Case Report

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Dentistry Section

Non-Syndromic Occurrence of Multiple Dental and Skeletal Anomalies: A Rare Case Report and Brief Literature Review

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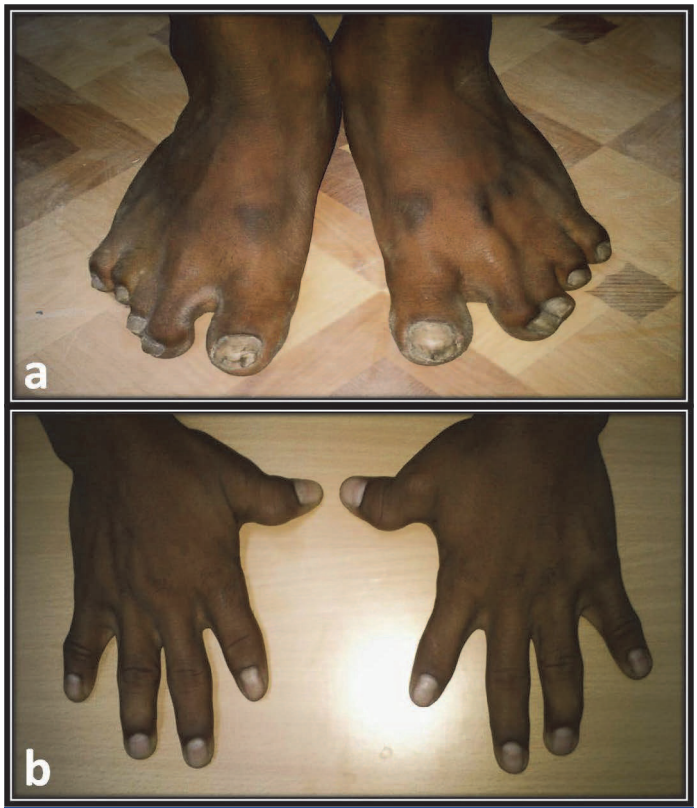
ABSTRACT

Abstract: Oral physician has always been challenged when it comes to diagnosing rare nonsyndromic cases because of the varied presentation of multiple dental abnormalities caused due to mutations in developmental regulatory genes. This coupled with skeletal abnormalities makes the task more difficult. But as we come across such rare constellation of findings, it makes the field more intriguing. Here, we report an extremely rare case of non syndromic occurrence of dental manifestations like multiple dens invaginatus, generalised microdontia, generalised hypoplasia, hypodontia, pulp stones and widening of pulp chamber along with skeletal findings of bilateral syndactyly of legs and brachydactyly of hands and legs. Although many non syndromic cases have been reported in the literature, the unusual occurrence of findings in the present case is being reported for the first time.

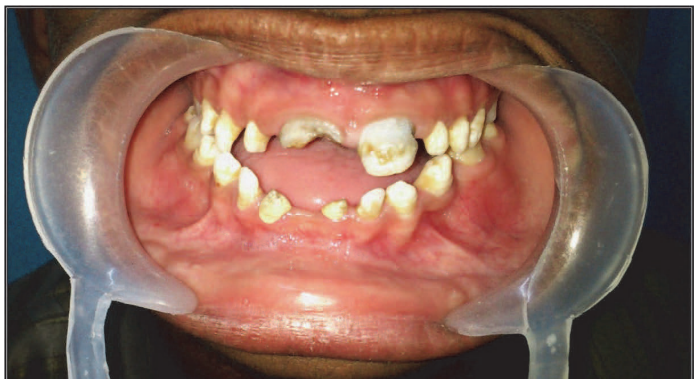
Keywords: Dens invaginatus, Microdontia, Non-syndromic, Syndactyly

