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Case Report

Prader Willi syndrome in a neonate with aspiration pneumonitis: a case report

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ABSTRACT

Prader Willi syndrome (PWS) is a rare genetic disease, even rarely diagnosed in neonates. PWS in a neonate presents with central hypotonia and feeding difficulty. Hereby, we report first neonatal case from India, who presented with aspiration pneumonitis. This case is presented to understand the subtle clinical features of PWS in neonates to facilitate early diagnosis and early intervention which improves quality of life. A term neonate presented at 9 days of life with complain of choking while feeding followed by rapid breathing. Apart from severe respiratory distress general examination revealed facial dysmorphology and bilateral cryptorchidism with scrotal hypoplasia. There was marked central hypotonia and weak cry. A provisional diagnosis of PWS with aspiration pneumonia was made and karyotyping along with DNA methylation was sent. Positive DNA methylation confirmed our diagnosis. Baby was managed with i.v antibiotics and parents were given basic information about PWS. They were educated about feeding techniques to prevent choking spells and informed about multidisciplinary care that baby will require. Prader Willi Syndrome should be considered in differential diagnosis in neonate presenting with central hypotonia, feeding difficulties and prompt diagnostic DNA testing should be done for confirmation.

Keywords: Aspiration pneumonia, Neonate, Prader Willi syndrome, Hypotonia, India

INTRODUCTION

PWS is rare imprinting disorder caused by gene inactivation of paternally inherited region 15q11-13 on chromosome 15 with a reported prevalence of 1/29,000 in newborns.[1] It poses a diagnostic challenge in neonates, most cases have been diagnosed only in infancy and older children.

The rarity of the disease and the late diagnosis due to inconspicuous clinical features are main factor responsible for high morbidity. Till date only few cases of PWS have been reported in neonates from Asia and no neonatal case has been reported from India.

CASE REPORT

A full term (40 week) male baby was born by elective cesarean section to a multigravida mother from a non-consanguineous marriage. Pregnancy was uneventful and there was no history of maternal hypothyroidism and decreased fetal movements. There was no history suggestive of birth asphyxia. Neonate presented on day 9 of life with rapid breathing just after feeding, vomiting, and choking. Baby was apparently normal for the first 9 days, accepting and tolerating feeds well. The other siblings were normal and there was no history of any fetal loss. At presentation vitals were, HR 138/minute, RR 70/minute, CRT was less than 3 seconds and SPO2 was 92%. Abdomen was scaphoid, bilateral cryptorchidism

and scrotal hypoplasia was also present. Chest examination revealed crepitations bilaterally. There was little limb activity with weak cry and poor suck and Moro's reflex was absent. Facial features noticed were: almond eyes, thin, convex upper lip and angle downward with narrow bifrontal diameter. Scrotum was hypoplastic with bilateral cryptorchidism (Figure 1). A provisional diagnosis of Prader Willi syndrome (PWS) with aspiration pneumonia was made. ABG showed mild respiratory acidosis (pH: 7.30, pO2: 72, pCO2:50, HCO3:29). Treatment was started with oxygen and IV antibiotics. Karyotyping and DNA methylation were sent. Karyotyping was normal. DNA methylation confirmed our diagnosis (Figure 2). Patient was discharged after 10 days of treatment on spoon feeding and proper counseling of mother. The infant was well with adequate weight gain after one month.

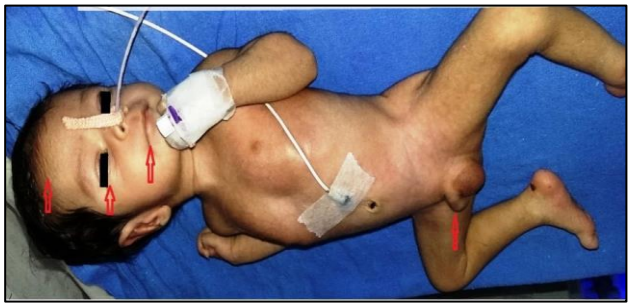
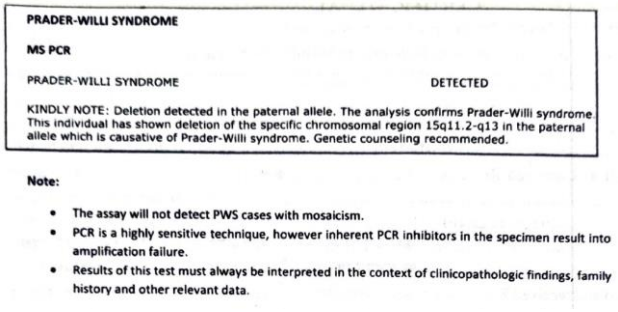


Figure 1: Arrow (1) Almond eyes, 2) thin and convex upper lip with agle pointing downward, 3) narrow bifrontal diameter (dolichocephaly) and 4) bilateral cryptorchidism with scrotal hypoplasia.



