SNORD116 Deletions Cause All Features of Prader-Willi Syndrome

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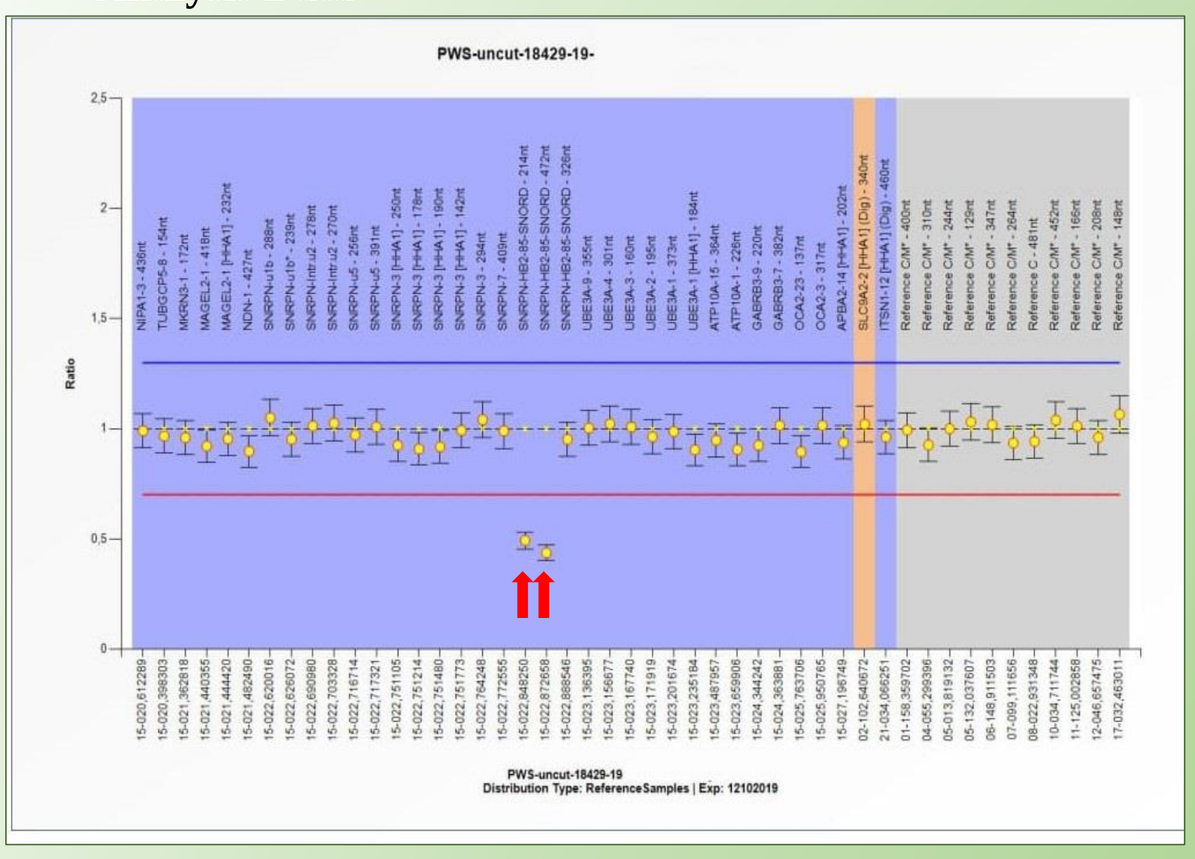
INTRODUCTION

