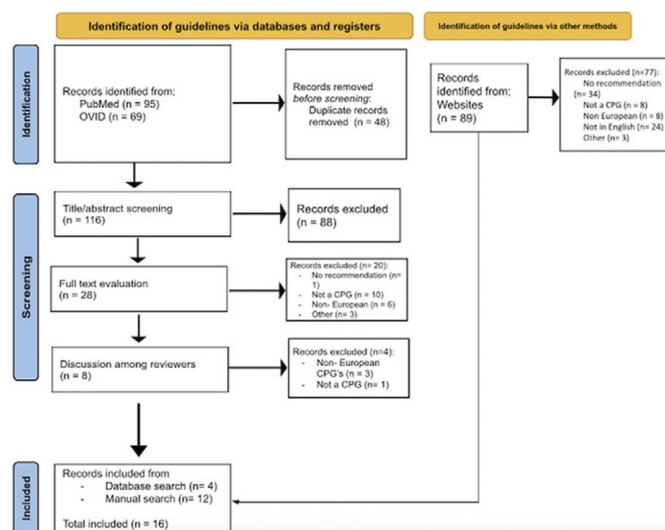


Methods: A scoping review was conducted across scientific databases (PubMed/MEDLINE and Ovid) and grey literature sources (Image 1. Flow diagram). Inclusion criteria centred on European CPGs published in English from 2010 onward with specific recommendations on genetic testing in mental disorders. Quality assessment was performed using the International Centre for Allied Health Evidence (iCAHE) checklist. Data extraction focused on guideline characteristics, target populations, and genetic test recommendations.

Results: Sixteen CPGs met the inclusion criteria, displaying considerable heterogeneity in quality and content, and covering a limited range of mental disorders. Six guidelines addressed neurodevelopmental disorders. Most recommended genetic testing in Autism Spectrum Disorder (ASD) when indicators such as intellectual disability (ID) or dysmorphic features were present; however, one guideline recommended routine testing. Only one guideline included recommendations for genetic testing in ID; routine access to Fragile X testing, chromosomal microarray, and whole genome sequencing was recommended as standard care.

Eleven guidelines provided recommendations on genetic testing in neurodegenerative disorders. In dementia, consensus on routine testing was generally limited to young-onset cases or those with distinct genetic profiles. APOE genotyping was generally discouraged. Guidelines for diagnostic testing for Huntington's Disease (HD) were consistent. Access to predictive testing with appropriate genetic counselling for at-risk adults was also recommended.

Image:



Conclusions: Based on our findings and the wider literature, we recommend considering genetic testing for: 1) all patients with ID, 2) patients with ASD exhibiting features suggestive of a genetic cause, such as ID and dysmorphic traits, and 3) patients with dementia with a young age of onset or a family history indicative of a Mendelian disorder. For HD, testing should be informed by phenotypic features and family history. Establishing harmonised, evidence-based guidelines is essential to integrate testing effectively. Key considerations include clinical utility, patient autonomy, and access to genetic counselling to ensure informed and supportive care.

Disclosure of Interest: None Declared

EPV0879

Season of birth/C-reactive protein gene interaction differentially affects negative symptoms domains in patients with schizophrenia

V. Golimbet^{1*}, T. Lezheiko¹, M. Gabaeva¹, N. Kolesina¹ and V. Mikhailova¹

¹Clinical Genetics Laboratory, Mental Health Research Center, Moscow, Russian Federation

*Corresponding author.

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Introduction: Schizophrenia is a severe psychiatric disease caused by genetic and environmental factors or their interactions that can contribute across multiple disease domains, including negative symptoms (NS), a core feature of schizophrenia.

Objectives: To study the association between season of birth (SOB), a well-replicated risk factor for schizophrenia, and NS domains avolition/apathy (AA) and diminished expression (DE) and to search for an interaction effect of SOB and rs2794521 genetic variants of the inflammatory marker C-reactive protein (CRP) on these domains.

Methods: The study included 2475 patients with schizophrenia. Patients born during the months of December to February were considered to be winter-born (n=636) and patients born in other months were considered to be non-winter-born (n=1839). Genotypes for CRP rs2794521 were obtained for 2437 patients. NS factors were calculated based on the Positive and Negative Syndromes Scale.

Results: There was a significant effect of SOB on AA scores (p=0.009), which remained after adjustment for sex and illness duration. Patients born in winter had higher scores compared with those born in other seasons. No significant effect of SOB on DE scores was observed. An association between the CRP rs2794521 G-allele and AA scores was found (p=0.044) in the winter-born group, with the carriers of the G-allele having higher scores. There was no effect of the G allele on DE scores in this group and on AA or DE scores in the non-winter group.

Conclusions: The results provide new evidence about the effect of SOB and SOB/CRP gene interaction on schizophrenia NS domains.

Disclosure of Interest: None Declared

EPV0880

The association between oxytocin receptor gene polymorphism, childhood adversity and negative symptoms of schizophrenia

T. Lezheiko¹, V. Golimbet^{1*}, V. MIKHAILOVA¹ and M. Gabaeva¹

¹Clinical Genetics Laboratory, Mental Health Research Center, Moscow, Russian Federation

*Corresponding author.

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Introduction: Neurohormone oxytocin plays an important role in the pathogenesis of mental illness, and also moderates the relationship between stress factors, especially those acting in the early stages of development, and the development of mental disorders. Literature data indicate that environmental risk factors significantly increase the risk of schizophrenia and the severity of its clinical presentation.