Pachyonychia Congenita

PC Advocate/Peer Coach Training
AGENDA—PC TRAINING MEETING

12:00 pm Luncheon and Introductions

12:30 pm What Is Pachyonychia Congenita? ............................................................... 1
Presented by PC Advocates

12:45 pm Skill Building for PC Advocates and Peer Coaches
  • Active Listening ...................................................................................................... 2
  • Challenges for Patient Advocacy ........................................................................... 3
  • Evaluating Information (team discussion) ............................................................ 4
  • Growing Your PC Vocabulary ............................................................................... 5
  • PC Care—Sharing Best Tips .................................................................................. 6

2:00 pm BREAK

2:15 pm PC Speed Learning ........................................................................................... 7

2:45 pm Creating Opportunities for PC Advocates ..................................................... 8
Discussion

3:30 pm Grand Rounds—The PC Way (Sample Booklet)

4:00 pm Meeting Summary
WHAT IS PACHYONYCHIA CONGENITA?
WHAT IS PACHYONYCHIA CONGENITA?
presented by PC Advocates

All PC Advocates are prepared to present information on any slide and have been randomly assigned for today’s presentation as follows:

<table>
<thead>
<tr>
<th>Slide</th>
<th>Duration</th>
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</thead>
<tbody>
<tr>
<td>1</td>
<td>1/2 minute</td>
</tr>
<tr>
<td>2—overview of terms</td>
<td>1/2 minute</td>
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<tr>
<td>3</td>
<td>skip</td>
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<tr>
<td>4—explanation of terms</td>
<td>1 minute (skip 3)</td>
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<tr>
<td>5—phenotype/genotype</td>
<td>1 minute</td>
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<tr>
<td>6—affect nails</td>
<td>1 minute</td>
</tr>
<tr>
<td>7—plantar callus</td>
<td>1 minute</td>
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<tr>
<td>8-9— cysts and FHK</td>
<td>1 minute</td>
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<tr>
<td>10—oral leukokeratosis</td>
<td>1 minute</td>
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<tr>
<td>11—Burden of Rare and the Burden of PC</td>
<td>2 minutes</td>
</tr>
<tr>
<td>12—My Story  How PC Impacts My Life The Ways I Manage No Treatment; Self Care The Difference PC Project Makes</td>
<td>2 minutes</td>
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</table>
Pachyonychia Congenita (PC) is a rare genetic skin disorder. It affects the keratin proteins in the skin, nails, feet, and tongue. The genetic cause is a mutation in one of five keratin genes: KRT6A, KRT6B, KRT6C, KRT16, or KRT17. The phenotype of PC varies across five major types: PC-K6a, PC-K6b, PC-K6c, PC-K16, and PC-K17. There is no effective treatment for PC, and the number of people affected is estimated to be between 2,000 and 10,000 in the USA.
### Major Clinical Findings of PC

#### Blisters and Calluses

**PC = Painful Calluses**

![Blisters and Calluses](image)

#### Cysts

**PC = Painful Cysts**

![Cysts](image)

#### Follicular Hyperkeratoses (FHK)

**Pachyonychia Congenita**

![Follicular Hyperkeratoses (FHK)](image)

#### Leukokeratosis

**Pachyonychia Congenita**

![Leukokeratosis](image)

### My PC Story

- The Burden of a Rare Disease
- Misdiagnosis and misunderstanding
- No treatment (unmet medical need)
- Isolation
- The Burden of PC
- Pain (limiting activities)
- Appearance
- Time for care

### My PC Story

- How PC impacts my life.
- The hardest thing about PC.
- The ways I manage PC.
  - No effective treatment for PC.
  - Patients rely on self-care to trim callus, treat blisters, FHK and cysts.
- The difference PC Project has made.
ADVOCATE AND PEER COACH SKILL BUILDING:

ACTIVE LISTENING
ADVOCATE AND PEER COACH SKILL BUILDING
ACTIVE LISTENING

This is the foundation of success. Without this skill nothing else really matters.

**Active Listening means:**
- Removing all distractions
- Listening to speaker’s signs & sounds
- Feeding back that you have understood

**SPEAK** in such a way that others love to **LISTEN** to you. **LISTEN** in such a way that others love to **SPEAK** to you.

One of the most **sincere** forms of **respect** is actually listening to what **another** has to say.

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**Whole Body LISTENING**

- **EYES**
  - Looking at person talking

- **FEET**
  - Quiet on the floor

- **EARS**
  - Both ears ready to hear

- **BODY**
  - Facing the speaker

- **MOUTH**
  - Quiet (no talking, humming or making sounds)

- **BRAIN**
  - Thinking about what is being said

- **HANDS**
  - Quiet in lap, pockets or by side

- **HEART**
  - Caring about what the person is saying

**“Most people do not listen with the intent to understand; they listen with the intent to reply.”**

Stephen R. Covey

**QUESTION:** Why is Mickey Mouse such a good listener?
ADVOCATE AND PEER COACH SKILL TRAINING:

CHALLENGES
In serving individuals around the world, every patient advocacy group faces enormous and frequent challenges that are mostly unknown to the patient population. These pitfalls have caused many groups to fail. We need PC Advocates and Peer Coaches to be aware of these obstacles and to assist in carrying a consistent and clear message of our goals and purposes.

