Keywords: Type 2 diabetes mellitus, dyslipidemia, obesity.

307. GENETIC ASPECTS OF PRADER-WILLI SYNDROME

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Introduction: PWS is a complex genetic disorder affecting appetite, growth, metabolism, cognitive function and behavior.

Characterized by: 1) Low muscle tone 2) Short stature (when not treated with growth hormone) 3) Incomplete sexual development 4) Cognitive disabilities 5) Behavioral problems 6) The hallmark characteristics – chronic feelings of insatiable hunger and a slowed metabolism that can lead to excessive eating and life-threatening obesity.

The syndrome is due to the loss of expression of several genes encoded on the long arm of chromosome 15 (15q11.2–q13). The complex phenotype is most probably caused by a hypothalamic dysfunction that is responsible for hormonal dysfunctions and for absence of the sense of satiety.

People with PWS have a flaw in the hypothalamus part of their brain, which normally registers feelings of hunger and satiety. While the problem is not yet fully understood, it is apparent that people with this flaw never feel full; they have a continuous urge to eat that they cannot learn to control. To compound this problem, people with PWS need less food than their peers without the syndrome because their bodies have less muscle and tend to burn fewer calories.

Materials and methods: While doing the review we screened worldwide literature and interactive sources. We tried to choose information that will be reliable and will explain the genetic aspects, clinical features and complications of PWS.

Discussion results: PWS is rarely seen worldwide disease especially in RM that imposible diagnoses without genetics tests in neonates. Increasing awareness to PWS can bring to increase use of genetic methods of diagnoses and less miss diagnoses.

Conclusion: Prader-Willi syndrome is a complex multisystem disorder. Patients can be affected by various problems; therefore precocious diagnosis is fundamental to guarantee optimal assistance. [illegible] visions and clinical follow-up need to consider all of these possible problems. A multidisciplinary team is required, made up of specialists such as neonatologists, geneticist, pediatricians, endocrinologists, orthopedic surgeons, psychologists, psychiatrists, physiotherapists, and urologists to deal with the numerous medical and psychological problems a PWS patient has to face. Only in this way we can improve quality of life, prevent complications, and prolong life expectancy in patients with PWS.

Key-words: Prader-Willi, Obesity.