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Prune Belly Syndrome: A Case Report in Barranquilla - Colombia

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Abstract: ***Prune Belly Syndrome is a rare disease, with incidence of 1 case per 30,000 live-born children, described since the 19th century by Frohlich, characterized by a set of congenital malformations with the triad of bilateral abdominal cryptorchidism, urinary tract malformations such as (megabladder, megaureters, urachus persistence, hydronephrosis and/or renal dysplasia) and weakness or partial or total absence of abdominal wall muscles. We report a case of a 36-week-old newborn with prenatal findings of alterations in the urinary tract via ultrasound. After birth, a very suggestive physical examination of prune belly syndrome was performed.***

Keywords: Prune belly syndrome, bilateral cryptorchidism and varus equine foot

1. Introduction

Eagle-Barrett Syndrome, also known as Prune Belly Syndrome (PBS) consists of a set of congenital malformations characterized by the triad of bilateral abdominal cryptorchidism, urinary tract malformations such as (megabladder, megaureters, persistence of urachus, hydronephrosis and/or renal dysplasia) and weakness or partial or total absence of abdominal wall muscles [1,2,3].
Described since 1839, when Frohlich showed the first case of a child who had a defect of the lateral abdominal muscles, chest in keel and lack of testicular descent, then in 1895 Parker associated this syndrome with malformations of the urinary tract (hydronephrosis, hydroureter and large bladder); in 1949 the American urologists Obrinsky, Barret and Eagle popularized the medical condition and Osler in 1961 described the wrinkled appearance of the abdominal wall with the term "Prune Belly", as it has been called at present [4,5].

The incidence of PBS in the world is about 1 per 30,000 births, it has been recorded that it predominates classically in men up to 95%, with a greater association in black newborns [6,7]. The cause of PBS is unknown, there are three theories suggested, which resembles the embryonic origin of the condition, as well as the appearance of associated genetic factors such as deletion in the factor HNF1β, necessary for visceral endodermal differentiation [8].

The initial diagnosis is clinical, with confirmatory imaging aids, it is important to make the diagnosis in a timely manner for its high mortality and to be able to provide adequate treatments and prevention of future complications that have a negative impact on the patient's quality of life [3].

Although the condition is generally known, socioeconomic factors, precarious health systems, lack of access to prenatal appointments with specialized personnel, late diagnosis, poor preparation of neonatal intensive care units, the absence of genetic counseling may be a consequence of high morbidity and mortality data in patients, which is why it is important to describe this type of syndromes in order to impact the medical scientific community and strengthen us even more to a better manage of PBS. We present a case report of Prune Belly syndrome identified in a third-level care institution in Barranquilla, Colombia.

2. Methodology

The methodology of this case report involves a comprehensive review of the medical history of a newborn diagnosed with Prune belly syndrome, combined with a review of relevant literature in databases such as PubMed, Medline and Embase to explain these findings.

3. Case Report

Newborn male of 36 weeks by Ballard, son of a 19-year-old first-time mother, blood type or rh-positive, no data of consanguinity with partner, no pathological or infectious history throughout pregnancy, during the controls was reported in the ultrasound of nuchal translucency high risk of aneuploidy by ultrasound markers, in addition to anatomical detail ultrasound showed bilateral renal multicystic dysplasia, megabladder and severe oligohydramnios (See Figure 1), however despite these prenatal diagnoses there was no documented follow-up by the gynecology team.

The patient is born in a third-level hospital in the city of Barranquilla, via Caesarean section indicated by anhydramnus and multicystic bilateral renal dysplasia, apgar score at 7/10 at 1 minute, at 5 minutes 8/10. Anthropometric measures suitable for gestational age, weight: 2,130 gr, size 47 cm. At the physical examination, the newborn's attention shows dysmorphic facies: low implantation ears, deficiency of the abdominal wall muscles, bilateral cryptorchidism, bilateral varus equine foot and clinodactyly of the fifth finger. Based on previous findings in ultrasound reports of

malformations of the urinary tract in utero and details of the physical examination, a diagnosis of PBS was made (see Figure 2).

Subsequently the patient presents an episode of hypotonia associated with widespread cyanosis, desaturation, extreme bradycardia, so advanced neonatal resuscitation is initiated, it is decided to ensure airway with orotracheal tube, adrenaline is administered, resulting in improved heart rate, coloration and muscle tone; once stabilized, the patient is transferred to the neonatal intensive care unit (NICU).

During his stay in the NICU chest X-ray was performed evidencing bilateral pneumothorax (see Figure 3), which is why the pediatric surgery service is called indicating bilateral thoracotomy and central venous catheter implantation due to difficult venous access, then the patient presents deterioration of ventilatory mechanics with new cardiorespiratory arrest, starting advanced cardiopulmonary resuscitation maneuvers without a satisfactory response so he is officially declared dead.

