compound Phenotype	Dual Diagnosis Prevalence (%)	DS Prevalence (%)	ASD Prevalence (%)	Effect Size
Cardiovascular	41.5	40.6	6	0.61
Developmental/Behavior	61	22.6	45.4	0.553
Endocrine and Metabolism	35.8	25.3	5.2	0.544
ENT	43.9	31.5	10.4	0.535
Ophthalmologic	32.4	17.3	5	0.502
Pulmonology and Sleep	41.8	35.4	11.7	0.479
Psychiatric	21.2	4.8	27.8	0.437
GI	43.7	28.5	15.3	0.434
Neurologic and Musculoskeletal	50.6	21.1	28	0.427
Dental	24.8	7.4	5.4	0.379

Results: Prevalence



Results: Logistic Regression



Discussion

- For all compound phenotypes
  - Dual diagnosis > DS
  - Dual diagnosis > ASD*
    - *Except psychiatric diagnoses
- Five compound phenotypes showed significant relationships with the three populations
  - Cardiovascular, D&B, Endo and Metabolism, ENT, and Ophthalmologic

Discussion

- For dual diagnosis compared to DS
  - Increased prevalence of Neuro/MSK, Developmental/Behavioral, and Psychiatric compound phenotypes
- For dual diagnosis compared to ASD
  - Multiple compound phenotypes showed significantly increased prevalence
    - Not psychiatric compound phenotype
  - Developmental/Behavioral OR 1.15

Discussion

- These direct comparisons between three of the populations allows for contextual comorbidity comparison
  - Dual diagnosis *medical comorbidity* appears roughly equivalent to DS alone, but much more complex compared to ASD alone
  - Dual diagnosis *psychiatric comorbidity* appears to be much more compared to DS alone, but less complex than ASD alone

Discussion

- Limitations (and advantages) of Cerner Health Facts approach
  - Sample size
  - Low precision (no context about how patient received diagnoses)
- Pressing research questions
  - Why does the ASD diagnosis confer an increased risk of neurologic disorders?

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