



Pachyonychia Congenita Project

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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.

Correspondence

Pachyonychia congenita type 2: abnormal dentition extending into adulthood

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SIR, Pachyonychia congenita (PC) is a group of hereditary disorders characterized by hypertrophic nail dystrophy and associated ectodermal features.¹ Two main clinical syndromes have been distinguished.² In PC type 1 (PC-1; Jadassohn–Lewandowsky syndrome, OMIM 167200) pachyonychia is associated with focal palmoplantar keratoderma (PPK), follicular keratoses and oral leucokeratosis. In PC type 2 (PC-2; Murray–Jackson–Lawler syndrome, OMIM 167210) associations include multiple pilosebaceous cysts. Focal PPK in PC-2 may be milder than in PC-1, and follicular keratoses, bushy eyebrows, angular cheilosis, hoarseness and (in children) unruly hair can also occur. Mutations in keratins have been found to be the molecular basis for both these disorders, affecting one of the paired keratins of specialized epidermis, K6a/K16 (PC-1) and K6b/K17 (PC-2).^{3,4}

One of the striking features of PC-2 is the frequent first presentation with primary dentition erupted at birth (prenatal or natal) or within the first 30 days of life (neonatal). Frontal teeth are typically affected; they are soft, friable, and said to be prone to caries, usually being lost within the first few months of life.^{5–7} In addition to natal teeth, early multiple tooth development at 4–5 months and, in one case, early natal tooth loss without immediate permanent tooth replacement, have been reported.⁴

In some cases, second primary teeth, nonpermanent teeth occurring after the loss of natal teeth, will develop in addition to the natal teeth but have been thought to be ultimately replaced by normal permanent teeth during childhood. Histologically, the polypean dental papillae of these natal teeth show mucosal hyperplasia and irregular rete ridge proliferation. Cytoplasmic vacuolization and oedema are seen in the upper and spinous layers of the mucosa. Irregular osteodentine-like structures with cell inclusions and interglobular dentine are seen.⁸ K17 expression has been described in the early stages of epidermal appendage development⁹ and in the predominantly hard portion of the hair shaft,¹⁰ but its expression in tooth progenitors is not known.

We now report for the first time possible effects on adult as well as infantile dentition of PC-2. We studied a multi-generation family with PC-2 due to a heterozygous missense mutation in the helix initiation motif of K17 (Asn92Asp)

presenting with classical features of dystrophic nails, focal plantar keratoderma, multiple epidermal cysts, abnormal eyebrow and body hair (pili torti) and natal teeth.³ Of 18 affected members over two generations whom we were able to interview, seven reported having either full or upper dentures fitted in their late teenage years due to ‘crumbling’ and exaggerated friability of their adult teeth. There was no history of increased caries. All affected patients had initially been treated with multiple crowns to prevent the disintegration of their teeth, without benefit. This history is suggestive of either enamel loss or attrition, but it has not been possible directly to assess any affected teeth. By contrast, none of their nine unaffected siblings had dentures or any similar dental problems.

Although the premature loss of primary teeth in conjunction with early eruption may be of no clinical significance, the loss of primary or permanent teeth in the absence of trauma should not be overlooked by the clinician.¹¹ Nontraumatic early loss of adult dentition is usually caused by caries or destructive periodontal disease. It is possible that the abnormal dentition in PC-2 may be a result of dentine abnormality, which is nonkeratin related, instead of a keratin-related enamel abnormality.

Abnormalities in adult teeth have not previously been described in PC-2. However, we were able to obtain a history only of unusual increased friability, suggesting a dentine abnormality, and not of caries. Dental friability has not previously been noted and as only some members were affected, it may be multifactorial with K17 mutation contributing indirectly. The effect might alternatively be specific to the Asn92Asp mutation.

Pending further evidence, we recommend that dermatologists caring for children with PC-2 should bear in mind a possible effect on permanent dentition and consider appropriate dental examination and care. Moreover, the presence in PC-2 of structurally abnormal, premature teeth and possibly adult teeth suggests that K17-expressing keratinocytes play an important role in odontogenesis and tooth eruption: this is an area for further study.³

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Conflicts of interest: none declared.