Single upper central incisor syndrome: Report of a rare case

A. Sanguida

Department of Paedodontics and Preventive Dentistry, Indira Gandhi Institute of Dental Sciences, Sri Balaji Vidyapeeth University, Pillayarkuppam, Puducherry, India

Correspondence
A. Sanguida, Department of Paedodontics and Preventive Dentistry, Indira Gandhi Institute of Dental Sciences, Sri Balaji Vidyapeeth University, Pillayarkuppam - 607 403, Puducherry, India.
Phone: +91-8056105873.
Email: sanguidaa@igids.ac.in

Received 25 April 2017; Accepted 20 May 2017

How to cite the article:

doi: 10.15713/ins.ijmdcr.64

Abstract
Solitary median maxillary central incisor (SMMCI) syndrome is considered as a microform of holoprosencephaly (HPE) spectrum. It is a unique developmental abnormality affecting midline structures of the body. Systemic abnormalities involve major systems of the body such as cardiovascular, gastrointestinal, renal, endocrine, genital, and skeletal systems. Abnormal features involving eyes, ears, nose, mouth, and skin are also present. SMMCI patients belong to the HPE spectrum, and hence are at risk of having children with a more severe form of HPE. Very often SMMCI patients may first be seen by a pedodontist or an orthodontist and it is important that the dentist performs a complete examination to rule out systemic abnormalities and involve a multidisciplinary team to manage the patient. This article reports about a 5-year-old male child who reported with dental abscess in whom SMMCI was an incidental finding. Further investigations revealed the association of SMMCI with anomalies such as chronic respiratory obstruction, short stature, abnormal clinical and radiographic craniofacial morphology, duplicated left thumb phalynx, and absent left testis.

Keywords: Incisor, nasal obstruction, single upper central incisor, syndrome, thumb

Introduction
Single median maxillary central incisor (SMMCI) means that the tooth is the only central incisor tooth in the maxilla, present exactly in the midline, occurring only in the maxilla, and though of unusual crown form it is not a mesiodens. SMMCI syndrome has been considered as a microform of the holoprosencephaly (HPE) spectrum. HPE (holos – undivided; prosencephaly – frontal brain) is a complex developmental field defect of the forebrain in which the cerebral hemispheres fail to separate into distinct halves.[1] HPE spectrum can be classified into five different types; cyclopia, ethmocephaly, cebophagephaly, median cleft palate, and short upper lip (with mid-palatal ridge and SMMCI).[2] Although SMMCI could be a predictor of HPE, in the majority of cases HPE occurs in the absence of SMMCI. The incidence of SMMCI is 1:50000 and that of HPE is 1:16000 live births.[1]

Most severe cases of HPE do not survive and may appear as spontaneous abortions, and less severe cases may be characterized by an SMMCI.[2] Hence, SMMCI may first be detected by a pedodontist or an orthodontist. SMMCI syndrome could be a developmental field effect involving midline structures of the body due to unknown events occurring between 35th and 38th days of intrauterine life. In the head, the defects involve cranial bones, maxilla, and its dentition, especially the central incisors, nasal airways, and sometimes the brain.[1] Several systemic abnormalities such as short stature, growth hormone deficiency, panhypopituitarism, hypothyroidism, pituitary gland morphological abnormalities, congenital heart disease, tracheoesophageal fistula, anal atresia, congenital ectopic anus, absent left kidney, undescended testis, slow learning abilities or ADHD have been found to be associated with SMMCI. Abnormalities involving facial structures such as visual defects, hypotelorism, slanting palpebral fissures, microphthalmia, iris coloboma, thick pine and small pit of ears, periauricular skin tags, nasal obstruction, depressed nasal bridge, anteverted nostrils, nasal groove, deviated nasal septum, and small nose are also common. Skin anomalies such as incontinentia pigmenti achromians, café au lait spots and fine axillary hair are other associated anomalies. It is also associated with syndromes such as CHARGE association, VACTERL association, ectodermal dysplasia, velocardiofacial syndrome, autosomal dominant HPE, and Duane retraction syndrome. It is also associated with chromosomal abnormalities such as del (18p), 47XXX, and mutation in the sonic hedgehog (SHH) gene.[3,4]

A single maxillary central incisor tooth is easily detectable, and hence relatives also should be examined carefully as it may indicate an expression of the HPE malformation. It is important, therefore, to perform complete analysis of the family pedigree of a patient with SMMCI so that we can explain the implications for future pregnancies of the parents and their children.[1]
The purpose of this paper is to emphasize to the dentist the importance of early identification of patients with single maxillary central incisor because the SMMCI may not be a concern for the patient; however, the patient might be suffering from other major or minor systemic anomalies and undergoing treatment for the same without a clue about the true nature of the disease.

