A 10-year-old girl with mixed Malaysian–Burmese parentage under follow-up for tuberous sclerosis complex (TSC), epilepsy and mental retardation was noted to have a slow enlargement of her right thumb and index finger over a 5-year duration (Fig. 1). A white patch on the head (poliosis) was also present (Fig. 2). The diagnosis of TSC was confirmed by the presence of hypomelanoctic macules, cortical tubers, subependymal nodules, periungual fibroma and poliosis. Although our molecular genetic analyses of TSC1 and TSC2 in this patient failed to identify any disease-causing mutation, information on genetic mutation is not mandatory in establishing TSC diagnosis. Magnetic resonance imaging scan of the brain showed cortical tubers (Fig. 3—white arrows) as well as subependymal nodules (Fig. 3—black arrows). X-ray of the right hand showed irregular periosteal new bone formation (Fig. 4). Her epilepsy is currently controllable by medication but she remains mentally retarded.

TSC has been associated with numerous tumor growths in various organs such as brain, retina, lung, heart, kidney, ovaries and testis. However, the involvement of new bone formation and soft tissue overgrowth has not been commonly described. We postulate that the macrodactyly is a form of tumorigenesis. Tumorigenesis has been associated with the pathogenesis of TSC. Our finding of macrodactyly has not been described in the current diagnostic criteria for TSC (2013) and thus could be an addition to the existing clinical features of TSC.

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