Prune Belly Syndrome

Dr Hasan Nugud
Consultant
Paediatric Surgeon
Prune Belly Syndrome

Prune belly syndrome, also referred to as abdominal muscle deficiency syndrome, congenital absence of the abdominal muscles, Eagle-Barrett syndrome, Obrinsky syndrome, Fröhlich syndrome, or, rarely, triad syndrome, is a rare, genetic birth defect affecting about 1 in 40,000 births. About 97% of those affected are male. Prune belly syndrome is a congenital disorder of the urinary system, characterized by a triad of symptoms (1/gross ureteric dilatation, 2/anterior abdominal wall underdevelopment (resulting in the "prune belly" appearance), 3/ Bilateral undescended testes (cryptorchidism)),

The syndrome is named for the mass of wrinkled skin that is often (but not always) present on the abdomen of those with the disorder.
Prune Belly Syndrome

Symptoms
• A partial or complete lack of abdominal wall muscles. There may be wrinkly folds of skin covering the abdomen.
• Cryptorchidism (undescended testicles) in males
• Urinary tract abnormality such as unusually large ureters, distended bladder, accumulation and backflow of urine from the bladder to the ureters and the kidneys (vesicoureteral reflux)
• Frequent urinary tract infections due to the inability to properly expel urine.
• Later in life, a common symptom is post-ejaculatory discomfort. Most likely a bladder spasm, it lasts about two hours.
• Ventricular septal defect
• Malrotation of the gut
• Club foot
Prune Belly Syndrome
Diagnosis
Prune belly syndrome can be diagnosed via ultrasound while a child is still in-utero. Frequent urinary tract infections often herald prune belly syndrome voiding cystourethrogram.
Diagnosing a baby with Prune-Belly Syndrome is done by doing or undergoing the following examinations:
• Physical examination
• Ultrasound examination
• Blood test or examination
• X-ray examination
• Voiding cystourethrogram or VCUG examination
• IVP exam or Intravenous pyelogram examination
• Further Imaging Examination such as Tc 99m DMSA scintigraphy
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Marked distention of the abdomen and bulging flanks secondary to a large urinary system and the absence of abdominal wall musculature.
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*Urinary tract abnormalities include*
- bilateral hydroureteronephrosis:
  - often with extremely dilated, tortuous ureters
  - varying degrees of renal dysplasia
- enlarged urinary bladder, often with *urachal diverticulum*
- vesicoureteral reflux is common
- poor bladder contractility
- dilated posterior urethra without urethral obstruction
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Ultrasound examination of the kidneys shows bilateral hyperechoic kidneys with poor development of the calyces and dilated tortuous ureters.
Prune Belly Syndrome

On CT, the ureter in this patient with prune belly syndrome fills the entire abdomen.
Babies diagnosed with Prune Belly syndrome have or manifest the following signs, symptoms and characteristics:

- Lungs are underdeveloped
- Anomalies or defects of the cardiac system
- Wrinkled prune like abdomen
- Renal collecting structures are dilated
- Bladder is dilated
- Anomalies in the gastrointestinal system
- Talipes or club foot
- Abnormal limbs
- Constipation
- Appearance of a Little Buddha
- Delayed walking and sitting
- Frequent urinary tract problem
- Undescended testicles (Cryptorchidism)
- Azoospermia
Prune Belly Syndrome

Complications
-Surgery is often required but will not return the organs to a normal size.
-Bladder reductions have shown that the bladder will again stretch to its previous size due to lack of muscle. - Complications may also arise from enlarged/malformed kidneys, which may result in renal failure and the child's going on dialysis or requiring a kidney transplant.
-With proper treatment, however, a longer, healthier life is possible.
-Musculoskeletal abnormalities include pectus excavatum, scoliosis, and congenital joint dislocations including the hip.
-Diagnosis of prune belly syndrome necessitates a thorough orthopaedic evaluation because of the high prevalence of associated musculoskeletal abnormalities.
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Complications
The common complications that are associated with Prune-Belly Syndrome are as follows:
• Deformities of the bone structures such as funnel chest, clubbing of foot, dislocation of the hips, missing digit or limb
• Urinary tract disease which may lead to kidney transplant or dialysis
• Constipation
• Undescending of the testicles, leading to cancer or infertility
• Lung is underdeveloped or medically termed as pulmonary hypoplasia
• Poor coughing mechanism which leads to a possibility of pneumonia
• Gastrointestinal tract abnormal fixation
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Treatment
One option is to perform a vesicostomy helping to prevent urinary tract infections. Similarly, consistent self catheterization, often several times per day, can be an effective approach to preventing infections. Surgical "remodeling" of the abdominal wall and urinary tract. Boys often need to undergo an orchiopexy.
Prune Belly Syndrome

Immediately after birth
A catheter was inserted to the bladder and well drained
achieving
Better stability of the newborn
Prune Belly Syndrome

Associations
aneuploidic syndromic associations
  trisomy 18
  trisomy 13
other associations
  congenital cardiac anomalies
    ventricular septal defect (VSD)
    atrial septal defect (ASD)
    tetralogy of Fallot
  intestinal malrotation
  imperforate anus
  polydactyly/syndactyly
  talipes equinovarus
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Differential diagnosis
For antenatal hydronephrosis with hydroureter, consider:
*posterior urethral valves*: may show a key hole sign and no evidence of cryptorchidism
*megacystis microcolon intestinal hypoperistalsis syndrome*: tends to have polyhydramnios and more females affected
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Prognosis & Life Expectancy
This kind of syndrome is a life threatening and needs to be treated appropriately. The prognosis varies from one person to another. However, reports say that the average life expectancy of patients with this kind of syndrome is not long after birth. It can range from either being stillborn or premature death within their first week, because of kidney or lung problems at birth. In general, the prognosis will greatly depend on the functioning of the patient’s renal system. Some patients die right at birth, while there are some who survive. Those who survive will continue to encounter more problems for as long as they live. The main reason for those who survive is because the fertility interventions given were modern and their bodies adapted to the kind of interventions given early on. In general, there is a poor prognosis of patients who are diagnosed with Prune Belly syndrome.
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Prevention
Since it is a fetal developmental syndrome, the prevention is targeted in a fetal anomalies screening, something which is routinely done during pregnancies. There is a possibility of terminating the pregnancy if given the choice or perhaps a possibility of an intrauterine surgical procedure in order to prevent the further development of prune-belly syndrome. This kind of surgical procedure, although rarely done, according to studies conducted, is considered promising. Surgical procedure is recommended for the earliest phase of the syndrome. It seems to be the only preventive way in extending the life of the patient suffering the disease.