**Case Report**

A 6-year-old male child reported to the pedodontics clinic with a chief complaint of pain in the left lower back teeth. During the examination, it was an incidental finding that the child had only a single maxillary deciduous central incisor located exactly in the midline. There was no history of trauma involving the maxillary incisor region. The medical history was significant in that the child had suffered from a nasal blockage at birth. He has also been suffering from chronic respiratory problems for which he takes medication prescribed by physicians. On physical examination, the child was of short stature; height 96.25 cm (below 3rd percentile), weight 15 kg (below 3rd percentile). No similar problems were noticed in the parents and the two siblings. The patient also had a duplicated thumb phalynx on the left hand [Figure 1]. Other notable extraoral features were frontal bossing, high hairline, depressed nasal bridge, arch-shaped upper lip, indistinct philtrum, and anteverted nostrils [Figure 2].

Notable intraoral features were a single symmetric deciduous central incisor exactly in the midline with hypoplastic defects, other primary teeth showing varying degrees of hypoplastic defects and dental caries, absent labial frenum of the upper lip, absent incisive papilla and a complete mid-palatal ridge [Figures 3 and 4]. Tooth number 74 and 75 had deep dental caries with chronic dentinoalveolar abscess, and hence root canal treatment was initiated after reviewing radiographs. Intraoral periapical radiograph, panoramic radiograph, and cephalometric radiographs were advised to confirm clinical findings and explore additional anomalies. The intraoral periapical radiograph revealed the presence of a single primary central incisor and a single permanent central incisor exactly in the midline with the absence of intermaxillary suture. Panoramic radiograph also confirmed this finding and revealed a very narrow nasal cavity [Figure 5]. Cephalometric radiograph was analyzed and showed retrognathic maxilla and mandible (SNA 78°; SNB 76°), short anterior and posterior cranial base length (S-N = 57.7 mm and S-Ba = 20.6 mm). Effective maxillary length was 59.3 mm. A deviant morphology of the sella was also noted [Figure 6]. After the initial management of the abscessed tooth the patient was subject to further systemic evaluation. A notable finding was the absence of left testis. Abdominal ultrasound was normal; no other midline structures were affected. The patient’s parents were made aware about the findings and were asked to report for further laboratory investigations, but they failed to report.

![Figure 1: Left hand showing duplicated thumb phalynx](image1.png)

![Figure 2: Face showing frontal bossing, high hairline, arch-shaped upper lip, indistinct philtrum, anteverted nostrils](image2.png)

![Figure 3: (a and b) Frontal view of occlusion showing single maxillary deciduous central incisor and absence of labial frenum of upper lip](image3.png)

![Figure 4: Maxillary occlusal view showing absent incisive papilla, complete mid-palatal ridge](image4.png)
Single maxillary central incisor syndrome  

Sanguida

Figure 5: Panoramic radiograph showing single primary and permanent central incisor, narrow nasal cavity

Figure 6: Abnormal sella morphology

Discussion

SMMC1 was first described by Scott in 1958 as an isolated defect in a 6-year-old girl.² SMMC1 could be considered either as an integral component of severe HPE or an anomaly that occurs alone and in other conditions unrelated to HPE. It could also be the only manifestation in some members of a dominantly affected family with variable expressivity for HPE and incomplete penetrance and rarely it can be an isolated dominant trait with an SHH mutation.⁵ The child affected in this report was a male and the history, clinical and radiographic examinations confirmed that the severe mental illness (SMI) was the only central incisor present in both the primary and permanent dentitions.

An anomaly that is positively associated with SMMC1 is congenital nasal malformation (either choanal atresia or mid-nasal stenosis or congenital nasal pyriform aperture stenosis). This child was suffering from continual nasal obstruction, and the panoramic radiograph showed a narrow nasal cavity. However, a computed tomography (CT) scan would have helped diagnose the exact nature of the nasal malformation.

