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

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Characterization of Comorbidities in Patients with a Dual Diagnosis of Down Syndrome and Autism Spectrum Disorder

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\textsuperscript{6-8}

ICD 9 and 10 Codes $\rightarrow$ PheCodes $\rightarrow$ 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 $\rightarrow$ 100s $\rightarrow$ 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

<table>
<thead>
<tr>
<th>Population (%)</th>
<th>Dual Diagnosis (N=1,075)</th>
<th>DS (N=21,187)</th>
<th>ASD (N=97,181)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Sex</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>65.3</td>
<td>53.4</td>
<td>77.8</td>
</tr>
<tr>
<td>Female</td>
<td>34.7</td>
<td>46.5</td>
<td>22.1</td>
</tr>
<tr>
<td><strong>Race/Ethnicity</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Caucasian</td>
<td>54.3</td>
<td>52.2</td>
<td>60.7</td>
</tr>
<tr>
<td>African American</td>
<td>11.4</td>
<td>12.5</td>
<td>14.9</td>
</tr>
<tr>
<td>Hispanic</td>
<td>3.2</td>
<td>3.9</td>
<td>2.4</td>
</tr>
<tr>
<td>Other</td>
<td>31.1</td>
<td>31.4</td>
<td>21.9</td>
</tr>
</tbody>
</table>
## Results: Prevalence and Effect Size

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

Prevalence of Compound Phenotypes

- Cardiovascular
- D&B
- Endocrine/Metabolism
- ENT
- Optho

Prevalence (%)

- Dual Diagnosis
- DS
- ASD

Compound Phenotype Grouping
Results: Logistic Regression

<table>
<thead>
<tr>
<th>Compound Phenotype</th>
<th>Cardiovascular</th>
<th>Dev/Beh</th>
<th>Endo/Metab</th>
<th>Neuro/MSK</th>
<th>Psychiatric</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adj. odds ratio and 95% CI</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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?
References


