Solitary median maxillary central incisor in association with hemifacial microsomia: A rare case report and review of literature

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Abstract

Solitary median maxillary central incisor (SMMCI) is a rare dental anomaly. It is estimated to occur in 1:50,000 live births. The SMMCI tooth differs from the normal central incisor in that the crown form is symmetric and it develops and erupts precisely in the midline of the maxillary dental arch in both primary and permanent dentitions. Presence of SMMCI with hemifacial microsomia (HFM) is a very rare clinical condition. We report a case of HFM in a male of Indian origin who presented with SMMCI in both primary and permanent dentitions. The association of HFM with SMMCI may be due to defective development of neural crest cells and/or lack of space in maxilla.

Keywords: Hemifacial microsomia, solitary median maxillary central incisor, anodontia

Introduction

Solitary median maxillary central incisor (SMMCI) is a rare dental anomaly.[1] Presence of a solitary symmetrical maxillary central incisor of normal crown dimensions situated precisely in the midline in both primary and permanent dentitions was apparently first reported by Scott.[2] Since then, SMMCI has been reported both as an apparently isolated dental finding and with a variety of midline developmental defects, e.g. holoprosencephaly (HPE), and/or pituitary dysfunction. The name SMMCI was originally suggested by Hall *et al*.[3] Various terminologies like "monosuperoincisivodontic dwarfism," "single central incisor syndrome," "single maxillary central incisor," and "single incisor," suggested by other authors,[4-7] do not adequately describe the peculiarly formed incisor tooth.[8] The SMMCI differs from normal incisor in that the crown form is symmetric and it develops precisely in midline of maxillary dental arch in both primary and permanent dentitions and is not a supernumerary tooth.[8] The incidence of SMMCI is around 1:50,000 live births.[8] SMMCI may occur in isolation or in association with other systemic abnormalities like short stature, pituitary insufficiency, microcephaly, choanal atresia, midnasal stenosis, and congenital nasal pyriform aperture stenosis.[9] It is considered as one of the most minimal expressions (microforms) of the HPE spectrum.[3] Deletions on chromosomes 7 and 18 (at 7q36.1 and 18p-), which are in chromosomal regions that harbor HPE genes, have been reported to be associated with SMMCI.[10-13] Some investigators found Sonic Hedgehog (SHH) mutation in patients affected by SMMCI.[1,9,14] Hehr et al.[15] emphasized the wide phenotypic variability in families with HPE and SHH mutation.

Hemifacial microsomia (HFM) is believed to be the second most common craniofacial anomaly following cleft lip and palate, involving first and second branchial arch derivatives.[16] An association has been described between HFM and hypodontia.[17] Barring a single case report,[18] the presence of single maxillary central incisor has not been reported in association with HFM. This article reports the case of a boy with HFM syndrome who presented with the classical signs of the syndrome and an SMMCI.

Case Report

A 6-year-old boy of Indian origin attended cleft and craniofacial clinic at Oral Health Science Centre, PGIMER, Chandigarh, with the chief complaint of asymmetry of face. The baby was born to non-consanguineous healthy parents of normal stature. There was no history of hereditary disease in his family. There was no history of medication during pregnancy and the delivery was atraumatic and full term.

The patient had undergone surgical procedure 1 year after birth for torticollis and had average built and normal mental development. There was no history of nasal malformation or stenosis.

The face was asymmetric with hypoplastic left side. Microtia was apparent with preauricular tags. The philtrum was

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Access this article online	
Quick Response Code:	Website: www.contempclindent.org
	DOI: 10.4103/0976-237X.91810

indistinct and occlusal plane was canted up to left. The masseter and temporalis muscles were hypolplastic and mastoid prominence was small [Figure 1].