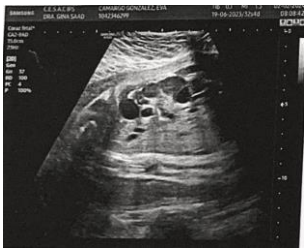

Figure 1: Intrauterine renal ultrasound, with multicystic renal dysplasia

Figure 2: Patient with Prune Belly syndrome, with absence of abdominal wall muscles, distended abdomen, thin wrinkled skin, bilateral cryptorchidism, low implantation of ears and varus equine foot

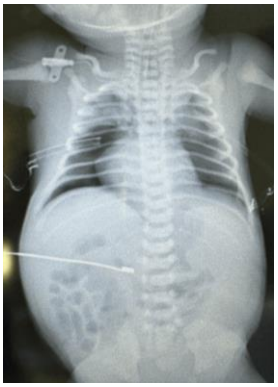

Figure 3: Bilateral pneumothorax is observed, with an atelectatic band in the upper lobe of the right lung.

4. Discussion

Prune Belly syndrome (PBS), also known as Eagle Berret syndrome, described since the 19th century [4,5], corresponds to a rare congenital alteration, with a characteristic triad that includes: hypoplasia or absence of abdominal wall muscles, moderate to severe dilation of the urinary tract and bilateral cryptorchidism [1,2,3]. It has an incidence of 1 case per 30,000 live births. It occurs mostly in males and less than 5% in females [6,7].

PBS is known as a very serious condition, with alarming mortality and complications. There are 20% of patients who are born without life, and 30% die during their initial hospitalization, such as our patient, also, the remaining 50% face urinary problems throughout their lives, with a high incidence of chronic renal failure (67%). Other worrying data include prematurity in 43% of live births, the need for intubation in 48%, and 25% of live births presenting congenital heart abnormalities [9,10,11,12].

Although various explanations have been suggested, the exact cause of prune belly syndrome has not yet been determined. There are three theories that have greater repercussion in the scientific community, one of them is the complex of obstruction and urethral malformation, which proposes that an obstruction in the urethra during embryonic development can lead to distension of the bladder, affecting the development of the urinary tract, abdominal wall, and testicular descent [13]. The second theory attributes this syndrome to a defect in the yolk sac [14], and the third one, raises a possible defect in the lateral plate of the mesoderm, which is the embryonic structure from which the ureters, the bladder, the prostate, the urethra and the gubernaculum are derived [15], data on genetic alterations, such as mutations in the factor HNF1β, necessary for endodermal visceral differentiation, have been described [8].

PBS can be diagnosed prenatal when there is a trained multidisciplinary team, there are even data that report therapeutic alternatives at this stage, as in cases of severe obstructive uropathy where vesical decompression is performed by placing percutaneous catheter in the bladder to prevent the development of oligohydramnios, the involvement of renal function and therefore decrease lung involvement [1, 2, 3, 16].

Prune belly syndrome can be diagnosed prenatally when there is a trained multidisciplinary team, there are even data that report therapeutic alternatives at this stage, as in cases of severe obstructive uropathy where vesical decompression is performed by placing percutaneous catheter in the bladder in order to prevent the development of oligohydramnios, the involvement of renal function and therefore decrease lung involvement [1,2,3,16]. Postnatal management will be aimed primarily at improving renal and lung function, which will have a variable impact on the health and quality of life of the child, thus the need for a surgical approach according to the need of the patient can be divided into three categories: urinary tract reconstruction, abdominal wall reconstruction and orchidopexy [16,17].

It is very important to emphasize that management must be adapted to everyone, achieving a balance between early intervention and unwanted effects to improve survival and limit sequelae.

Although our patient dies during the first 24 hours of life, there are reports of cases such as those described by Arias et al [18] and Urquieta Maldonado et al [19], where patients are treated early, performing follow-up protocols from prenatal and postnatal stages, something that differs with our patient, where they find anomalies of the urinary tract by ultrasound and unfortunately there is no multidisciplinary follow-up of the condition.

5. Conclusions

Prune belly syndrome is a rare disease, often unknown to doctors and primary care teams. This condition shows a variable severity from the neonatal to postnatal period, requiring an early intervention and a multidisciplinary team for the adequate care of these patients who are born with a very high risk of mortality.

6. Future Scope

Our case of PBS aims to analyze the complexity of this pathology in the neonatal period, that often the outcome is fatal, so it is necessary to know the pathology and know how to implement a protocol of directed care in all the delivery rooms and neonatal intensive care units. This information provides the presentation of a PBS case and updated knowledge of the pathology.

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