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Rare case report "Cooks syndrome, Mammary Digit Nail syndrome(MDN)"

KEYWORDS: Mammary Digit Nail syndrome2, Cooks syndrome1, Anonychia, Juvenile hypertrophy of breasts.

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ABSTRACT *We describe a case with congenital absence of all nails and distal phalanges of all fingers and toes. This syndrome was first describe by cooks et al in 1985, in addition to this she also had juvenile hypertrophy of breast which describes asMammary Digit Nail syndrome2(MDN) by Mira Genzer et al. This is to be first time reported from India as per our knowledge.*

Introduction:
Congenital diseases are very rare and amongst them very few cases from all over the Globe has been reported so far. Of such congenital diseases, here we would like to report one of the congenital disorder which fits into Cooks syndrome[1], Mammary Digit Nail syndrome[2] as a first case of its kind from India. Such cases also reported earlier from the other countries.

Case history
Case report:
45 year old female came for consultation for her mother's diabetic status, With no pertaining any complaint to her. We accidently found that she didn't have all nails and distal phalanges. On further evaluation and examination, she was born as a second child of her parents of non consanguineous marriage with normal full term delivery without any complication.

However since birth all nails and distal phalanges of fingers and toes were absent. She achieved all her social and motor milestones at normal age without any mental regression and retardation completed higher secondary education.

At the age of 13 years before menarche, she developed rapid enlargement of both breasts in span of 4 months upto umbilicus for that she was undergone breast reduction surgery and approximately 7-10 kgs tissue removed from left breast and 8-10 kgs from right breast suggestive of juvenile hypertrophy of breast, at the age of 15 years she attained menarche. She was unmarried because of social stigma associated with condition. Her menstruation cycle was even normal up to the age of 35 years. When she was diagnosed to have fibroid uterus, for which she underwent hysterectomy.

On examination her weight and height was normal, no any other malformation except the above findings. No associated skin changes were present.

(1) Image 1

(2) Image 2

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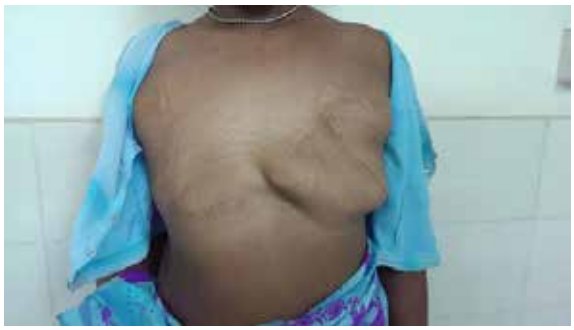
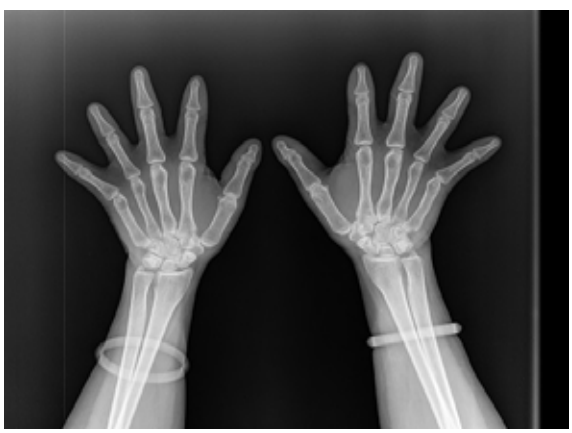
(3) Image 3

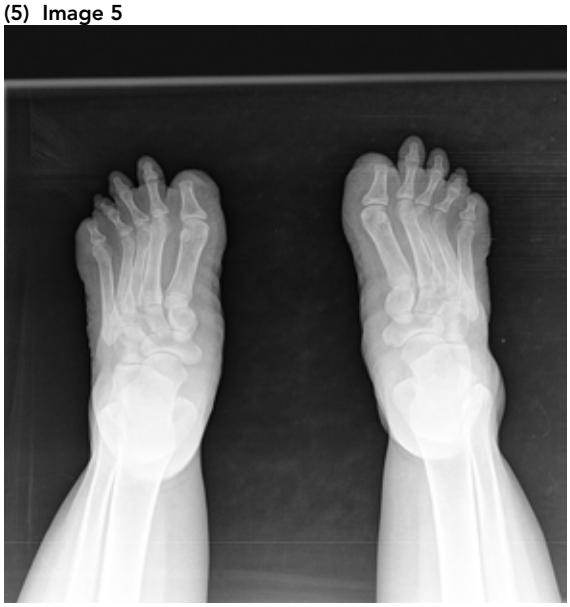


Radiological findings:
X-Rays of both hands and feet showed absence of distal phalanges in most of the digits.no other abnormalities were found on the xrays. Chest xray findings were within normal limits.

(4) Image 4



(5) Image 5



Haematological findings:
All haematological findings were within normal limits, however S. TSH level was falling into range of subclinical hypothyroidism however there were no clinical signs of hypothyroidism.

Discussion :
Congenital anonychia/ onchodystrophy with absent distal phalanges and juvenile hyperthrophy of brest appears to be similar with MDN syndrome.[2] reported by Mira et al and cooks reported by cooks et al.[1] the main positive findings were JHB. Various other syndromes like the ulnar mammary syndrome, the limb mammary syndrome and the holtolam syndrome are genetic syndromes affecting both mammary glands and limbs, however anonychia, associated with JHB has been documented only as MDN syndrome.[2]

Previous reported cases were either autosomal recessive[4] and autosomal dominant[5] mode of Inheritance, however we were not able to trace the case further because of unmarried status. But she didn't have any past family history with such findings.

This case has no other skin changes or any mental retardation or any hearing impairment as described by "Levick et al[3]"in their case.

Anonychia may be present with additional anomalies like absence of patella, (cless et al, 1957) lymphoedema (maisels, 1966). However in our case report it was only associated with JHB.

Anonychia and associated anomalies (cases were previously reported from Russia , Germany, Holland and Iraq and now first case with such findings to be reported from India.

Conclusion:
Rare condition associated with congenital anonychia with juvenile hypertrophy of breast with normal mental progression is first time to be reported from india which fits into Mammary Digit Nail syndrome[2](MDN syndrome) and cooks syndrome[1].

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