for family psychoeducation, psycho-social interventions, and cognitive-behavioural education treatment approaches in individuals with CdLS.

Disclosure of Interest: None Declared

### **EPP0804**

# TRAPPC9 deficiency's implication in "secondary" autism spectrum disorders

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**Introduction:** Autism spectrum disorder (ASD) is a highly heterogeneous neurodevelopmental disorder with many contributing risk genes. Multiple intellectual disability (ID) susceptibility genes have been identified in ASDs to date. The trafficking protein particle complex subunit 9 *TRAPPC9* (OMIM#611966) in an autosomal recessive intellectual disability (ID) gene associated with not fully penetrant phenotype combining secondary microcephaly, dysmorphic facial features, obesity, autism spectrum disorder (ASD) and attention-deficit hyperactivity disorder (ADHD).

**Objectives:** The aim of this study is to consider *TRAPPC9* deficiency in autosomal recessive ID with ASD.

**Methods:** We present the observation of two siblings, born to Tunisian consanguineous and healthy parents, followed up for syndromic intellectual disability (ID) associated ASD and microcephaly. A clinical exome sequencing was performed to one child using a Trusight One kit of Illumina. We used sanger sequencing to validate the suspected variant for the other child and to specify the parental segregation.

**Results:** A homozygous pathogenic variant in the *TRAPPC9* (NM\_001160372.4) gene, c.1414C > T (p. Arg472Ter) were identified in one child. Sanger sequencing confirmed the homozygosity profile of this variant for the other child while the parents were both heterozygous carriers.

**Conclusions:** Repetitive behaviours, especially hand-flapping, were the mean ASD feature in our patients. The current variant is known in the Tunisian population. It is described to lead to the creation of a premature stop codon and a truncating protein causing a *TRAPPC9* deficiency. The impairing neuronal NFkB signalling due to *TRAPPC9* deficiency has been suggested to be implicated in ASD. Due to the profound ID seen in both patients, we suggest the classification of ASD related to *TRAPPC9* deficiency as "secondary" rather than "primary".

Our results support the implication of *TRAPPC9* in secondary ASD and shed the light on the possibility of screening p. Arg472Ter in Tunisian patients with this form of ASD as it is a recurrent mutation in the Tunisian population.

Disclosure of Interest: None Declared

#### **EPP0805**

# Bipolar Disorder and Parkinson disease: a 123I-FP-CIT SPECT study

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**Introduction:** Bipolar Disorder (BD) has been suggested to be a risk factor for development of Parkinson Disease. Psychiatric drugs used as standard treatment of BD includes many drugs that are known to induce drug-induced parkinsonism (DIP).

**Objectives:** Clinical differentiation between PD and DIP is a clinical and scientific crucial result. It might be aided by functional neuroimaging of the dopaminergic nigrostriatal pathway.

**Methods:** Twenty consecutive BD patients with parkinsonism were clinically assessed and underwent <sup>123</sup>I-ioflupane dopamine transporter SPECT. Imaging data of BD patients with pathological nigrostriatal pathway were further compared to a population of *de-novo* PD patients.

**Results:** Four BD patients had abnormal scans; they had higher putaminal binding ratio and putamen-to-caudate ratios than PD patients, despite similar motor symptom burden.

**Conclusions:** in our initial results, up to 20% of BD patients with parkinsonism might have an underlying dopaminergic deficit, which is higher than excepted in the general population. This evidences supports that BD represents a risk factor for subsequent development of neurodegenerative parkinsonism.

Disclosure of Interest: None Declared

#### **EPP0806**

### Chronic obstructive pulmonary disease and comorbid psychiatric disorders: preliminary results of an 8-year retrospective study using real data

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**Introduction:** Chronic obstructive pulmonary disease (COPD) is the third leading cause of mortality worldwide. In Portugal, it is estimated to afflict 14.2% of the population over the age of 45 and is one of the most common causes of morbidity, with a significant

social impact and excessive expenses. Moreover, COPD is associated with high levels of psychological distress and diverse psychiatric disorders that heighten the disease burden as they are associated with increased risk of exacerbations and frequent hospitalizations. Despite this overview, psychiatric conditions remain understudied compared to comorbid general medical conditions, and few studies have assessed their effect on COPD hospitalization outcomes.

