CLINICAL CASE

Prune belly syndrome: a case report and literature review

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Prune belly syndrome; Abdominal muscle deficiency; Cryptorchidism; Urinary tract anomalies

Abstract
Prune belly syndrome is a rare malformation that presents in 1 out of every 40,000 live births. It occurs almost exclusively in males (> 95%) and is characterized by the following triad: urinary tract anomalies, abdominal muscle deficiency, and bilateral cryptorchidism. The protruding abdominal wall resembles a dried prune. A 28-month-old boy presented with a prominent abdomen, visible outline of the intestinal segments, central abdominal muscle deficiency, and undescended testes. He had grade V bilateral vesicoureteral reflux, a large-capacity bladder, and megaureters. We performed abdominal wall reconstruction with the Monfort technique, bilateral orchidopexy, and ureteral remodeling with a bilateral Cohen ureteral reimplantation. Reduced abdominal circumference, reduced ureteropelvic ectasia, corrected ureteral reflux, and retention of the testes in the scrotum were observed in the postoperative progression.

One-third of these patients progress to renal failure. The grade of dysplasia, cystic disease, and baseline serum creatinine > 0.7 mg/dl are risk factors for renal deterioration. Abdominal wall reconstruction is performed for esthetic purposes, but improved bladder and bowel emptying and pulmonary function have been suggested. Early orchidopexy is recommended.

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Presentamos el caso de un paciente varón de 2 a˜nos 4 meses. El abdomen era prominente, con dibujo de asas intestinales, deficiencia de la musculatura central abdominal y testículos no descendidos. Presentaba reflujo vesicoureteral bilateral grado v, vejiga de gran capacidad y mega-uréteres. Realizamos reconstrucción de la pared abdominal tipo Monfort, orquidopexia bilateral y remodelación con reimplante ureteral bilateral tipo Cohen. En la evolución posquirúrgica observamos una reducción de la circunferencia abdominal, disminución de la ectasia urétero-piélica, corrección del reflujo ureteral y permanencia de los testículos en escroto.

Un tercio de los pacientes progresarán a falla renal. El grado de displasia, la enfermedad quística y una creatinina sérica basal > 0.7 mg/dl son factores de riesgo para el deterioro renal. La reconstrucción de la pared abdominal tiene un fin estético; se ha observado también mejoría en el vaciamiento vesical, intestinal y en la función pulmonar. Se recomienda la orquidopexia temprana.

Criptorquidia; Anomalías del tracto urinario

Introduction

Prune belly syndrome (PBS) is a rare malformation that presents in one out of every 40,000 live births, almost exclusively in males (> 95%). It is characterized by the triad of: urinary tract anomalies, abdominal muscle deficiency, and bilateral cryptorchidism. The protruding and hypoplastic abdominal wall resembles a dried prune, thus the syndrome’s name. Urinary tract malfunctions are due to smooth muscle dysplasia in the pelvis and ureters. Depending on type and severity, there are 3 different clinical presentation forms: 1) nonviable oliguric form with severe renal dysplasia, 2) marked renal dysplasia, megaureters, progressive renal failure, and 3) the most favorable form with moderate renal dysplasia and different grades of ureteral and bladder dilation.1-3

Case presentation

A 28-month-old male was diagnosed with bilateral renal ectasia in the second trimester of gestation and he was delivered through cesarean section. He underwent a vesicostomy at one month of age and presented with bilateral hip dysplasia. Physical examination revealed a prominent, distended abdomen bulging at the flanks. The intestinal segments were visible on the skin, muscle deficiency was palpated at the midline of the abdominal wall, and no testes were palpated in the scrotum. The patient presented with grade V bilateral vesicoureteral reflux and the cystogram showed a megabladder, megaureters, and bilateral hydronephrosis. Upon arrival at our service, the child presented with a creatinine level of 1.9 mg/dl, urea 143.4 mg/dl, creatinine depuration of 17.6 ml/min/1.73 with the Schwartz formula, and grade IV renal insufficiency. The intra-abdominal testes of the patient were located and one-stage bilateral orchidopexy was performed (figs. 1 and 2). Posteriorly, abdominal reconstruction was carried out through the Monfort technique (figs. 3 and 4) and ureteral remodeling with bilateral Cohen ureteral reimplantation. The patient’s postoperative progression was satisfactory; both the abdominal circumference and ureteropelvic ectasia were reduced, bilateral ureteral reflux was corrected, bladder voiding was adequate, and the testes remained in the scrotum.

