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PRADER-WILLI SYNDROME – A CASE REPORT

Paediatrics

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ABSTRACT

Prader -willi syndrome is a rare multi-systemic genetic disorder, in which there is either deletion of genes in the proximal arm of paternal chromosome 15 or maternal disomy in the proximal arm of chromosome 15. It usually affects both males and females equally with a large preponderance for boys. [1] we present a rare case of a 4 months old infant presented with respiratory distress, feeding difficulty, generalized hypotonia, microcephaly, dysmorphic facial features, delayed development, bilateral undescended testis and failure to thrive. Patient was treated with intravenous antibiotics, oxygen support and symptomatic treatment. Genetic testing was done by mlpa (multiplex ligation dependent probe amplification) for confirmation of diagnosis which was suggestive of prader-willi syndrome. After proper feeding training, genetic counselling to parents and satisfactory weight gain, patient was subsequently discharged on the 12th day of admission.

KEYWORDS

Prader-willi syndrome, hypotonia, failure to thrive.

INTRODUCTION

Prader-willi syndrome (pws) is a rare genetic disorder of chromosome 15 affecting approximately one in every 10,000-30,000 births. It is characterized by low birth weight, neonatal hypotonia, failure to thrive, delayed development, short stature, childhood obesity, hyperphagia, hypogonadism causing genital hypoplasia and pubertal insufficiency. Patient also has characteristic dysmorphic facial features and intellectual disability. Characteristic dysmorphic features include narrow bifrontal diameter, almond shaped palpebral fissures, thin upper lip, downturned mouth [2]. The majority of newborns with pws present with marked neonatal hypotonia, this is associated with decreased movements, weak crying, and poor reflexes, including poor sucking, often resulting in failure to thrive. During infancy many PWS children display a range of behavioral problems that become more noticeable in adolescence and adulthood and interfere mostly with quality of life. The complex phenotype is most probably caused by hypothalamic dysfunction that is responsible for hormonal dysfunctions and for absence of the sense of satiety. Genetic testing is used to confirm pws diagnosis [3].

Primary treatments include dietary management, growth hormone, regular monitoring of comorbidities and behavioral and cognitive therapies [2].

CASE STUDY

A 4 months old male child came to the emergency department of a tertiary care hospital with complaints of fever since 7 days, cough and decreased oral intake since 4 days and difficulty in breathing since 2 days. Patient also had a history of poor sucking and difficulty in feeding, delayed development and not gaining weight. Patient was a full term normally delivered male child with birthweight 2990 gm and had cried immediately after birth. There was a significant past history of nicu admission at the first hour of life for transient tachypnea of newborn. Patient was also diagnosed with posterior urethral valve. At 15th dol patient was again admitted for feeding difficulty and dehydration and was also operated for puv during this time.

On examination, patient's weight was 3.8 kg which was suggestive of failure to thrive. Patient also had absent neck holding, dysmorphic facial features in form of hypertelorism, almond shaped eyes, microcephaly, fullness of dorsum of hands and feet and bilateral undescended testes. On systemic examination, patient had a frog-like posture (see figure 1), marked generalized hypotonia and adductor and popliteal angles were increased. On axillary suspension there was a feeling of 'slipping through' and on ventral suspension there was an inverted "u" position (rag doll posture). Along with this, patient also had tachypnea and increased work of breathing with bilateral crepitations on auscultation.

Patient was admitted to picu. On admission, patient had respiratory distress so oxygen therapy was started via nasal prongs. Patient was treated with first line antibiotics, iv fluid & o2 support in the picu. Blood investigations were suggestive of raised total count and raised inflammatory markers. [see table 1] abdominal ultrasound suggested possibility of multiple calculi in right kidney. Pelvic ultrasound showed both testes in the inguinal region for which pediatric surgery consultation was done. After pediatric neurology consultation, genetic testing was done by mlpa, which showed prader-willi syndrome. Patient was given oxygen support for 11 days which was gradually tapered. Feeding was started on the 4th day of admission with a nasogastric tube. Breastfeeding followed by spoon feeding with formula milk was started on the 9th day of admission. Patient was able to take spoon feeding well and weight gain was recorded from the 9th day of admission. Patient was discharged after 12 days of admission with proper feeding and immunization advice and genetic counselling to parents.

