Potter’s syndrome: A story of the rare, rarer and the rarest

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ABSTRACT

Potter’s syndrome is a rare condition affecting one in 2000-5000. We present here two autopsy cases of Potter’s syndrome, with the rare finding of discoid adrenals and the even rarer finding of in situ neuroblastoma in one of the cases.

KEY WORDS: Discoid adrenals, in situ neuroblastoma, Potter’s syndrome

INTRODUCTION

Potter’s syndrome was first described by Edith Potter in 1946 at the Chicago Lying IN Hospital in U.S.A. Its incidence varies between one in 2000 and one in 5000.[1] Males are affected more commonly. The recurrence risk rate is as high as 3% and 6%. Here we present two cases of Potter’s syndrome with discoid adrenals, diagnosed on autopsy. In addition, one of the cases also had in-situ neuroblastoma.

CASE REPORT

We present here autopsy findings of two male neonates, born to primigravida mothers (aged 24 years and 26 years) at 32 and 35 weeks respectively. One case was booked and was detected to have oligohydramnios on prenatal ultrasound scans with no other significant antenatal history. There was no history of teratogenic drug intake. The other case was an unbooked one.

At birth, the babies weighed 2.9 kg and 2.75 kg. The neonates suffered from severe respiratory distress and were put on ventilation. Both neonates died due to respiratory failure, at 10 h and 15 h after birth, respectively. Autopsy was performed after taking consent.

The external examination of the babies revealed dysmorphic facies with low set ears, flat nasal bridge with broad flat nose, widely separated eyes, compressed chin, and prominent inner canthus of each eye (Potter’s facies). They also had broad short fingers, clubbed feet with equinovarus in the first and equinovalgus[2] deformity in the second baby [Figure 1a]. One of the neonates had micropenis and left sided undescended testes along with imperforate anus.

Lungs of both the neonates were severely hypoplastic and weighed 18 g and 13.9 g respectively. The lung/body weight ratios were 0.006 and 0.005 respectively (normal lung/body weight ratio at term is 0.222 ± 0.002). Microscopic examination of lung tissues from both cases showed collapsed alveoli, but some of them were lined by tall columnar epithelial cells. The alveolar septa were thickened and congested with reduced radial bronchiole-alveolar count in both cases, confirming the gross diagnosis of hypoplastic lungs [Figure 1b and c]. No congenital anomaly was detected in the heart.

On further dissection, both the kidneys were found to be absent. The bladder washypoplastic. The adrenals were enlarged and discoid in shape, and the pair of adrenals weighed 6.6 g and 7 g respectively [Figure 1d]. The sections from one adrenal of the second baby revealed the presence of large nests of small round neuroblastic cells in the medulla. The cell clusters were surrounded by thin fibrovascular septae. There was no evidence of the ganglionic differentiation. Evidence of necrosis, calcification or hemorrhage was lacking. The cells stained positive for synaptophysin. The adrenal cortical tissue was unremarkable [Figure 2a-d]. Based on these features, a diagnosis of in situ neuroblastoma was made.

DISCUSSION

Classic Potter’s syndrome occurs in a setting of bilateral renal agenesis. Potter’s syndrome or sequence has been divided into 4 distinct subgroups [Table 2].[3] The average incidence is one in 4000. It usually
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The development of kidney is crucial for the formation of amniotic fluid. In Potter’s syndrome there is a failure of the metanephros to develop into kidney, leading to bilateral renal agenesis, resulting in oligohydramnios. Oligohydramnios causes restricted movement and growth, leading to numerous physical deformities. Bain, Smith and Gault concluded that the Potter facies can be found associated with any urogenital abnormality which results in failure of fetal micturition in utero. Studies by DeMyer and Baird support the widely accepted view that in the human infants with Potter’s syndrome, the structural abnormalities of the face, lungs and limbs are secondary to oligohydramnios. Oligohydramnios was recorded in our booked case.

Classic Potter’s syndrome is always fatal. It is, therefore, important to pick it up during prenatal ultrasound at an early stage and hence that a pregnancy can be terminated. The absent kidneys may be difficult to pick up, especially in view of the compensatory adrenal hyperplasia. In cases of renal agenesis, the adrenal gland takes an ovoid, elongated or discoid shape and maybe seen as a reniform structure on parasagittal and transverse scans in the renal fossae. The term “lying down adrenal sign” has been ascribed to the elongated appearance of the adrenal, not normally moulded by the adjacent kidney. This important radiological finding has been reported in prenatal scans of Potter’s syndrome. The presence of discoid adrenals, as seen in both of our cases, has rarely been mentioned in autopsy series of Potter’s syndrome. However, it provides an important radiological clue for obstetric management.

The second baby had neuroblastic nodules in the adrenal. Persistence of primitive chromaffin cells migrating from primitive neuroectoderm to adrenal during fetal life gives rise to in situ neuroblastoma. The neuroblastic cells proliferate and reach a peak between 15 and 18 weeks of gestation and then begin to regress. They were first named ‘in-situ neuroblastoma’ by Beckwith and Perrin.

Table 1: Clinicopathological findings of the two autopsy cases

| Features                                     | Case I                      | Case II                    |
|----------------------------------------------|-----------------------------|----------------------------|
| Gestational age/sex of baby                  | 32 weeks/male               | 35 weeks/male              |
| Birth weight                                 | 2990 g                      | 2750 g                     |
| Lung/body ratio                              | 0.006                       | 0.005                      |
| USG findings prenatally                      | Oligohydramnios             | Unbooked                   |
| Duration of extrauterine life                | 4 h                         | 10 h                       |
| Autopsy finding                              | Both kidneys absent, adrenal hyperplastic and discoid, undescended testis, micro penis, imperforate anus, equinovarus | Both kidneys, ureter, urinary bladder aplastic, adrenal hyperplastic and disc shaped, equinovalgus |
| Microscopy                                   | No renal tissue identified Bilateral hypoplastic lung with reduced bronchial-alveolar ratio | No renal tissue identified Bilateral hypoplastic lung with reduced bronchial-alveolar ratio |
| Presence of foci of in-situ neuroblastoma in the adrenals |                             | Presence of foci of in-situ neuroblastoma in the adrenals |

Table 2: Type of Potter’s syndrome

| Classic Syndrome                          | Bilateral renal agenesis          |
|-------------------------------------------|----------------------------------|
| Potter’s syndrome type 1                  | Autosomal recessive polycystic kidney |
| Potter’s syndrome type 2                  | Renal dysplasia                  |
| Potter’s syndrome type 3                  | Autosomal dominant polycystic kidney |
| Potters’ syndrome type 4                  | Obstruction either in kidney or ureter leading to kidney disease |
in 1963. In situ neuroblastoma has been reported in 0.4-2.5% of autopsy cases. The majority of these foci undergo spontaneous involution in the postnatal life and rarely progress to neuroblastoma. These neuroblastic rests represent a persistence of developmental variation rather than a true neoplasm. Urinary level of catecholamines is a useful noninvasive investigation for followup of these patients. Absence of chromosome 1 deletions and n-myc amplifications can also help to distinguish these cases from neuroblastoma. To the best of our knowledge, the association of in situ neuroblastoma with Potter’s syndrome has not been reported before.

Potter’s syndrome itself is a rare condition and we take the opportunity of reporting two cases of Potter’s syndrome associated with the rarer finding of discoid adrenals and presence of the rarest microscopic finding of in situ neuroblastoma.

GUARANTOR

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