Intraoral examination revealed the presence of single deciduous central incisor. The incisor was placed in midline and had symmetrical right and left contours, which resembled distal contour of central incisors. There was no labial frenum and incisive papilla. The palate had characteristic v shape, with fine bony ridge running along the length. The patient had poor oral hygiene and multiple carious teeth. The arches were narrow in size with the left side more collapsed than the right [Figure 2].

Intraoral periapical x-ray views of the patient's maxillary incisor region confirmed the presence of solitary incisor. The permanent incisor was seen above the deciduous and erupting in midline. The intermaxillary suture was distinctly visible on the radiograph [Figure 3].

The patient required a multidisiciplinary management and long-term follow-up. In the first phase, oral hygiene was monitored and oral hygiene instructions reinforced. This was followed by restoration of all carious lesions by a pediatric dentist.

The patient was reviewed every 6 months and radiographs for regular monitoring of facial growth were done. The facial asymmetry aggravated as the age advanced due to differential growth of right and left sides [Figures 4–6]. At the age of 8½ years, the permanent maxillary central incisor which has the morphology of fused central incisors erupted in midline. The size of this central incisor was comparable to normal central incisor [Figure 7]. Panoramic view showed that the coronoid process, condylar process, and mandibular ramus were hypoplastic on the left side and eruption of teeth was delayed due to lack of space in arch [Figure 8].

As a part of dental treatment, maxillary expansion can be commenced in late mixed dentition and the space created after moving SMMCI to one side of the arch can be replaced by a single tooth implant or crown by a prosthodontist. Mandibular distraction and reconstruction of face can be planned in multiple stages. Consultation with ENT specialist for auricular defects should be done. Genetic counseling is required, as some individuals with SMMCI, normal intelligence, and normal brain image have had children with HPE[19] and SMMCI has been recognized as a risk factor for holoprosencephalic offspring.

Discussion

To the best of our knowledge, this is the second case report in literature in which SMMCI was found in association with HFM. The first was reported by Garcia de Paula e Silva *et al.* in a 10-year-old male patient.[18]

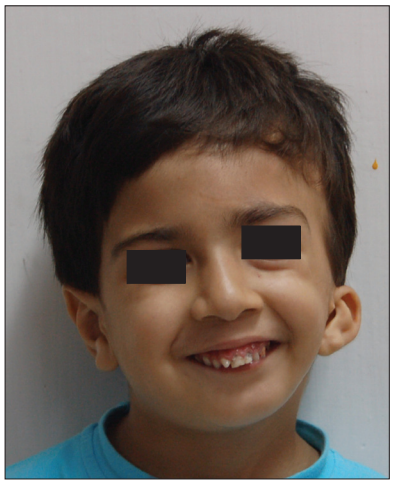

Figure 1: Frontal view of the patient at 6 years showing facial asymmetry, indistinct philtrum and SMMCI

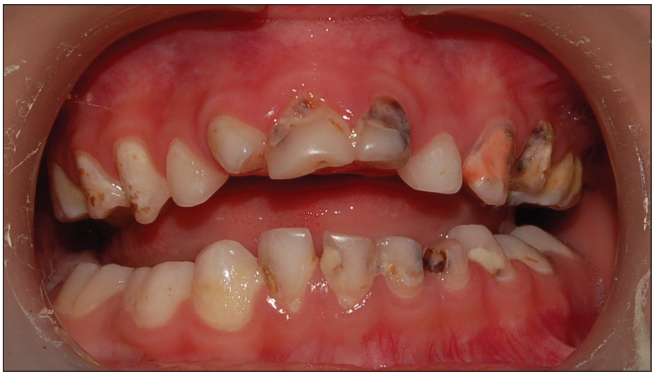

Figure 2: Intraoral view showing the presence of SMMCI (primary central incisor) and absence of labial frenum

