Case Report

Pediatric Renal Autopsies — A Report of 3 Cases

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Abstract

Although the contribution of autopsies in modern day pathology has been substantial, yet its role is hardly appreciated. We present 3 interesting cases of pediatric autopsies highlighting distinct renal disorders of diverse etiopathogenesis such as autosomal dominant polycystic kidney disorder, bilateral renal dysplasia and partially differentiated cystic nephroblastoma.

Keywords

autosomal dominant polycystic kidney disorder, bilateral renal dysplasia, partially differentiated cystic nephroblastoma

Introduction

"Only the living have problems with death, death is a mystery only to the living and the living looks to the dead for help", according to an old Chinese proverb.[1] Despite the importance of autopsies as a diagnostic tool, support for autopsies have remained inappropriately and dismally low, particularly due to greater availability of advanced imaging techniques. However, the ultimate fate of autopsy will depend on the organized effort of pathologists as autopsy remains the singular opportunity for a comprehensive diagnostic and medico-legal examination.

Renal cystic disorders(RCD) are a heterogenous group of disorders in which cystic change may be unilateral or bilateral ; may affect part or whole of the kidney. The cysts may represent a primary disorder of renal perenchyma, or a secondary change in portions of the nephrons or collecting ducts that originally differentiated normally. Cystic kidneys may be isolated abnormality, or there may be associated abnormalities in the urinary tract or in other systems, resulting in a recognizable pattern of multiple abnormalities. The clinical significance may be trivial or grave, and the fact that some varieties are genetically determined makes their accurate identification important in terms of genetic counselling.

Case Reports

Case 1 : A still-born 28 weeks' twin male fetus weighing 2.3 kg each was delivered by lower segment caesarean section (LSCS) following prolonged and preterm labour in a 20 year primigravida . Ultrasonography (USG) revealed twin pregnancy with both the fetus showing bilateral renal cysts along with oligohydraminos . On external examination, both the fetus showed bilateral club foot while abdominal dissection of both the fetus showed bilateral enlarged kidneys each measuring 10×5×3 cm with the cut-section showing identical features of multiple cysts of variable sizes filled with clear fluid with intervening solid areas (**Fig. 1a**). Histopathological examination (HPE) showed features of autosomal dominant polycystic kidney disorder (AD-PKD) including the presence of the characteristic glomerulocystic spaces (**Fig. 1b**). All other organs including the liver showed no gross and microscopic abnormalities. There was no suggestive family history.

Case 2: A still-born 26 weeks' female fetus weighing 2kgs was delivered by LSCS following prolonged and preterm labour in a 26-year primigravida. USG revealed oligohydraminos with bilateral enlarged, cystic kidneys of the fetus. External examination of the fetus showed facial dysmorphism such as cleft palate, and low set ears.

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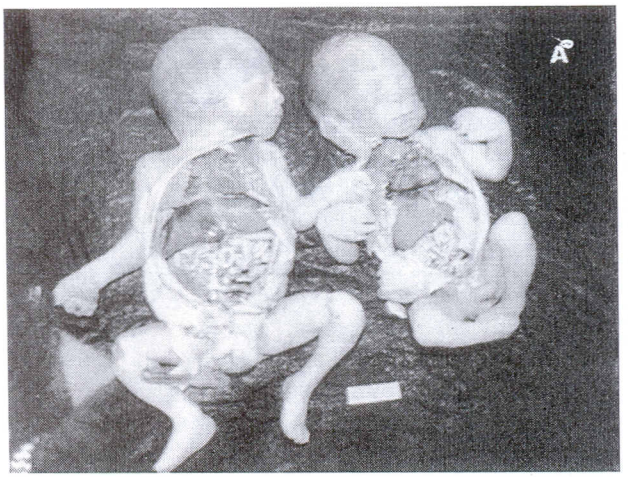

Fig 1(a)

Gross photograph of case 1

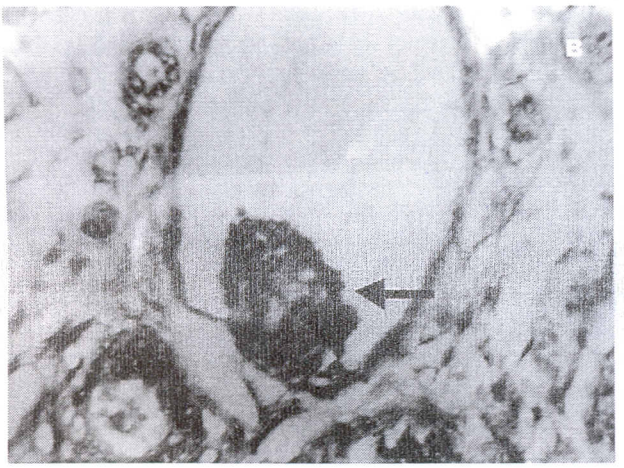

Fig 1(b)

Microphotograph of case 1

Abdominal dissection showed bilateral enlarged kidneys each measuring 8x4x2 cm. C/S of both the kidneys showed multiple cysts of variable sizes filled with clear fluid. HPE showed mesenchymal tissue including cartilage suggestive of bilateral renal dysplasia(**Fig. 2**). All other organs showed no gross and microscopic abnormalities. Family history was not suggestive in our case.

