Facial Angiofibromas and Periungual Fibromas in Tuberous Sclerosis

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Picture 1.

Picture 2.

A 78-year-old woman with chronic kidney disease visited our emergency department due to pain in her right flank. Abdominal computed tomography revealed perirenal hematoma and angiomyolipomas, and transcatheter arterial embolization was performed. Angiography also showed bilateral renal aneurysms. A physical examination revealed several red papules (facial angiofibromas) (Picture 1) and skin-colored rubbery nodules (periungual fibromas) in the proximal nail fold of the left annular finger (Picture 2) and in the lateral nail groove of the right toe. A diagnosis of tuberous sclerosis complex (TSC) was made based on the characteristic clinical features described above (1, 2). Genetic testing was not performed. Further investigation revealed a retinal hamartoma but no pulmonary lymphangioleiomyomatosis. She had no history of epilepsy or intellectual disability.

With the development of novel treatment strategies, the importance of an accurate diagnosis of TSC is increasing.

The recognition of these characteristic skin features is important for making an early diagnosis.

The authors state that they have no Conflict of Interest (COI).

References

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