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Brief Clinical Report: Prune Belly Syndrome in an Anencephalic Male

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Abstract

We describe a postmature anencephalic infant with atrophy of the abdominal musculature (prune belly syndrome). Other associations of these conditions are noted.

Keywords

anencephaly; prune belly

INTRODUCTION

Both prune belly syndrome (PBS) and anencephaly (AN) are well-recognized entities [Smith, 1982; Woodard, 1978; Warkany, 1971; Lemire, Beckwith, and Warkany, 1978] but have not been reported together. Each may occur alone or in association with other major defects. Thus, neural tube defects (NTD), oral clefts, diaphragmatic hernia, and omphalocele may occur together ["schisis-association," Czeizel, 1981]. Anterior-wall defects (omphalocele, gastroschisis, and PBS) may be associated with conditions such as urinary tract obstruction not a direct consequence of the primary diagnosis [Baird and MacDonald, 1981]. We have not found PBS and AN among these reported associations. We describe here a male infant with both conditions. The possibilities of chance association of AN and PBS as independent events (1/750 × 1/35,000 = 1/26,250,000) and of casual or syndromic relationship are discussed. No conclusion is possible.

CASE REPORT

An anencephalic male infant (case 1) with evidence of the prune belly syndrome (PBS) was born after a 42-week gestation to a 17-year-old unmarried PO G1 AbO white female. The father was not a relative. The only medications taken were vitamins and Actifed ®, although at the end of the pregnancy she used small amounts of antibiotics for a sinus infection. The mother smoked a few cigarettes at the beginning of the pregnancy. Ultrasonography during the pregnancy did not reveal a fetal head. The mother's family history was unremarkable for neural tube or other defects. Very little was known about the father's family except for a questionable history of seizures as a baby and "slow" uncles and aunts.

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Examination revealed a cystic mass (6 cm × 3.75 cm) in the frontal area, absent right upper eyelid, and semicircular skin defect in the center of the forehead. There was a deficiency in the supraorbital bone and the ears were slightly malformed and possibly low set (Fig. 1). The maxilla was prominent. The abdominal muscles were atrophic and the abdomen was grossly distended. Bilateral cryptorchidism was noted. The nails were long and there was bilateral clubfoot.

Postmortem examination documented anencephaly with absence of the anterior pituitary and hypoplasia of the adrenal cortex secondary to this. The neurohypophysis was absent. Muscles of the anterior abdominal wall were absent and there was dysplasia of the kidneys, which weighed 5.5 gm together. Both appeared multicystic grossly and islands of cartilage, fibrous tissue, and cysts were noted in sections (Fig. 2). There was bilateral cryptorchidism and hypertrophy of the wall of the urinary bladder. Several small cystic spaces were noted beneath its mucosa. The urethra was patent but the external opening in the foreskin measured less than 1 mm in diameter and was difficult to enter or exit with a probe. Postmaturity was also evident. There was a patent ductus arteriosus. The lungs were grossly atelectatic and sections showed septal congestion and multiple focal areas of atelectasis. There was evidence of extramedullary hematopoiesis in the liver sinusoids.

The brain weighed 400 gm. The cerebral hemispheres were grossly abnormal although two hemispheres and a falx were recognized. The corpus callosum was absent. Frontal, parietal, and occipital areas could be recognized. Malformed, rudimentary temporal lobes were present. A thin, rather tenuously attached brain stem was seen at the base of the cerebral hemispheres. Cerebellar hemispheres were not easily recognizable and Purkinje cells were not seen. A moderate degree of astrocytosis was noted in the brain stem section. The corticospinal tracts were minimally developed. At the base of the brain, the basilar artery and right posterior communicating artery were recognizable. Both carotid arteries and middle cerebrals were also present. The remainder of the tissue at the base of the brain consisted of brownish granular membranous debris. Histologically, no anterior pituitary tissue was present.

DISCUSSION

Several aspects of this case warrant note. Hydramnios is observed in 30–50% of cases of anencephaly and there is a tendency to prematurity. However, the urinary obstruction noted with PBS generally leads to oligohydramnios. Neither was noted in this case.

Anencephaly has also been associated with postmaturity [Warkany, 1971] and this was indeed the case here as evidenced by dates and by the overgrowth of the fingernails. This child is male; a preponderance of females among anencephalic infants and a limitation of PBS almost exclusively to males has been found [Warkany, 1971]. Cryptorchidism, which was noted in this case, occurs in 95% of prune belly cases [Pagon, Smith, and Shepard, 1979] and is common in AN. Clubfoot has been described in both anencephaly and PBS [Warkany, 1971].

Prune belly itself is thought to be caused by bladder distention secondary to urethral obstruction [Pagon, Smith, and Shephard, 1979]. While the absence of polydydramnios in this case may be due to PBS, the loss of posterior pituitary tissue and resultant excess urine formation [Benirschke and McKay, 1953], even without complete urinary tract obstruction as postulated here, may have been responsible for the PBS appearance of this infant.

