EPV0798

Dissecting the Heterogeneity of Autism: Focus on Phelan-McDermid Syndrome

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Introduction: Autism spectrum disorders (ASD) are a group of neurodevelopmental disorders that show delays and deficits in the development of multiple brain functions, which are characterized by social communication, poor language development, and restricted and stereotyped patterns of interests and behaviours. ASD affects about 1-2 % of the population and are considered to be highly genetic in nature. Structural variations of chromosomes have been identified in some ASD individuals, most common on chromosome 7q, 15q and 22q.
Objectives: 1-To present a systematic literature review of the natural history of individuals with 22q13.3 deletion syndrome, Phelan-McDermid syndrome (PMS). PMS, increase awareness of different phenotypes 2- Correlation of clinical manifestations of PMS with hypothesized underlying biological mechanisms 3-Rational for novel treatments is inferred through translational neuroscience approaches.
Methods: We have conducted a systematic literature review of the natural history of individuals with PMS, including both cross-sectional and long-term longitudinal analyses and correlation with hypothesized underlying biological mechanisms, including roles in regulation synaptic development, function, and plasticity. This systematic review includes the basis for a promising common pathway for ASD pathogenesis and the clinical implications of novel therapeutic strategies inferred through translational neuroscience approaches.
Results: This systematic review, therefore, outlines the: (1) Pathophysiological basis and clinical manifestations of PMS; (2) PMS pre-clinical models and applications to ASD; and (3) clinical implications of novel therapeutic strategies.
Conclusions: A promising common pathway for ASD pathogenesis and rational for novel treatments is inferred through translational neuroscience approaches. Neurobiological basis for lithium treatment is indeed supported by experimental results and current clinical findings.

Disclosure: No significant relationships.
Keywords: Autism; Phelan-McDermid Syndrome; Phelan-McDermid Syndrome; autism

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Clinical and psychological features of the brain organization of mental activity in children with autistic disorders

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Introduction: Autism spectrum disorders (ASD) – nosological continuum of genetically and clinically heterogeneous mental disorders united by complex violation of mental development, social interaction and behavior.
Objectives: Identify the features of brain functioning in children with ASD.
Methods: Neuropsychological diagnostics (Zh. Glozman), results acoustic brainstem evoked potentials (ABEP), mathematical methods. The study involved 48 children with diagnoses (ICD-10): F84. 0, F84. 1, F84.5, aged 3 to 8 years (average age 4.18 years).
Results: Neuropsychological examination revealed the main Indices of the functioning of brain blocks: the Index of activation and energy components of activity (I Index), the Index of the right-hemisphere holistic strategy of information processing (Index II-rights), the Index of the left-hemisphere analytical strategy of information processing (Index II-left), the Index of programming, regulation and control of activity (III Index). Then the children were divided into groups ($p<0.001$): 1 group – 10 children with high I Index ($W = -6.03$); 2 – 20 children with high Index II-rights ($W = -5.74$); 3 – 18 children with high III Index ($W = -2.32$). Correlation analysis showed: for group 1 difficulties in the perception of auditory information are characteristic; for group 2 – reduced level of control over the course of mental activity, difficulties in automating thinking and speech, coordination of movements; for group 3 – relationship is not manifested.
Conclusions: Phonemic perception causes the greatest difficulties in children of group 1. Children of group 2 are characterized by difficulties in the formation of motor speech skills, a decrease in voluntary activity and control over it.

Disclosure: No significant relationships.

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I am the best and I have everything in life

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Introduction: Megalomanic ideas in a patient with limited intellectual functioning may be due to the psychotic clinic or be the result of their disability.

