Skin hyperpigmentation in Indian population: insights and best practice

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Pachyonychia Congenita: New Classification and Diagnosis

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Sir,

We read with interest the article “Pachyonychia congenita Type 1: Case report and review of the literature,” by Rathore et al.[1]

We would like to emphasize the importance of genetic testing to confirm a clinical diagnosis, draw your attention to the revised classification of pachyonychia congenita (PC) and provide further information regarding this rare skin disorder. Historically, PC was subdivided, with the two major clinical forms being PC-1 and PC-2. With an improvement in the understanding of the disease based on detailed clinical and molecular data collected by the International PC Research Registry (IPCRR), a new classification system was proposed, that divides PC into five subtypes based on the keratin gene affected.[2] The authors referred to this publication, but they did not mention the new classification, which is more scientific and has mutational analysis as the discriminator. Reference of the disease as PC-1 and PC-2 is obsolete and should be revised in the published literature. Analysis of the IPCRR dataset showed considerable phenotypic overlap between cases classified as PC-1 and PC-2.[3]

There are a number of other rare genetic skin disorders including Clouston syndrome (due to mutations in GJB6) that share clinical features with PC, making clinical diagnosis challenging and difficult. The importance and value of genetic testing have been demonstrated in several cases enrolled in the IPCRR where molecular data has excluded a diagnosis of PC and another diagnosis was identified and confirmed.

The case cited by the authors and also in many other publications as “recessive PC,”[4] is another example where molecular data have been extremely important and has finally identified the genetic defect in this family. With the advent of whole exome sequencing, the family was analyzed and a mutation identified in the CAST gene thereby confirming that they do not have PC but have a different disorder, called PLACK syndrome.[5] To date, there are no reported cases of recessive PC confirmed by genetic testing.

For patients, confirmation of their diagnosis by genetic testing allows for accurate genetic counseling and management of their disorder. We recommend the clinical diagnosis of PC in this case should be confirmed by genetic testing. PC Project (www.pachyonychia.org) provides genetic testing and a number of other free services to PC patients worldwide that enroll in the IPCRR. The PC Project also provides patient support and information and advice on the care of PC. The continual addition of clinical and molecular data from newly diagnosed individuals to the IPCRR database is driving the design and development of future treatments and clinical trials for PC.

We urge authors and editors to rely on data based on genetically confirmed patients. Reporting cases based merely on clinical diagnosis may add incorrect information to the existing knowledge about this ultra-rare disease.

Research is the key to cure and PC Project works with this goal and provides assistance in the diagnosis, treatment, and research efforts for physicians from different parts of the globe.

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Conflicts of interest
There are no conflicts of interest.

References

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