Congenital Absence of Abdominal Muscles: The Prune - Belly Syndrome

A Case Report

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The clinical syndrome known as congenital absence of abdominal muscles was first described in 1839 by Frohlich.[1] In1895 Parker[2] noted marked urogenital anomalies in a patient with deficient abdominal muscles. Subsequently, additional cases have been reported with increasing frequency, so that more than 120 cases are now documented.[3]

This clinical entity is characterized by hypoplasia or absence of distinct groups of abdominal muscles. There are usually associated urogenital anomalies, such as hydronephrosis and hydro-ureter, distended bladder with thickened, trabeculated wall, obstructing lesions in the lower urinary tract, congenital renal dysplasia and bilateral cryptorchism. Skeletal anomalies of the hips and pectus excavatum may also be present; however, malrotation of the intestinal tract and cardiac abnormalities are less often encountered. Respiratory tract infections are frequent among infants with this syndrome, probably because of their inability to give a forceful cough. The diaphragm is flattened and the lower ribs are flared outwards due to lack of countertraction from the abdominal muscles. Nearly 60 per cent of the patients reported have died early in infancy primarily from renal insufficiency or respiratory failure.[4] This condition is almost entirely limited to male subjects, only seven cases having been reported in girls.[5]

The purpose of this paper is to describe the first such case to be reported from Hacettepe Medical Center.

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Case Report

H. K. (68/64286), a three-month-old boy, was admitted to this hospital with bulging abdomen and a history of constipation since birth.

The history of pregnancy and delivery was uneventful. The parents and four siblings were well and healthy, and no consanguinity between the parents was found.

The patient's weight was 5,500 gm, height 58 cm, and the head, chest and abdominal circumferences were 41.5 cm, 39.5 cm and 42 cm respectively. The thorax was deformed, the lower ribs flared outwards and fine rales were heard over both lung fields. The liver extended two centimeters below the right costal margin, but the spleen was not palpable. The testes were undescended. The abdomen was bulging and without any consistency, and the peristaltic movements of the intestine could easily be seen on both sides, especially the right (Figures 1 and 2). The

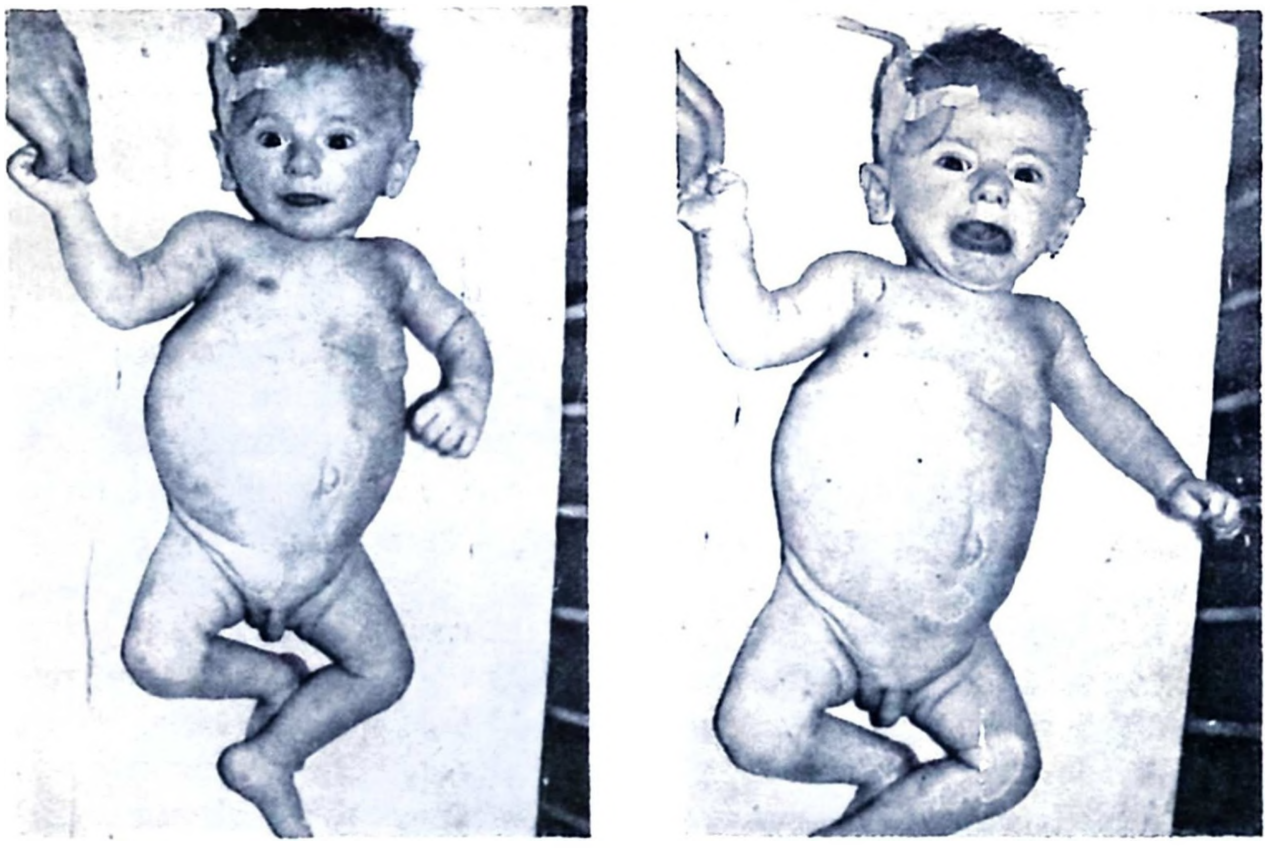

Figure 1

muscles were not palpable at all on the right side, and those on the left were poorly developed. When the child cried the umbilicus was not displaced upwards and the abdominal wall was relaxed.

