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Psychotic disorders in young patients with Prader-Willi syndrome: A case report and literature review

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Introduction: Prader-Willi syndrome (PWS) is a genetic disorder with an estimated prevalence of 1:25,000. PWS results from defective gene expression on the paternal copy of chromosome 15. In 70% of the cases it is a deletion that means that part of the paternal chromosome 15 is missing. Maternal uniparental disomy (mUPD) is present in 25% of cases. Typical clinical features of PWS are dysmorphism, hypotonia, hyperphagia, hypogonadism and developmental delay. In addition, the syndrome is accompanied by various psychiatric symptoms that are often insufficiently known within the psychiatric field. Regarding the relationship between PWS and schizophrenia spectrum disorders (SSDs), individuals with mUPD appear to have a 3 to 4 times higher risk of psychotic symptoms than those with the deletion subtype. Psychotic episodes have an atypical presentation with recurrent episodes of confusion and rapidly fluctuating psychotic and mood symptoms.

Objectives: To describe an unusual clinical case in order to determine the management regarding clinical approach, and provide an overview of psychotic episodes in patients with PWS for the general practitioner with the most up-to-date information on workup and management.

Methods: We report a case involving a 13-year-old woman with PWS (mUPD of chromosome 15) and mild intellectual disability (IQs 59), who presented psychotic symptomatology in the form of disorganized behavior, delusional ideation, auditory hallucinations, self-referentiality and suspicion. Parents reported that these symptoms started two days prior the day of consultation. No environmental stressors were identified and no recent treatment changes were made. Patient's medication consists in 150 mg sertraline per day due to anxiety control and aid in emotional and behavioral regulation.

Results: Given the diagnostic approach of a psychotic episode (PE) in a patient with PWS, it was decided to offer 0.5mg risperidone per day, in an increasing pattern until reaching a final dose of 1.25 mg per day, presenting a global remission of the psychotic symptomatology.

Recommendations for patients with PWS presenting PE are based upon systematic reviews. Patients with PWS, especially mUPD subjects, are at risk for SSDs and mood disorders. Antipsychotics (APs) are the gold standard in the treatment of SSDs, and some authors have suggested that APs protect patients with previous psychotic symptoms from relapse. It is unknown whether there is a protective effect of APs in mUPD patients who have not previously exhibited psychotic signs.

Conclusions: PWS represents a good example of a genetic disease with behavioral and psychiatric symptoms that may be challenging

to treat with psychotropic medications. For a better understanding of psychiatric problems in adults with PWS, longitudinal studies with careful and standardized follow-up of psychiatric symptoms in PWS are necessary.

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Cognitive characteristics in women with mastectomy, mental symptoms and marriage compliance

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Introduction: Although mastectomy is a treatment for the cleaning of cancer cells in the breast, it can also cause psychological changes in women.

Objectives: This research was carried out to investigate the cognitive distortions, mental symptoms, and marital adjustment relationships in women with mastectomy.

Methods: The descriptive and comparative study was conducted in a public hospital with 90 mastectomies and 90 healthy female samples. The data were collected using The Cognitive Distortions Scale (CDS), The Brief Symptom Inventory (BSI), and Marriage Adjustment Scale (MAS) and were evaluated by descriptive statistical analyzes, t test, Mann-Whitney U test and Spearman Correlation Analysis.

Results: Women with mastectomy scored high in the healthy control group in all sub-dimensions of CDS and BSI and this difference is highly significant ($p < 0,5$). Women with mastectomy scored higher in total agreement sub-dimensions than the healthy control group and the difference between them was statistically significant ($p < 0,5$). There are positive, very strong and statistically significant relationships between all sub-dimensions of the CDS and the total sub-dimension scores of the BSI ($r = ,769$ and $,919$). There are positive, strong and statistically significant relationships between the sub-dimensions of the CDS and the sub-dimension of the agreement and the total score of MAS ($r = ,681$ and $,734$). There are strong and statistically significant relationships between the total and sub-dimension scores of the BSI and the sub-dimension and total score of the MAS's agreement ($r = ,672$ and $,778$).

Conclusions: Cognitive distortions and mental symptoms are significantly higher in women with mastectomy, and cognitive distortions are associated with mental symptoms. Cognitive distortions and psychological symptoms scores, marriage adjustment scores and agreement sub-dimension scores increase in women.

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