GENETICS AND GENODERMATOSES

PACHYONYCHIA CONGENITA : A CLINICAL AND GENETIC EVALUATION

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Background: Pachyonychia Congenita(PC) is a rare genetic keratinizing disorder characterized by thickened nailbeds of all fingers and toes, palmoplantar keratoderma, calluses with underlying blisters and sometimes accompanied by oral leukokeratosis, cysts, follicular hyperkeratosis, palmoplantar hyperhidrosis and natal teeth.
PC is caused by a mutation in one of five keratin genes: KRT6A, KRT6B, KRT6C, KRT16 or KRT17 and is divided into 5 types based on the specific keratin gene involved.

Observation: A young boy of 18 years,born to non consanguineous parents presented with a history of repeated blistering occurring intermittently over the soles of the feet since birth. On examination,we observed thickened discolored finger and toe nails ,and painful calluses with fissures on the soles of the feet.On further evaluation, natal teeth were seen along with hyperhidrosis of the palms and soles , white patches over the tongue,follicular keratosis over elbows,knees and waistline and a cyst over the arm pit.Hence,a clinical diagnosis of PC was considered and in view of this,a genetic opinion was sought.A mutation analysis was done where a heterozygous missense variation in exon 1 of the KRT17 gene that results in the amino acid substitution of Serine for Asparagine at codon 92 (p.N92S) was detected.
Pachyonychia Congenita(PC-K17) was thus confirmed.
The observed variation has previously been described in patients affected with PC but the N92S variant has not been reported in both the 1000 genomes and ExAC databases.

Key message: We wish to highlight this case in view of the relatively rare occurrence of Pachyonychia Congenita and the paucity in case reports of genetic work-up in this disease owing to cost and feasibility factors,and restricted access to such laboratory facilities.