Laboratory examination revealed a normal blood picture. Hb was 12 gr/100 ml, WBC 10,200/ccm, total serum protein 5.6 gm/100 ml (albumin 3.2 gr/100ml, globulin 2.4 gr/100 ml) and NPN was 40 mgr/100 ml.

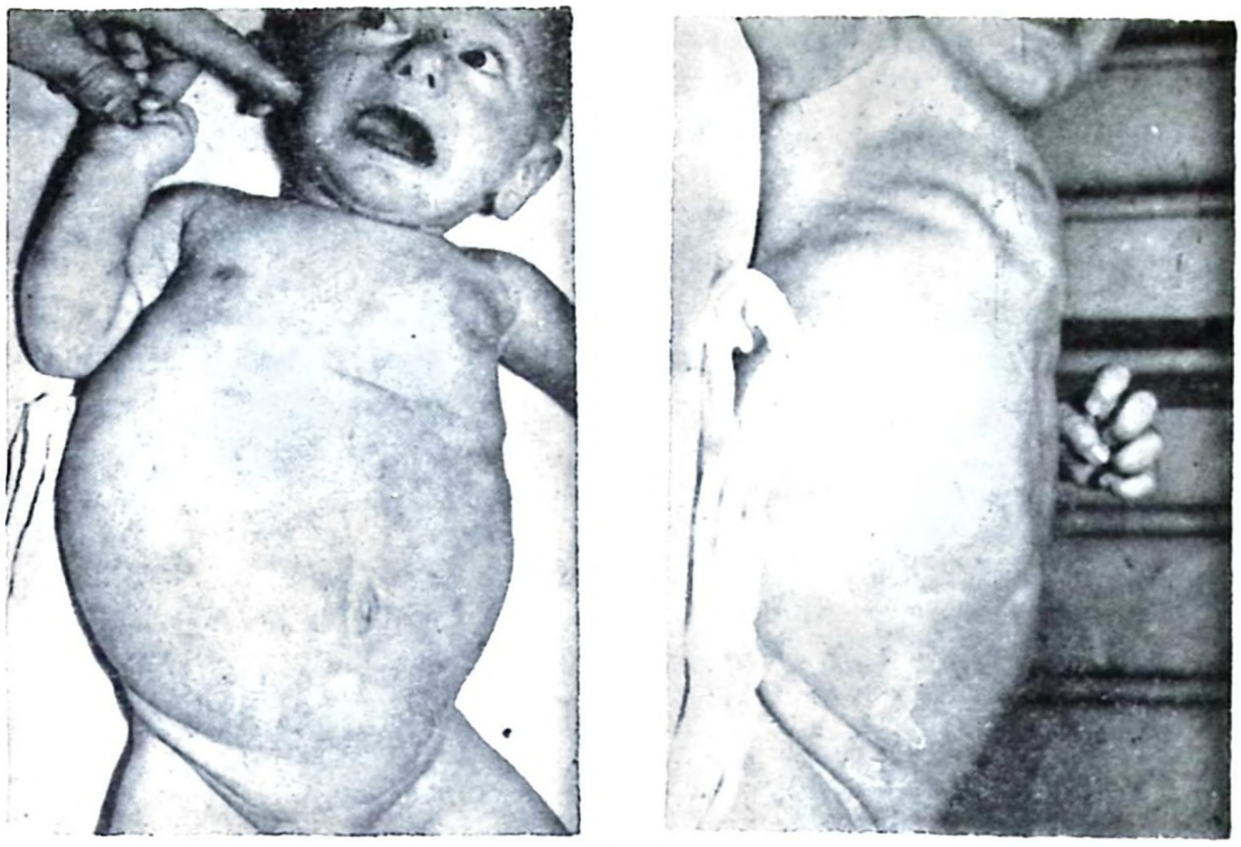

Figure 2

X-ray studies showed IVP to be normal, a barium enema showed no abnormalities, and chest roentgenogram revealed mild bronchopneumonic infiltration.

The patient died from paralytic ileus and dehydration due to severe diarrhea acquired on the 26th day after hospitalization; Shigella flexneri was cultured from the stools.

Only a partial post-mortem examination could be performed. The right superior rectus abdominus muscle was found to be absent; the obliquus externus and rectus inferior muscles on the same side, and all the muscles of the abdominal wall on the left, consisted only of a few muscle bundles. The liver was displaced on the left, probably due to lack of attachment and pressure exerted by the paralyzed intestines. The kidneys, urethers and bladder were normal in size.

Comment

The pathogenesis of the common malformations in this syndrome has been discussed at great length in the literature. The more recent explanations have favored a general disturbance in embryogenesis between the sixth and tenth weeks of gestation, which would result in multiple system abnormalities.

Osler stated[6] that owing to the lack of resistance in the abdominal wall normally met by the urinary bladder, it expands rather than contracts. Normally micturition is effected by the combined forces of contraction of the detrusor muscles of the urinary bladder and contraction of the abdominal muscles and diaphragm. A similar mechanism is believed to exist in intrauterine life. Thus the lack of the accessory forces from the abdominal muscles permits accumulation of urine in the bladder, which becomes dilated and hypertrophied, causing secondary hydronephrosis and hydro-ureter to occur. However, this was not observed in our patient.

Summary

A case of congenital absence of abdominal muscles is presented. The patient had bulging abdomen, the lower ribs were flared outwards, and the testes undescended. There were no urinary tract abnormalities. The literature concerning this malformation is briefly reviewed.

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