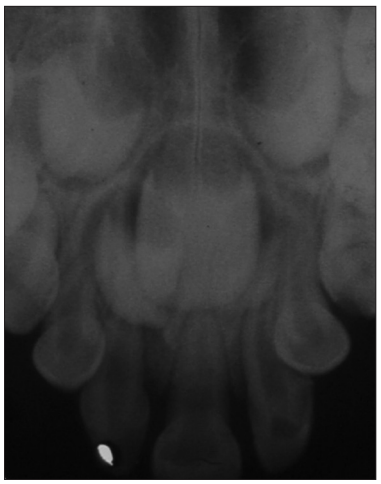

Figure 3: Intraoral periapical radiograph confirming the presence of SMMCI and intermaxillary suture

The presence of SMMCI has been correlated to HPE. It is likely that a number of mechanisms can give rise to SMMCI syndrome, some of which may also cause HPE. Hall *et al.*[3] reported a series of 21 consecutive cases of SMMCI, the

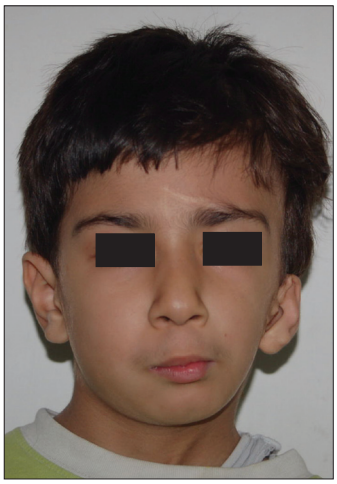

Figure 4: Extraoral frontal view at 8 years showing exaggerated asymmetry and retarded growth on the left side

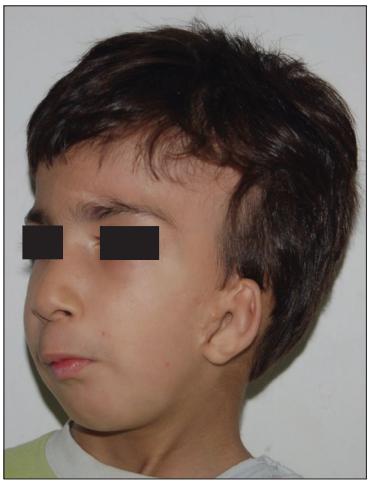

Figure 5: Right lateral view showing normal development

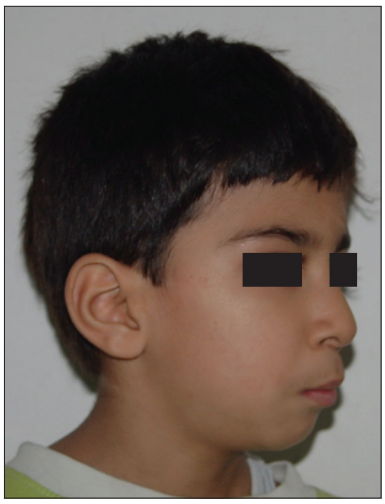

Figure 6: Left lateral view showing stunted growth

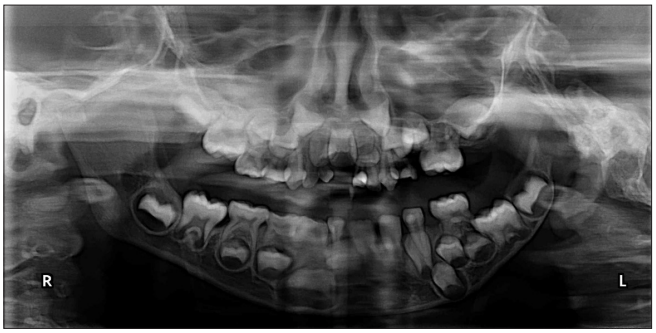

Figure 8: Panoramic radiograph at 8 years of age showing HFM, SMMCI, and delayed eruption on the left side

