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Case Report
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Prune-Belly Syndrome in A Libyan Boy with Down Syndrome: A case report

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ABSTRACT

Prune belly syndrome is a rare congenital anomaly of uncertain etiologies. It is characterized by deficiency of abdominal muscles, undescended testes, and severe urinary tract abnormalities. The association between prune belly syndrome and Down syndrome is very rare. We report the first case of 12 hours old term Libyan male who was found to have features of Down syndrome and Prune-belly anomalies.

Key word- *Down syndrome; Prune Belly syndrome; Obstructive uropathy.*

NTRODUCTION

Prune belly syndrome (PBS), also known as "Eagle- Barrett Syndrome" was first described by Frolich in 1839. Although it was thought to be more common in people of African descent in USA, there were few reports of this syndrome from African countries.[1-3] It has been reported in siblings indicating an Autosomal pattern of inheritance.[4] Mainly boys were affected, only 3-4% of non-familial forms were females which suggested X Linked or autosomal recessive pattern. PBS is a group of birth defects that involve three main problems; Poor development of abdominal muscles, causing skin of the belly area to wrinkle likes a prune, cryptorchidism, and urinary malformations which may include hydro ureter and hydronephrosis, and/or vesicouretral reflux. The margins of the liver and spleen, as well as bowel loops are visible. Thoracic deformity with flaring of the lower ribs and impaired diaphragmatic functions have been reported.[5] Antenatal ultrasound may show maternal oligohydramnios, hydronephrosis and distended urinary bladder. In some cases, antenatal ultrasound may help to detect other anomalies like ventriculo septal defect, atrial septal defect, and tetralogy of fallot have been reported in 10% of patients. Musculoskeletal abnormalities, clubfoot, gastrointestinal tract problems, malrotation with mesenteric defect, imperforate anus, gastroschisis, and Hirschsprung disease and or underdeveloped lungs; hypoplasia and atelectasis were reported.[4] PBS affects 1 per 30,000-40,000 live births, predominance of male, and 4% of all cases were products of twin pregnancies.[6-8] Prune belly syndrome has been reported to be associated with Trisomy 18 and Trisomy 21.[9-11] In India in 1987, two infants with rare manifestations of prune belly syndrome were described, besides the usual features, the first infant had Down syndrome, and the second infant had arthrogryposis multiplex congenita, bilateral talipes equinovarus and low set malformed ears.[12] Unfortunately, 20% of babies with Prune Belly syndrome may die before birth (Still birth), and 50% die from renal disease in the first two years of life. The remaining 30% of infants had varying degrees of urinary problems.[13] There is no cure for this syndrome, but intensive antibiotics for urinary tract infection and early surgery for abdominal muscles, urinary tract problems, and undescended testicles are recommended.[13]

CASE REPORT

A 12 hours old Libyan male baby referred from peripheral hospital to Special care baby unit at Tripoli Medical Centre as Down syndrome with generalized hypotonic, poor sucking, abdominal distension, and anuria.

Birth history: Post term baby boy delivered by emergency cesarean section due to fetal distress to 38 years old mother G9 P6 + 2 abortions, no antenatal care, birth Wight of 4.300Kg and delayed crying after delivery.

Physical Examination: Term male baby with clinical feature of DS (Figure 1), global hypotonia, not tachypnic, not distressed with no cyanosis, and no skeletal deformity. The baby has pan-systolic murmur with good perfusion, a complete AV canal confirmed by echocardiography. Abdomen appears distended with deficient muscle wall, bulging flanks, mild wrinkled abdominal skin (Figure 2,3), visible peristalsis, supra pubic mass up to umbilicus with dullness on percussion (full urinary bladder), bilateral undescended testes and normal anus (Figure 2).

After emptying the bladder ultrasound of abdomen showed mild hydronephrosis of both kidneys with few tiny cortical

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cysts and mild ascites.
Micturating-cystouretrography study showed grade V bilateral vesicoureteric reflux.
Investigations: Blood sugar, blood gases, CBC and chest x ray were normal, C-reactive protein and viral screen were negative, urea was 42 mg/dl, creatinine was 1.3 mg/dl, Sodium and Potassium both were normal, 24 hours urine collection for creatinine 40 (NR 14-36 mg/kg/24hours), and chromosomal study revealed ~ 47XY + 21. The baby was referred to pediatric nephrologist but unfortunately lost in follow up.