CASE REPORT

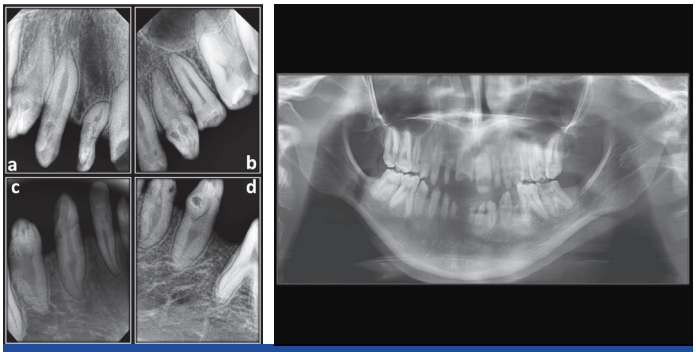
An 18-year-old male patient reported with the complaint of pain in maxillary anterior teeth. Pain was localised and dull aching. There was no significant medical history and family history was also non contributory. General physical examination of the patient revealed short fingers of hand and legs (brachydactyly) and bilateral syndactyly in fingers of leg [Table/Fig-1]. Intraoral examination revealed deeply carious 11, moderately carious 21 and 47, microdontia in relation to 12, 13, 22, 23, 31, 32, 33, 41, 42, 43, conical shaped lateral incisors, and generalised spacing. Generalised hypoplasia with yellowish discoloration and diffuse pitting was noted [Table/Fig-2]. Radiographic evaluation showed congenitally missing 41, dens invaginatus in 12, 13, 14, 23, 24, 25, 34, 35, 42, 43, 44, 45, widened pulp chambers, pulp stone in 34, complete agenesis of the maxillary and mandibular third molars [Table/Fig-3]. Periapical involvement with 11 and 21 was present. Orthopantomograph showed decreased height of the body of the mandible [Table/Fig-4]. PA Skull radiograph revealed no abnormality. Hand wrist radiograph of patient was suggestive of skeletal age more than 11yrs as all carpal bones were visualised. In index and little finger only two phalanges were seen bilaterally thus accounting for shortness of fingers [Table/Fig-5]. X-ray foot showed only soft tissue fusion between 2nd and 3rd toe. Distal and middle phalanges were not present in all toes bilaterally [Table/Fig-6]. Complete blood examination and hormonal assay were carried out to rule out any associated syndrome but no significant changes were observed. A multidisciplinary approach was planned to treat this case. Extraction of grossly carious 11 was carried out and endodontic treatment of 21 and all the teeth affected by dens invaginatus was done in follow-up visits. Along with this oral prophylaxis was carried out. Finally prosthodontic rehabilitation was carried out to enhance occlusion, mastication and aesthetics.



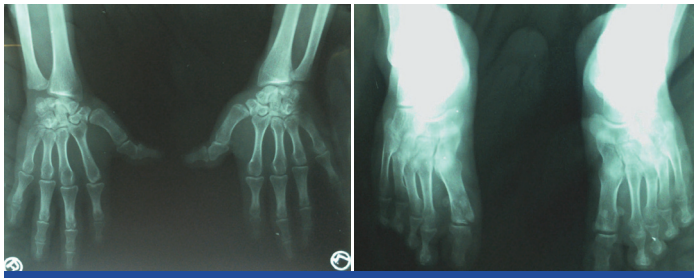
[Table/Fig-1]: Clinical photograph showing brachydactyly of fingers of hands and toes of legs, also seen syndactyly of 2nd and 3rd toe bilaterally



[Table/Fig-2]: Intraoral photograph showing microdontia, carious teeth and generalized hypoplasia of dentition



[Table/Fig-3]: Intraoral periapical radiographs showing multiple dens invaginatus in maxillary and mandibular teeth
[Table/Fig-4]: OPG showing decreased height of body of mandible



[Table/Fig-5]: Hand wrist radiograph showing missing phalanges in index and little finger
[Table/Fig-6]: X ray foot of patient with missing distal and middle phalanges in all toes and only soft tissue fusion between 2nd and 3rd toe bilaterally

DISCUSSION

Genetic disturbances or environmental factors during tooth morphogenesis may lead to odontogenic anomalies. Combination of multiple dental anomalies is usually associated with specific syndromes. Cases with non-syndromic occurrence of multiple dental anomalies have been reported in the literature [1,2]. Syndactyly of feet also known as webbed toes usually involves skin connection between the two areas, but involvement of the underlying bones may be noted. It is often inherited and is not unusual. It is commonly associated with Down syndrome and oculodentodigital syndrome. Rarely, it is seen as a component of Apert syndrome, Carpenter syndrome, Cornelia de Lange syndrome, Pfeiffer syndrome, Smith-Lemli-Opitz syndrome, and use of the medication hydantoin during pregnancy (fetal hydantoin effect) [3].

Dens invaginatus also known as dens in dente and dilated compound odontome is a malformation of teeth which occurs due to an infolding of the dental papilla during tooth development. Maxillary lateral incisors are most commonly affected teeth and bilateral occurrence is not uncommon. This condition is known to be associated with Ekman-Westborg-Julin syndrome, Williams syndrome and Nance Huran syndrome [4].

Variations in size of a tooth arise when the form of the tooth is being determined by the enamel organ and the sheath of hertwig at the bell stage of enamel organ. The term microdontia is defined as having abnormally small teeth. It is classified into 1) Microdontia involving only a single tooth, 2) Relative generalized microdontia due to relatively small teeth in large jaws and 3) True generalized microdontia, in which all the teeth are smaller than normal. True generalized type occurs in some cases of pituitary dwarfism otherwise its occurrence is very rare. Some syndromes associated with microdontia are Gorlin-Chaudhry-Moss syndrome, Williams's syndrome, Ullrich-Turner syndrome, trisomy 13, Rothmund-Thomson syndrome, Hallermann-Streiff, Orofaciodigital syndrome, Oculo-mandibulo-facial syndrome, Tricho-Rhino-Phalangeal, type1 Branchiooculo- facial syndrome [5].

