Abstract

Introduction
The Bourneville tuberous sclerosis is a rare hereditary disease affecting several organs and tissues of the human body. It originates as neurological, cutaneous and visceral expressions in multiple hamartoma localisations.

Case report
We report the case of a Moroccan male of 51 years of age that presented bilateral lumbago, urinary tract manifestations without CT scan preparation that objectified the presence of bilateral renal lithiases. The abdominal CT scan diagnosed the Bourneville tuberous sclerosis considering the presence of angiomyolipoma. The patient was abstained from any therapeutic approach in the follow-up except if he presented complications. We discuss mechanisms responsible for the lithiase formation.

Conclusion
Our Bourneville tuberous sclerosis case with bilateral lithiases has never been reported in the literature. We incite the practitioners evoke the Bourneville tuberous sclerosis whenever lithiases of the high urinary organs are revealed.

Introduction
The Bourneville tuberous sclerosis (BTS) is characterised by the occurrence of benign tumours such as hamartoma. It is a rare hereditary disease characterised by dominant autosomic transmission with variable depth. The renal affection by BTS represents the second cause of death after neurological affection. The skin, eyes and the heart are also affected.

We report the case of a patient that presented bilateral lumbago and urinary tract affection without preparation that showed the presence of bilateral lithiases. To our knowledge, this BTS association with bilateral lithiases has never been reported in the literature. Hence, we discuss the mechanisms responsible for lithiase formation and incite practitioners to evoke the BTS whenever lithiases of the high urinary organs are revealed.

Case report
A Moroccan male patient was 51 years old without significant pathological history. The patient presented bilateral lumbago without any associated signs. The clinical examination found the patient to be of a good general condition with a light sensitivity of bilateral lumbar vertebra. The dermatological examination (Figure 1) found multiple wheals of pink to red colour with a smooth brilliant surface and measuring 1 to 10 mm. Almost the same lesions were found in the daughter of the patient (Figure 2). The biological assessment was without particular findings. Without preparation, the urinary tract CT scan objectified multiple bilateral renal lithiases. The abdominal ultrasound showed a totally dedifferentiated angiomyolipoma (AML) on both kidneys. The abdomino-pelvic CT scan objectified a big lithiasic kidney containing a lithiase of calcic density with the presence of multiple masses of fatty density on both kidneys.

Figure 1: Cutaneous angiofibroma in the reported case.

Figure 2: Cutaneous angiofibroma of the daughter.
transformation of a gene positioned either on the long arm of chromosome 9 or on the arm of the chromosome 16. The incidence of BTS in the general population at birth is estimated to be 1/6,000 while the general prevalence is about 1/10,000.\(^1\)

The BTS frequently affects the kidney by manifesting AML, renal cysts and renal cancer; the BTS also affects the brain tissue such as cortical tubers and ependymoma nodules; the skin is affected through hypomelanic, angiofibroma and Koënen tumours. Finally, the heart and eyes are also affected.

The BTS is a multisystemic disease. The renal affection represents 60% of all cases and reveals different aspects. Frequently, AML is found while renal cysts, renal carcinomas, segmented and focal glomerulosclerosis and the interstitial fibrosis are rare. The renal affection has to be systematically detected and regularly.

The AML is the most frequent tumour that is found in 75–80% of patients of more than 10 years old. It is a benign tumour with lesions which are often multiple, bilateral, increasing in size, with age being able to originate haematuria.\(^3\) Lumbago and abdominal pains, or tangible masses at palpation, are usually revealed. The AML might remain asymptomatic for a long time, being multiple and bilateral without particular gender differences.\(^4-6\)

These lesions are nevertheless characterized by their haemorrhagic risk in 5–25% of cases. Patients might present with haematuria and spontaneous break in the retroperitone. The importance of the clinical symptomatology is correlated with the tumour volume. Indeed, 90% of the symptomatic tumours have a volume over or equal to 4 cm. The cystic lesions are also frequent and might be unique cysts or renal polykystosis.\(^7\)

The risk of renal carcinoma in BTS patients is the same compared to the normal population. Ultrasound and CT scans are the main diagnostic tools of AML. The cutaneous

**Discussion**

The BTS is a rare phacomatous characterised by dominant autosomic transmission with a variable entrance. This disease is caused by a

---

**Figure 3:** (A), (B) Renal bilateral angiomyolipoma.

This also showed a renal AML aspect of both kidneys (Figure 3A and 3B). The patient was withdrawn from any therapeutic decision except if he had any complications.
manifestations are frequent and represented by achromic spots and angiomyolipoma. The neurological anomalies during the BTS constitute the first cause of morbidity and mortality and are connected to the presence of cerebral hamartoma, which might be expressed as cortical tubers, nodules under ependyma and astrocytoma of huge cells. The diagnosis of BTS in our patient was demonstrated by evidencing the AML in the CT scan and on the skin levels by direct clinical examination. Only bilateral AML associating bilateral lithiasis was demonstrated by the CT scan in our patient. No other neurological infraction was demonstrated. The BTS associated with bilateral lithiasis was never reported in the literature. The calculi are a compact load of one or several crystallised substances. The main mechanism involved in the calculus formation consists of an excessive concentration of components which are not really soluble in the urines. These excessive compound concentrations are precipitating such crystals which aggregate to form calculi. We suppose that the big volume of these masses is compressing the excretive tract and originates from a urinary stasis, hence causing formation of calculus. The authors indicate the renal biopsy in the case of AML suspicion in the CT scan to eliminate the presence of a malignant tumour. The therapeutic abstention is indicated by most authors; however the patients must be informed about the risks and the clinical haemorrhagic signs related to the AML such as acute pain and shock state. The embolisation should be suggested in cases of spontaneous haemorrhagic break of an AML. Surgery should be required for cases where the embolisation is not available in the emergency department. Indeed, our patient was closely followed-up.

Conclusion
The BTS is a hereditary familial disease. The renal affection including various complications might originate in increased morbidity of the disease. Cases of BTS-associating bilateral lithiasis were never reported in the literature. We incite the practitioners to evoke BTS while lithiasis of the high urinary organ findings are revealed.

Consent
Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editor-in-chief of this journal.

Abbreviations list
AML, angiomyolipoma; BTS, Bourneville tuberous sclerosis.

References