Use of Articles in the Pachyonychia Congenita Bibliography

The articles in the PC Bibliography may be restricted by copyright laws. These have been made available to you by PC Project for the exclusive use in teaching, scholarship or research regarding Pachyonychia Congenita.

To the best of our understanding, in supplying this material to you we have followed the guidelines of Sec 107 regarding fair use of copyright materials. That section reads as follows:

Sec. 107. - Limitations on exclusive rights: Fair use
Notwithstanding the provisions of sections 106 and 106A, the fair use of a copyrighted work, including such use by reproduction in copies or phonorecords or by any other means specified by that section, for purposes such as criticism, comment, news reporting, teaching (including multiple copies for classroom use), scholarship, or research, is not an infringement of copyright. In determining whether the use made of a work in any particular case is a fair use the factors to be considered shall include - (1) the purpose and character of the use, including whether such use is of a commercial nature or is for nonprofit educational purposes; (2) the nature of the copyrighted work; (3) the amount and substantiality of the portion used in relation to the copyrighted work as a whole; and (4) the effect of the use upon the potential market for or value of the copyrighted work. The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.

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.
Pachyonychia Congenita

Synonym
Jadassohn-Lewandowsky Syndrome, PC-1
Jackson-Lawler, PC-2

Inheritance
PC-1: autosomal dominant; K16 and K6a gene on 17q12-21 and 12q13, respectively
PC-2: autosomal dominant; K17 and K6b gene on 17q12-21 and 12q13, respectively

Prenatal Diagnosis
DNA analysis

Incidence
Over 100 case reports; increased in Slavic and Jewish populations; M>F

Age at Presentation
Birth (if natal teeth); otherwise first few months of life

Pathogenesis
Keratin gene defects disrupt normal intermediate filament assembly and may play a role when frictional stressors are applied to epithelial cells

Key Features
Nails
All 20 nails affected—fingers > toes; subungual hyperkeratosis with increased transverse curvature (“pincer nail effect”) and distal elevation of nail plate; brownish-yellow discoloration; painful paronychial infection with Candida, Staphylococcus; may shed nail plate
Skin
Focal symmetric palmoplantar keratoderma with/without hyperhidrosis, pain, bullae
Follicular hyperkeratosis of elbows, knees, extensor extremities
Steatocystoma multiplex (PC-2), epidermoid cysts (PC-2)
Mouth
Natal teeth (PC-2)
Oral leukokeratosis (PC-1, not promalignant)—tongue, buccal mucosa
Eyes (less common)
Corneal dystrophy, cataracts

Differential Diagnosis
Dyskeratosis congenita (p.180)
Other palmoplantar keratodermas
Weber-Cockayne syndrome (p. 242)
Psoriasis
Onychomycosis

Laboratory Data
Oral biopsy (or diagnosis equivocal)
Nail biopsy

Management
Referral to dermatologist/podiatrist—nail paring with electric file, urea or salicylic acid in petrolatum under occlusion to nail plate, avulsion with matrix destruction, antibiotics, orthotics; keratolytics, emolliation to hyperkeratosis lesions

Prognosis
Lesions persist throughout life; rarely blindness secondary to corneal dystrophy
Babies can suffer from recurrent painful staphylococcal paronychia... Treat infection promptly... Dental filers work well in paring the nail... Podiatrist can be tremendous help... I've seen a few adults attending the EB clinic who were misdiagnosed... The presence of natal teeth and nail changes is very suggestive of Pachyonychia congenita... It is important to remember that type that begins after birth called the tardive variety... BK

The nail defect markedly affects function making it difficult to pick up small objects and impairs fine touch... Most patients have been male... Although there's not a lot to offer them, you may try 40% urea in petrolatum applied to nail plate for 7 to 10 days under occlusion with subsequent physical debridement... Total nail avulsion surgically may be necessary... RS
# Pachyonychia Congenita

**Synonym**
- Jadassohn-Lewandowsky syndrome, PC-1
- Jackson-Lawler, PC-2

**Inheritance**
- PC-1: autosomal dominant; K16 and K6a gene on 17q12-21 and 12q13, respectively
- PC-2: autosomal dominant; K17 and K6b gene on 17q12-21 and 12q13, respectively

**Prenatal Diagnosis**
- DNA analysis

**Incidence**
- Over 100 case reports; increased in Slavic and Jewish population; M > F

**Age at Presentation**
- Birth of natal teeth; otherwise first few months of life

**Pathogenesis**
- Keratin gene defects disrupt normal intermediate filament assembly and may play a role when frictional stresses are applied to epithelial cells

**Key Features**

**Nails**
- All 20 nails affected—fingers > toes; subungual hyperkeratosis with increased transverse curvature ("pincer nail effect") and distal elevation of nail plate; brownish-yellow discoloration; paraxial paronychial infection with Candida; Staphylococcus; may shed nail plate

**Skin**
- Focal symmetric palmoplantar keratodermas with/without hyperhidrosis, pain, bullae
- Follicular hyperkeratosis of elbows, knees, extensor surfaces
- Stratocystoma multiplex (PC-2); epidermoid cysts (PC-2)

**Mouth**
- Natal teeth (PC-1)
- Oral leukokeratosis (PC-1), not premalignant—tongue, buccal mucosa

**Eyes (less common)**
- Corneal dystrophy, cataracts

**Differential Diagnosis**
- Dyskeratosis congenita (p. 180)
- Other palmoplantar keratodermas
- Weber-Cockayne syndrome (p. 242)
- Poromas
- Onychomycosis

**Laboratory Data**
- Oral biopsy (of diagnosis equivocal)
- Nail biopsy

**Management**
- Referral to dermatologist/podiatrist—nail paring with electric file, urea or salicylic acid in petrolatum under occlusion to nail plate, avulsion with matrix destruction; antibiotics; orthotics; keratolytics; emollient to hyperkeratotic lesions

**Prognosis**
- Lesions persist throughout life; rarely blindness secondary to corneal dystrophy

---

**Clinical Pearls**

- Babies can suffer from recurrent painful staphylococcal paronychia...
- Treat infection promptly...
- Dental files work well in paring the nail...
- Podiatrist can be tremendous help...
- I've seen a few adults attending the EB clinic who were misdiagnosed...
- The presence of natal teeth and nail changes is very suggestive of pachyonychia congenita...
- It is important to remember the type that begins after birth called the tardive variety...

- The nail defect markedly affects function making it difficult to pick up small objects and impairs fine touch...
- Most patients have been male...
- Although there's not a lot to offer them, you may try 40% urea in petrolatum applied to nail plate for 7 to 10 days under occlusion with subsequent physical debridement...
- Total nail avulsion surgically may be necessary...
Skin

Associated Findings

- Oral leukokeratosis
- Stratocystoma multiplex
- Pilosebaceous cysts
- Follicular Hyperkeratosis
- Painful Palmoplantar keratoderma Bullae
- Subungual hyperkeratosis
Skin

Associated Findings

- Oral leukokeratosis
- Steatocystoma multiplex
- Bullae
- Subungual hyperkeratosis
- Palmoplantar keratoderma
- Hyperhidrosis
- Follicular hyperkeratosis

10.23. Fingernails with yellow-brown discoloration, marked subungual hyperkeratosis, increased transverse curvature, and distal elevation of nail plate. (20)

10.24. Benign oral leukokeratosis on tongue. (20)

10.25. Follicular hyperkeratosis on knees. (20)

10.26. Steatocystoma multiplex in patient with PC-2. (1)