It is said that “pituitaries” are always present in anencephalics [Angevine, 1938, quoted in Benirschke, 1953; Lemire, 1978], whereas the neurohypophysis is usually reduced or absent [Warkany, 1971]. One of Benirschke’s cases manifested polyhydramnios, a hypertrophied bladder and dilatation of the ureters and renal pelves that could not be attributed to mechanical obstruction of the urethra. These authors also noted hypertrophy of the bladder and dilatation of the ureters and pelves in a number of anencephalics in whom polyhydramnios was noted. The authors suggested that the urologic changes might have been caused by excess amniotic fluid with increased hydrostatic pressure as a result of the polyuria caused by the lack of fetal antidiuretic principle. This in turn interferes with the normal emptying of the fetus’s bladder. Or the lack of the oxytocic action of the posterior pituitary secretion might itself affect normal tone of the smooth musculature of this system. Our case had no definite polyhydramnios but we cannot rule out this mechanism. In the same paper, note is made of the case of Landing [1950] in which oligohydramnios and amnion nodosum of the placenta were associated with anencephaly and hypoplastic polycystic kidneys. Benirschke [1953] suggested that the endocrine abnormality ordinarily would have resulted in intrauterine diabetes insipidus but that obstruction to urine flow prevented both polydydramnios and excess amniotic fluid. Apparently our case falls in between.

The associations of prune belly syndrome and a variety of other conditions were summarized by Warkany [1971]. Among the less obvious associations (ie, those not involving the gastrointestinal or urinary systems) are cleft palate, Ullrich-Turner syndrome (also reported recently by Lubinsky, Doyle, and Trunca [1980]), multiple pterygia, Wiedemann-Beckwith syndrome [Knight et al, 1980], choanal atresia [Brierre, 1963], and gastroschisis [Willert et al, 1978]. One case of macrogryrus has been described [Ralis and Forbes, 1971]. There is only one published instance of neural tube defect-spina bifida, meningomyelocele, and hydrocephalus [Mathieu et al, 1953].

In addition to the anencephalic child reported here, we are aware of a stillborn male infant (case 2) with prune belly, arhinencephaly but otherwise normal brain, and multiple congenital anomalies consisting of left cleft lip, bilateral cleft palate, postaxial polydactyly, incomplete closure of the foramen ovale, patent ductus arteriosus, high interventricular septal defect, anomalous origin of the right subclavian artery, and dilated hypertrophic bladder. There is no mention of lower urinary tract obstruction, nor do we know whether this was sought. Unfortunately, no chromosome studies were done on that infant.

The question immediately arises whether the two conditions, PBS and AN, represent a chance association or are they part of a syndrome. When this paper was first submitted, a reviewer suggested both case 1 and 2 may represent Meckel syndrome as redefined by Fraser and Lytwyn [1981]. These authors posit kidney dysplasia as the sine qua non, and at

least two other defects of those commonly attributed to the syndrome as required for diagnosis. Case 2 had polydactyly and cleft lip and palate, but we carefully reviewed the kidney sections and found no dysplasia despite the presence of hydronephrosis. Case 1 had dysplastic kidneys but no findings other than the anencephaly. Thus neither of our case meets Fraser and Lytwyn's criteria for Meckel syndrome. Additionally, it must be noted that Wigger and Blanc [1977] found reports of renal dysplasia in 25 of 265 cases of PBS and in 11/14 of their own patients. Genitourinary abnormalities are common in AN. Renal weight is usually reduced, but there is little consistency in the pattern of malformations noted by Lemire, Beckwith, and Warkany [1978]. These authors list 19 instances of genitourinary abnormalities, many multiple, among the 68 anencephalics they describe, but only 1 of these had cystic dysplasia of one of the kidneys.

However, this does not lay the matter to rest. Seller [1981] unaware of Fraser and Lytwyn's work, also grappled with the problem of the variable, pleiotropic expression of the Meckel gene. She concluded that in the absence of other sibs, no definite diagnosis can be made when a patient manifests only one of the major abnormalities of Meckel syndrome. Omphalocele, the finding common to Meckel syndrome, the schisis-association [Czeizel, 1981] and "anterior-wall defects" [Baird and MacDonald, 1981], was not present in either of our cases. However, the possibility remains that these two cases, or one of them, are examples of the Meckel syndrome, forme fruste.

We are left with two male infants with prune belly syndrome, one with an associated neural tube defect and one with faulty development of the brain plus other anomalies. In neither is a syndromic diagnosis possible, nor is it entirely clear what led to the prune belly. We think that absence of antidiuretic hormone [Benirschke and McKay, 1953] is most likely in the first case but are unable to document pituitary damage in case 2. We would urge careful dissection and thorough study of the posterior pituitary in cases of neural tube defects with associated malformations and of the urinary system in PBS, especially when other anomalies are present.

