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Case Report

A Rare Occurrence of Prader Will Syndrome in a 9-Month-Old Tanzanian Girl: A Case Report

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Abstract

Prader-Will syndrome is a disarray caused by a depletion or disruption of genes in the proximal arm of chromosome 15 or by maternal disomy in the proximal arm of chromosome 15. In sub-saharan Africa the prevalence of Prader will syndrome is estimated to be rare genetic disorder with a birth incidence of 1/10,000 to 1/30,000, and an estimated prevalence of approximately 10,000 to 20,000 living individuals in the United States and Globally it is estimated to be for almost 2% of the paediatric patients with the Prader Will syndrome. It affects males and females equally, as well as all races and ethnicities in Tanzania the epidemiology of Prader Will syndrome is not yet to be documented and thus We report the first case being presented in sub-sahara Africa. To Our Patient since before diagnosed with (PWS) was well till the age of two months when she gradually started increasing in weight with time, as days elapsed the patient was constantly gaining weight, 2.9 kg at birth to 14 kg at 7 months. This Case Report discuss about the Rare incidence of occurrence of Prader Will syndrome in Tanzania and its properly Diagnostic criteria interventions and treatment Management plan.

Keywords: Prader-Willi Syndrome; Paediatric; Tanzanian; Growth Hormone Replacement

Introduction

Prader-Willi syndrome is a disarray caused by a depletion or disruption of genes in the proximal arm of chromosome 15 or by maternal disomy in the proximal arm of chromosome 15 [1]. Commonly associated characteristics of this disorder include diminished fetal activity, obesity, hypotonia, intellectual disability, short stature, hypogonadotropic hypogonadism, strabismus, and small hands and feet [2]. In sub-sahara Africa the prevalence of Prader will syndrome is estimated to be is a rare genetic disorder with a birth incidence of 1/10,000 to 1/30,000, and an estimated prevalence of approximately 10,000 to 20,000 living individuals in the United States and globally it is estimated to be for almost 2% of the paediatric patients with the Prader William syndrome [3]. It affects males and females equally, as well as all races and ethnicities in Tanzania the epidemiology of Prader will syndrome is not yet to be documented and thus we report the first case being presented in sub-sahara Africa. Prader-Willi syndrome is caused by the absence of paternally-expressed, imprinted genes on chromosome 15q11-13. Loss of activity can occur by one of three major genetic mechanisms [4]. Microdeletion of the paternally inherited chromosome (60% - 70%) of cases, further divided into type I and

type II, 7 megabases (Mb) and 5 Mb in size, respectively); maternal uniparental disomy subsequently to trisomic rescue (30% - 40% of cases) or mutation/epimutation of the Prader-Willi syndrome/ Angelman syndrome (PWS/AS) imprinting center 3% of cases with the Prader will syndrome [5].

To our patient since before diagnosed with (PWS) was well till the age of two months when she gradually started increasing in weight with time, as days elapsed the patient was constantly gaining weight, 2.9 kg at birth to 14 kg at 7 month with time the mother and nurses at the clinic noticed that the child had doubled the weight of her fellows but the mother was not really concerned by the time the child was 8 month her weight was 17 kg and she was neither speaking two words like mama/baba nor could she could be able to crawl but easy fatigue on crawling a short distance, now she is 9 month but she cannot even sit without a support nor can she stand. This case report discuss about the rare incidence of occurance of Prader William syndrome in Tanzania and its properly diagnostic criteria interventions and treatment management plan.

Case Report

A mother presented into our clinic with a 9 month-year-old girl with the chief complaints of a drastically, increasing in body weight. The patient was well till the age of 2 months when she gradually started increasing in weight with time, as days elapsed the patient was constantly gaining weight, 2.9 kg at birth to 14 kg at 7 month and with time the mother and nurses at the clinic noticed that the child had doubled the weight but the mother was not really concerned, by the time the child was 8 month a heading to 9 month her weight was 17 kg and she was neither not able to speak common children words but with easy fatigue on crawling a short distance. The mother reported that the child was low birth weight when she was born and was admitted to the ward for almost 1 week prior to discharge, the baby cried for 15 minutes late only after she was resuscitated and gave a weak cry, also had poor sucking ability. From the patient history a final conclusion was made to the paediatric that had poor or developed a slow milestone however she completed immunization and attended all clinics respectively, despite of that she had no any history of blood transfusion or any surgery being attended. On general examination the patient was awake, afebrile - T.36 C, pallor, jaundiced, symmetrical facial appearance, flabby cheeks enlargement, normal oral mucosa, tooth and tongue, Obese with normal head appearance, almond-shaped eyes, slightly depressed nasal bridge, thin upper lip, normal eyes and ear crease, but multiple skin folds around the neck, hands and abdomen, gynaecomastia noted and umbilical hernia noted, palpable LN/ obvious bone deformities or edema (sacral/limb) as seen in figure 1. Although the anthropometrics measures were presented as shown in table 1. Systemic examination to our patient revealed: hypotonia, hyporeflexia and reduced muscle bulkiness lower motor neurone lesion, normal cardiovascular, gastrointestinal or respiratory system and to this the final diagnosis was reached as (PWS) Prader William syndrome with the similar subjective findings as revealed to our patient, poor muscle tone as shown to figure 2. Delayed milestones/intellectual delay, speech delay, poor physical coordination, prominent nasal bridge, small hands and, feet with tapering of fingers, the laboratory findings were unremarkable. The further investigation such as, genetic testing: DNA methylation analysis and fluorescent *in situ* hybridisation (FISH) techniques, body composition analysis by dual-energy X-ray absorptiometry (DEXA), anthropometry or another method. DEXA is used to assess fat mass rather than bone mineral density in this case were suggestive to be performed as to proceed with the further management treatment plan of the patient.

