Images of the month

Musculoskeletal involvement in Tuberous Sclerosis

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Keywords: bone lesions, soft tissue swelling, Tuberous sclerosis complex.

Word counts: 237 words

Figures: 2

Tables: zero

Abstract

This patient is a 9-year-old girl who had a diagnosis of tuberous sclerosis complex (TSC), based on clinical features (facial angiofibromas, hypomelanotic macules and cortical dysplasia). She had a swollen left ring finger, which has become more evident as she grew older. The swelling has been proportionally increasing with her size. It has not limited her function.

TSC is a neurogenetic condition caused by mutations in the tumour suppressor genes TSC1 and TSC2. [1,2] Mutations in either TSC1 or TSC2 lead to overactivation of the mTORC 1 pathway and relatively uncontrolled cell growth. [3] This, in turn, causes growth of hamartomas in various organs. Bone lesions are often incidental findings on X-rays or CT scans of patients with TSC but these lesions are generally over-looked as it is assumed that they are clinically insignificant, asymptomatic and nonspecific. However, they can cause significant pain and physical disability, particularly when there is spinal involvement. Their natural history is unknown as not many patients with TSC are screened, or receive routine follow-up for bone lesions. [4] We do not routinely screen for bone lesions and it is not recommended by the international consensus guidelines for TSC management. It would not be practically possible to routinely screen all patients with TSC for these lesions because they can occur in any bone, as illustrated in this case. However, in our practice, we image or X-ray patients if they are symptomatic. mTOR inhibitors, such as rapamycin, have been shown to be effective in halting the growth of bone lesions and improving function. [5] of mTOR inhibitors may alleviate the patient's symptoms. However, currently mTOR inhibitors do not have licensing approval in the UK for this purpose (figures 1 and 2).

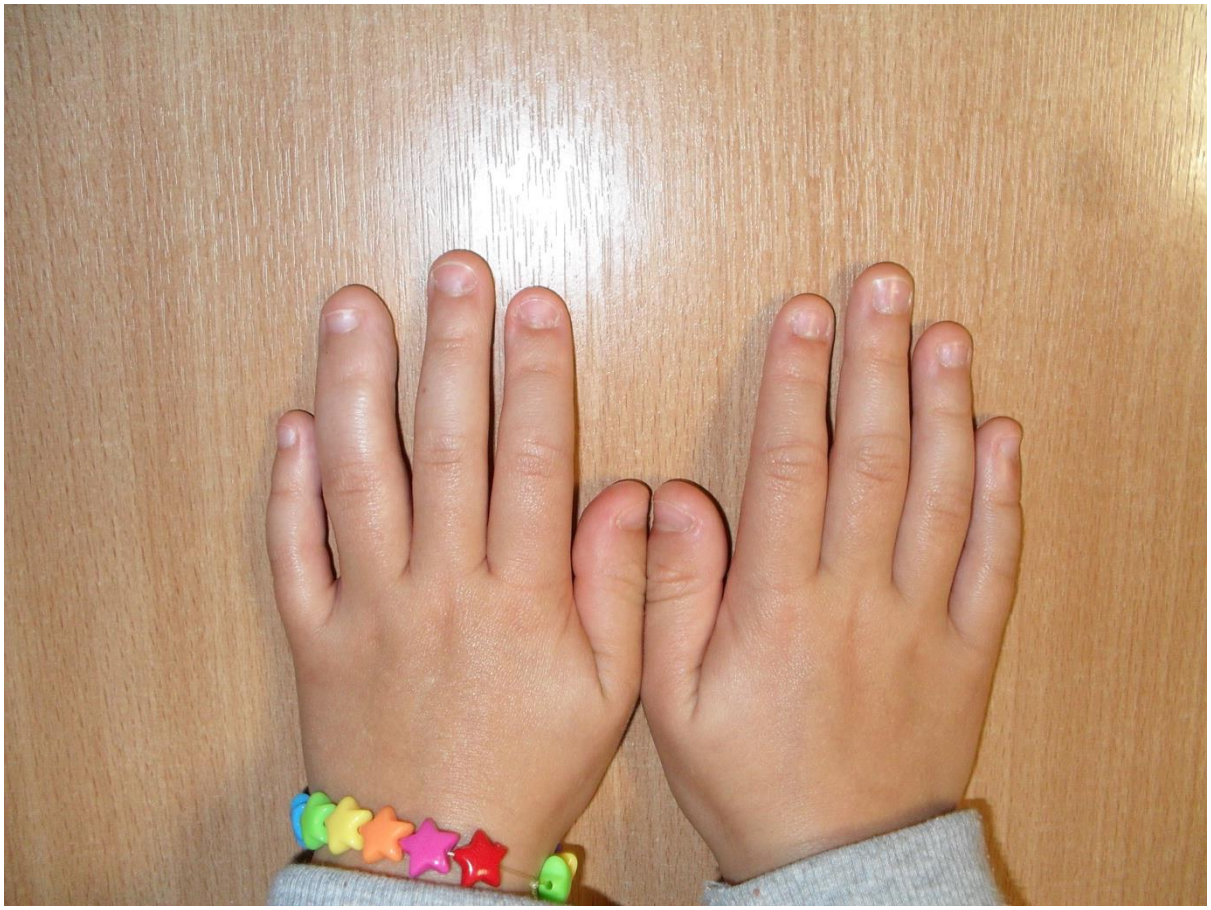

Figure 1: photo of both hands which shows swelling of the left ring finger.

