

Marie-Sainton syndrome (cleidocranial dysplasia): early diagnosis is the key

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DESCRIPTION

Marie-Sainton syndrome or cleidocranial dysplasia is a rare, autosomal dominant skeletal disorder (OMIM number 119600) caused by a mutation in the *RUNX2* (CBFA1) gene encoding a runt-related transcription factor required in the differentiation of osteoblasts and chondrocytes.¹ The affected patients have a short stature, skull deformity, mid-face hypoplasia, clavicular hypoplasia, delayed tooth eruption and multiple impacted supernumerary teeth.² The skull abnormalities are characterised by patent fontanels or with delayed closure, open sutures and the presence of multiple Wormian bones.¹ Root resorption is delayed in the primary dentition; hence there is retention of deciduous teeth. Root resorption is delayed, with a lack of cellular cementum, and the permanent teeth are impacted. There is pseudo-mandibular prognathism, non-union of symphysis menti in children, reduced facial height, high arched palate and malocclusion owing to a hypoplastic maxilla.³ Rarely even supernumerary teeth are not seen with this condition due to a missense mutation p.R131C (c.391C>T) in the *RUNX2* gene.⁴ When the disorder is detected early, then the extraction of the retained deciduous teeth, along with surgical exposure and subsequent orthodontic movement of the permanent teeth, could be attempted, ensuring a better quality of life.³

A 20-year-old female patient reported with the complaint of missing teeth. History revealed that her deciduous teeth did not exfoliate timely and her mother also suffered from a similar problem. On physical examination, short stature,



Figure 1 Clinical pictures showing shoulders approximating towards the midline, (A) Brachydactyly with wide thumb (B) and intraorally, multiple over-retained deciduous teeth, missing permanent teeth and a high arched, narrow palate were seen (C,D).

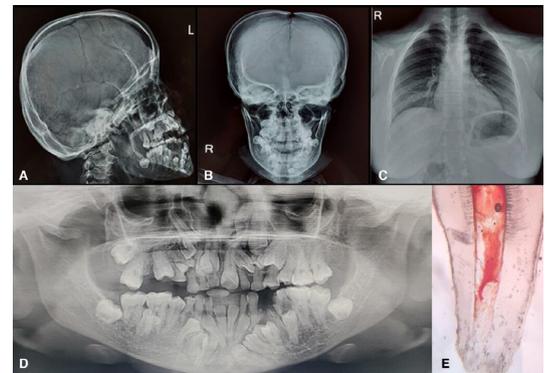


Figure 2 Skull radiographs showing widened cranial sutures with Wormian bones, frontal bossing and mid-face hypoplasia (A,B), hypoplasia of the clavicles bilaterally on chest radiograph (C) and multiple impacted teeth, and over-retained deciduous teeth on panoramic radiograph (D). Ground section of deciduous tooth revealed absence of cellular cementum in the root (E).

brachycephaly, frontal bossing, hypertelorism, depressed nasal bridge, limbs with hyperlaxity, shoulders appeared slanting and could be approximated towards the midline (figure 1A). Brachydactyly with wide thumbs (figure 1B), flat foot and genu valgo were also observed. Intra-orally, multiple over-retained deciduous teeth, missing permanent teeth and a high arched, narrow palate were seen (figure 1C,D). The patient was subjected to radiological investigations. Widened cranial sutures with Wormian bones, frontal bossing and mid-face hypoplasia were seen on skull radiographs (figure 2A,B). Chest radiograph revealed hypoplasia of the clavicles bilaterally (figure 2C). Panoramic radiograph revealed multiple impacted teeth, impacted supernumerary teeth and over-retained deciduous teeth (figure 2D). The deciduous teeth

Learning points

- ▶ Marie-Sainton syndrome or cleidocranial dysplasia is an autosomal-dominant skeletal dysplasia affecting intramembranous ossification.
- ▶ The condition can be diagnosed from the clinicoradiological features like open fontanelles, hypoplastic clavicles and multiple supernumerary teeth.
- ▶ The management is multidisciplinary and requires long-term follow-up.



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were extracted, and ground section of the tooth revealed absence of cellular cementum in the apical third region of the root (figure 2E). The patient underwent extraction of deciduous teeth and was advised for orthodontic treatment.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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