PRADER-WILLI SYNDROME

MS PCR

PRADER-WILLI SYNDROME DETECTED

KINDLY NOTE: Deletion detected in the paternal allele. The analysis confirms Prader-Willi syndrome. This individual has shown deletion of the specific chromosomal region 15q11.2-q13 in the paternal allele which is causative of Prader-Willi syndrome. Genetic counseling recommended.

Note:

- The assay will not detect PWS cases with mosaicism.
- PCR is a highly sensitive technique, however inherent PCR inhibitors in the specimen result into amplification failure.
- Results of this test must always be interpreted in the context of clinicopathologic findings, family history and other relevant data.

Figure 2: DNA methylation report.

DISCUSSION

Prader Willi syndrome (PWS) is a rare genetic disorder. It was first described in 1956 by Prader and Willi et al.[2] The phenomenon of gene inactivation on selected chromosomal regions is known as genomic imprinting. This was the very first genetic disease described due to genomic imprinting.[3] Additionally, it is the most common genetic cause of obesity.[4] The characteristic feature of PWS in neonates is marked central hypotonia associated with decreased limb activity, weak cry and feeding difficulties. Therefore, it should be considered in the differential diagnosis of all hypotonic newborns.[5] Our patient presented with aspiration pneumonia secondary to feeding difficulty consistent with previous reports. The most common cause of central hypotonia in neonates is birth asphyxia leading to hypoxic ischemic encephalopathy. Other common differentials are intracranial hemorrhage, cerebral malformations, chromosomal abnormalities (like Down's syndrome), and congenital as well as acquired infections. Nevertheless, typical dysmorphology, dolichocephalism, almond-shaped eyes, thin upper lip, and narrow bifrontal diameter and cryptorchidism or mild scrotal hypoplasia are the key features. These physical findings conjointly with central hypotonia help to clinch diagnosis. In our case, these features led to suspicion of PWS.

Though diagnosing a neonate with a rare genetic disease is distressing for parents and challenging for clinician. Still establishing the diagnosis of PWS early is very important because it gives the opportunity of providing accurate and timely management. It also helps in decreasing associated morbidities. Diagnosis of PWS is based on clinical findings that change with age.[6] The clinical diagnostic criteria in neonates are central hypotonia with feeding problems, typical facial dysmorphology and hypogonadism. This disorder is two staged with early infantile and latter childhood phase. The infantile phase occurs from birth, to two years and characterized by failure to thrive and developmental delay. The second stage occurs around two years with predominantly neuroendocrinal (delayed development, short stature, hypogonadism, childhood obesity) and behavioral problems (sleep disorders, obsessive compulsive behavior, and mental retardation.[5]

Presently, methylation studies are considered the best screening as well as the gold standard tests. Methylation should be the first-line test to perform.[7] FISH and UPD (Uniparental disomy test) tests are indicated to determine the exact genetic subtype of defect. In majority of cases (75%), paternal deletion has been found. Maternal deletion accounts for one fourth (20-25%) of cases only. Micro deletions are noted rarely (1%). Multidisciplinary management (pediatrician, endocrinologist, orthopedic specialist, dietician, cardiologist, psychiatrist etc.) is necessary. Diabetes, cardiac failure, and respiratory disorders (respiratory insufficiency or infections) are major causes of morbidity and mortality in PWS patients.

CONCLUSION

To conclude, PWS should be considered as one of the differential diagnoses in a neonate presenting with central hypotonia with feeding difficulties. Early diagnosis and multidisciplinary team management will reduce morbidity and better clinical outcome can be achieved. In future, more studies from India (Asia) are required for clinical analysis of neonates with PWS and to understand the impact of early diagnosis (less than 28 days) and multidisciplinary intervention on quality of life, morbidity, and mortality in these children.

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