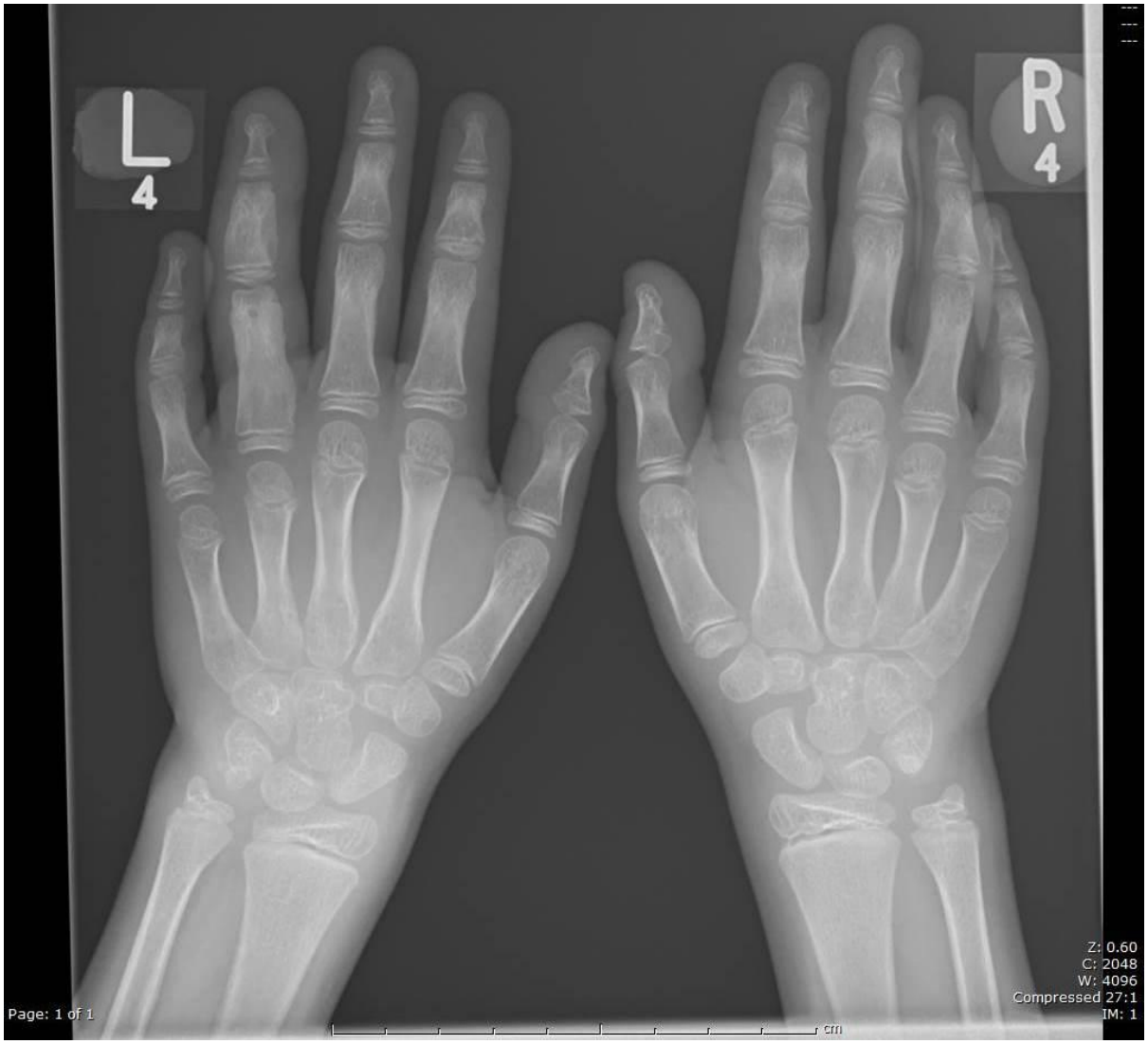

Figure 2: x-ray shows a combination of diffuse soft tissue thickening and bony thickening and sclerosis of the shafts of the middle and proximal phalanges of the left ring finger.

Competing interests: None.

Funding: not applicable

No additional data

References

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