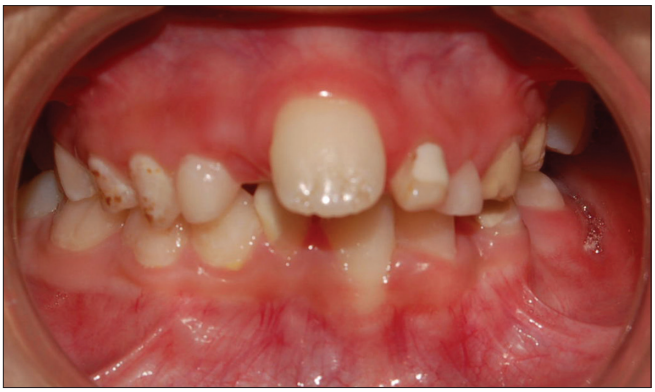

Figure 7: Intraoral view at 8 years showing SMMCI (permanent central incisor)

spectrum of anomalies, and associated features present in these cases, and defined SMMCI syndrome. SMMCI can also be a feature of recognized syndromes or associations or a finding in patients with specific chromosomal abnormalities.

Nanni *et al*.[9] reviewed the extensive number of anomalies in addition to HPE with which SMMCI had been reported. The correlation has been found with,Velocardiofacial (VCF), DiGeorge syndrome, HPE, ectodermal dysplasia, and Duane retraction syndrome, but not with HFM.

HFM is a syndrome which affects craniofacial structures to varying degree. This syndrome occurs at a higher rate in males than in females.[20] The incidence of this condition, also known as oculoauriculovertebral dysplasia, is about 1 in 5000 to 25,000 live births.[21] The phenotype is highly variable and the features include unilateral deformity of the external ear and small ipsilateral half of the face with epibulbar dermoid and vertebral anomalies. Coloboma of the upper eyelid is frequent. The ear deformities range from preauricular tags of cartilagenous masses to atresia of the external auditory canal, anomalies in the size and shape of the external auricle, and even anotia.[22] In addition to the craniofacial anomalies, there may be cardiac, vertebral, and central nervous system defects.[16] Commonly observed malformations include microtia, macrostomia, and failure of formation of mandibular ramus and condyle. The patient had classical features of HFM.

A factor aggravating asymmetry at an earlier age might be torticollis, but due to early release (within 1 year), the effect on growth of face is expected to be minimum.

The etiology of both the conditions remains uncertain although various theories have been proposed. The SMMCI may be due to a congenitally missing tooth bud with agenesis of the incisor and the remaining incisor erupts in the midline.[23] It has been hypothesized that the formation of one instead of two teeth could result from a disturbance in the mitotic potential of the incisor tooth bud, which could be under genetic and environmental determinants.[24] It has also been proposed that for SMMCI to form, the dental lamina must have fused prematurely in midline, resulting in apposition and fusion of forming tooth buds.[3,9,25] This prevents normal formation of intervening bone and associated soft tissue. It has also been suggested that space limitation in maxillary arch or deficiency of lateral growth from midline would result in premature fusion of spreading lamina from the right and left sides.[25] In this patient, the solitary incisor might be a result of premature fusion in midline as in HFM there is lack of space in jaws.

Although genetic and environmental pathogenic mechanisms have been proposed for HFM, it is likely that the cause is multifactorial. In a review of various pathogenic models, the authors underscore studies of the genetic component of HFM.[26] Several investigators favor a vascular basis for the development of this anomaly.[27-31] Another theory is based on disruption of migration of neural crest cells during craniofacial development.[17,32-34] If a disturbance in neural crest cell development or migration plays a role in HFM and hypodontia, it would follow that a correlation between the two conditions should occur. It is generally accepted that normal odontogenesis requires the presence and interaction of neural crest ectoderm and mesenchymal cells. Disturbances in the odontogenic process can produce abnormal or incomplete dental development.[35] In a previous study, an association has been found between smaller size of dentition and HFM.[36]

It should be noted that the SMMCI may occur alone or in other conditions bearing no relationship to HPE.[30] Cohen[37] states that rather than thinking about a single central incisor as a microform of HPE, it is better to think of it as

1. An integral component of severe HPE,
2. An anomaly that occurs alone and in other conditions unrelated to HPE,
3. The only manifestation in some members of a dominantly affected family with variable expressivity of HPE and incomplete penetrance, and
4. Rarely as an isolated dominant trait with an SHH mutation.

