Psychiatric disorders in Prader-Willi syndrome: a case report about psychotic symptoms

Marta Herrera Durán, Carlos Gómez Sánchez-Lafuente, Rocío Reina González
Psychiatrist, Hospital Universitario Virgen de la Victoria, Málaga
Resident physician in the psychiatry department at Hospital Universitario Virgen de la Victoria, Málaga
Resident in family medicine at Hospital Regional Universitario, Málaga

INTRODUCTION

Prader-Willi syndrome (PWS) is a complex neurodevelopmental genetic disorder due to paternal loss of imprinted genes on chromosome 15 and characterized by a range of mental and physical findings.

The features of PWS were first documented in an adolescent female by J. Langdon Down in 1887, but the syndrome went unrecognized until 1956 when Prader, Labhart, and Willi reported nine individuals with similar clinical findings.

The incidence is approximately 1/10,000 to 25,000 live births and it occurs in both sexes and all races.

PWS is caused by lack of expression of genes on the paternally inherited chromosome 15q11.2-q13 region. There are three main genetic subtypes in PWS: paternal 15q11-q13 deletion (65-75 % of cases), maternal uniparental disomy 15 (20-30 % of cases), and imprinting defect (1-3 %).

The main phenotypic features include intrauterine growth retardation, neonatal hypotonia and failure to thrive during infancy. Clinical manifestations change with age, and other features such as hyperphagia with food seeking, usually 2 to 3 years old, and progressive obesity, which is related to complications such as cardiopulmonary failure, sleep apnea, hypertension and diabetes mellitus type 2. Short stature, hypogonadism, developmental delay, cognitive disability and behavioural problems become evident.

In the Prader-Willi Syndrome the psychiatric symptoms are fundamentally behaviour disorders and obsessive-compulsive symptoms in relation with food.

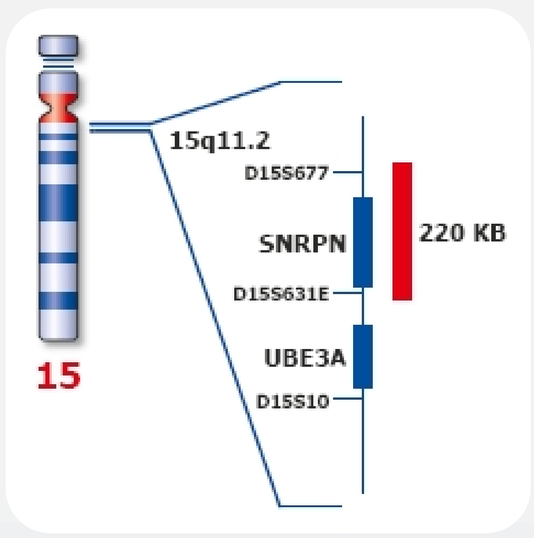
A characteristic behavioural profile becomes evident in early childhood, with temper tantrums, stubbornness, controlling and manipulative behaviour, obsessive-compulsive characteristics, and difficulty with change in routine. Lying, stealing, and aggressive behaviour are common.

In addition to the syndrome's characteristic hyperphagia and food seeking, individuals with Prader-Willi syndrome also have increased risks of nonfood, compulsive behaviours. These include skin picking, which is highly prevalent, as well as more variable rates of hoarding, redoing and concerns with symmetry, exactness, cleanliness, ordering and arranging.

They also have an increased risk of psychotic disorder or affective illness with a psychotic component, especially young adult patients and those with the maternal uniparental disomy as opposed to paternal deletion. True psychosis is evident in young adulthood in approximately 5-10% of patients. Behavioural and psychiatric problems interfere the most with quality of life in adulthood.

CASE REPORT

20-year-old woman. Diagnosed with Prader-Willi syndrome with 2-year-old, mother subtype. It presents phenotype corresponding to this syndrome, with obesity, low size, almond-shaped eyes, small head, thin upper lip and small hands and feet. Menarche with 17 years, presented delay in development of secondary sex characteristics. At the age of fifteen was diagnosed with Diabetes Mellitus type II, being treated with anti-diabetic oral since then. It has an IQ boundary, with test of WAIS 65.



Concerned parents described, from age 3, she was a nervous and capricious child with frequent temper tantrums if they contradict her. Presence of hyperphagia with obsessive thoughts and excessive preoccupation with food, search for it and even history of small thefts in supermarkets. She has daytime sleepiness, tending to stay asleep during the day, basically if she takes an activity that she finds boring. With 18 years she initiates tracking in Mental Health by presenting clinic consistent in lability and emotional instability that responds favourably to treatment with fluoxetine 20 mg per day.

Reason for consulting: in the last three weeks she is more anxious and nervous than usual. Sometimes she talks about any inconsistencies and she is scared. She has also a noticeable decrease in the usual hours of sleep in her, not sleeping during the day and presenting conciliation insomnia at night. She refuses to attend occupational day centre, which assists for 5 years.

Psychopathological examination found that the patient has vague and type discriminatory, auditory hallucinations (call her silly and useless), and that this makes her display suspicious and nervous with her surroundings, even with her parents.

It is guideline treatment with 5 mg Aripiprazole per day, plus lorazepam 1 mg every 12 hours (the latter is phased out at 10 days to quit), obtaining remission of psychopathology in 10 days. Currently she maintains such treatment for six months.

The choice of antipsychotic is made according to the organic pathologies of the patient (obesity and type II diabetes), in order to avoid to worsen them with other psychoactive drugs of worse metabolic profile.

CONCLUSION

Behavioural and psychiatric problems interfere the most with quality of life in adulthood. These should be detected early and treated appropriately with parental education and psychotropic medication if it was necessary. Serotonin agonists have been the most successful in reducing temper outbursts and improving compulsivity. Psychosis is treated in a standard manner.

BIBLIOGRAPHY:
Lo ST, Collin PJ, Hokken-Koelega AC. Psychiatric disorders in children with Prader-Willi syndrome-Results of a 2-year longitudinal study. Am J Med Genet A. 2015 May; 167 A(5):983-91.
Larson FV, Whittington J, Webb T, Holland AJ. A longitudinal follow-up study of people with Prader-Willi syndrome with psychosis and those at increased risk of developing psychosis due to genetic subtype. Psychol Med. 2014 Aug; 44(11):2431-5.
Sinnema M, Boer H, Collin P, Maaskant MA, van Roozendaal KE, Schrander-Stumpel CT, Curfs LM. Psychiatric illness in a cohort of adults with Prader-Willi syndrome. Res Dev Disabil. 2011 Sep-Oct; 32(5):1729-35.
Soni S, Whittington J, Holland AJ, Webb T, Maina E, Boer H, Clarke D. The course and outcome of psychiatric illness in people with Prader-Willi syndrome: implications for management and treatment. J Intellect Disabil Res. 2007 Jan; 51(Pt 1):32-42.