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Case Series

Literature Review of Solitary Median Maxillary Central Incisor (SMMCI) with Presentation of Case Series

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Abstract

Background: Solitary median maxillary central incisor (SMMCI) is a rare oddity, which may be associated with a complex of multiple developmental abnormalities. Sometimes, it is related to background systemic features, but may also be a separate localized anatomical aberration. Its occurrence has been estimated in 1:50,000 live births. SMMCIs were described together with cleft lip and palate (CLP), Velo Cardio Facial Syndrome (VCFS), exaggerated midline maxillary torus, distorted soft palate, and absence of the uvula and the midline labial frenum.

Aim: We present occasional sporadic cases of otherwise normal children.

Design: 5 cases of children are described and their clinical and radiographic examination are presented.

Results: Two peculiar findings, unreported till now, were the tendency of the pulp canal close to the apex to split and the lack of visible incisive papilla.

Conclusion: SMMCI occurs in both primary and permanent dentitions of the same child. It may be associated with systemic conditions or as a sporadic appearance. Splitting of the radicular canal close to the apex may affect root canal treatment and should be taken into consideration.

Keywords: *Solitary Median Maxillary Central Incisor (SMMCI); Maxillary Torus; Cleft Lip and Palate*

Introduction

Solitary median maxillary central incisor (SMMCI) as an isolated occurrence and SMMCI as part of a syndrome are rare dental anomalies [1]. The incidence has been estimated as 1:50,000 live births [2]. These anomalies are caused by heterozygous mutation in the Sonic Hedgehog Gene (SHH; 600725) on chromosome 7q36 [3].

The awareness of these anomalies goes back to 1958, where Scott reported "Absence of upper central incisor" [3]. Since then, the scientific literature regarding SMMCI is characterized by single or sporadic case reports, as well as efforts to investigate the etiology and delineate associated developmental and genetic aberrations.

Short stature and deficient growth hormone were described in patients with solitary maxillary central incisors in the primary and consecutive permanent dentitions. In some cases, with this feature, growth hormones were normal. No similar or associated oral and dental aberrations were found in families of those patients. Superoincisivodontic dwarfism, also known as 'monosuperoincisivodontic dwarfism' was used then to describe such phenomena [4,5]. Choanal atresia/mid-nasal stenosis and holoprosencephaly (HPE) was added to the short stature to a series of 21 consecutive cases of SMMCI syndrome in 1997 [2].

The holoprosencephaly spectrum (HPE): HPE stands for a teratologic series of varying severity, expressed by median malformations of the face and brain. There are a variety of chromosomal causes, yet most cases are sporadic and of unknown etiology. Less severe cases of Autosomal Dominant (AD) HPE may present with mild manifestations of hypotelorism, anosmia, hyposmia (reduced ability to smell), microcephaly, mental deficiency, or midface hypo-

plasia, and SMMCI. Ocular colobomas may also be included.

HPE is a specific malformation complex resulting from incorrect cleavage of the embryonic prosencephalon, (forebrain) with concurrent facial anomalies (midline facial structures) ranging from cyclopia to mild forms such as hypotelorism and SMMCI, with other clinical manifestations such as anophthalmia to mild hypotelorism, coloboma, hyposmia, dysplasia of olfactory bulbs and optic nerves [6].

The inheritance of HPE is Autosomal Dominant - the parents might be with minor manifestations such as single central incisor, hypotelorism, hyposomia, but offspring may be with severe manifestations, which emphasizes the importance of genetic counseling.

SMMCI syndrome, previously considered a simple midline defect of the dental lamina is "a possible predictor of HPEs of varying degrees in the proband, in members of the proband's family, and in the family's descendants" [2].

Most cases of HPE occur sporadically, yet occasionally a single upper central incisor associated with HPE may be found in close relatives (kindred). These findings "may represent evidence for a less severe form of HPE that may be transmitted in an AD fashion" [7].

Midline craniofacial malformations in association with hypothalamic-pituitary disorders has been observed in a girl with true precocious puberty associated with a SMMCI in both primary and permanent dentitions, and a hypothalamic hamartoma [8].