Case 3: A 10-month old child was referred to our hospital from a rural clinic with complaints of severe breathlessness, cough, fever along with generalized edema since last 7 days. He was the 3rd child of non-consanguienous marriage with normal delivery and was duly vaccinated. On general examination , palour was present. Chest examination showed

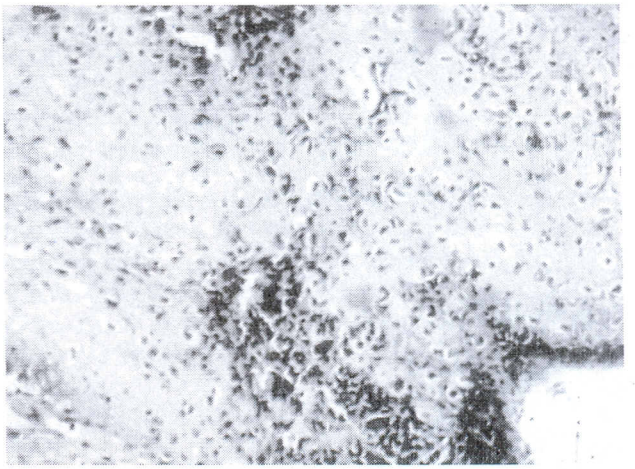

Fig. 2

Microphotograph of case 2

bilateral crepts and rhonchi on auscultation. P/A showed large, ballotable, intra abdominal mass measuring 10×8 cm involving the left hypochondruim, lumbar and umbilical regions. Routine blood investigations showed normocytic, normochromic anemia with neutrophilic leucocytosis. ECG showed sinus tachycardia and chest-X-ray showed massive, bilateral pleural effusion. Unfortunately, the child developed sudden deterioration in condition and expired. Consent for autopsy was obtained and performed. On gross examination, bilateral edema of the lungs with areas of consolidation were noted and confirmed by microscopy as bilateral pulmonary edema and bronchopneumonia. Abdominal dissection showed normal right kidney while the left kidney was enlarged and measured 15×8×3 cm. C/S showed multiple cysts of variable sizes filled with clear fluid. No solid, nodular or papillary areas were noted . Normal kidney was present at the periphery. HPE showed features of cystic partially differentiated nephroblastoma (CPDN) without anaplasia or heterologus differentiation

Discussion

Although the very suggestion of autopsy evokes indifference and sceptism, medical autopsy retains its importance for various reasons. The autopsy has a pervasive influence on medical care, medical science, society in general and the family of the deceased. The influence ranges from intangible understanding of the life-cycle and the relationship between life and death to the facilitation of organ retrieval and transplantation[2].

Cysts are the common renal abnormalities that, on direct inspection of the kidneys, are usually visible to the naked

eye. Several theories have been suggested regarding the pathogenesis of cystic disorders including : 1) all cysts , whether acquired or inherited, develop from pre-existing renal tubule segments; 2) after achieving a size of a few millimeters, most cysts lose their attachments of their parent-nephron segment; 3) the cysts –lining epithelium generally shows abnormal cellular differentiation and sustained prolofreation; 4) there is appropriate remodeling of the extracellular matrix to accommodate the enlarging cysts[3].

ADPKD affects around 1 in every 1000 individuals and is found in all continents and in all racial and ethnic groups. It is inherited as an autosomal dominant trait with complete penetrance[4]. The most common genotype is mutated PKD1 which affects around 85% of the cases, those involving PKD 2 is 15% . About one half of the young adults with ADPKD have cysts of the liver, and the prevalence increases with age to as high as 90% in patients whose life is prolonged by renal transplantation. These cysts, like renal cysts in ADPKD, are products of " second hit somatic mutation"[5]. About 10% of patients have cysts and abnormal duct proliferation in the pancreas[4]. Fewer than 5% have cysts in spleen. About 5% have arachnoid cysts. Cysts of the thyroid, ovary, endometrium and seminal vesicle have been also rarely reported[4].

Recent cytogenetic studies indicate at least 2 mutant genes for ADPKD exist, which have demonstrated that 1st mutation in PKD1 occurs at a locus (16p 13.3) on chromosome16, and a 2nd mutation in PKD2 occurs in a locus 4q13-q23 on chromosome 4. The protein product of the PKD1 gene is predicted to be along glycoprotein with trans-membrane domains and a cytoplasmic tail[6].

Bilateral total renal dysplasia is fatal in the newborn period in association with the Poter syndrome of oligohydraminos and pulmonary insufficiency[7]. The so called multi-cystic variety of total renal dysplasia is about 3 times more common than the non-cystic atrophic, or hypodysplastic variety and is the most common type of renal cystic disease diagnosed throughout childhood[8]. The incidence is 0.02 to 0.05 per 1,00, 000 hospital admissions[8]. Although few features of facial dysmorphism was present in our case but no suggestive family history was available and hence we could not reach a definite diagnosis of Meckel, Zellweger, Jeune and oral-facial –digital syndrome.

CPDN is usually unilateral, multiloculated and circumscribed lesion[9]. The recent discovery of papillonodular type of CPDN has led to modification of its diagnostic criteria , which are : 1) Septa and papillonodules, when present, are the only solid portions of the tumor and contain blastemal cells admixed with their normal and aberrant derivatives. 2) A discrete entirely cystic tumor containing luminal papillonodules in some cases. 3) The tumor without and with papillonodules is classified as a convential and papillonodular type of CPDN, respectively[9].

The clinico-pathological differential diagnosis includes other pediatric renal lesions, such as cystic mesoblastic nephroma, cystic renal dysplasia, cystic nephroma and Wilm's tumor with cystic change[10]. This can be differentiated from CPDN microscopically and hence pathologic examination of the resected specimen is mandatory.

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