Coffin-Siris syndrome 7 (CSS7, OMIM #618027) is an extremely rare multisystemic autosomal dominant genetic disease caused by heterozygous mutation in the double PHD fingers 2 (DPF2), characterised by global developmental delay with mild to moderate intellectual disability, speech impairment, behavioural abnormalities, growth failure, coarse facial characteristics, and hypoplastic fifth toenails [1]. CSS7 is exceptionally rare, with only 10 cases reported worldwide [1–3].

At the time of presentation, the patient’s height was 99.0 cm (–3.55 SD) and weight was 13.5 kg (–3.10 SD). The patient exhibited self-injurious behaviour in the form of hair pulling but did not experience pain from this action. Characteristic features of the patient included microcephaly, small eye fissure with esotropia, microtia, low nasal bridge, broad nose, and high palatal arch. Additionally, brachydactyly was observed in the patient’s second and fifth fingers. Furthermore, there was local hyperpigmentation of the skin (Fig. 1).

Due to her parents demanding height intervention, after carefully weighing the benefits against the risks, recombinant human growth hormone (rhGH) therapy was initiated at the age of 5 years and 7 months with a daily dose of 2 IU. No adverse effects related to the use of rhGH were observed. During treatment, the growth velocity was recorded as 9.8 cm/year, 6.5 cm/year, and 4.7 cm/year in the first, second, and third year, respectively. After 3 years and 3 months of rhGH treatment, the height standard deviation score (HtSDS) increased by 1.53 SD and weight standard deviation score (WtSDS) increased by 1.74 SD.

More than three years’ treatment response of recombinant human growth hormone in a patient with Coffin-Siris syndrome 7

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as microcephaly, microtia, low nasal bridge, and broad nose [2–4]. However, the skeletal abnormalities documented in prior cases, including craniosynostosis and clinodactyly [2–4], were not observed in this patient. Distinctively, bilateral microphthalmia and local skin pigmentation were found in this patient. It is worth noting that the patient also carries a mutation in ASH1L gene, suggesting that the intellectual disability may be a result of the combined expression of the 2 genes.

Short stature is a common clinical feature in patients with CSS7, affecting approximately half of the patients diagnosed with this condition, yet the underlying mechanisms remains unclear. In previous reports, 80% of individuals with short stature were also found to have feeding difficulties [2–4]. Due to the desire to improve his height level, the parents sought growth hormone therapy. However, her growth velocity decreased annually. Despite this challenge, following the treatment, the height of the child increased from –3.55 SD to –2.02 SD, indicating that rhGH may indeed offer some degree of height improvement in patients with CSS7. However, BA progression accelerated during treatment — the difference between BA and chronological age was advanced from 0.7 years to 2.2 years, but because this individual carried a genetic variant that causes abnormal phalangeal development, the Greulich and Pyle method may not be applicable to her BA.

**Ethics statement**
Ethical approval is not required for this study.

**Author contributions**
Data collection: L.Y. and W.Q. Drafting manuscript: L.Y. and W.Q. G.C.X. critically reviewed the manuscript and provided significant input. All authors read and approved the final version of the manuscript.

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**Conflict of interest**
The authors have no conflicts of interest to declare.

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