Pachyonychia Congenita: Brief Appraisal of History and Current Classification

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Sir,

We read with great interest the informative case report “Pachyonychia congenita with late onset (PC tarda)” by Sravanthi et al. in the July-August 2016 issue of the Journal.[1] However, we would like to emphasize certain points regarding the history and current classification system of this rare disorder of keratinization [a Mendelian disorder of cornification (MeDOC)].

1. It is almost universally believed that the disorder was first described by Müller in 1904.[1] However, the first case of PC (with cutaneous horns) was originally described by St George Ash in an Irish girl as early as 1685 (quoted in[2]).[3]
2. A large amount of clinicogenetic information, based on the data collected from the International Pachyonychia Congenita Research Registry (IPCRR), has become available in the last several years. This has formed the basis of a new classification system for PC, gradually replacing earlier classifications, and is based almost exclusively on the mutated genes. This classification was proposed at the 2010 IPC Symposium,[4] and has recently been incorporated in the Rook's Textbook of Dermatology,[5] as well as in several papers.[3,6] The new classification is as follows:
   a. (a)PC-K6a (caused by mutation of KRT6A); (b) PC-K6b (caused by mutation of KRT6B); (c) PC-K6c (caused by mutation of KRT6C); (d) PC-K16 (caused by mutation of KRT16); and (e) PC-K17 (caused by mutation of KRT17).

To the best of the current author’s literature search, many recent Dermatology textbooks do not follow this classification system, as it is a recent one and some pioneers in the field have used the term PC-U, where PC is suspected, but no mutation has either been found (or could not be looked for). PC-U may be used as an “umbrella term” to simplify the classification of PC patients in the 6th type until a genetic testing may help in further classification. It may also be noted that PC-K6c (caused by the mutation of the KRT6C gene) has been least represented (17/535 cases; 3% only).

To conclude, the new classification revealing a broad spectrum of overlapping clinical and pathologic features has closely correlated phenotype to the specific keratin genotype in PC patients.[3-4] However, limitations of classifying PC patients in India, as per the new classification system, is definitely challenging as diagnostic genetic testing may not be easily available, as was the case in our recent report.[3]

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Conflicts of interest

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References


