Renal Histopathology of Laurence-Moon-Biedl Syndrome: Tubulointerstitial Nephritis without Specific Glomerular Changes

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Dear Sir,

Laurence-Moon-Biedl syndrome (LMBS) is known to be accompanied not infrequently by renal impairment. As to the renal histology of LMBS, conflicting results have been reported. Faulkner et al. [1] described subendo-thelial scalloping of the glomerular basement membrane (GBM) with swelling of the endothelial cells. Price et al. [2] documented marked ultrastructural alterations of the GBM composed of effacement of the trilaminar architecture, segmental irregular thickening, and accumulation of granular or fibrillar material within the inner third of the GBM. They suggested that these ultrastructural changes might be the primary glomerular abnormality in LMBS. On the contrary, Tieder et al. [3] reported four cases of LMBS in which tubulo-interstitial lesions were noted without specific ultrastructural changes in the GBM. Here we present a patient with typical LMBS. In this case, tubulo-interstitial and vascular lesions were the most remarkable abnormalities.

Fig. 1. Light micrograph of the kidney demonstrating diffuse interstitial fibrosis, tubular atrophy, and severe sclerotic change of the arterioles. There were no proliferative changes in the glomeruli. HE, ×95.

Case Report

Patient Y.M., a 13-year-old girl, was admitted with proteinuria and moderate renal failure. LMBS was diagnosed on the basis of polydactyly (supernumerary fingers of both hands were removed at birth), obesity, mental retardation, and retinitis pigmentosa. There was no family history. The parents were not consanguineous.

Her height was 139.0 cm, the weight 45.5 kg and blood pressure 166/110 mmHg. Intravenous pyelography showed no morphological abnormalities. BUN was 12 mg/100 ml, serum creatinine 1.3 mg/ 100 ml, creatinine clearance 42.8 ml/min, and 24-hour urine protein excretion 0.5 g. Urinary sediment was normal, and urina culture was sterile. Serum total cholesterol was 190

mg/100 ml, triglyceride 109 mg/100 ml, total protein 6.8 g/100 ml, and fasting blood glucose 80 mg/100 ml with normal response to glucose tolerance test. There was no apparent hypogonadism. Renal biopsy showed widespread tubular atrophy and diffuse interstitial fibrosis with small foci of lymphocytes infiltration. One half of the glomeruli were globally sclerotic (nearly 50 glomeruli were obtained in total), while another half showed almost normal appearance. There was no cellular proliferation. In some glomeruli, segmental mesangial sclerosis with adhesions to Bowman's capsule or pericapsular fibrosis were observed. Vascular changes were remarkable; in every arteriole, medial fibrosis and intimal sclerosis with hyaline deposition were conspicuous. Some arterioles were completely occluded (fig. 1).

Immunofluorescence studies revealed no significant deposition of immunoglobulins or complement components. On electron microscopy, there were no important ultrastructural abnormalities in the GBM except for some foot process effacement. Electron-dense deposits were not demonstrated (fig. 2).

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Fig. 2. Electron micrograph of the glomerulus showing no ultra-structural abnormalities except for some foot process effacement. x 4,600.

Discussion

Renal histological findings in our patient were almost identical with those of Tieder's cases. The main histological changes were found in interstitium and vessels. Therefore, the suggestion of Price et al. [2], viewing the ultra-structural changes of the GBM as a primary abnormality, appears not to be applied to every case of LMBS.

It should be impossible to consider that each renal involvement of the LMBS is caused by the same mechanism. Although terminal features of chronic renal failure may be common to every patient, early renal lesion and the course to the end-stage renal failure seem to be diverse in each patient. In some patients, ultrastructural changes of the GBM may be the initial renal lesions. In others, tubulointerstitial disease and/or vascular lesion is assumed to be the main feature of the renal disease. In connection with the latter, regular urine cultures and blood pressure measurements are recommended to prevent progression to terminal renal failure [4]. However, the relation between deteriorating renal function and urinary tract infection or hypertension has not been clarified in detail. Further studies are required for understanding the renal involvement of LMBS.

References

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