To the best of our knowledge, this is the first non-syndromic case with the occurrence of multiple dental anomalies like multiple dens invaginatus, generalised enamel hypoplasia, microdontia, hypodontia, pulpstones, pulp widening along with syndactyly of toes and short fingers. The aetiology behind such presentation is probably related to genetic mutations. Various systemic conditions were included in the differential diagnosis like hypoparathyroidism, pseudohypoparathyroidism, vitamin D resistant rickets, hypophosphatasia, oculodentoosseus dysplasia, dystrophic epidermolysis bullosa, trichodentoosseous syndrome, tuberous sclerosis, dwarfism, Russell silver syndrome, Seckel syndrome, William's syndrome and amelogenesis imperfecta.[6]. Oculodentoosseous syndrome is characterized by syndactyly and clinodactyly of fourth and fifth fingers also has dental manifestations like generalized enamel hypoplaisa, thin dentinal walls, extensive caries leading to early pulp involvement and open apices. As the patient did not show any other abnormal manifestations like CNS involvement, short roots, taurodontia, etc., all the syndromes associated with the dental anomalies were ruled out.

Sedano et al., reported a case of 15-year-old female patient with five dens invaginatus along with permanent mandibular left mulberry molar, molarization of some premolars, several microdontic conoid teeth, retention of five primary teeth, absence of several permanent teeth germs, a macrodontic molar with abnormal roots and several periapical radiolucencies associated with dens invaginatus [4]. But the authors did not find any skeletal abnormalities as reported in the present case.

Dens invaginatus is a developmental anomaly and usually detected on routine radiographs. The invagination is a pathway for micro-organisms and irritants. Constant irritation results in pulp necrosis and periapical abscess. This can be prevented by sealing the invagination with restorative material. But if periapical area is involved then treatment of choice is root canal treatment as was done in the present case.

An unusual combination of idiopathic generalised short root anomaly associated with microdontia, taurodontia, multiple dens invaginatus, obliterated pulp chambers and infected cyst has also been reported by Desai et al., [7]. Suprabha et al., reported a case with multiple dens invaginatus, shovel shaped incisors, generalised enamel hypoplasia, hypodontia, generalised microdontia, cup shaped premolars, root resorption and multiple periapical lesions, and supernumerary teeth [8]. Casamassimo et al., reported a case of a 12 and a half year-old boy who presented with microdontia, taurodontia of mandibular molar teeth and dens invaginatus in multiple teeth [9]. Takeda et al., reported a case of bilateral multiple dens invaginatus involving the maxillary central incisors, mandibular canines, and first premolars in an 11-year-old boy who also had steeple-head and mild mental retardation [10].

Our case differs from the above cases as there was an additional finding of skeletal abnormalities i.e. bilateral syndactyly in legs and short fingers of hand and legs along with dental abnormalities. Patients consent was taken regarding all the investigations and procedures to be carried out. Syndromes/conditions that constitute some of the manifestations but not all of the present case are discussed in [Table/Fig-7].

CONCLUSION

The combination of dental anomalies, as seen in this case, probably indicates a common unknown genetic factor in the aetiology giving rise to different phenotypic manifestations. The constellation of dental findings seen in this case is certainly rare and differs from previously reported cases. The case is also sporadic, with no positive family history. For the clinician, the difficulty lies in diagnosis, as a number of syndromes and pathologies need to be ruled out, due to large constellation of dental findings. Such a case needs to be diagnosed early and requires multidisciplinary treatment approach.

Syndromes/ conditions	Skeletal features	Orofacial and Dental features
Taurodontism, microdontia and dens invaginatus [5]		Generalized microdontia, taurodontism of first permanent molars, multiple dens invaginatus
Oculo-dento-digital dysplasia [11]	Bilateral micropthalmos, abnormally small nose, hypotrichosis, 5th finger camptodactyly, syndactyly of fourth and fifth fingers, missing toe phalanges	Enamel hypoplasia, thin dentinal walls, extensive caries, open apices
Symphalangism kantaputra type [5]	Distal symphalangism, hypoplastic carpal bones, brachydactyly, syndactyly of fingers and toes, cone shaped epiphyses, absent/small nails	Microdontia, dental pulp stones, narrowed zygomatic arch
Cranio ectodermal dysplasia [12]	Dolichocephaly, sparse slow growing fine hair, epicanthal folds, brachydactyly, narrow thorax	Hypodontia and/or microdontia
Trichodento-osseous syndrome [13]	Kinky curly hair, increased cranial thickness obliterated diploe and no visible mastoid pneumatization, thin brittle nails, dolichocephaly	Enamel hypoplasia, taurodontism
Russell silver syndrome [14]	Low birth weight, asymmetric growth of body, Clinodactyly, precocious puberty, short stature, Wide forehead with a small triangle-shaped face and small, narrow chin, café au lait birth marks, short arms fingers and toes	Mandibular micrognathia, deep bite, dental crowding, microdontia, high arched palate

[Table/Fig-7]: Comparison with syndromes and conditions constituting some (but not all) manifestations of present case

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