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An unusual occurrence of Karsch–Neugebauer syndrome with orodental anomalies

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Abstract

Karsch–Neugebauer syndrome is a rare congenital anomaly characterized by split foot, split hand anomalies in association with congenital nystagmus. It is an inherited condition and often occurs in both the hands and the feet. Its pattern of inheritance is autosomal dominant, though occasionally it can skip a generation. It affects about 1 in 90,000 babies, with males and females equally being affected. This rare syndrome may be associated with additional abnormalities such as hypodontia, delayed eruption, variation in morphology and position of teeth, but it is not widely documented. In view of the low incidence of this syndrome and of the limited number of reports on this in the literature, the objective of this article is to highlight the oral findings of this syndrome.

KEY WORDS: Split hand deformities, ectrodactylia of hands, lobster claw malformation, karsch neugebauer syndrome

Karsch–Neugebauer syndrome is a congenital anomaly of the hand where the middle digit is missing and the hand has a cleft where the metacarpal of the finger should be. This syndrome occurs in association with congenital nystagmus and is named after J. Karsch and H. Neugebauer. [1,2] The split in the hands, commonly known as ectrodactyly, gives the appearance of lobster claw. Ectrodactyly is an autosomal dominant disorder.[1] It may also be known as “lobster claw hand,” “split hand deformity,” “split hand/foot malformation (SHFM),” “cleft hand,” and “ectrodactylia of the hand.”

Oral findings of this syndrome include hypodontia, conical or peg-shaped teeth, anodontia, retained deciduous teeth with facial manifestations like frontal bossing, sunken cheeks, thick everted lips, thinning of eye brow, and congenital nystagmus with hypertelorism.

This case report is the first of its kind in the dental literature.

Case Report

A 38-year-old male patient reported with a complaint of carious tooth. On physical examination, the patient was found to be healthy and of normal height and weight. Examination of the upper limb [Figure 1] revealed abnormalities in the form of missing 2nd digit and clefting of the hand on both sides since birth. Similarly, in the lower limb [Figure 2], abnormalities in the form of fusion of 2nd and 3rd toes on the right foot with absence of clefting since birth were seen. The left foot was normal. There was no family history of similar deformity. History of consanguinity of parents, drug intake, and exposure to radiation was ruled out. No one in the family, i.e., elder sister and younger brother, was affected. On extraoral examination, congenital nystagmus, thick everted lips, and hypertelorism were evident [Figure 3]. On intraoral examination, many permanent teeth were found to be missing [Figures 4 and 5] and history of delayed eruption was given.



Figure 1

Photograph showing upper limb of the patient



Figure 2

Photograph showing lower limb of the patient



Figure 3

Extraoral examination showing congenital nystagmus, thick everted lips, and hypertelorism

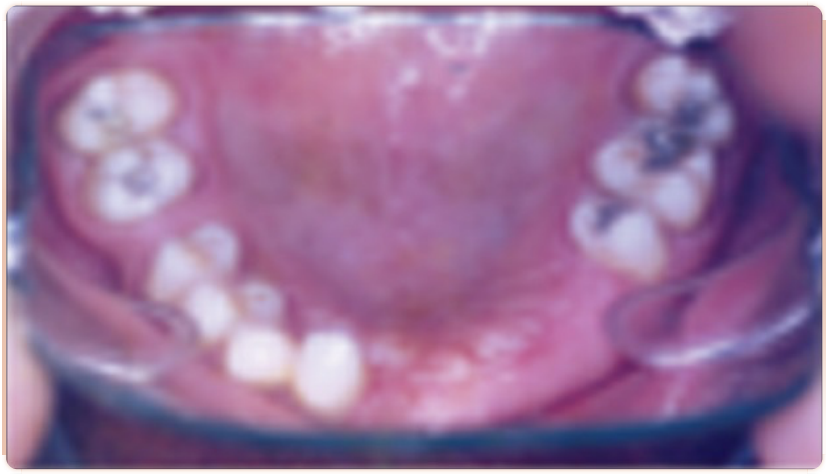


Figure 4

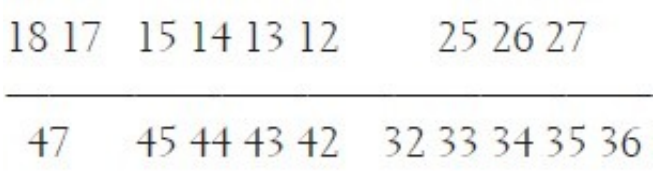
Maxillary occlusal view



Figure 5

Mandibular occlusal view

Number of teeth clinically present: 19



Rotated permanent both lower and upper incisors were seen along with deep bite. Restoration was done in 17, 18, 25, 26, 27, 36, and 47. Variation in tooth size and its position was highly appreciated. Examination of nails and mucosa did not reveal any abnormality. A skin biopsy showed normal sweat glands.

