Research Days   GME Research Days 2021

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

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☐**Medical Student**
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IRB Number: 17080492

Describe role of Submitting/Presenting Trainee in this project (limit 150 words):
The submitting author served as the primary investigator for the project. He developed the clinical question and objectives for the project. He identified Cerner Health Facts as a data source and coordinated initial meetings with the data scientist to begin the project. He also completed the background research, identified the phenotype code methodology from this background reading, and developed the concept of combining phenotype codes into compound groupings. He worked with the other authors to finalize the compound phenotype classification system as well.   He coordinated research meetings with the other members of the team. He will also serve as the presenter and develop supplementary materials to facilitate the presentation.

<u>**Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words**</u>

Background:
Up to 19% of patients with Down syndrome (DS) meet diagnostic criteria for autism spectrum disorder (ASD) (Channell, et al, 2019). While the medical and psychological comorbidities for patients with DS or ASD are well characterized, comorbidities and outcomes for patients with a dual diagnosis (DS-ASD) are poorly understood. A large cohort of patients with DS-ASD, as well as those with ASD and DS alone, are needed to better understand this. Methods for grouping and analyzing complex diagnostic phenotypes are also needed.

Objectives/Goal:
Our objective is to utilize Cerner Health Facts, a multi-institutional healthcare database, to identify large populations with DS, ASD, and DS-ASD. This will allow for characterization and comparison of their ICD9/10 diagnoses. A secondary objective is the development of a higher order classification system based on ICD9/10 diagnoses to allow for identification of meaningful differences in body system dysfunction across populations.

Methods/Design:
Patients birth to 18 years with at least one encounter in Cerner Health Facts and diagnoses of ASD, DS, or DS-ASD were identified. Medical and psychological diagnoses in the form of ICD9/10 codes were extracted and combined into phenotype codes (Denny, et al, 2013; Wu, et al, 2019). Phenotype codes were then grouped by physiologic system into compound phenotypes. Prevalence rates for these compound phenotypes were then computed and compared across the DS, ASD, and DS-ASD samples.

Results:
1,087 patients with DS-ASD, 22,862 patients with DS, and 98,979 patients with ASD were identified. Thirty-two compound phecode groupings were developed from 1,886 phecodes. Prevalence rates for each of these compound phenotype groupings were calculated. As an example, 47.9% of DS-ASD patients were noted to have diagnoses in the Pulmonology/Sleep grouping, similar to those with DS. However, Pulmonology/Sleep diagnoses were over three times more prevalent compared to those with ASD. In the DS-ASD population, Neurologic/Musculoskeletal diagnoses were nearly 2.5 times more prevalent compared to those with DS, and over 1.75 times more prevalent when compared to those with ASD.

Conclusions:
Patients with DS-ASD had higher rates of a wide range of medical and psychological diagnoses compared to those with DS or ASD alone. The compound phenotype classification scheme is a viable method for comparing diagnoses between distinct populations, as well as aggregating differences to produce interpretable phenotypic trends. These trends can both inform clinical practice and provide the basis for future work, such as investigating the link between mortality and comorbidities in those with DS-ASD or determining if a relationship exists between medical comorbidities and behavioral challenges in the DS-ASD population.