In this case, SMMCI along with HFM may be considered as an anomaly that has occurred alone and is unrelated to HPE. It is also possible that in patients presenting with HFM, the SMMCI may be more than a chance finding. A further critical evaluation of patients with HFM and association between the two might be helpful in providing insight into the unknown etiology of the two conditions.

References

1. Garavelli L, Zanacca C, Caselli G, Banchini G, Dubourg C, David V, *et al*. Solitary median maxillary central incisor syndrome: clinical case with a novel mutation of sonic hedgehog. Am J Med Genet A 2004;127:93-5.
2. Scott DC. Absence of upper central incisor. Br Dent J 1958;104: 247-8.
3. Hall RK, Bankier A, Aldred MJ, Kan K, Lucas JO, Perks AG. Solitary median maxillary central incisor, short stature, choanal atresia/midnasal stenosis (SMMCI) syndrome. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 1997;84:651-62.
4. Rappaport EB, Ulstrom R, Gorlin RJ. Monosuperocentroincisivodontic dwarfism. Birth Defects Orig Artic Ser 1976;12:243-5.
5. Maréchaux SC. The single maxillary central primary incisor: report of case. ASDC J Dent Child 1986;53:124-12.
6. Mass E, Sarnat H. Single maxillary central incisors in the midline. ASDC J Dent Child 1991;58:413-6.
7. Parker PR, Vann WF Jr. Solitary maxillary central incisor: clinical report. Pediatr Dent 1985;7:134-6.
8. Hall RK. Solitary median maxillary incisor syndrome. Orphanet J Rare Dis 2006;9:1-12.
9. Nanni L, Ming JE, Du Y, Hall RK, Aldred M, Bankier A, *et al*. SHH mutation is associated with solitary median maxillary central incisor: a study of 13 patients and review of the literature. Am J Med Genet 2001;102:1-10.
10. Aughton DJ, AlSaadi AA, Transue DJ. Single maxillary central incisor in a girl with del(18p) syndrome. J Med Genet 1991;28: 530-2.
11. Boudailliez B, Morichon-Delvallez N, Goldfarb A, Pautard JC, Lenaerts C, Piussan C. Solitary upper incisor, hypopituitarism and monosomy 18p chromosome aberration. J Genet Hum 1983;31:239-242.
12. Frints SG, Schrander-Stumpel CT, Schoenmakers EF, Engelen JJ, Reekers AB, Van den Neucker AM et al. Strong variable clinical presentation in 3 patients with 7q terminal deletion. Genet Couns 1998; 9:5-14.
13. Roessler E, Muenke M. Holoprosencephaly: a paradigm for the complex genetics of brain development. J Inherit Metab Dis 1998;21:481-97.
14. Marini M, Cusano R, De Biasio P, Caroli F, Lerone M, Silengo M, *et al*. Previously undescribed nonsense mutation in SHH caused autosomal dominant holoprosencephaly with wide intrafamilial variability. Am J Med Genet A 2003;117:112-5.
15. Hehr U, Gross C, Diebold U, Wahl D, Beudt U, Heidemann P, *et al*. Wide phenotypic variability in families with holoprosencephaly and a sonic hedgehog mutation. Eur J Pediatr 2004;163:347-52.
16. Hemifacial Microsomia;HFM. Available from: http://www.ncbi.nlm.nih.gov/omim/164210.
17. Maruko E, Hayes C, Evans CA, Padwa B, Mulliken JB. Hypodontia in hemifacial microsomia. Cleft Palate Craniofac J 2001;38:15-9.
18. Garcia de Paula e Silva FW, Carvalho FK, Diaz-Serrano KV, Freitas AC, Borsatto MC, Queiroz AM. Solitary median maxillary central incisor in association with Goldenhar's syndrome: a case report. Spec Care Dentist. 2007;27:105-7.
19. Nanni L, Ming JE, Bocian M, Steinhaus K, Bianchi DW, Die-Smulders C, *et al*. The mutational spectrum of the Sonic hedgehog gene in holoprosencephaly: SHH mutations cause a signifcant proportion of autosomal dominant holoprosencephaly. Hum Mol Genet 1999;8:2479-88.
20. Rollnick BR, Kaye CI, Nagatoshi K, Hauck W, Martin AO. Oculoauriculovertebral dysplasia and variants: phenotypic characteristics of 294 patients. Am J Med Genet 1987;26:361-75.