Discussion

Karsch–Neugebauer syndrome is a congenital anomaly characterized by the absence of part or all of one or more fingers or toes.[2] Although Karsch–Neugebauer cases occur sporadically, familial forms have been observed with the majority showing autosomal dominant inheritance,[1] while some autosomal recessive patterns of inheritance have been reported. Chromosomal or physiological defects during the formative stage of limbs and orodental structures are responsible for this rarely occurring syndrome.[3]

This rare syndrome has many forms and one of the commonest forms is associated with a specific region of human chromosome 7 that contains two homeobox genes, *DLX5* and *DLX6*.[4] These genes are similar to a gene in insects called distal-less that controls limb development. When this gene is defective in the fruit fly, the distal part of the insect limb is missing. Ectrodactyly may be present alone or may be part of a number of birth defects. Hand deformation alone is unlikely to affect health.

SHFM is a congenital malformation of the hands and feet, which involves fusion of digits (called syndactyly), missing or underdeveloped bones, and a median cleft of the hand or foot.[3] When the hand or foot is split, it gives the appearance of lobster claws.[5] When there is no splitting (either a single finger or a single toe is present), it is referred to as monodactyly. Gradations between these types occur and cases of each type sometimes are found in the same family. SHFM results from failure of formation of parts of hands, feet, or both due to a variable deficiency of central rays of the autopad. Previously known as split hand/foot or ectrodactyly, the most acceptable term nowadays is “congenital central limb ray deficiency.” Usual clinical manifestations include presence of congenital syndactyly, median clefts of hands and feet, along with hypoplasia or aplasia of carpal, tarsal, metacarpal, metatarsal, and phalangeal bones. In milder forms, there is partial or incomplete deficiency leading to the absence of middle fingers or toes. One of the examples of the milder forms is the Karsch–Neugebauer syndrome. While severe forms may leave a two-digit or even a one-digit hand, feet are less commonly involved and bilateral involvement of hands and feet is often associated with positive family history.[6]

Ectrodactyly is clinically heterogeneous, presenting in both an isolated form (non-syndromic ectrodactyly) and associated with other organ systems including the cranio-facial, genitourinary, and ectodermal structures (syndromic ectrodactyly).[7] One of the examples of syndromic ectrodactyly is the Karsch–Neugebauer syndrome (ectrodactyly, congenital nystagmus, and hypodontia). Ectrodactyly may also be associated with focal dermal hypoplasia. The syndromic form has a variable degree of expression.[8] The other syndromes in which ectrodactyly is a prominent feature are EEC (ectrodactyly, ectodermal dysplasia, clefting),[9] multiple synostoses syndrome,[10] amniotic band syndrome,[10] cleft hand and absent tibia syndrome,[10] Adams–Oliver syndrome,[11] and Lobster-claw syndrome.[12]

The apical ectodermal ridge is a critical signaling center that directs the outgrowth and patterning of the developing limb. The fundamental developmental defect in ectrodactyly is the failure of the apical ectodermal ridge to maintain this activity.

The diagnosis of ectrodactyly syndrome can be complex because of the overlap of symptoms with other ectodermal dysplasia syndromes. Transvaginal ultrasonography performed at 14 weeks gestation demonstrates deformities of hands and feet and can help us with the diagnosis of this syndrome. Currently, there are several treatments which can normalize the appearance of the hands, yet they will not function precisely the same way as regularly formed hands. The prognosis for most individuals with ectrodactyly syndrome is very good. Some people with ectrodactyly use prosthetic hands to avoid the rude stares of others. Ectrodactyly is an inherited circumstance which can be treated surgically to better role and show. Early physical and occupational therapy can help those with ectrodactyly adapt, and learn to write, pick up things, and be fully functional. Genetic findings could have great implications in clinical diagnosis and treatment of not only ectrodactyly, but also many other related syndromes.[13]

Conclusion

From this case report, we learn that orodental anomalies like hypodontia, missing permanent teeth, and variation in tooth size and its position may also be associated with Karsch–Neugebauer syndrome. Early diagnosis is important for proper and better management of orodental problems since these are not as easily diagnosed signs of this syndrome as compared to other signs such as hand and foot defects.

Footnotes

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Conflict of Interest: None declared.

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