Figure 1: Feature of Prune-belly syndrome and Down syndrome.

Figure 2: FPS (Abdomen distended with deficient muscle wall, bulging flanks, mild wrinkled abdominal skin, and bilateral undescended testes).

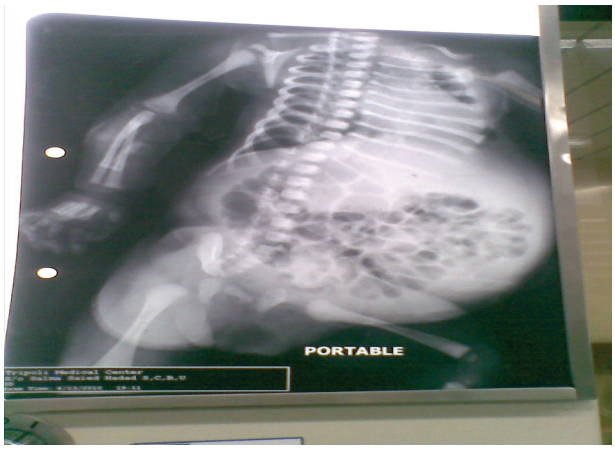

Figure 3: Plane X ray (Abdomen distended, dilated bowel, and bulging flanks).

DISCUSSION

The association between PBS and Down syndrome was reported in few cases.[8-14] The cause of this association is still unknown. Down syndrome has been associated with renal hypoplasia, hydroureter hydronephrosis; ureterovesical and ureteropelvic junction obstruction, posterior urethral valve and vesicoureteric reflux.[15-17]
The etiology and pathogenesis of PBS are still not clear, despite very intensive investigations performed in that field.[17] Various theories have been proposed to explain clinical features of PBS; fetal outlet obstruction theory, theory of mesodermal arrest and yolk sac theory. Unfortunately none of above explains the entire components of PBS.[18]
Clinical and pathologic experience with several cases of prune-belly syndrome indicates that urogenital anomalies can be attributed to a functional urethral obstruction which is the results of prostatic hypoplasia (mechanical cause).[19] In1991 report of two cases of Prune Belly syndrome (female and male newborns) were reported with urethral obstruction and other associated malformations: imperforate anus, vaginal septae and bicornate uterus in female case; unilateral anorchia and hyaline membrane disease in male. These findings support mechanical pathogenic theory. In cytogenetic studies no chromosomal abnormalities were detected. Both karyotypes were normal.[12] PBS has been reported without genetic alteration or other genetic syndromes.[8-10, 14-20]
In 2003 Al Harbi reported first case of a girl born to a diabetic mother who was found to have Down syndrome and Prune Belly anomalies[12], in 2005 one case of Prune Belly syndrome in a Nigerian Child with Down syndrome was reported[14], in 2008 a report of an Egyptian infant with Down syndrome and prune belly syndrome.[11]
In Japan 2007 a report of two siblings diagnosed as having Beckwith-Wiedemann syndrome, in addition to Beckwith-Wiedemann syndrome, one of the siblings was also diagnosed with Prune Belly syndrome and the other sibling suffered from obstructive uropathy and unilateral cryptorchidism, which are also seen in prune belly syndrome. These two cases point to a potential association between Beckwith-Wiedemann syndrome, Prune Belly syndrome, and urinary tract anomaly.[21] In 2007 PBS with VATER/VACTERL association was reported. This association is extremely rare.[22]
It has been recognized recently that many genes involved in renal nephrogenesis either reappear or are expressed to a markedly greater degree in renal disease. Siebert and Walker reported on the recurrence of urethral stenosis/atresia in 2 sibling fetuses with bladder outlet obstruction, severe oligohydramnios, pulmonary hypoplasia, and prune belly syndrome, a micro deletion of 6p25.3 was identified in the mother and one fetus, but noted that it is not associated with a gene known to be involved in urethral development and was therefore of unknown significance.[23]

CONCLUSION

Down syndrome may be associated with prune belly syndrome. Both had renal complications, which may suggest that prune belly sequence occurs secondary to obstructive uropathy.

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ETHICAL CONSIDERATION

A verbal permission from mother was granted before taken photographs and for possible publication.

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