Figure 1: Patient presented with increasing in body size and weight and poor developmental milestones.

Discussion and Conclusion

Prader-Willi syndrome is associated with a constellation of symptoms that significantly negatively impact quality of life for af-

S.NO	Items	Objective findings value
01	Body Weight	17.5 kg
02	Mid- upper arm circumference	14 cm both arm
03	Occipitofrontal Circumference	52 cm
04	Percentile	4.6
05	Height	93.4 cm
06	Waist	15 cm
07	Body Mass Index	14.5

Table 1: Anthropometric measurements as objective findings.

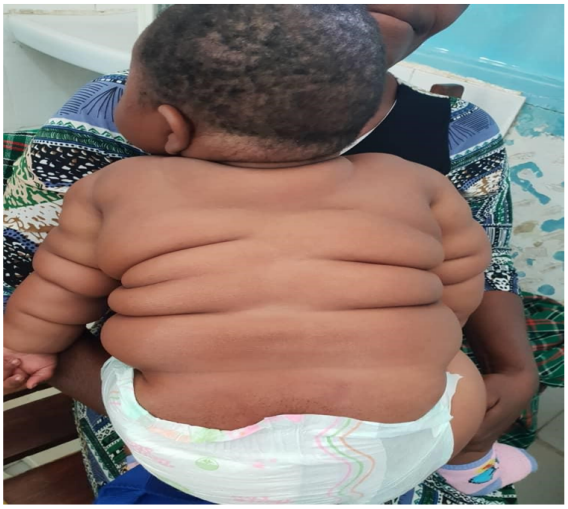

Figure 2: Our patient presented significance (PWS) symptoms with the poor muscle tone from backside view.

fected individuals and their families [6]. The initial clinical course of PWS is characterized by hypotonia in infants, with decreased movement, lethargy, feeding difficulties, and failure to thrive [7]. A defining feature of Prader-Willi syndrome is the change in appetite over time, with the onset of hyperphagia an unrelenting, pathologically excessive appetite sometime after early childhood [8]. To our patient she presented with the skin discoloration or rashes, symmetrical facial appearance, flabby cheeks enlargement, normal oral mucosa, tooth and tongue.

Obese with normal head appearance, almond-shaped eyes, slightly depressed nasal bridge, thin upper lip, normal eyes and ear crease, but multiple skin folds around the neck, hands and abdomen, gynaecomastia noted and umbilical hernia noted as presented and to this her mother reported to have noticed increase in body weight but the feeding habits have not increased as well. On physical examination the 9-month old girl was found to have on skin systemic with the normal skin coloration but multiple skin folds per limb, the upper limb was with joint and bulk muscles mass with hypotonia tone, hyporeflexia.

In this the same case which was diagnosed in Colombia in the case report observed by Estaphania., *et al.* which their findings was at birth, he was noted to have neonatal hypotony. During a follow-up visit at 3 months, psychomotor delay, strabismus and short stature were noted and the progressive the child deteriorated in mentally while his speech was noted to be deteriorating. Most cases of Prader-Willi syndrome are not inherited, particularly those caused by a deletion in the paternal chromosome 15 or by maternal uniparental disomy [9]. When left untreated obesity typically begins after 1 - 2 years of age and later exacerbated by hyperphagia resulting in lack of sense of satiety. The major cause of morbidity and mortality is morbid obesity [10]. PWS is caused by the loss of function of genes in a particular region of chromosome 15. People normally inherit one copy of this chromosome from each parent. Some genes are turned on active only on the copy that is inherited from a person's father (the paternal copy). This parent-specific gene activation is caused by a phenomenon called genomic imprinting [11]. In another 25 percent of cases, a person with Prader-Willi syndrome has two copies of chromosome 15 inherited from his or her mother (maternal copies) instead of one copy from each parent. This phenomenon is called maternal uniparental disomy [12,13]. Despite of that to our patient presented with this clinical features and with to this condition was characterized by lethargy and hypotonia causing poor feeding and failure to thrive, developmental and intellectual (mean IQ: 60 - 70s) disability, hypogonadism (small external genitalia and pubertal insufficiency), hyperphagia leading to morbid obesity [14]. A typical behavioural phenotype that includes temper tantrums and compulsive trait. Management is focused on anticipatory guidance and addressing the consequences of the syndrome and is very age-dependent family support is essential to cope with the behavioural difficulties throughout childhood [15].

The child must be kept away from excessive food intake to guard against subsequent obesity. Input from a paediatric gastroenterologist, endocrinologist, psychologist, psychiatrist, clinical dietician, occupational therapists, speech therapists, exercise advisors and orthopaedic consultants may be helpful [16]. Regular monitoring of physical parameters, psychological factors, behaviour, diet and exercise should be part of the treatment package [17]. Growth hormone replacement in Prader-Willi syndrome has resulted in dramatic benefits to the phenotypic health and self-image of those treated. However, till date no pharmacological therapy is effectively against the management of the disease. There is need for the regularly care and frequently check-up as to improve the overall diet and exercises for the paediatric patients.

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