_Fighting for a cure._

*Connecting & helping patients.*

_Empowering research._

Here are just a few of the problems mentioned in the last six months on various patient advocacy blogs. How will you help us prevent these disruptive events for the PC community?

**DISCUSSION**

- Researchers funding research directly with patients without peer review
- Physicians signing up individual patients without peer review
- Incorrect and embedded ideas about PC, genetics, gene therapy, drug development
- Understanding why enrolling every family member is so important
- The need for large dollars in funding and the importance of every small donation/action
- Splinter groups
- Posting medical advice on social media including Facebook
- Using anecdotal experience as evidence
- Expecting personal ‘perks’ (such as physician consultations on non-PC related topics)
ADVOCATE AND PEER COACH SKILL BUILDING:

EVALUATING INFORMATION
ADVOCATE AND PEER COACH SKILL BUILDING
EVALUATING INFORMATION

1. Social media posts and patient reported experience—
2. News stories—example “CRISPR” (PC NewsBrief February 2016)
3. Published articles— (PC NewsBrief April 2016)
ADVOCATE AND PEER COACH SKILL BUILDING
SOCIAL MEDIA

A surprising number of people who post in social media sites especially Facebook groups provide anecdotal experience for evidence. This can be confusing and misleading. Monitoring these groups and chiming in with quality information when needed is an essential service to dispel misinformation.

Facebook—Patient Chat  www.facebook.com/groups/47155151452/
Facebook—Organization Page  www.facebook.com/pachyonychia/
Twitter @Pachyonychia  twitter.com/Pachyonychia
Instagram  www.instagram.com/pachyonychia
LinkedIn  www.linkedin.com/company/pc-project
Pinterest  www.pinterest.com/pachyonychia/
YouTube  www.youtube.com/Pachyonychia

Step 1—Become active for PC on Social Media sites. Interact with members and posts. Welcome new members. Direct members to the website or PC Project. Be an example.

Step 2—Watch & Gather. Be on the look out for sensitive or dangerous posts. Some posts may need to be removed or answered not in social media. Watch for good posts and gather information to be added to the PC Website or PC Wiki. Help encourage members to maintain the guidelines (see Facebook PC Patient Chat Guidelines on next page).

To reach more of the group wait for “critical mass” to build in the message thread. If you respond to the initial post, only the questioner will see it. Wait until 15 or 20 people (if possible) have chimed in and then drop it on ‘em. Thread participants will ALL get a notification of another post to the thread, and they’ll all benefit... and from an organizational marketing point of view, they will all see your organization in action at its best. (obvious exception would be for a critical item where time is of the essence notify immediately.)

Step 3—Create & Post. Develop ideas of things needed on social media.
ADVOCATE AND PEER COACH SKILL BUILDING
EVALUATING INFORMATION—NEWS STORIES

EXAMPLE: News articles on the CRISPR technology.

The quiet revolutionary: How the co-discovery of CRISPR explosively changed Emmanuelle Charpentier’s life
Nature - 17 hours ago
Emmanuelle Charpentier, a key inventor of the gene-editing technology CRISPR-Cas9

Scientists solve CRISPR's 'Energizer bunny' problem
STAT - 17 hours ago

God's Red Pencil? CRISPR and the Three Myths of Precise Genome Editing
CounterPunch - 22 hours ago

More news for CRISPR

QUESTION: Here is a link about CRISPR. Do you think we are far from the application of this technique?

ANSWER: CRISPR is only a new tool to aid gene sectioning. We have demonstrated the effectiveness of siRNA which is another tool with a similar ‘interference’ function. Whatever tool is used for creating the drug, delivery to the cells of the skin is still the main obstacle we have not yet solved.
Activity — Team Discussion.

Work with your specialist partner to compare and contrast the strengths and weaknesses of two recently published articles on PC.


INCORRECT INFORMATION
There is a massive amount of incorrect information about Pachyonychia Congenita online and in printed materials. Our PC website (www.pachyonychia.org) is a reliable source of correct and up-to-date information on PC.

However, on the PC website, we include all articles published about PC and related disorders. The best articles are highlighted on the ‘Select Lists’ on the right panel. Some keys to help you evaluate information and articles.

1. Is genetic testing information included? If not, the report may not really be about PC! An example is an article on ‘recessive’ PC. All of the information on our website states that PC is a dominant disorder. This is important. After nearly 8 years, genetic testing showed that the patients said to have ‘recessive PC’ actually have a completely different disorder.

2. Does the information contradict information listed on ‘What Is PC?’ on the PC website? Many single case (or single family) articles incorrectly connect things found in that family with PC. For example, although testing is conducted to establish PC, the article says that hair loss or deafness or some other characteristic is associated with PC. You can rely on the PC website information. Or, feel free to send us an email for an answer to PC questions you may have.