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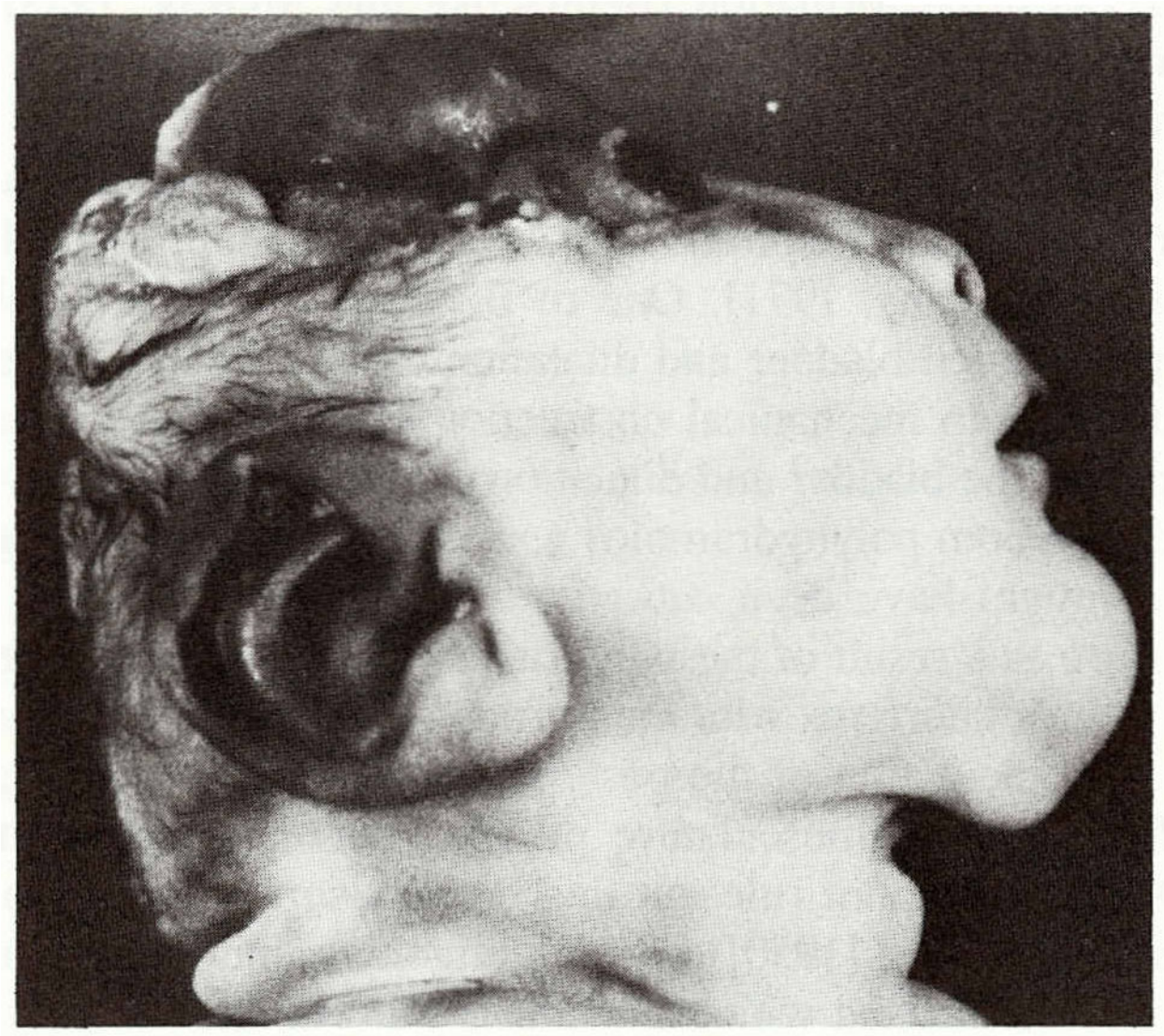

Fig. 1.
Profile of head of case 1.

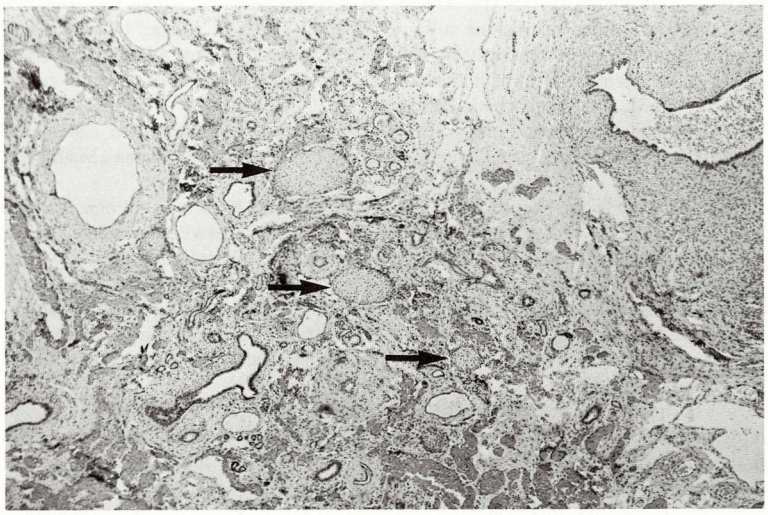

Fig. 2.

Photomicrograph of left kidney, case 1, showing immature stroma and islets of cartilage (arrows) characteristic of renal dysplasia.