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Case Report

Dentistry Section

Marie-sainton Syndrome Through the Lens of CBCT: A Rare Case Report and Literature Review

UMAPATHY GOWDA[1], NIDHI S SHETTY[2], GADIGI SAMPREETHA[3]

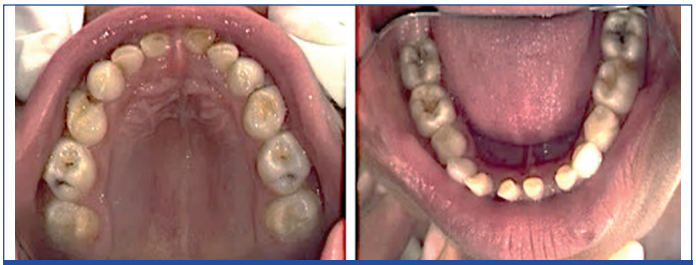
ABSTRACT

Marie-Sainton Syndrome (MSS), also known as Cleidocranial Dysplasia (CCD), is a rare autosomal dominant skeletal disorder primarily caused by mutations in the RUNX2 gene, which is essential for osteoblast differentiation and skeletal morphogenesis. The syndrome is characterised by generalised skeletal dysplasia, including hypoplastic or absent clavicles, delayed ossification of cranial sutures, and short stature. Distinct craniofacial features such as frontal and parietal bossing, midface hypoplasia, and brachycephaly are commonly observed. Dental anomalies are among the most prominent clinical signs and include delayed exfoliation of primary teeth, delayed or failed eruption of permanent teeth, and the presence of multiple supernumerary teeth. This report describes the case of a 13-year-old female presenting with retained primary teeth, delayed eruption of permanent teeth, and characteristic craniofacial features consistent with MSS. Clinical examination revealed frontal bossing, midface retrusion, and a high-arched palate. Cone-beam Computed Tomography (CBCT) was performed, revealing multiple unerupted permanent teeth and six supernumerary teeth distributed across all four quadrants. In addition, CBCT imaging identified other skeletal anomalies not clearly evident on conventional panoramic radiographs. This case highlights the critical role of CBCT in the comprehensive assessment and treatment planning of patients with MSS. Its ability to provide high-resolution, three-dimensional visualisation enables more accurate localisation of impacted and supernumerary teeth, aiding in surgical and orthodontic management. Early diagnosis and interdisciplinary care are essential to mitigate functional and aesthetic complications associated with this complex syndrome.

Keywords: Clediocranial dysplasia, Hyperdontia, Retained deciduous teeth, 3D imaging

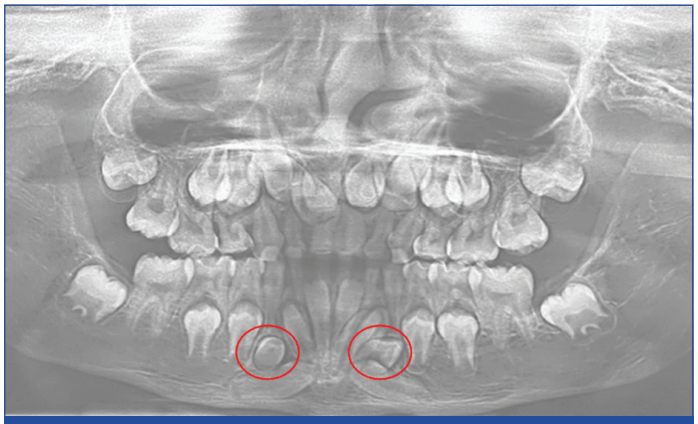
CASE REPORT

A 13-year-old female patient reported to the Department of Paediatric and Preventive Dentistry with the chief complaint of retained deciduous teeth in both the upper and lower arches. The patient had no prior dental history. Notably, her mother exhibited similar craniofacial and oral features, although she was not ready to undergo medical evaluation. On general physical examination, the patient presented with characteristic features suggestive of a syndromic condition, including short stature, frontal bossing, and midfacial hypoplasia. Additional findings included dry skin, sparse eyebrows, and clubbing of both fingernails and toenails. Hypoplastic clavicles were noted, contributing to excessive shoulder mobility. However, the patient's vital signs and systemic health were within normal limits. Intraoral examination revealed the complete retention of primary teeth in both arches, along with the presence of the permanent first molars (teeth 16, 26, 36, and 46) [Table/Fig-1]. Extraoral examination further confirmed features such as frontal bossing, sparse eyebrows, dry skin, and digital clubbing.



