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cortex, hippocampus, hypothalamus, dorsal striatum, nucleus Accumbens, ventral tegmental area and amygdala) using quantitative PCR and ELISA methods in the different groups of mice (ad libitum, ad libitum with wheel, food restriction and food restriction with wheel). Statistical analysis will compare the measures for different samples by one-way or two-way ANOVAs depending the group of animals or brain regions and blood.

**Results:** To date, no difference of the level of transcription for *Bdnf* was observed between the different groups of mice (ad libitum, ad libitum with wheel, food restriction and food restriction with wheel) in the prefrontal cortex, hippocampus and hypothalamus. We expect significant differences of Bdnf expression in the other brain regions of interest for the food restricted animals with or without the wheel compared to ad libitum animals. We expect also differences in the level of expression of Bdnf in fasted animals compared to the refeed animals.

**Conclusions:** The BDNF could represent a potential biomarker of AN for the diagnostic and the prognosis in the evolution to the remission when weight recover and thus will allow a better understanding of the aetiology of AN. This study is supported by Fédération pour la Recherche sur le Cerveau.

Disclosure of Interest: None Declared

#### **EPP0458**

# Behavioral signs of CHARGE syndrome and CHD7 mutational spectrum

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**Introduction:** CHARGE syndrome is a genetic entity caused by mutations in the chromodomain helicase DNA-binding protein 7 gene (CHD7) at 8q12.1. There are pleiotropic signs among individuals with this disorder. Diagnosis is clinical using medical criteria. CHD7 gene mutations are usually found in 90% of affected patients.

**Objectives:** The aim of this study was to report behavioral signs of CHARGE syndrome and their phenotype-genotype correlations.

**Methods:** Four Tunisian males from Sfax (Tunisia) with clinical features suggestive of CHARGE syndrome were examined at our genetic counselling at the medical University of Sfax. Assessment of facial dysmorphic and behavioral features, karyotyping using RHG banding and molecular screening of CHD7 mutations were performed. Molecular analysis was made using direct Sanger sequencing of the entire CHD7 gene.

Results: Molecular genetic analysis revealed two deletions of the CHD7 gene at exon 3 for the first patient and at exon 8 for the second. The two genetic alterations were associated to retarded growth development and genital hypoplasia. Sensory impairments included for the first visual defects and for the second auditory and olfactory defects. Besides constant delayed psychomotor development, the two patients shared receptive and expressive communication disorders, anxiety, attention deficit, cognitive impairment and intellectual disability. There were no aggressive traits nor major autistic features. Learning disabilities were also present for the two patients.

**Conclusions:** The CHD7 gene controls the developmental pathways as a transcriptional regulator in the nucleoplasm through chromatin organization. Mutational alterations lead according to the affected domains, and the structure of the nonfunctional CHD7 protein, to the perturbation of the regulation of the developmental pathways' genes expression. CHD7 is demonstrated as an important component of neurogenesis through two neuronal determination factors: Sox4 and Sox11. While nonsense, frameshift and missense mutations are most common, deletions and duplications are less frequent. Moreover, while exon 3 is commonly altered, mutations of exon 8, which is related to the CHD7 protein chromodomain, are very rare. Phenotype-genotype correlations according to the type of genomic alteration of CHD7 gene are rarely published, particularly concerning behavioral and psychological features of CHARGE association. Here, physical disorders of our two patients seem to be different but behavioral features seem to be common. Multidisciplinary care is thus required for CHARGE syndrome and molecular analysis must be indicated because the type of the genomic alterations may be a key step for a more accurate management of physical and behavioral disorders.

Disclosure of Interest: None Declared

### **EPP0460**

"... wise, amazed, temp'rate, and furious, Loyal and neutral, in a moment": first heritability analysis of affective temperaments reports remarkably high SNP-based heritability

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Introduction: Depression shows a moderate heritability of 37-42%, which can be up to 75% in severely depressed samples 75%. At the same time SNP-based heritability of depression in GWAS-s is around 8-9%. Heterogeneity of the depressive phenotype may contribute not only to the lack of understanding its genetic background but may also hinder the identification of novel targets. Thus clinically relevant intermediate endophenotypes are needed for. The affective temperaments in the Akiskal model may be considered high-risk states or subclinical manifestations of mood disorders. Considering their strong genetic and biological background, high heritability in family studies, and their temporal stability, they may prove to be relevant endophenotypes for depression.

**Objectives:** The aim of the current study was to investigate the genetic determinants and heritability of affective temperaments based on a GWAS approach.