Genetics: Dolan., *et al*. (1981) described a 28-month-old boy, and Aughton., *et al*. (1991) reported a 7 years old girl with SMMCI [9,10]. Through genetic examination, both were found to have (18p) deletion syndrome. Due to the rarity of reporting of the anomaly at that time (1981 - 1991), SMMCI were evaluated as fused maxillary central incisors. It is associated with some degree of HPE in some cases with SMMCI in the mild manifestation of the HPE [10].

Occasionally, SMMCI with semilobar HPE, a median cleft lip, flat nose with a single nostril, hypotelorism, and normal chromosomes, may be an incidental mutation in otherwise healthy close relative, with no apparent dental or facial anomalies. Indeed, the presence of SMMCI in a newborn baby may be significant in genetic counseling as an indicator of potential HPE in the next generation, even if other relatives are apparently normal. The reason behind this suggestion is that the penetrance and expressivity of the AD form of HPE vary widely [11].

Winter., *et al*. (1988) [12] and Buntinx and Baraitser (1989) [13] described a SMMCI in patients with a form of ectodermal dysplasia (ED). Although this is an unusual manifestation of ED, it should be considered as significant in possible gene carriers. In the family presented there, the inheritance pattern is likely to be autosomal recessive (AR) [12,13]. Artman and Boyden (1990) [14] described a 5 years 1 month old girl with 'slow growth', periodic severe headaches, intermittent nasal congestion, and near complete blindness secondary to microphthalmia. Her height and weight were both well below 3 SD and at the 50th centile for an 18 month and 9 months respectively. Her bone age was of 2 years 5 months and CT scan showed a small sella turcica with a hypoplastic pituitary gland. A small bony ridge was noted in the roof of the hard palate. The growth hormone was below that of normal controls. The girl has been defined as having isolated growth hormone deficiency, microphthalmia and isolated growth hormone deficiency in association with SMMCI [14].

The association of SMMCI with nasal obstruction was recognized [15]. Congenital nasal pyriform aperture stenosis (CNPAS) was described by Brown in 1989 [16] as a cause of nasal airway obstruction in the newborn, but there was no mention of SMMCI [16].

SMMCI is a non-specific sign, it may occur as an isolated abnormality or as microform of HPE with autosomal dominant transmission [15]. Those patients noted to suffer from congenital nasal pyriform aperture stenosis (CNPAS) must be observed for eruption of a SMMCI, which has been described as "megaincisor" together with maxillary bony overgrowth. These patients must be assumed to have a microform of HPE, which necessitates chromosomal analysis, evaluation of relatives, counseling of parents, CT scan to check for CNS malformations, and assessment of the hypothalamic-pituitary-thyroid-adrenal axis. Indeed, CNPAS may suggest a midfacial dysostosis with associated endocrine and central nervous system abnormalities, more than an isolated congenital abnormality of the airway.

SMMCI may occur as an isolated finding or in association with other systemic abnormalities like short stature, pituitary insuf-

ficiency, microcephaly, choanal atresia, midnasal stenosis, and CNPAS. It can also be a feature of recognized syndromes with specific chromosomal abnormalities or with no chromosomal abnormality [17].

Masuno., *et al*. (1990) [18] described 2 unrelated cases of SMMCI with 7q terminal deletion. They had mental retardation, microcephaly, hypotelorism, short stature, and normal levels of plasma growth hormone. One patient had bilateral caudal ectopic kidneys, double renal pelvises, and dilated ureters. The other had bilateral hydroureteronephrosis. The authors suggested that 7q terminal deletion is one of the causes of SMMCI [18].

Nanni., *et al*. (2001) [17] performed a molecular study of 13 patients with SMMCI who did not have HPE. They studied 2 genes, SHH (600725) and SIX3 (603714), in which mutations had been reported in patients showing SMMCI as part of the HPE spectrum. They found a new missense mutation in SHH (I111F; 600725.0014) which may be specific for the SMMCI phenotype since it had not been found in patients with HPE or in normal controls [17].