Figure 1 Location of the intra-abdominal testes and their descent.

Figure 2 Orchidopexy.
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Discussion

Beta factor 1 nuclear hepatocyte gene implication in the etiology of this syndrome has recently been suggested, based on the reports of 2 cases. Granberg et al. ruled out the possibility of this gene’s implication when they detected it in only one of 32 patients studied and therefore the genetic base of this disease is still undetermined.4

Close to one-third of the patients that survive to the postnatal period will progress to renal failure, requiring future kidney transplantation. The early identifiable risk factors are the grade of renal dysplasia and cystic disease. The most specific marker for significant dysplasia is baseline creatinine, which is the lowest level of creatinine in the first 12 months of life. Noh et al. reported that a baseline creatinine > 0.7 mg/dl and clinical pyelonephritis are prognostic factors for renal failure.5,6 In the case of our patient, he already had a serum creatinine level of 1.9 mg/dl, signifying poor renal function prognosis.

Orthopedic malfunctions vary between 30-40% of the patients and the musculoskeletal system is the second most affected in frequency after the genitourinary system in relation to this syndrome. Hip dysplasia and atrophy of a given limb are common.1

Dysplasia and hypoplasia of the central abdominal muscles cause the particular appearance of the abdomen. A disorder of the myofilaments and Z-line disorganization have been observed under the electron microscope. The 3 muscle layers under the umbilicus are especially affected, and the muscle can be completely replaced by fibrous tissue, whereas the peripheral muscles are partially or totally spared.5

Advancement of the well innervated and vascularized peripheral musculature is the basis of abdominal wall reconstruction, with or without excision of the affected muscles and fascia.5 Abdominal wall reconstruction is performed for esthetic purposes, but consequent improvement in bladder and bowel emptying has also been observed, as well as better pulmonary function due to the greater intra-abdominal pressure that the reconstruction may exert. However, further studies are needed to provide a better basis for these latter benefits. There are different techniques for performing abdominal reconstruction. The Monfort technique involves an elliptical incision from the xiphoid process to the pubic symphysis. A second incision is made at the umbilicus to keep it in situ. The dissection of the fascia and muscle is extended to the anterior axillary line. Two vertical incisions are made on both sides of the fascia over the central bridge, increasing the abdominal wall thickness and reducing the redundant tissue. This technique was first described in 1991 and performed on 9 patients with satisfactory results in them all.7-9

Ureteral reconstruction is indicated when there is progressive deterioration of renal function or persistent urinary tract infections, for the purpose of reducing urinary stasis.1

The renal collecting system is characteristically dilated, but the grade of dilation is not related to the grade of dysplasia. Vesicoureteral reflux is present in 75% of the children with PBS.9

Bilateral cryptorchidism with abdominal testes is one of the main characteristics of PBS. The mechanical obstruction produced by bladder and urinary system dilation, added to abdominal wall deficiency, appear to be key factors in the absence of testicular descent. The gubernaculum, equally important in relation to descent, has been observed to be atresic or abnormally adhered to the pubic tubercle.9

Early orchidopexy, around 6 months of age, at the time of performing the other necessary corrective surgery, is recommended in these patients.1,9

Conclusions

The genetic base of PBS has yet to be determined. Progression to renal failure is common in these patients. The presence of serum creatinine > 0.7 mg/dl is an important prognostic factor for renal function. Orthopedic abnormalities are second in frequency, after those of the genitourinary system and the abdominal wall. At present,

Figure 3 Abdominal reconstruction with the Monfort technique. Advancement of the peripheral musculature.

Figure 4 Abdominal reconstruction with the Monfort technique. Approximation of the skin.
abdominal wall reconstruction is performed for reasons of esthetics, but benefits in bladder and bowel emptying and in pulmonary function have been suggested, requiring further studies for their confirmation. The Monfort technique is a good reconstruction option. Ureteral reconstruction is indicated when there is progressive renal function deterioration or persistent urinary tract infections, to reduce urinary stasis. Early orchidopexy, at around 6 months of age, and together with other corrective surgery, is recommended in these patients.

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References


Ethical responsibilities

Protection of persons and animals. The authors declare that no experiments were performed on humans or animals for this study.

Data confidentiality. The authors declare that they have followed the protocols of their work center in relation to the publication of patient data.

Right to privacy and informed consent. The authors have obtained the informed consent of the patients and/or subjects referred to in the article. This document is in the possession of the corresponding author.

Financial disclosure

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Conflict of interest

The authors declare that there is no conflict of interest.