Retinitis pigmentosa and Aortic Regurgitation in a Patient with Adult Polycystic Kidney Disease

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Dear Sir,

Although many hereditary renal diseases have been described in association with retinitis pigmentosa [1,2], the combination of adult polycystic kidney disease and retinitis pigmentosa has not yet been reported. We herein report the first case of retinitis pigmentosa in an adult patient with polycystic kidney disease who also had aortic regurgitation.

This patient, a 30-year-old male, was admitted to our hospital for aortic valve replacement after a recent episode of endocarditis. Aortic valve incompetence was diagnosed 20 years ago while polycystic kidney disease was discovered by ultrasonography at the age of 21. Family history revealed that an aunt of the patient also had polycystic kidney disease and underwent chronic hemodialysis treatment for end-stage renal failure. Visual weakness had started during childhood when a decrement in visual acuity and night blindness was confirmed. There was no history of eye trauma or inflammation.

Ophthalmological examination confirmed severe visual loss (visual acuity 1/20 in the left eye and 1/40 in the right eye) while fundoscopy showed retinitis pigmentosa (fig. 1). Abdominal CT scan showed bilateral increased kidney size with multiple cysts as well as liver cysts (fig. 2).

Cardiac catheterization revealed severe aortic regurgitation. During the cardiac operation no calcifications were found on the aortic valve. The mitral valve and the aorta were normal.

Retinitis pigmentosa, the ocular component of the hereditary renal retinal syndrome, has been described occasionally in patients

Fig. 1.

Patient's retina.

Fig. 2.

Abdominal CT scan of the patient.

Although adult polycystic kidney disease has been well described as to its autosomal dominant mode of transmission, we cannot draw conclusions concerning the mode of transmission of retinitis pigmentosa in our patient. It is also unknown if our case represents a distinct syndrome unreported up to now or if it is a matter of coincidence.

References

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