Marini., *et al*. (2003) [19] studied a family, previously described by Camera., *et al*. (1992) [20] in which the mother presented with a SMMCI and mild hypotelorism and her daughter and 2 fetuses were diagnosed with HPE. Sequencing of DNA in this family identified a nonsense mutation in the SHH gene [19,20]. In this paper we present cases, otherwise healthy children with this phenomenon and one suspected with systemic aberrations.

About 30 years ago (1991), one of us (EM) came across a boy with SMMCI phenomenon, which proved to be unfamiliar to most peer colleagues across the country (Figure 1) [21]. By now, this boy, now a father, has 2 otherwise healthy children. Like his mother 30 years ago, he refuses systemic or genetic examinations of the family. Other two cases (Figure 2 and 3) were provided then by general practitioners.

Only one additional child, with obvious features, suggesting a syndrome (case 5-EM), has been examined for dental treatment by one author (ME), but the mother had refused further comprehensive examinations, which prevented detailed information. Hence, the purpose of this review and presentation of further small case series is to draw attention to the rarity of this phenomenon, sometimes referred to as a syndrome, or a part of a genetic feature with, or without variable associated anomalies.

Case Series

First three cases (Figure 1-3) were presented in detail in Mass and Sarnat 1991. In the first two cases splitting of the pulp canal close to the apex (type IV according to Weine's classification) can be observed (Figure 1 and 2). In case no 3 no apical splitting was observed (Figure 3) [21].

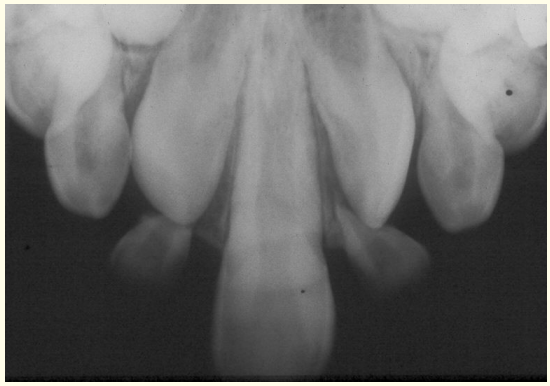

Figure 1: *TA - Anterior periapical radiograph of early mixed dentition [21]; Note splitting of the pulp canal close to the apex (type IV according to Weine's classification) [22].*

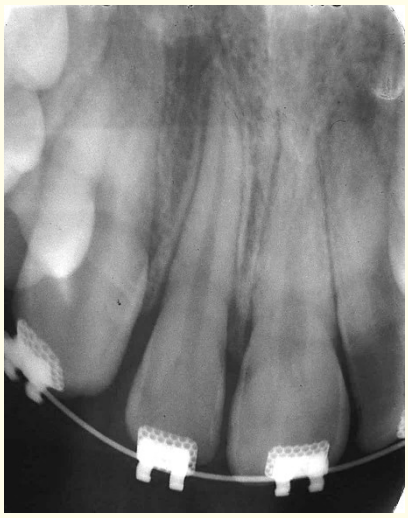

Figure 2: *RM - Anterior periapical radiograph of permanent dentition during orthodontic treatment [21]. Note splitting of the pulp canal close to the apex (type IV according to Weine's classification) [22].*

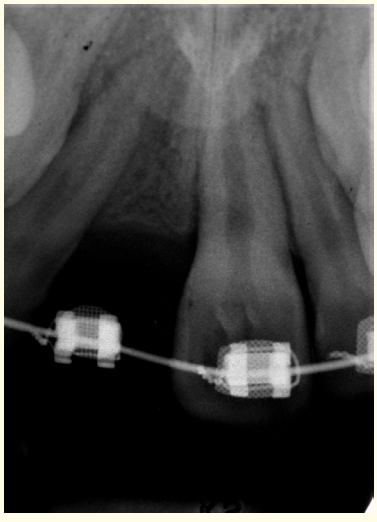

Figure 3: *KM- Anterior periapical radiograph of permanent dentition during orthodontic treatment [21]. No splitting of pulp canal at the apex can be observed.*

Case no 4

KS: A 7 years old boy was referred by a general practitioner for consultation, due to "Delayed dental development and a peculiar phenomenon in the maxillary incisal area". Other than the characteristic appearance in the primary and the following permanent SMMCI and exaggerated midline palatal torus in a maxillary cohesion, medical and dental examinations were inconclusive (Figure 4). Only a radiograph of the deciduous dentition was available.