[Table/Fig-1]: Intraoral pictures of the maxillary and mandibular arches showing the presence of all the primary teeth along with first permanent molars in both arches.

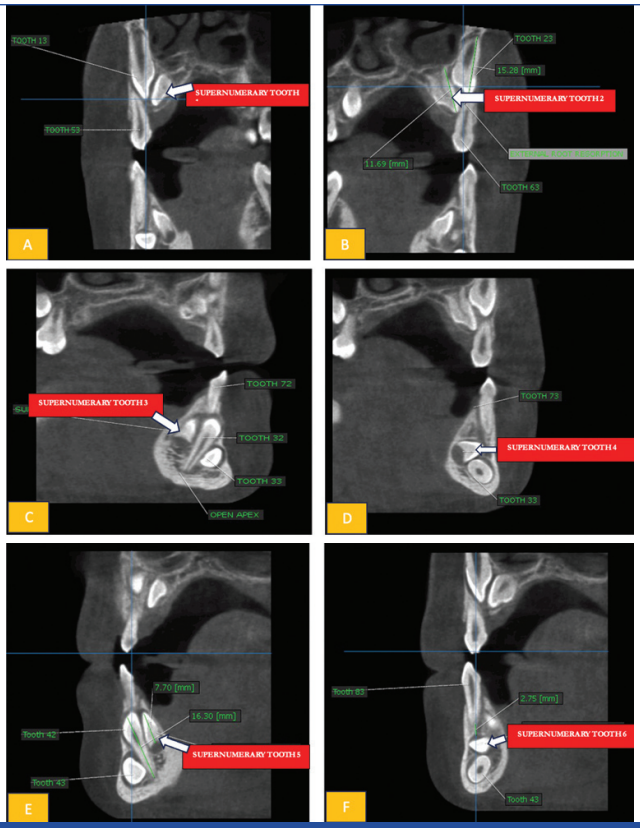
Patient was advised Orthopantomograph (OPG) to check for the development of permanent teeth. OPG revealed impacted tooth buds of 11, 12, 13, 14, 15, 17, 21, 22, 23, 24, 25, 27, 31, 32, 33, 34, 35, 37, 41, 42, 43, 44, 45, 47 along with two supernumerary teeth in relation to 33 and 43 impacted [Table/Fig-2].



[Table/Fig-2]: OPG revealing all the retained primary teeth and erupted permanent first molars, along with the presence of only two supernumerary teeth in the mandibular anterior region.

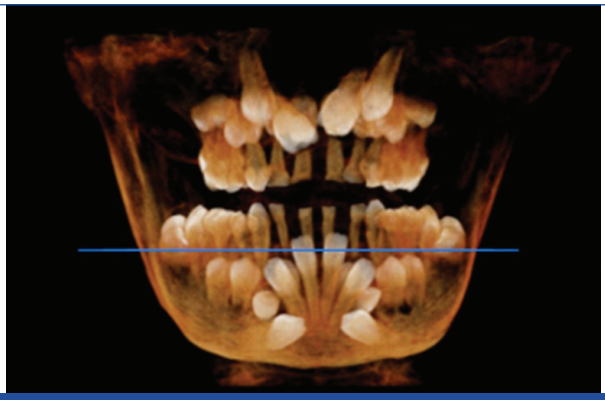
To know the status and location of all permanent and impacted teeth, CBCT was advised and the report revealed that there were over retained deciduous teeth in relation to 51, 52, 53, 54, 55, 61, 62, 63, 64, 65, 71, 72, 73, 74, 75, 81, 82, 83, 84, 85. Dental caries in relation to 52, 53, 54, 61, 62, 63, 64, 85. Impacted teeth irt to 11, 12, 13, 14, 15, 21, 22, 23, 24, 25, 31, 32, 33, 34, 35, 41, 42, 43, 44, 45. Horizontally impacted permanent teeth in relation to 33 and 43. Vertically impacted teeth in relation to 31, 32, 41, 42. Mesioangular impaction in relation to 11 and 21 [Table/Fig-3].