Objectives: This case is intended to highlight the importance of a joint approach between psychiatrists and psychologists to assess functionality before and after the psychotic episode.
Methods: 34-year-old woman with no mental health history. She came to the emergency department for an episode of aggression at home. Her parents report that they have observed strange behaviour, she is more aggressive, speaks alone, changing voices and global insomnia in the last few days. Her language is incoherent and disorganised, with a long response latency. Megalomaniacal and catastrophic delusions. Possible auditory hallucinations and thought control phenomena.
Results: During admission, antipsychotic treatment was introduced with good tolerance and response on the part of the patient. She has been distancing herself from the ideas and has become somewhat critical. A psychological evaluation was carried out with different scales that showed borderline IQ.
Conclusions: It is important to make a good assessment of the patient's symptoms in order to make a differential diagnosis. In this case, it is advisable to carry out a control and follow-up, as well as a neuropsychological assessment before and after the acute episode. In addition, a multidisciplinary approach with psychologists, psychiatrists and social workers is important.

Disclosure: No significant relationships.
Keywords: borderline intelligence; megalomaniacal ideas; psychology; Psychosis

EPV0799

Influence of assisted outpatient living in people with intellectual disabilities on individual quality of life and resilience – design of a doctoral thesis

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Introduction: The research field of disability and mental health is politically and socially relevant, because the psychological well-being of people with intellectual disabilities has often been trivialized in therapy and legislation. Since the pursuit of mental health is coming into focus due to removal of stigma and emerging awareness, people with disabilities must have equal opportunities to choose their place of residence and get suitable psychosocial support.
Objectives: Our aim is to investigate the influence of assisted outpatient treatment (AOT) on the quality of life and resilience of people with intellectual disabilities who are living self-determined.
Methods: A participatory mixed-methods design is chosen as it enables the greatest possible standardization and allows a high flexibility. The project will be divided into three parts: A systematic literature search to gain knowledge about the field and to estimate the sample size, a pre-post-comparison of the WHOQOL-BREF to evaluate AOT in terms of self-perceived quality of life and a focus-group of handicapped people to reflect the study results with attention on resilience.
Results: To foster the doctoral thesis, several questions can be discussed: 1) How is the relation of disability and mental health to be described? 2) What might be the pros and cons of self-determined living? 3) Which steps need to be taken to implement AOT more often?
Conclusions: The topic is relevant in the public health sector and the results could help to sensitize professionals and the general society regarding to participation in everyday life. The recommendations developed may serve to implement comparable forms of housing.

Disclosure: No significant relationships.
Keywords: intellectual disability; assisted outpatient living; resilience; Quality of Life

EPV0801

Assessment of intellectual disability in Klinefelter's Syndrome associated with other Prenatal and Perinatal risk factors. A case report

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Introduction: Klinefelter's syndrome (KS) is considered as a genetic risk factor for intellectual disability and specifically for impairment on verbal skills. Prenatal and perinatal morbidity concurrent with this diagnosis determine the entity and typology of neurodevelopmental deficits.
Objectives: To describe the intellectual and adaptive functioning of a patient with KS and prenatal and perinatal difficulties, assessed in the Service of Psychiatry, Clinical Psychology and Mental Health at La Paz University Hospital (Madrid).
Methods: A descriptive study was conducted of a 6 year and 7-month-old boy with diagnosis of KS (karyotype XXY), extremely low weight at birth (752 gr.), Arnold Chiari type I malformation with post-hemorrhagic hydrocephalus and periventricular leukomalacia, bronchopulmonary dysplasia, intra-atrial septal defect, left kidney agenesis with iatrogenic adrenal insufficiency, and acute ventriculitis due to E. Coli. Neuropsychological evaluation was carried out in October 2021. Leiter-3 scale was selected due to the absence of expressive language; and his parents completed the questionnaire Adaptive Behavior Assessment System-II.
Results: The nonverbal intelligence quotient was observed in the very low range (full-scale IQ = 69). The result of general adaptive behavior (z = -2.83), showed very high functional disability both in the conceptual, social, and practical domains.
Conclusions: The high number of causes of disability in this patient is consistent with a high degree of functional disability. Efficient evaluation sessions of intellectual performance, adaptive functioning, and necessary supports, due to the absence of expressive language and limited receptive language, are required. A specific neuropsychological evaluation profile should be established for KS.

Disclosure: No significant relationships.
Keywords: Klinefelter syndrome; prenatal and perinatal difficulties; intellectual disability; nonverbal intelligence