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Images

Macrodactyly as a rare manifestation of tuberous sclerosis

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1. Case report

A 5-year-old girl with tuberous sclerosis presented with congenital macrodactyly of the first and second fingers of the left hand, which became more evident as she grew older. It was painless and without significant functional impairment.

A radiograph of her hands (Fig. 1) showed bone expansion, cortical thickening, undulating periosteal reaction, and irregular periosteal new bone formation involving the first and second fingers of the left hand. Additionally, the soft tissues of the affected fingers were enlarged, with increased density.

2. Discussion

Tuberous sclerosis complex (TSC) is a genetic disease that is responsible for the growth of hamartomas in various organs, including the brain, skin, kidneys, and heart.[1] Bone cysts are found in up to two-thirds of patients and often in hand phalanges.[2] Another common skeletal manifestation of TSC is the presence of patchy sclerotic lesions in the pelvis, vertebral bodies, and neural arches.[2]

Congenital macrodactyly is an extremely rare TSC manifestation that has been described in few case reports,[1,3–5] with an incompletely understood underlying mechanism.[1] Notably, the presence of localized tissue overgrowth and macrodactyly is seen in other neurocutaneous disorders, including neurofibromatosis type 1, von Hippel-Lindau, and Sturge–Weber syndrome, which increases the possibility of a similar macrodactyly mechanism.[3] Somatic mosaicism for PIK3CA has been described in several individuals with Klippel–Trenaunay syndrome, who may also present with macrodactyly.[1] The phenotype of the PIK3CA-related overgrowth spectrum ranges from isolated macrodactyly to syndromes that are defined by tissue overgrowth and vascular malformations.[6] The coexistence of somatic mosaicism for PIK3CA and TSC gene mutation may explain the macrodactyly in patients with TSC. Other authors have previously hypothesized that this phenotype is explained by the presence of somatic mosaicism for the loss of heterozygosity at one or more of the TSC loci.[1]

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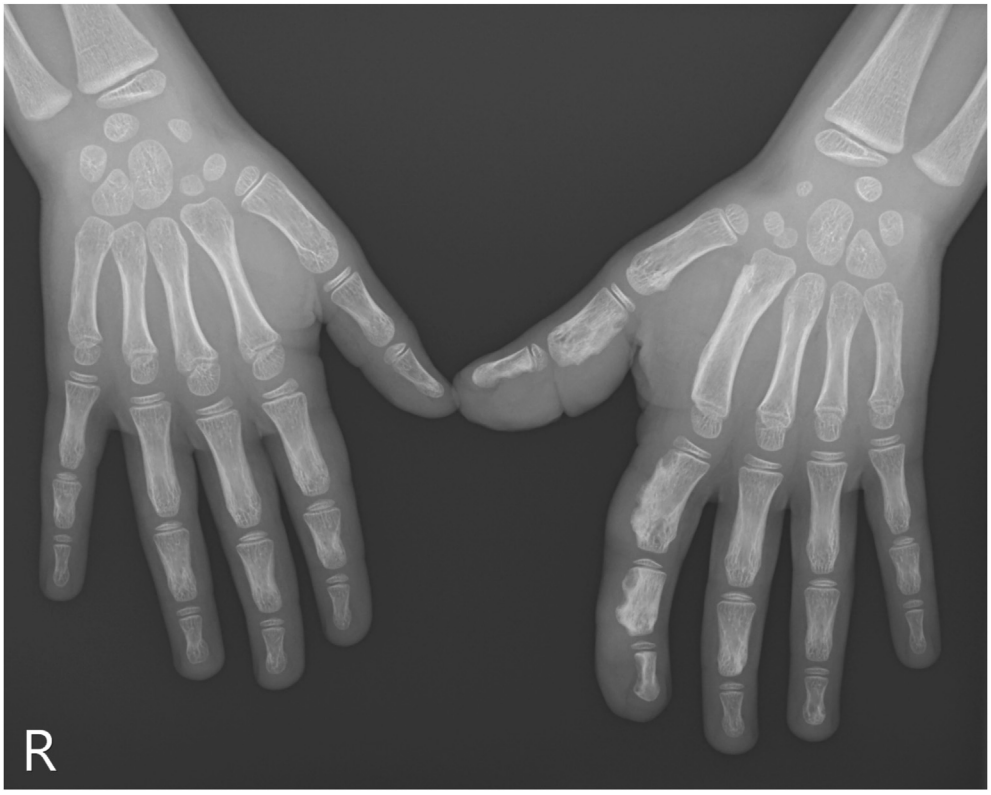

Figure 1 Radiograph of the hands showing macrodactyly involving the first and second fingers of the left hand.

3. Patient consent or parental/guardian consent

Obtained.

Declaration of competing interest

The author has stated explicitly that there are no conflicts of interest in connection with the article.

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