43 THE ROLE OF ECHOCARDIOGRAPHY IN THE MANAGEMENT OF PEDIATRIC RENAL PATIENTS (PTS)

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Accurate assessment of cardiac function (CF), fluid status and pericardial effusion (PE) in children with renal failure is often difficult. The role of echocardiography in the management of these PTS was evaluated by analyzing 112 echograms (ECHO) obtained because of cardiopericardiac enlargement (CPE) on chest x-ray, cardiac murmur not attributable to anemia, changing fluid balance or possible bacterial endocarditis (BE) in 34 PTS. 12 PTS not requiring dialysis (NO DIAL) and 22 PTS on dialysis (DIAL) were studied. The CF and cardiac size were acceptable in all of the NO DIAL PTS and required no change in their therapy. However, 19/22 DIAL PTS had CPE. By ECHO 15/19 had left ventricular volume overload (LVVO) that was not clinically apparent, 5 had PE alone and 10 had LVVO and PE. Routine dialysis, antihypertensive drugs and diet restriction resulted in improvement of the clinical and ECHO findings in only 8. The remaining 11 PTS had persistent decreased CF on ECHO, with 6 requiring intensive ultrafiltration, 5 digoxin, and 1 of the 5 closure of an A-V fistula to improve CF and PE. One DIAL PT in whom an ECHO was done to rule out BE had hypertrophic cardiomyopathy without evidence of vegetation. An ECHO in another PT with rapid deterioration revealed a large mitral valve vegetation that had developed in 30 days, identified as Candida albicans at autopsy. Hence, echocardiography provided valuable information about fluid status and CF, and significantly aided the clinician in caring for the children with renal failure.

44 CONGENITAL HEPATIC FIBROSIS AND POLYCYSTIC KIDNEY DISEASE.

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From 1940 to 1975, congenital hepatic fibrosis was diagnosed in 18 patients who were less than 18 years old. Only patients with histologic confirmation of congenital hepatic fibrosis were included. Although the symptoms and clinical courses differed among the age groups, the histologic liver changes showed little variation. All four patients who presented before the age of 3 months had enlarged kidneys, and all four died in respiratory failure.

Fourteen patients were seen after the age of 3 months because of symptoms associated with portal hypertension. Of the 14 patients, 6 died after massive upper gastrointestinal hemorrhage and 1 died after a shunt operation. All 7 patients in this age group who died had renal insufficiency. Of the seven patients still alive at the time of this study, only two had mild renal insufficiency, but all seven had evidence of portal hypertension.

Polycystic renal disease was diagnosed in all patients. Fourteen had renal tissue diagnosis, and four had urographic evidence of the disease. Review of the histologic specimens of eight patients revealed that, in the four patients less than 3 months old, the findings were typical of infantile polycystic disease, and in the four older patients, cortical and medullary cysts were present--findings consistent with infantile polycystic kidney disease.

45 TUBEROUS SCLEROSIS PRESENTING AS POLYCYSTIC KIDNEYS IN INFANCY.

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Polycystic kidneys may rarely be a feature of the tuberous sclerosis complex in adults. A 3 month old female presented at the emergency room with a 2 day history of diarrhoae and vomiting. Physical examination revealed bilateral renal masses which by pyelography and ultrasonography were polycystic kidneys. The mother was noted to have adenomata sebacium on her forehead. At age 5 months the patient developed myoclonic seizures. Computerized tomography demonstrated the presence of paraventricular calcifications. Over the following 2 years other features of tuberous sclerosis have been observed.

A 3 month old male presented for routine immunization and was noted to have bilateral renal masses. At age 5 months he developed myeloclonic seizure activity and computerized tomography demonstrated paraventricular calcifications. A biopsy of renal cystic lesions was typical of that described for renal cysts of tuberous sclerosis. Since that time both children have had progressive mental deterioration and have been extremely hypertensive though renal function have remained satisfactory. The cases serve to illustrate that though not a previously recognized phenomenon tuberous sclerosis can present as renal polycystic disease in infancy and indicates that care should be taken to outrule the presence of this entity when genetic advice is sought in cases of polycystic renal disease in infancy.