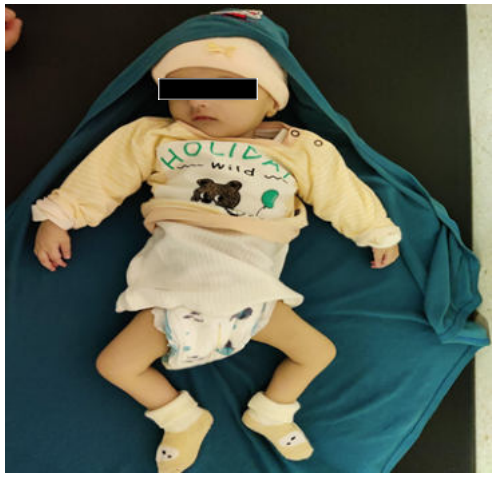

Figure 1: Frog like posture in Prader-Willi Syndrome

Table 1: Laboratory parameters at the time of admission

Investigations	Reference Range	Patient Result
Complete Blood Counts		
Hemoglobin	12-18 g/dl	12.5
Total White Blood Cells	4000-10000/mm3	14070
Hematocrit	36-55%	39.3
Platelet count	150000-450000/mm3	393000
D-Dimer	0.5 mcg/ml	0.84
CRP	<0.6 mg/dl	Negative
S. Ferritin	10-282 ng/dl	>1650
S. Alanine transaminase	<42 IU/L	43
Ionized Calcium	1.12-1.32 mmol/L	1.19
Urea	15-45 mg/dl	37.7
S. Creatinine	0.7-1.3 mg/dl	0.36
S. Sodium	135-145 mmol/L	137
S. Potassium	3.5-5.5 mmol/L	5.2
S. Calcium	8.6-10 mg/dl	10
Troponin-I	<2.0 ng/L	0.006
S.Free T4	0.8-2 ng/dl	0.82
S.Free T3	2.1-4.4 pg/ml	2.39
S. TSH	0.35-5.5 microIU/ml	7.55
S. Iron	65-175 mcg/dl	22
Urine RM		Calcium Oxalate Crystal seen
Blood Culture		No growth
AFB of Gastric aspirate		No MTB detected
CPK Total	22-198 U/L	61
Arterial blood gas analysis		
pH	7.35-7.45	7.46
PCO2	35-45 mmHg	27.3
PO2	80-100 mmHg	195
Bicarbonate	22-26 mmol/L	21.6
SO2	96-97 %	99.8

DISCUSSION

PWS is a hereditary disease associated with genomic imprinting. The several mechanisms of PWS include: 1) a paternally derived large deletion of 15q11-q13, accounting for 70% of all patients 2) a maternal uniparental disomy, accounting for approximately 25-29% 3) a defect in the genomic imprinting mechanism, about 5%. Consensus diagnostic criteria for pws includes major criteria, minor criteria & supportive criteria. Major criteria include neonatal /infantile hypotonia, feeding problems and failure to thrive as an infant, weight gain at 1-6 years, obesity; hyperphagia, characteristic dysmorphic facial features, small genitalia; pubertal delay and insufficiency, developmental delay/intellectual disability. Minor criteria include, decreased fetal movement and infantile lethargy, typical behavior problems, sleep apnea, short stature, hypopigmentation for the family, small hands and feet for height, narrow hands, straight ulnar border, esotropia, myopia, thick and viscous saliva, speech articulation defects, skin picking4. Supportive criteria include decreased vomiting, scoliosis and /or kyphosis, early adrenarche, osteoporosis[5]. The mainstay of diagnosis is dna methylation testing to detect abnormal parent specific imprinting within the prader-willi critical region (pwcr) on chromosome 15; dna methylation-specific testing is important to confirm the diagnosis of pws in all individuals[6].

CONCLUSION:

Prader-willi syndrome is a complex multisystem disorder. Patients can be affected by various problems; therefore, early diagnosis is fundamental to guarantee optimal assistance. 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 & psychological problems. Only in this way we can improve quality of life, prevent complications and prolong life expectancy in patient with pws[7].

REFERENCES:

1. Tayal E, Indushekar K, Saraf BG, Sheoran N, Doda A. Prader-Willi syndrome: a case report. Public Health–Open Journal. 2016;1(1):12-5. https:// www.nationwi dechildrens org / Document/Get/126690 Accessed on 20/11/2021.
2. Cheon CK. Genetics of Prader-Willi syndrome and Prader-Will-like syndrome. Annals of pediatric endocrinology & metabolism. 2016 Sep;21(3):126.
3. Kliegman R. Nelson Textbook of Pediatrics. 21st ed. Vol. 1. Philadelphia: Elsevier; 2020.
4. Holm VA, Cassidy SB, Butler MG, Hanchett JM, Greenswag LR, Whitman BY, Greenberg F. Prader-Willi syndrome: consensus diagnostic criteria. Pediatrics. 1993 Feb;91(2):398.
5. Driscoll DJ, Miller JL, Schwartz S, Cassidy SB. Prader-Willi Syndrome. GeneReviews®[Internet]. 2017 Dec 14.
6. Elena G, Bruna C, Benedetta M, Stefania DC, Giuseppe C. Prader-Willi syndrome: clinical aspects. Journal of obesity. 2012 Jan 1;2012.