There were multiple impacted supernumerary teeth in the maxillary and mandibular arches. A total of 6 supernumerary teeth were seen. Supernumerary tooth 1 was palatal to 13, with the crown fully formed. It was 7.9 mm long, beneath the nasal floor, with thin bone separation from 13 and surrounded by follicular space. Supernumerary tooth 2



[Table/Fig-3]: Presence of all six supernumerary teeth revealed on CBCT examination.

was palatal to with complete crown and 1/3 root. It was 11.69 mm long, beneath the nasal floor, with thin bone separation from 23, and surrounded by follicular space. Supernumerary tooth 3 was lingual to 32, conical, 6.47 mm long, with crown complete with early root formation. There was thin bone over the incisal surface, surrounded by follicular space. Supernumerary tooth 4 was horizontal below 73, above the impacted canine, crown was complete with early root, 6.32 mm in length. The crown faced labially, surrounded by follicular space. Supernumerary tooth 5 was lingual to 42, conical, 7.7 mm in length, crown complete with early root, with thin bone over the incisal surface, surrounded by follicular space. Supernumerary tooth 6 was horizontal below 83, 2.75 mm above impacted canine. The crown faced labially, 7.53 mm in length, crown complete with early root and surrounded by follicular space. Three-dimensional (3D) images show the various impacted and retained teeth in both the jaws [Table/Fig-4].



[Table/Fig-4]: Digital volume tomography showing all the retained primary and impacted permanent teeth.

At the time of reporting, the patient and parents were counselled regarding the staged surgical and orthodontic treatment plan. The family was advised to report for the extraction of retained deciduous teeth and surgical exposure of impacted permanent teeth, followed by orthodontic guidance. However, despite repeated reminders and counselling, the patient did not return for the proposed intervention. Hence, no active treatment could be initiated, and long-term follow-up data are unavailable

DISCUSSION

Marie-Sainton Syndrome (MSS), also known as cleidocranial dysplasia or dento-osseous dysplasia, is a rare, autosomal dominant skeletal condition characterised by short stature, frontal and parietal bossing, delayed cranial suture closure, absent or hypoplastic clavicles, hypoplastic maxilla, and brachydactyly [1,2]. Dental anomalies include multiple supernumerary teeth, primary tooth retention, delayed eruption, and permanent dentition impaction. These issues rarely impact main tooth development, but often lead to low height, midface dysplasia, and sloping shoulders, prompting treatment [3,4].

In the presented case, a 13-year-old female patient reported with retained primary teeth and absence of permanent teeth eruption. On clinical examination, characteristic craniofacial features of MSS were noted, including midface hypoplasia, sloping shoulders, and brachycephaly. A CBCT scan was initially performed to check for the position of the two supernumerary teeth, which were seen on OPG examination, but later, along with those, four other supernumerary teeth were revealed, along with delayed eruption patterns.

The utility of CBCT in this case was pivotal and decisive. Unlike conventional radiographs, CBCT offers high-resolution, three-dimensional visualisation of the craniofacial complex. It allows precise evaluation of buccolingual and mesiodistal angulations; exact spatial positioning of supernumerary and impacted teeth; proximity to adjacent structures (nerves, sinuses, and roots); cortical bone integrity and thickness; Eruption pathways and obstructions

In this case, CBCT unveiled four additional supernumerary teeth not visible on the OPG, bringing the total to six, distributed across all four quadrants. The images also revealed delayed eruption patterns and the complex orientations of the impacted permanent teeth. These insights were critical in guiding treatment planning, especially for the surgical approach and orthodontic guidance of erupting teeth. The volumetric data generated by CBCT provided detailed, slice-by-slice information—essential in a condition like MSS where skeletal and dental anomalies coexist in three dimensions.

The findings in this case align with those reported in previous literature, where the presence of multiple supernumerary teeth (often fewer than ten) and delayed eruption are hallmark features of MSS [5,6].

Although genetic testing for the RUNX2 mutation, known to be involved in the pathogenesis of MSS, was advised, financial constraints limited further molecular investigations [7]. However, the diagnosis was clinically and radiographically confirmed based on characteristic skeletal and dental features consistent with previously reported cases.

Counselling of the patient and parents was carried out to explain the nature of the syndrome, possible genetic implications, and the importance of long-term interdisciplinary management. Emphasis was placed on the need for staged treatment involving surgical, orthodontic, and possibly prosthetic interventions [8]. However, due to financial and logistical limitations, treatment had not been initiated at the time of report submission. The patient is currently under periodic follow-up, and early surgical intervention is planned based on eruption monitoring.