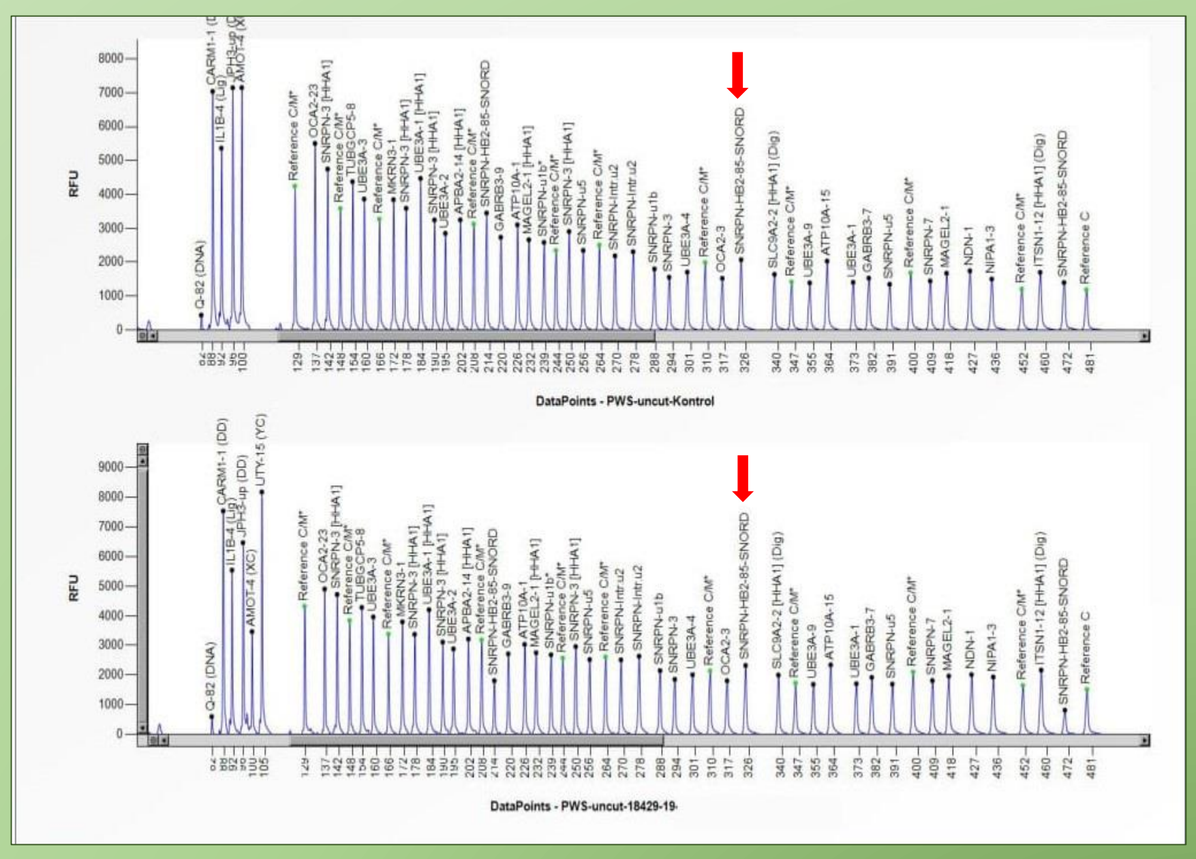
Prader-Willi syndrome (PWS) is characterized by hypotonia, feeding difficulties in infantile period, excessive eating in early childhood, intellectual disability, behavioral problems and growth hormone deficiency. Approximately 65-75 % of patients with PWS have a deletion 15q11.2-q13, which can be detected using fluorescence in situ hybridization (FISH) or arrayCGH. This ratio increases up to 99% with Methylation-specific multiplex ligation-dependent probe amplification (MS-MLPA). MS-MLPA not only provides a high rate of diagnosis, but also gives the clinician a better genotype phenotype correlation, giving the deletion site information more specific. With current report, we aimed to compare the clinical findings of our case with PWS syndrome in whom *SNORD116* gene deletion was detected by MS-MLPA method with the clinical findings of very few cases reported in the literature.

METHODS

CLINICAL FINDINGS: Prenatal history revealed polyhydroamnios and decreased fetal movements. The patient was born 3800 g with C/S at term and remained in the incubator for 3 weeks due to pulmonary hypertension. The patient with congenital hypotonia started to hold his head when he was 6 months old. He had difficulty feeding during the transition to solid foods. He started walking around 2 years old. His first seizure occurred at the age of 3.5 years in the febrile period as generalized tonic clonic contractions. His second seizure recurred nine months later. The patient's EEG was abnormal and Keppra was started. He had no seizures for 1.5 years. The patient gained weight after 4 years of age, and his height and weight were over 97 percentile when he was 5.5 years old. The patient, who was delayed to speak, could say about 50 words at the age of 5 and could form sentences of 2-3 words. In his examination, almond-shaped eyes, full cheeks, micrognathia, inverted nipple, abdominal obesity, pes planus, muscular hypotonia and small penis were observed.

Analysis Data





RESULTS

The FISH study performed using the SNPRN / CEP 15 / PML probe was evaluated normal. In MS-MLPA analysis, deletion of snoRNA cluster region in *SNORD116* gene was observed. Images of both analyzes were shown in Figures.

DISCUSSION

In this case report, we compared the clinical findings of our case with *SNORD116* deletion in the literature. Common findings include obesity, speech retardation, intellectual disability and typical facial findings, which are common findings for PWS. Therefore, we thought that the deletion of *SNORD116* is a very effective cause for PWS phenotypes. We also think that the *SNORD116* deletion is a process that needs to be focused more on PWS treatment.