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.

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Dear Sir,

PACHYONYCHIA CONGENITA

Pachyonychia congenita is a rare genodermatosis which is autosomal dominant in inheritance. We describe a Malay toddler who presented with leukaemia and this condition.

A 20-month-old Malay boy was referred to our hospital for the management of acute lymphoblastic leukaemia. He had been noted to have abnormal nails at infancy and had been initially diagnosed to have onychomycosis. His nail clippings grew *Aspergillus niger* and he had been treated with itraconazole for several months. However, there was no improvement in the condition of his nails. He was then labelled as having yellow-nail syndrome.

On examination at our centre, he was found to have hyperpigmentation and hyperkeratosis of all his fingernails as well as toenails. His mother had similar findings (Fig. 1) and she also had hyperkeratosis of her soles. No one else in the family was affected. Apart from the nail changes, the child did not have any oral lesions and his palms and soles were also normal. A diagnosis of pachyonychia congenita type 1 was made clinically and his mother was counselled regarding the disease. The child has completed two years of chemotherapy without complications and is currently well and in remission.

Classic type 1 pachyonychia congenita (Jadassohn–Lewandowski syndrome) is due to mutations in the gene for keratin 16. It can cause a variety of ectodermal defects affecting the nails, skin, oral mucosa, larynx, hair and teeth.[1] Nail dystrophy is the most striking feature, with thickened, tubular nails which can present either at birth or early in life as in our patient. The second most prominent feature is palmoplantar keratoderma, which was present in this child's mother. Other features include onychogryphosis, follicular keratosis and oral leucoplakia. Type 2 pachyonychia congenita is characterised by natal teeth and the development of epidermal cysts or steatocysts.

Fig. 1 Hyperpigmentation and hyperkeratosis of all 10 fingernails of both mother and child.

Onychomycosis is also a cause of hyperkeratosis. However, it rarely affects all 20 nails. The most common cause of tinea unguium is the dermatophyte *Trichophyton rubrum*. In Malaysia, it has been found that moulds, in particular *Aspergillus niger,* cause a significant proportion (35%) of onychomycosis.[2]

The yellow nail syndrome is a triad of thickened, excessively curved, slow-growing yellow nails, primary lymphoedema and chronic respiratory disease. It has previously been classified as a dominantly inherited condition with variable expressivity. However, this has been challenged recently by findings that suggest that the majority of cases are sporadic and that the nail changes may remit.[3] Our patient had no previous medical history of note; he had never had lymphoedema or respiratory problems.

The yellow nail syndrome is known to be associated with malignancies, in particular Non-Hodgkin's lymphomas and carcinomas.[4] In contrast, pachyonychia congenita has not been associated with cancers although squamous cell carcinoma has been reported in a patient who had chronic plantar ulcerations.[5] It is likely that the leukaemia in our patient was a chance association.

The treatment of pachyonychia congenita consists of keratolytic agents and lubricants for the palmar and plantar hyperkeratosis. At present, there is no satisfactory treatment for the nail changes.

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