46 PATHOLOGICAL STUDIES, INCLUDING MICRODISSECTION, OF RENAL CHANGES IN THE MECKEL SYNDROME

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This study is part of a continuing project on the classification of renal cystic disease using both clinical and pathological criteria. In this paper we present pathological studies, including microdissection, of cystic kidneys from 10 patients with a condition which is well defined by clinical and genetic criteria, the Meckel syndrome.

The renal cysts were bilateral in 9 patients and unilateral in one. In the bilateral cases, there were numerous cysts up to 1 cm in diameter present in both cortex and medulla, while in the unilateral case cysts were both few in number and small in size (most were less than 1 mm in diameter) and present only in the cortex. Most of the cystic kidneys were markedly enlarged (average 10 times normal weight) and, in all but one of the bilateral cases, the right and left kidneys were of comparable size.

Histologically there were reduced numbers of renal corpuscles with many cysts of varying sizes present within a moderately cellular interstitial tissue. Many of the cysts contained intraluminal polypoid projections, similar to the picture presented by the intracanalicular fibroadenoma of the breast.

Microdissection displayed rounded cysts which were characteristically localized to the terminal proximal convoluted tubules, and to the crests of the loop of Henle.

This combination of renal pathological features is, to the best of our knowledge, diagnostic of the Meckel syndrome.

47 ULTRASTRUCTURAL LESIONS OF THE GLOMERULAR BASEMENT MEMBRANE IN LAURENCE-MOON-BIEDL-BARDET SYNDROME (LMBBS).

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Although renal involvement is common in LMBBS, glomerular ultrastructural (EM) changes have not been delineated. We have studied glomeruli from 3 patients with LMBBS and have found similar EM changes in the glomerular basement membranes (GBM). Each had retinopathy, obesity and polydactyly; 2 were retarded. Two had decreased renal function and hypertension; one had normal function and blood pressure; none had proteinuria. Intravenous urography revealed reduced concentration and distorted calyces in two cases. The third was normal.

Light microscopy revealed segmental increases in mesangial cells and matrix of all glomeruli; this ranged from mild proliferation to sclerosis. Marked changes in the GBM were seen by EM. There was diffuse effacement of the trilaminar architecture and segmental and irregular thickening alternating with thinner regions, and rarefaction. There were also accumulations of granular and fibrillary material in the inner third of the GBM. Spherical dense particles were also seen. In some areas the GBM was split and mesangium was interposed. No deposits were seen by EM or immunofluorescence.

These GBM changes were found in each case and were independent of the degree of uremia or hypertension; these changes may therefore be the primary or earliest glomerular abnormality seen in LMBBS.

48 Congenital Renal Abnormalities in Treacher Collins Syndrome.

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Renal abnormalities are not a usual feature of Treacher Collins Syndrome - three cases of hydronephrosis have been reported.

A 39 year old man presented with symptoms of renal failure. High dose urogram did not visualize the kidneys. Retrograde pyelograms showed a dysmorphic (R) pelvis and the (L) pelvis consisting only of a single calyx. Ten weeks after receiving a cadaver transplant he died from the complications of C.M.V. infection. Post mortem examination revealed a (R) kidney of 6 cms and (L) kidney of 3 cms. The (L) kidney consisted of a single pyramid. Histopathology showed glomerular sclerosis, periglomerular fibrosis and interstitial infiltrate with lymphocytes and plasma cells.

His 14 year old son had normal renal function and blood pressure. I.V.P. showed both kidneys had dysmorphic pelvicalyceal systems; (R) kidney was 8.5 cms in length and (L) kidney 10.5 cms.

It would seem unlikely that these most unusual renal abnormalities were not related to the Treacher Collins Syndrome in this family, so that renal abnormalities should be suspected in this syndrome.