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A CASE REPORT ON PRADER WILLI SYNDROME

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ABSTRACT **Background:** Prader–Willi syndrome (PWS) is a rare, complex genetic disorder caused by the absence of paternally expressed genes on chromosome 15q11-q13, most commonly due to a deletion or maternal uniparental disomy. It is characterized by neonatal hypotonia, feeding difficulties in infancy, followed by hyperphagia, obesity, developmental delays, hypogonadism, short stature, and behavioral abnormalities. Endocrine dysfunction, including growth hormone deficiency, contributes to metabolicand growth issues. Early diagnosis with molecular genetic testing enables timely interventions such as growth hormone therapy, dietary management, behavioral support, and multidisciplinary care, improving quality of life and reducing obesity-related complications.

KEYWORDS : Prader–Willi syndrome, chromosome 15q11- q13, hyperphagia, obesity, hypotonia, genetic disorder, growth hormone therapy, behavioral management

INTRODUCTION:

PWS is a genetic disorder usually caused by chromosomal deletions on chromosome 15q11 or by uniparental disomy (UPD). Absence of a paternal allele of chromosome 15q11, due to chromosomal deletion or the presence of two imprinted copies due to maternal UPD, results in PWS. In addition to the large de novo deletions of 15q11 on the paternal chromosome, PWS can be caused by aberrant methylation of the 15q11 locus.

A 4 year male child presented with presented with morbid obesity, hypogonadism, dysmorphism, infantile hypotonia, feeding difficulties at neonatal period, small testis, developmental delay.

Physical Examination :

Flat nasal bridge , tooth decay , short stature, small hands and tapered fingers ,small feet, marked obesity.

Investigations:

Thyroid profile :free t3 : 8.5 :free T4 :17.8 ,TSH :3.060, Chest x ray : no abnormal lung opacities Usg abdomen :no significant sonological OAE : b/l refer

Genetic studies: No deletion or duplication were detected.The methylation ratio of 1.0 indicating 100% methylation was detected within the detection limits of MS-MLPA, in the SNRPN and MAGEL2 domains in 15q11 of PWS region represents Prader-Willi Syndrome (PWS) with maternal uniparental disomy.

Figure 1: child with prader wili syndrom

MANAGEMENT :

Growth hormone therapy is beneficial in improving growth, muscle mass, and cognitive function. Nutritional management, including strict caloric control, is critical to prevent obesity. Behavioral interventions are necessary to manage food-seeking behaviors and other psychological challenges.

CONCLUSION:

Prader-Willi Syndrome is a complex genetic disorder that requires a lifelong, multidisciplinary approach to management.Early growth hormone therapy, strict dietary control, and behavioral support can significantly improve outcomes. Families should receive genetic counseling to understand the inheritance patterns and the likelihood of recurrence in future pregnancies.

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