**Objectives:** This study aimed to describe the occurrence of a vast array of psychiatric comorbid diagnoses in COPD hospitalizations and to understand their impact on hospitalization outcomes.

**Methods:** A retrospective observational study was conducted. All inpatient episodes from 2008 to 2015 of patients with at least 40 years and a primary diagnosis of COPD (ICD-9-CM codes 491.x, 492.x and 496) were selected from a national administrative database that included all hospitalizations in mainland public hospitals. From these sampled episodes, secondary psychiatric diagnoses were identified (ICD-9-CM codes 290.x-319.x). Age at hospitalization, sex, psychiatric comorbidities, length of stay (LoS) in days, admission type and date, destination after discharge, in-hospital mortality and hospital charges were analyzed.

**Results:** From a total of 66,661 COPD hospitalizations, 17,652 (26.5%) corresponded to episodes with a secondary psychiatric diagnosis. Patients with a comorbid psychiatric diagnosis were on average younger at admission (70.3 vs. 75.9 years, p<0.001), had a longer median LoS (9.89 vs. 9.33 days, p<0.001) and higher urgent admission rates (96.2% vs. 95.7%, p=0.009). There was also a significant association between discharge destination and psychiatric diagnoses (p<0.001).

**Conclusions:** These findings suggest that mental disorders have an adverse and quantifiable impact on COPD hospitalization outcomes. With this in mind, to provide optimal treatment for patients with both conditions, psychiatric disorders should become a matter of routine evaluation and follow-up.

Disclosure of Interest: None Declared

#### **EPP0807**

## Clinical and biochemical parameters associated with substance-induced psychotic disorder: which differences between alcohol, cannabis and psychostimulants

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**Introduction:** According to DSM V, substance-induced psychotic disorder is a mental health condition in which the onset of psychotic symptoms can be traced to the use of a psychotropic substance. The pathogenesis of this disease is still poorly understood; current literature traces its causes back to genetic predisposition and early traumatic events (i.e. child abuse).

**Objectives:** The present study aims to identify specific clinical features and biochemical markers which could be addressed as predictors for the long-term prognosis of this disease. Moreover, we aim to identify specific correlations between the clinical phenotype and the underlying substance abuse, in order to allow the early start of a tailored treatment.

**Methods:** Between 2020 and 2022 we recruited 218 patients referring to the Policlinico Hospital in Milan and the San Gerardo Hospital in Monza, Italy. All the patients were diagnosed with substance induced psychotic disorder: 31 reported alcohol abuse (14,2%), 71 psichostimulants (32,6%), 116 cannabis, (53,2%). For each patients, we collected demographic data, medical records and a comprehensive psychometric assessment (GAF, PANSS, BPRS, Modified Sad Person Scale-MOAS). Furthermore, we collected a blood sample for dosing Na+, K+, Na+/K+, hemogram with formula and platelets, glucose , urea, creatinine, uric acid, transaminases,  $\gamma$ GT, bilirubin, plasma proteins, albumin, LDH, CPK, PCHE, cholesterol, HDL, LDL, Tg, TSH.

**Results:** Chi squared test  $(\chi^2)$  has been used to compare qualitative variables between the 3 subgrous (alcohol-, psychostimulants- and cannabis-induced psychotic syndromes) (fig.1). One way ANOVA test has been used to compare quantitative variables between the same 3 subgroups (fig.2). After removing one of the subgropus (alcohol-induced psychotic symptoms), the same analysis have been repeated. Significant variables have been included in a binary logistic regression model in order to confirm their validity as predictors for cannabis- and psychostimulants-induced psychotic disorders (fig 3). Finally we performed Omnibus test and Hosmer-Lemeshow test in order to verify the validity of these regression models.

Image:

