CONGENITAL ANONYCHIA TOTALIS: A RARE OCCURRENCE IN A PAKISTANI FAMILY

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INTRODUCTION

Complete anonychia means absence of all nails of fingers and toes. It may be congenital or acquired. Anonychia is commonly associated with hypoplastic or completely absent underlying phalanges; however, a rare form exists which is without any associated bony defect. Simple anonychia may be inherited in an autosomal recessive pattern or may result from a new mutation. We present here a middle-aged woman having complete absence of all nails, abnormal teeth, dry skin and left ectropion. She was educated about the disease and managed for the backache on rehabilitation guidelines. Gene mapping of her whole family was planned, but she was lost to follow up. This case provides evidence of the presence of this condition in Pakistani population. There is a need for further investigation and pooling of cases to build clinical data about phenotypes and the inheritance patterns of this disorder in our population.

ABSTRACT

Anonychia totalis is a rare disease characterized by complete absence of finger and toe nails. Possible associations include anomalies of bones, teeth, skull, hair, skin and sweat glands besides hearing and intelligence impairments. We report here a 60-year-old woman with low back pain and tenderness at the lumbosacral spine who was found to have complete absence of all nails, abnormal teeth, dry skin and left ectropion. She was educated about the disease and managed for the backache on rehabilitation guidelines. Gene mapping of her whole family was planned, but she was lost to follow up. This case provides evidence of the presence of this condition in Pakistani population. There is a need for further investigation and pooling of cases to build clinical data about phenotypes and the inheritance patterns of this disorder in our population.

Key Words: Anonychia, Anonychia totalis, Complete anonychia, Congenital

CASE PRESENTATION

A 60-year-old woman presented in the rehabilitation department of our hospital for complaint of lower backache of six years’ duration. There was no history of associated trauma, lifting heavy weights, fever, weight loss, disturbed sleep or appetite and urinary or bowel complaints. She had an absence of all finger and toe nails since birth. She suffered from occasional spontaneous blistering that healed with minimal scarring and hypopigmentation. Purpura and easy bruisability were also reported. Her hair growth was normal and she did not complain of reduced sweating, hearing loss or hyperkeratosis of palms and soles. Her unaffected parents were consanguinely married. She had one brother with the same condition out of a total of six siblings (three males and three females). She had two daughters and one son who were all unaffected. Her affected brother had two sons and one daughter with anonychia while two other daughters were phenotypically normal. The issues of rest of her siblings were unaffected.

Her examination revealed complete absence of nails as well as nail beds in all fingers and toes (figure 1). An ecchymotic patch was present on the left second toe, and a similar small lesion was present on the left big toe. Well margined, irregularly bordered depigment-
ed macules and patches were seen on distal part of fingers that were negative for vitiligo on Wood’s lamp examination. She had dry skin all over the body but signs of ichthyosis were absent. There were a few blisters on the left second and fifth toe with sclerosis of skin and partial syndactyly of second and third toes (figure 2).

Her musculoskeletal system was unremarkable except for mild tenderness over the spinous processes of lower lumbar spine. She had malformed teeth especially the upper left incisor with immature formation of enamel and generalized dental attrition along with stomatitis due to sloughing of oral epithelium (figure 2). Examination of the eyes revealed left sided ectropion and bilateral arcus senilis (figure 2). X-ray of the lumbosacral spine revealed reduced intervertebral disc space at LV4/ LV5 and LV5/SV1 levels and grade-I spondylolisthesis at LV5/SV1 level with defect in pars interarticularis of LV5 (figure 3). X-rays of both hands showed normally outlined phalanges with periarticular osteopenia (figure 3).

Keeping in view her history and physical examination, she was diagnosed to have simple congenital anonychia totalis with secondary complications. Due to lack of facilities and financial assistance, chromosomal studies and gene mapping could not be undertaken.

### DISCUSSION

The development of a normal nail requires the underlying bone to be normoplastic. An abnormality occurs when the bone is either absent or not normally developed. These abnormalities range from slight change to complete absence of finger nails and toe nails (complete anonychia). This may appear as an isolated abnormality or as a feature of genetic syndromes. Complete anonychia generally follows an autosomal recessive pattern and has been mapped to chromosome 20p135. Recently, mutations in the RSPO4 gene including a novel missense mutation c.178C>T (p.R60W) were identified in three Pakistani families presenting with combinations of anonychia and hyponychia.

Complete anonychia may occur in association with a wide variety of other congenital abnormalities, such as abnormally spaced teeth, absence of third molars and upper lateral incisors, absence of patella, microcephaly, curved digits and single transverse palmar crease, bizarre flexural pigmentation and hair abnormalities, and mental retardation with sensorineural deafness.

The features found in our patient closely resemble the cases described by Freire-Maia and Pinheiro. They  

Figure 1: Figure showing complete absence of nails as well as nail beds in all fingers and toes
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described five individuals (two females and three males); all inbred; with segregation ratio of approximately \( \frac{1}{4} \) among eighteen siblings who had normal parents. This strongly suggested an autosomal recessive mode of inheritance. All four living patients with anonychia also had hypoplastic upper lateral incisors while three of them also had agenesis of (one or more) third molars and widely spaced teeth to different degrees. X-rays of hands and feet showed no bone malformations. Our patient had abnormally spaced teeth and missing teeth especially the upper left incisor. She had normal hand and foot X-rays. She also had partial syndactyly of second and third toes, left sided ectropion, reduced intervertebral disc spaces and a spondylolisthesis at lumbo-sacral level. The parents were first-cousins and two out of six siblings were affected in her generation whereas none of her own offsprings were affected. Although three out of five children of her affected brother had anonychia, we unfortunately could not follow his wife’s lineage and consanguinity.

CONCLUSION

Our case provides some insight into the presence of this condition in the Pakistani population and emphasizes the need for further investigation and accumulation of data about the phenotypic patterns and mode of inheritance of this disorder in our population.

REFERENCES

Figure 2: Figure showing (a & b) Reduced intervertebral disc space at LV4/LV5 and LV5/SV1 levels, grade-I spondylolisthesis at LV5/SV1 with defect in pars-interarticularis of LV5 (c) Periarticular osteopenia in hand phalanges