Another anomaly associated with SMMC1 is short stature. Rappoport used the term “monosuperoentoicisivodontic dwarfism” when SMMC1 occurred with growth retardation. It should be noted that short stature is seen in only 50% of the cases and growth hormone deficient short stature in only 33% cases. Wesley et al. reported two children with unpaired maxillary central incisor, but who were normal in height and had normal growth hormone levels which might indicate a spectrum in this kind of an association of SMI and short stature.⁶ In the present case, there was no family history of short stature. It is also emphasized that before starting a detailed endocrine evaluation of a child with SMI, one has to observe height and growth rate and further laboratory evaluation should be undertaken only if the height is 2SD below the mean or if linear growth plateaus.⁶ Lo et al. reported 40 cases on SMMC1 and found that 69% had short stature, 48% had growth hormone deficiency/hypopituitarism, 23% had pituitary absence/hypoplasia, and 17% had del[18p] or r[18].⁵

Parentin and Perissutti reported a 4-year-old male child with SMMC1, growth hormone deficiency, Duane retraction syndrome in the right eye and a duplicated thumb phalnxx on the right hand.⁸ The child in the present case report showed a duplicated thumb phalnxx in the left hand. To the author’s knowledge, this is the second report of this anomaly being associated with SMMC1. Another notable finding in the present case report was the absence of left testis. Yassin and El-Tal reported a 9-year-old child with SMI, short stature with growth hormone deficiency, microcephaly, cryptorchidism, and deep nasal bridge.⁵

Kjær et al. reported on clinical features and craniofacial morphology of 10 patients (8-17 years) with SMMC1 (9 girls and 1 boy). These children had indistinct philtrum, arch-shaped upper lip, absence of labial frenum of upper lip, complete/incomplete mid-palatal ridge, SMMC1, nasal obstruction, narrow nasal cavities, septum deviation, retrognathia and posteriorly inclined maxilla and mandible, larger cranial base angle, reduced anterior cranial base length, and maxillary length and a deviant morphology of sella turcica in five out of ten patients.⁵ Tabatabaie et al. also reported similar findings in a group of thirteen patients with SMMC1.⁹ Apart from the presence of few of the above-mentioned anomalies the child in the present report also had abnormal sella morphology.

Diagnosis of SMMC1 is possible using ultrasound at 18-22 weeks but is rarely made prenatally. At birth, an infant with SMMC1 syndrome will appear normal, may be born preterm with low birth weight and neonatal nasal obstruction sometimes requiring surgical intervention. Once, an SMMC1 is identified along with the presence of any of the major or minor systemic anomalies the dentist should arrange for a multidisciplinary evaluation of the child and family members by a team of specialists comprising a geneticist, endocrinologist, otorlaryngologist, pediatric cardiologist, general surgeon, ophthalmologist, speech pathologist, thoracic physician, and allergist.¹¹ Patients with SMMC1 belong to HPE spectrum, and hence are at risk of having children with more severe form of HPE.¹⁰

Management in the simplest case of SMMC1 only and mild nasal airway narrowing involves only providing genetic counseling and good dental care.¹¹ Management of the primary dentition in SMMC1 involves only monitoring of facial development and...
occlusion.\(^4\) If there is the absence of internasal suture and parts of intermaxillary suture this will prevent transversal expansion of the palate, and hence maxillary expansion should not be done but only tooth movement with insertion of dental implants may be done. In some cases, the central incisor can be extracted and mesialization of laterals can be done. Orthodontic treatment is not required if the patient and parents are happy with the dentition and if there are no functional or esthetic problems.\(^9\)

In the permanent dentition use of expansion appliance to widen the palate will provide room for the SMMCI tooth to be moved to one side of the midline thus enabling placement of an artificial contralateral central incisor. The SMMCI tooth may then be recontoured to create the anatomical form of the appropriate side.\(^1\)

The child in this case requires further investigations such as CT scan to find the nature of nasal malformation, magnetic resonance imaging brain to rule out pituitary malformation, growth hormone assay, wrist radiograph to calculate bone age, and karyotyping. The family has to be provided with genetic counseling and be advised to come for routine dental visits to monitor growth and development of the child.

References

1. Hall RK. Solitary median maxillary central incisor (SMMCI) syndrome. Orphanet J Rare Dis 2006;1:12.