Absence of Abdominal Muscles with Urinary Tract Abnormality and Cryptorchidism

**Alternative Names**
Prune Belly Syndrome  
Eagle-Barrett Syndrome  
Triad Syndrome  
Abdominal Muscular Deficiency Syndrome

**WHO International Classification of Diseases**
Congenital malformations, deformations and chromosomal abnormalities

**OMIM Number**
100100

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

**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**
The etiology and pathogenesis of prune-belly syndrome are still quite vague, despite very intensive investigations performed in that field. According to data in the literature, disturbances responsible for the occurrence of PBS can be induced by genetic susceptibility, chromosomal anomalies, and other factors. The condition similar to PBS has been induced experimentally in animals by gene mutation (Danforth's short-tailed rats).

**Epidemiology in the Arab World**

**Lebanon**
Afifi et al. (1972) described an affected offspring with prune-belly syndrome born to first-cousin parents

**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

Contributors
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