

Alström Syndrome Scientific Workshop Morrisburg Ottawa 2001

Pathology

Four patients were autopsied, one male and 3 female, all dying between 32 and 42 years of age. All had moderate to severe renal glomerulosclerosis and interstitial fibrosis. In the most severely affected kidneys approximately 50% of glomeruli had undergone complete fibrosis as shown by positive trichrome staining. Affected glomeruli were scattered among more normal glomeruli. All kidneys also had mild to moderately severe interstitial fibrosis. Two patients had severe cirrhosis of liver with bridging fibrosis between portal areas and regenerative nodules. One patient had portal fibrosis with bridging but no regenerative nodules. The fourth patient had a normal liver. Two female patients had dilative cardiomyopathy. Myocardial fibers were widely separated from one another thick bands of fibrous connective tissue. The male patient who had a normal heart, glomerulosclerosis and cirrhosis had very severe interstitial fibrosis of lung. The entire lung was replaced by dense fibrous tissue. This seemed most dense around obliterated bronchi. However the lesions were not those of classic bronchiolitis obliterans-organizing pneumonia (BOOP). Inflammation was not present. This patient also had atrophic fibrotic seminiferous tubules, as previously described in Alström patients.

Overall, multiple terminal lesions of Alström patients share the common feature of fibrosis, but patients differ from one another in which organs are affected and how severely each is affected. These findings suggest that one possible function of the normal Alström gene is to suppress the formation of scar tissue in organs that are exposed to pathogens such as hepatotoxins, renotoxins or cardiotoxic viral infections.