**Methods:** 775 subjects aged between 18-60 years recruited in Budapest, Hungary provided genetic samples and completed

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questionnaires including the TEMPS-A (Temperament Evaluation of Memphis, Pisa, Paris and San Diego) scale. A genome-wide association analysis was performed with the five affective temperaments as outcome variables. Age, gender, the top 10 principal components of the genome, and the other 4 phenotype were added in the model as covariates. Summary statistics derived from the GWAS analyses were used to estimate the heritability, i.e. the genetic variance explained by the different affective temperaments. LD score regression using LDPred2 [4] was performed to estimate heritability from the beta values and effect size in case of all 5 affective temperament phenotypes.

**Results:** rs3798978 showed a genome-wide significance (p=4.44x10-8) for anxious temperament, and several other variants showed suggestive significances for all five temperaments. The highest estimated heritability (h2 = 0.5224) was observed for the depressive temperament, and similarly high heritability was observed for the hyperthymic temperament (h2 = 0.4956). Anxious and cyclothymic temperaments showed almost the same heritability (cyclothymic h2 = 0.1651, anxious h2 = 0.1663), whereas for the irritable temperament, we got negative heritability estimation (h2 = -0.0567), which means that all of the phenotypic variance is explained by environmental factors.

**Conclusions:** Our analyses yielded remarkably high heritability values for depressive and hyperthymic temperaments explaining 52% and 50% of phenotypic variances. In contrast to the 8-9% SNP-based heritability in depression studies our findings suggest that these temperaments may be relevant endophenotypes for mood disorders.

Disclosure of Interest: None Declared

### **Mental Health Care 02**

## **EPP0461**

## Mental Illness Stigma among professionals at a Portuguese Medical Center

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**Introduction:** Mental Illness Stigma is a barrier in access to health-care. Stigma also influences population health outcomes by worsening, undermining adequate processes. The healthcare professionals show several stigmatising behavirous and cognitions, which may impair the adequate provision of care of this population with mental illness.

**Objectives:** We aimed to measure mental health stigma in health-care professionals at a portuguese hospital center.

**Methods:** A cross-sectional study of health profissionals was performed using a survey that included socio-economic and job related questions, personal and familiar questions regarding mental health, and Attribution Questionnaire 27 (AQ-27), a translated and validated stigma questionnaire with nine stigma sub-scales (Responsability, Pity, Anger, Dangerousness, Fear, Help, Coercion, Segregation and Avoidance).

**Results:** The sample included a total of 388 participants. The majority of the respondants were female (82,5%). The age ranged

from 22 to 69 (mean = 40,05). According to the job place distribution, we found statistically significant differences in various stigma subscales among several healthcare settings within our center. The inpatient unit professionals showed lesser stigmatising attitudes in anger, coercion, segregation and avoidance domains; and higher stigmatising attitudes in pity and help domains. However, professionals who work at surgery room showed higher stigmatising attitudes in danger and fear, but lesser levels of help domains. We also found differences in five stigma subscales among various health professions. The study didn't show differences in stigma domains regarding personal or professional contact with mental illness, neither academic studies in mental health.

**Conclusions:** Our findings suggest that workplace environment and profession may impact mental ilness stigma levels in healthcare professionals. We propose that future studies could be done to investigate methods to mitigate mental illness stigma, tailored to address different stigma domains in different workplace settings.

Disclosure of Interest: None Declared

### **EPP0463**

# Quality of Life (QoL) among medical students in Tunisia: a study using the WHOQOL-BREF instrument

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**Introduction:** Mental health problems such as stress, anxiety and depression have been described among medical students and are associated with poor academic and professional performance. That's why having a satisfying quality of life (QoL) is one of the main sources of motivation for students for their future.

**Objectives:** Our objectives were to assess the QoL of medical students and residents in Tunisia and to explore the influencing factors on this one.

**Methods:** This was a cross-sectional study among medical students and residents in Tunisia, all universities included, where they completed a questionnaire which comprised the WHOQOL-BREF instrument in its french version and several socio-demographics questions, in September 2022. Statistical analysis was performed by SPSS 26.0.

**Results:** One hundred twenty-five medical students and residents were included in our study. The mean age was  $26.10(\pm 3.41)$  years and most of them were female (73%). Mean scores of the WHOQOL-BREF in the physical, psychological, social and environmental domains were 36.51 ( $\pm 11.54$ ), 45.22 ( $\pm 15.71$ ), 37.19  $(\pm 18.61)$  and 52.94  $(\pm 14.84)$ , respectively. Students and residents had a relatively higher environmental mean score and a lower physical health mean score. The lowest mean score of the physical domain was observed in the 6th year students while the lowest mean scores of the psychological, social and environmental domains were observed in the medical students. Besides, we found a higher score of social and environmental domains in the residents group. In addition, we found a high correlation between psychological and environmental domains (p=0.000), psychological and social domains (p=0.021). We also found a correlation between age and social domain (p=0.034), in fact, the higher the age was the