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Perceived Stress and Access to Care in Parents of Children Living a Diagnostic Odyssey in Puerto Rico

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OBJECTIVES/GOALS: Diagnostic odyssey is the time it can take to a patient for receiving a diagnosis. Diagnostic process in rare diseases can be complex due to the heterogeneity of symptoms and lack of access to care. We aim to evaluate the association between diagnostic odyssey, perceived stress, and access to care, in parents of Puerto Rican patients with a rare disease. METHODS/STUDY POPULATION: We propose a cross-sectional study in parents of 100 children who received an uninformative whole exome sequencing (WES) report during a scheduled appointment with their geneticist. Discussion of WES results during clinical session, followed by a Perceived Stress Scale (PSS-10) and semi-structured interview to explore the experience of access to care during the diagnostic process will be arranged. Observation and interviews will be recorded. Data analysis and descriptive statistics will be calculated using STATA. Statistical associations (OR) will be estimated using generalized linear models at a 5% significance level. RESULTS/ANTICIPATED RESULTS: We expect to find high perceived stress in parents of Puerto Rican pediatric individuals having rare diseases, especially among single mothers. We will be able to identify limited access to care in Puerto Rico, especially in the testing pre-authorization process and long waits for geneticist appointments. Demand for advanced diagnostics is above the number of medical geneticists available in Puerto Rico, which triggers delayed diagnosis, management, and counseling. Therefore, these could affect the health disparities in our population with rare diseases. DISCUSSION/SIGNIFICANCE: This descriptive study will evaluate perceived stress in parents of pediatric patients living a diagnostic odyssey in Puerto Rico. No study has described perceived stress and access to care in this Hispanic population with undiagnosed conditions. Findings will contribute to a deep understanding of diagnostic process and limited access to care.

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Predictors of Substance Use Initiation by Late Childhood: Findings from the Adolescent Brain Cognitive Development (ABCD) Study

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OBJECTIVES/GOALS: Adolescence represents a critical period for substance use initiation. Various factors may contribute to trying a sip or single puff of a substance, that could lead to more frequent use. However, less is known about how predictors from multiple domains converge to impact risk for general substance use initiation. METHODS/STUDY POPULATION: The Adolescent Brain Cognitive Development (ABCD) study is a multi-site longitudinal study following youth into early adulthood. The present study included 7,644 ABCD children who reported no lifetime substance use (including any experimentation) at baseline (ages 9–10). Our primary aim was to use a random forest classification model to predict binary substance use initiation, defined as trying any non-prescribed substance (e.g., alcohol, tobacco, cannabis, non-prescribed medications), during a 2-year follow-up after baseline. A total of 402 variables from the following categories were examined as predictors: demographics, peer substance use and availability, mental and physical health, culture and environment, biospecimens, neurocognitive functioning, and structural neuroimaging variables. RESULTS/ANTICIPATED RESULTS: Over a two-year follow-up, 751 (9.8%) of substance-naÃ¯ve children reported trying a substance by age 11. The most common substance was alcohol, followed by cannabis and tobacco. Mean Decrease Accuracy (MDA) values were used to assess the relative importance of each predictor. The overall accuracy of the model in accurately predicting group membership (no substance use initiation vs. substance use initiation) was 57.66%. Of the top 5 predictors, the most important predictor was intent to use alcohol (MDA = .002). The following top predictors were structural neuroimaging variables: volume and surface area of right lateral occipital lobe (MDA = .0009 and .0008, respectively), surface area of right inferior temporal lobe (MDA = .0007), and surface area of left superior frontal lobe (MDA = .0007). DISCUSSION/SIGNIFICANCE: A combination of intent to use alcohol and structural neuroimaging indices were among the top predictors of substance use initiation. Understanding predictors of early substance use experimentation is important for identifying at-risk youth that may require targeted intervention approaches.

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Radiopathomics: Integration of Advanced Neuroimaging and Molecular Pathology Features in Meningiomas

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OBJECTIVES/GOALS: Our overall objective is to investigate the relationship between radiologic features of meningioma with recently identified histopathological and molecular biomarkers, and to apply a machine learning (ML) approach to further demonstrate their utility in predicting clinical outcomes. METHODS/STUDY POPULATION: We have enrolled a cohort of 84 patients with meningioma diagnosed on the basis of conventional gadolinium-enhanced MRI imaging features since September 2019. Each patient has demographic and clinical data, Ga-68-DOTATATE MRI/PET SUV and dynamic metrics, DCE-MRI perfusion parameters, and histopathologic data. Various tumor subregions will be segmented semi-automatically and later confirmed by experienced neuroradiologist. Histopathologic data will include histologic grade, mitotic rate, Ki67 proliferative index, and presence of WHO established atypical histologic features, immunohistochemical

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