R. Confounders included ancestry, BMI, age, sex, and 6 cell types. We tested both scores in our sample: smokers and non-smokers.
Results: In contrast to our expectations, none of the regression analyses showed a significant association with depression (MADRS-score). Nonetheless, a significant association was seen with biological sex for both analysis (overall: p=0.036, non-smokers: p=0.026). A reduced model with only this predictor explained 5% and 4% of the variance of the summary score calculated (R^2), respectively (overall: p=0.013; non-smokers: p=0.019). One of the ancestry components was marginally significant too in the non-smoker summary score (p=0.065). This was not the case anymore in the reduced model.
Conclusions: Our results show that caution is still in place when using methylation risk scores as specificity and sensitivity might not yet be optimized. The score built for depression incidence does not seem fitting for depression severity at this moment. The use of DNA methylation, a marker that is generally sensitive to confounding factors, for a risk score, might pose more challenges in the context of reliable summary statistics, in particular also for cross-trait examination, which is currently a typical use of polygenic risk scores.

Disclosure of Interest: None Declared

EPV0537

Behavioral and neurocognitive phenotypes in Crigler-Najjar syndrome in Tunisia

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Introduction: Crigler-Najjar 1 (CN1) due to exon 3 mutations of the UGT1A1 gene is a not rare genetic disease in Tunisia with a founder effect. CN1 syndrome is very severe, and most of CN1 Tunisian patients die soon after birth, within a maximum of one year, due to kernicterus. Liver transplantation, which is the only available therapeutic method for CN1, remains unreachable.
Objectives: The aim of this study was to report behavioral and neurocognitive phenotypes in CN1 patients who survived to school enrollment.
Methods: We have selected all patients evaluated from 2004 to 2010, both clinically and molecularly, for a deficiency of bilirubin-UGT enzyme activity leading to a pathological elevation of unconjugated bilirubin with a suspicion of CN1 syndrome. Direct sequencing of targeted PCR amplification products was performed for molecular analysis of UGT1A1. Behavioral and mental features of patients were studied through our genetic counselling.
Results: We identified 15 patients with the homozygous c.1070 A>G Tunisian mutation. Their age at diagnosis ranged from one week to 9 months for 13 patients. Six of them died within a month of molecular investigation. Only two boys were of school age, i.e. 6 and 9 years. The first had been hospitalized at 3 months year-old for a prolonged jaundice treated with phenobarbital and phototherapy. His psychomotor and neurological development was normal, with school attendance at the age of six. The second patient presented with an unexplored jaundice at the age of 3 days, which was later complicated by seizures and treated with phenobarbital. Despite neurological and motor sequelae associated to language impairments with slurred speech, he attended school at the age of six.
Conclusions: The neurological and behavioral profile of CN1 patients depends on familial and medical management. Quick diagnosis, close follow up and early liver transplantation can improve prognosis.

Disclosure of Interest: None Declared

EPV0540

Interaction analysis of monoaminergic polymorphisms and childhood environment related to personality functioning in patients with Borderline Personality Disorder

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Introduction: Neurobiological studies have shown that genetic variations affecting the intensity of monoamine neurotransmission play an important role in aggressive behavior and borderline personality traits. Also, the effect of family environment has been repeatedly shown on aggressive behavior and interpersonal functioning. Population-based longitudinal studies pointed out interactions between the so-called monoaminergic sensitivity alleles and childhood adversities.
Objectives: Our study aimed to analyze the associations between the most studied variable number tandem repeats of monoaminergic genes and the different psychological factors in adult patient and healthy control groups, checking for the moderating effects of the parental occupation and education, childhood abuse and trauma.
Methods: The recruited 73 patients with BPD diagnosis and 98 healthy controls were assessed by the Structured Clinical Interview for DSM-5. Participants filled out online questionnaires including the Level of Personality Functioning Scale – short version (LPFS-SR) and the Buss-Perry Aggression Questionnaire (BPQ). Childhood social environment and traumatic experiences were assessed by the Barratt Simplified Measure of Social Status and the Early Trauma Inventory or the Childhood Trauma Questionnaire. Genomic DNA samples were obtained either from peripheral blood, saliva or buccal swabs using the desalting technique. Functional dopaminergic and serotonergic polymorphisms were chosen based on previous findings, implicating them as sensitivity gene variants, e.g., the variable-number tandem-repeats of the dopamine D4 receptor, serotonin transporter and the monoamine oxidase-A (MAO-A) genes. Since the MAO-A gene is located on the X chromosome, sex-stratified analyses were also carried out.
Results: Family environment indexed by the Barratt Simplified Measure Social Status had significant effect on anger, hostility and interpersonal functioning ($p < 0.01$). In the pooled sample of patients and controls, individuals carrying the high activity alleles of MAOA had elevated scores on the BPQ subscales. When analysis