A 28-year-old patient was admitted because of painless, massive hematuria (Figure 1), accompanied by severe hypotension and hemodynamic instability. It was the fifth episode in the last 3 months. He has been previously treated for epilepsy, diagnosed in childhood and had moderate to severe cognitive impairment. The skin exam of the patient revealed several lesions: angiofibromas, over the nose, nasolabial folds, cheeks and chin (Figure 2), brown colored connective tissue nevi and periungueal fibromas. A magnetic resonance angiography (MRA) revealed multiple echogenic, nodular areas with a cystic component in both kidneys, structural disorganization of the right kidney, a supernumerary right renal artery and early bifurcation of the left renal artery. Due to the recurrent hematuria, the patient was submitted to an arteriography with embolization of nodules located in upper and lower poles.
of right kidney, and to embolization of microaneurysms in the same location, with preservation of the adjacent parenchyma (Figure 3). The intervention was performed without complications and the patient made an uneventful recovery, with no further evidence of bleeding. Kidney biopsies revealed angiomyolipomas.

Tuberous sclerosis complex (TSC), also referred as Bourneville disease, is an autosomal dominant neurocutaneous syndrome characterized by the development of multiple hamartomas distributed at various sites throughout the body, especially the skin, retina, kidney, brain, heart and lung. The prevalence is not well defined with reported incidence of from 1:6000 to more than 1:100 000. TSC arises from inactivating mutations of either TSC1 (chromosome locus 9q34.3) or TSC2 (16p13.3), which encode hamartin and tuberin, respectively. Although the classic triad of TSC is seizures, mental retardation and angiofibromas, this occurs approximately in only one-third of cases. Renal angiomyolipomas can lead to potential complications that include hemorrhage, the risk of which increases with the size and vascularity of the lesion, and mass effects. The current therapy to both control active bleeding as well as to prevent lesions with aneurysms from bleeding in the future is angiographic arterial embolization. The mammalian target of rapamycin complex 1 (mTORC1) is a downstream target of the hamartin/tuberin complex, and use of an mTORC1 inhibitor such as sirolimus is likely to serve as targeted therapy and ultimately affect the angiomyolipoma volume. A multicenter Phase 2 trial of 36 TSC patients using sirolimus for 52 weeks showed regression of kidney angiomyolipomas, brain tumors and liver angiomyolipomas. Future studies are needed to determine benefits and risks of longer duration treatment of patients with TSC.

Figure 3. Arteriographic aspect of nodules located in upper and lower poles of right kidney (a and b); arteriographic view after embolization of microaneurysms in the same location, with preservation of the adjacent parenchyma (c and d).
References