The proposed treatment plan included surgical removal of all retained primary teeth and impacted supernumerary teeth, surgical exposure of permanent teeth based on their eruptive sequence, and orthodontic guidance to align erupting teeth into occlusion, followed by long-term monitoring for craniofacial growth and dental development.

Internationally, studies by Park TK et al., [9] have described comprehensive management strategies for MSS, incorporating early

diagnosis, extraction of retained deciduous and supernumerary teeth, surgical exposure of permanent teeth, orthodontic alignment, and orthognathic interventions like Le Fort I advancement. However, such full-scope treatment is often resource-intensive. In contrast, the present case adopts a conservative, stage-based approach due to socioeconomic constraints, yet remains guided by CBCT data for optimal timing and planning.

In this case, six supernumerary teeth were observed across all four quadrants. Their deep impaction and variable orientations (buccolingual and mesiodistal) presented additional surgical challenges and features that have similarly been reported by Asan MF et al., [10] and Ramalingam S et al., [11] in Indian patients with MSS.

Compared to other Indian reports from the past decade, the current case displays typical skeletal and dental characteristics but with additional dermatological findings like sparse eyebrows, dry skin, and clubbing of nails—rarely emphasised in prior Indian literature. Most previous Indian cases lacked advanced imaging, relying only on orthopantomograms (OPGs), which underreported the total number and extent of impacted and supernumerary teeth [10-13]. Only a few, such as the report by Asan MF et al., [10] employed CBCT to reveal a total of 58 teeth, underlining its superiority over conventional modalities [Table/Fig-5].

Author (Year)	Age/Sex	Clavicular anomaly	Dental findings	CBCT used
Asan MF et al., (2022) [10]	Adolescent female	Hypoplastic clavicles	Multiple retained, impacted, and supernumerary teeth (~58 teeth)	Yes
Gupta NS et al., (2015) [12]	Adolescent	Hypoplastic/ absent clavicles	Multiple impacted and supernumerary teeth	Yes
Chaturvedi S et al., (2022, series) [13]	Nine CCD subjects	Hypoplastic clavicles	Dental anomalies + airway volume assessment	Yes
Bhagat V. (2019) [14]	16-year-old male	Hypoplastic clavicles	Multiple impacted and supernumerary teeth	No
Vishnurekha C et al., (2019) [15]	10-year-old female	Hypoplastic clavicles	Retained primary, unerupted permanent teeth, supernumerary teeth	No

[Table/Fig-5]: Comparative analysis of various case reports on MSS [10, 12-15].

This case emphasises the diagnostic and planning utility of CBCT in MSS and reinforces the need for early, multidisciplinary intervention. Comparative analysis shows consistent skeletal and dental findings across Indian and global cases, with CBCT enhancing accuracy and clinical outcomes. Further studies, especially involving genetic confirmation and long-term follow-up, are warranted to better understand the full phenotypic spectrum of MSS and refine treatment protocols.

CONCLUSION

MSS, though rare, presents distinct diagnostic and therapeutic challenges. Early diagnosis through clinical and radiological assessment, patient-family counselling, and a well-structured multidisciplinary treatment approach are essential for optimising both functional and aesthetic outcomes. CBCT proves to be an indispensable imaging modality in such cases, supporting precise diagnosis and comprehensive treatment planning. Continued follow-up and staged intervention remain key to successful management in resource-limited scenarios.

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PARTICULARS OF CONTRIBUTORS:
1. Professor, Department of Paediatric and Preventive Dentistry, Rajarajeswari Dental College and Hospital, Bangalore, Karnataka, India. **ORCID ID: 0000-0003-2426-5057.**
2. Postgraduate Student, Department of Paediatric and Preventive Dentistry, Rajarajeswari Dental College and Hospital, Bangalore, Karnataka, India. **ORCID ID: 0009-0000-6311-0141.**
3. Postgraduate Student, Department of Paediatric and Preventive Dentistry, Rajarajeswari Dental College and Hospital, Bangalore, Karnataka, India. **ORCID ID: 0009-0001-4467-087X.**

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Umapathy Gowda,
Professor, Department of Paediatric and Preventive Dentistry, Rajarajeswari Dental College and Hospital, No. 14, Ramohalli Cross, Kumbalgodu, Mysore Road, Bangalore-560074, Karnataka, India.
E-mail: umapathygowda@gmail.com

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