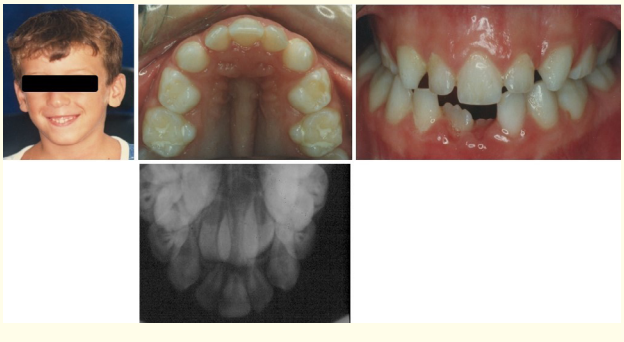

Figure 4: *KS- Facial and dental clinical pictures and upper anterior periapical radiograph of early mixed dentition. Note the exaggerated midline palatal torus, lack of anterior labial frenum and incisive papilla.*

Case no 5

EM: A 5 years old girl was referred for dental examination and treatment. The examination revealed mid nasal bulge; mild hypotelorism (in contrast, her brother had mild hypertelorism); atypical soft palate and lack of uvula; lack of anterior frenum; prominent mid palatal torus, without incisive papilla (Figure 5). Only a radiograph of the deciduous dentition was available.

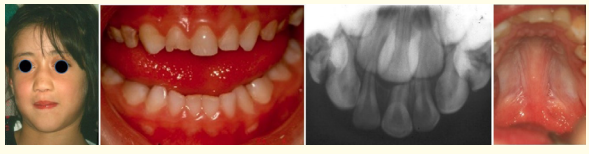

Figure 5: *EM– Facial and dental clinical pictures and upper anterior periapical radiograph of deciduous dentition. Note the exaggerated midline palatal torus, lack of anterior labial frenum, incisive papilla, posterior uvula with flabby soft palate.*

Upon suggestion to perform further systematic examination, the mother refused, leaving the clinic, claiming that she brought the child for "fillings".

Discussion and Conclusion

SMMCI and SMMCI syndrome is a rare phenomenon presented in the scientific literature sporadically. Due to its rarity, it was felt that any additional reports may contribute to contemporary awareness of the importance of comprehensive case examinations.

This article describes a case series of SMMCI with two rare features. The two new features, which were not mentioned in previous reports, are tendency of the pulp canal close to the apex to split (type IV according to Weine's classification) and the lack of visible incisive papilla. Of these cases, none went through genetic and/or systemic examination, due to unawareness of its genetic and systemic implications by general practitioners, or due to refusal of the parents for further comprehensive genetic examinations. Indeed, SMMCI it is now recognized as a possible predictor of holoprosencephalies of varying degrees in members of the proband's family, and in the family's descendants. Hence, it is hoped that this literature review and the presented cases will help to clarify understanding of solitary median maxillary central incisor as a sporadic entity or as a syndrome, and prompt pediatric dentists to conduct thorough tests upon its detection.

Bullet Points:

1. Solitary median maxillary central incisor (SMMCI) as an isolated occurrence and SMMCI as part of a syndrome are rare dental anomalies.
2. In SMMCI there is a tendency permanent incisor there is a tendency of the root canal to split close to the apex.
3. A lack of incisive papilla is an additional clinical finding that should be taken into consideration.

Authors Contributions

E.M. and U.Z. conceived the ideas, E.M. and A.O. collected the data, E.M. and U.Z. analysed the data, E.M, A.O, and U.Z led the writing.

Conflict of Interest

All authors have no conflict of interest.

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