# Abdominal Muscles, Absence of, with Urinary Tract Abnormality and Cryptorchidism

## Alternative Names
Prune Belly Syndrome
Eagle-Barrett Syndrome
EGBRS

## Record Category
Disease phenotype

## WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Congenital malformations and deformations of the musculoskeletal system

## Incidence per 100,000 Live Births
6-10

## OMIM Number
100100

## Mode of Inheritance
Autosomal dominant; autosomal recessive suggested by some reports

## Gene Map Locus
1q43

## Description
Prune-belly syndrome is characterized by a triad of abnormalities that include, absence of the lower portion of the rectus abdominis muscle and the inferior and midportions of the oblique muscles (causing the skin of the abdomen to wrinkle like a prune), undescended testicles (a condition seen in newborns whereby one or both of the male testes has not passed down into the scrotal sac), an abnormal, expanded bladder and problems in the upper urinary tract, which may include the bladder, ureters, and kidneys. Prognosis may vary from death in utero to a near-normal life expectancy.

Prune-belly anomalies are rare in children and occur mostly in boys. Several cases have been reported in girls (incomplete prune-belly syndrome) and are usually milder than in boys. As males suffering from PBS are sterile, they cannot transfer pathological genes, that is to say the disease itself.

## Molecular Genetics
There is evidence that absence of abdominal muscles with urinary tract abnormality and cryptorchidism is caused by homozygous mutation in the CHRM3 gene on chromosome 1q43. The M3 cholinergic receptor mediates autonomic neurotransmission in the ocular iris pupillary sphincter and the detrusor muscle in humans.

## Epidemiology in the Arab World
**Lebanon**
Afifi et al. (1972) described an affected offspring with prune-belly syndrome born to first-cousin parents.

**Oman**
Rajab et al. (1996) conducted a retrospective study to determine the incidence of posterior urethral valves (PUV) in Oman and investigate the effect of genetic and environmental factors on the condition. As all infants with congenital urinary obstruction were referred to a single pediatric surgery unit in the Royal Hospital for management, this unit's register was used, from which data of all Omani patients with PUV who were managed during the period of eight years (1987 to 1994) was obtained. During this period, 84 Omani individuals with PUV were fulgurated. Over 90% of these patients presented just after birth or within the first six months (probable type I PUV). The calculated incidence of PUV by using this data was 1 in 2375 males, which was higher than that reported in UK. Upon analyzing the dates of conception of these children, no seasonal variation was detected, as similar numbers of cases were conceived in the hot season and in the cooler season. On the other hand, this study showed marked regional variation with the highest incidence in the capital, Muscat (1 in 2000 males) and in the mountainous interior district, Dakhilya (1 in 1700 males) while the lowest incidence was seen in the southern district, Dhofar. Out of the 47 families in which

information about consanguinity was available, consanguinity was present in 87% of the parents, with 54% being first cousins (versus 24% in the general population), 11% were second cousins (similar to the general population) and 24% of the parents were from the same tribe (versus 22 in the general population). Analyzing the tribal names in 78 cases with available relevant information revealed that 11 patients (14%) came from one of the larger tribes, and six patients (7%) came from a second tribe, while 33% of the cases, each came from a single tribe. In 34% of the 84 children, other congenital anomalies were detected, with the commonest being genitourinary malformations in 17% (crytorchidism, hypospadias, meatal stenosis, ureteric diverticulum, absent kidney, duplex kidney, dysplastic kidney and PUJ obstruction), followed by gastrointestinal abnormalities in 9% (inguinal hernia, anal stenosis, Hirschprung disease, and high anorectal anomaly), and spinal lesions in 7% (spina bifida, hemivertebrae, spondylo-thoracic dysplasia, and scoliosis). In many children, several malformations occurred, but no specific syndromes were recognized. The segregation analysis with a p value of 0.013 suggested that autosomal recessive inheritance was not the causative factor of increased incidence in the Omani population. Rajab et al. (1996) recommended screening of the brothers of affected patients to prevent the irreversible renal deterioration and to use these data in further studies to determine the genetic factors predisposing to the malformation.

Sawardekar (2005) conducted a study to establish the prevalence of major congenital malformations in children born during a 10-year period in an Omani hospital in Nizwa. Of the 21,988 total births in the hospital, two children were born with Prune Belly Syndrome.

**Qatar**

Akl and Zayyoud (1983) reviewed the medical records of 83 children who presented with renal disorders in Qatar during a single year period. One patient with chronic renal failure was found to have the Prune Belly syndrome.

**Saudi Arabia**

Al Harbi (2003) reported the first case recorded in the literature of a girl born to a diabetic mother who was found to have Down syndrome and prune-belly anomalies (bilateral gross hydronephrosis, megaureter, and megacystis with abdominal muscle deficiency). The girl also had an atrioventricular septal defect. Diagnoses were confirmed with a cytogenetic study and micturating cystourethrography. She died at 29 days of age with a sudden collapse, most likely due to sepsis.

**Tunisia**

Boutheina et al. (2000) carried out 43 prenatal diagnoses of lethal urinary tract abnormalities during a five-year-period. The abnormalities detected included bilateral renal agenesis (56%), autosomal recessive polycystic kidney disease (16%), autosomal dominant polycystic kidney disease (14%), Meckel-Gruber syndrome and Prune-Belly syndrome (4%). The pregnancy was interrupted in 35 cases (81.4%).

**United Arab Emirates**

Al Talabani et al. (1998) studied the pattern of major congenital malformations in 24,233 consecutive live and stillbirth at Corniche hospital, which is the only maternity hospital in Abu Dhabi, between January 1992 and January 1995. A total of 401 babies (16.6/1,000), including 289 Arabs, were seen with major malformation. Single gene disorders accounted for 24% of the cases, 76% were due to autosomal recessive disorders. In their study, Al Talabani et al. (1998) observed one case of prune-belly syndrome born to first cousin parents from the United Arab Emirates. No recurrence was reported in the family. Al Talabani et al. (1998) concluded that their study was very close to representing the true incidence of congenital abnormalities in the whole United Arab Emirates, as they investigated over 98% of deliveries in Abu Dhabi, the capital of United Arab Emirates.

## References


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Akl K, Zayyoud M. The spectrum of childhood kidney disease in Qatar. Qatar Med J. 1983; 4(2):95-7.

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Sawardekar KP. Profile of major congenital malformations at Nizwa Hospital, Oman: 10-year

review. J Paediatr Child Health. 2005; 41(7):323-30. PMID: 16014135

## Related CTGA Records

Down Syndrome

## External Links

http://www.emedicine.com/radio/topic575.htm
http://www.nlm.nih.gov/medlineplus/ency/article/001269.htm
http://www.prunebelly.org/prune2.html
http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Prune%20Belly%20Syndrome

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