21. Pinheiro AL, Araújo LC, Oliveira SB, Sampaio MC, Freitas AC. Goldenhar's syndrome - case report. Braz Dent J 2003;14:67-70.
22. Gorlin RJ, Jue KL, Jacobsen V, Goldschmidt E. Oculoauriculovertebral dysplasia. J Pediatr 1963;63:991-9.
23. Yassin OM, El-Tal YM. Solitary maxillary central incisor in the midline associated with systemic disorders. Oral Surg Oral Med Oral Pathol 1998;85:548-51.
24. Osborn JW, Ten Cate AR. Advanced dental histology. 4th ed. Bristol, London, Boston: Wright PSG; 1983. p. 35-45.
25. Becktor KB, Sverrild L, Pallisgaard C, Burhøj J, Kjaer I. Eruption of central incisor,the intermaxillary suture and maxillary growth in patients with a single median maxillary central incisor. Acta Odontol Scand 2001;59:361-6.
26. Cousley RR, Calvert ML. Current concepts in the understanding and management of hemifacial microsomia. Br J Plast Surg 1997;50:536-51.
27. Braithwaite F, Watson J. A report on three unusual cleft lips. Br J Plast Surg 1949;2:38-49.
28. Keith A. Three demonstrations of congenital malformations of palate, faceand neck. Br Med J 1909;2:483.
29. Gorlin RJ, Jue KL, Jacobsen UL, Goldschmidt E. Oculoauriculovertebral dysplasia. J Pediatr 1963;63:991-9.
30. Poswillo D. The pathogenesis of the first and second branchial arch syndrome. Oral Surg Oral Med Oral Pathol 1973;35:302-28.
31. Johnston MC. The neural crest in abnormalities of the face and brain. Birth Defects Orig Artic Ser 1975;11:1-18.
32. Johnston MC. Embryology of the head and neck. Plast Reconstr Surg 1990;4:2451-95.
33. Sulik KK, Johnston MC, Smiley SJ, Speight HS, Jarvus BE. Mandibulofacial dysostosis (Treacher Collins syndrome): a new proposal for its pathogenesis. Am J Med Genet 1987;27:359-72.
34. Thomas JT, Frias JL. The heart in selected congenital malformations. A lesson in pathogenic relationships. Ann Clin Lab Sci 1987;17:207-10.
35. Shafer WG, Hine MK, Levy BM. A Textbook of Oral Pathology. 5th ed. Philadelphia: WB Saunders; 1983. p. 45-7,63-4.
36. Seow WK, Urban S, Vafaie N, Shusterman S. Morphometric analysis of the primary and permanent dentitions in hemifacial microsomia. J Dent Res 1998;77:27-38.
37. Cohen MM Jr. Problems in the definition of holoprosencephaly. Am J Med Genet 2001;103:183-7.

How to cite this article: Utreja A, Zahid SN, Gupta R. Solitary median maxillary central incisor in association with hemifacial microsomia: A rare case report and review of literature. Contemp Clin Dent 2011;2:385-9.

Source of Support: Nil. **Conflict of Interest:** None declared.