3. Another key is to look at the dates of the publications that are cited. Remember, the genes that cause PC were discovered by Smith/McLean about 1995. Before that date, there were many assumptions and guesses as each case was examined. Most of the best publications on PC have come after we had more than 200 patients in the IPCRR (about 2008 or after).
Pachyonychia Congenita Type 1: Case Report and Review of the Literature

Praveen Kumar Rathore, Varun Khullar, Anupam Das

Abstract
The case of an 8-year-old boy is hereby reported, who presented with nail dystrophy, subungual hyperkeratosis, oral leukokeratosis, and numerous follicular papules all over the body. The features were consistent with a diagnosis of pachyonychia congenita type 1. The case is being reported for its rarity. We also discuss in a nutshell, the literature till date.

Key Words: Follicular papules, leukokeratosis, nail dystrophy, subungual hyperkeratosis

Introduction
Pachyonychia congenita (PC) is a rare autosomal dominant disorder of keratinization.

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What was known?
Pachyonychia congenita is a rare genodermatosis with a wide array of cutaneous manifestations including nail dystrophy, subungual hyperkeratosis, follicular papules, oral leukokeratosis, palmoplantar keratoderma, etc.

Pin-head sized follicular papules over the entire body, concentrated over the face, back, buttocks, abdomen, and gluteal region [Figures 4-6]. Besides, hyperkeratotic lesions were distributed throughout the body. Marked hyperhidrosis of the palms and soles was observed. Palmoplantar keratoderma was present, along with painful hyperkeratotic plaques [Figures 7 and 8]. Mucosal examination was significant for the presence of asymptomatic oral leukokeratosis over the dorsum of the tongue [Figure 9]. Routine laboratory investigations including complete hemogram, hepatic profile, and renal profile were within normal limits. KOH microscopy and culture of nail clippings was negative. Skin biopsy from a hyperkeratotic lesion around elbow showed orthokeratosis and acanthosis [Figure 10]. No evidence of any malignancy was found during the thorough work up. Genetic and molecular biological studies could not be carried out due to lack of infrastructure facilities. Based on the above findings, he was diagnosed as pachyonychia congenita type 1. The child was prescribed Vitamin A and E along with emollients and keratolytics. He was prescribed oral Vitamin A at a dose of 25,000 IU, under a multidisciplinary approach after consultation with...
Department of Pediatrics and Ophthalmology. The patient is under stringent follow-up every 2 weeks. He has been referred to the Department of Physical medicine and rehabilitation for weight control exercises.

**Discussion**

PC is a rare genodermatosis with autosomal dominant mode of inheritance. Heterozygous mutations involving keratins K6a or K16 are associated with PC-1 whereas those involving K6B and K17 are associated with PC-2.⁶ Although, autosomal dominant mode is the most common mode of inheritance, there are reports of autosomal recessive inheritance as well.⁶
PC has been classified into four types, the common clinical findings in all of them being painful and debilitating plantar keratoses, nail dystrophy and hypertrophy, oral leukokeratosis, palmoplantar hyperhidrosis, and a variety of epidermal cysts.[11]

Patients with type 1 PC (Jadassohn–Lewandowsky syndrome) are characterized by the presence of nail dystrophy since birth. This may be accompanied with painful pachyonychia, hyperkeratosis of palms and soles over the pressure sites, oral leukokeratosis, palmoplantar hyperhidrosis, and follicular keratotic papules distributed throughout the body.[7] Besides, painful blisters also develop over the palms and soles. Another characteristic finding is the presence of verrucous lesions over the elbows, knees, popliteal fossae, and ankles. In addition, hoarseness of voice is also an important feature of PC type 1.[9,10]

In addition to the above mentioned findings, type 2 PC (Jackson–Lawler syndrome) has the features of natal teeth, hair anomalies including pili torti, unruly hair, and bushy eyebrows. Oral leukokeratosis and palmoplantar keroderma is milder in comparison to type 1 PC, but the development of epidermal cysts or steatocysts are the hallmark findings in type 2 PC. PC type 3 (Schafer–Branuer) has features of corneal leukokeratosis.

Type 4 PC is termed as PC tarda, which is manifested in the second or third decade of life. It results from mutations in the keratins 16 and 17 genes.[11]

PC with unusual features has been noted. Rare cases of pachyonychia congenita tarda have been reported with symptoms developing in the fifth decade of life.[12] Recently, a case of pachyonychia congenita associated with B-cell lymphoma has been reported.[13] Cases with unusual dental findings have also been reported.[14] An interesting case of PC with woolly hair in a 10 month old patient has been also reported.[15] Cases with isolated involvement of nails have also been described.[16] Apart from the numerous oral manifestations, median rhomboid glossitis in association with PC has been documented.[17]

The clinical features in our case are suggestive of PC type 1 with characteristic subungual hyperkeratosis and follicular papules over the entire body since birth. Histological findings of orthokeratotic hyperkeratosis and acanthosis confirm our diagnosis. The patient was
prescribed Vitamin A and E along with emollients and keratolytics. Mahajan et al. reported a case of PC, which was treated with oral Vitamin A and E.[18] Vitamin A stimulates differentiation and leads to normalization of accelerated epidermopesis of pathological keratinocytes of epidermis of skin and nail. With this background, we have prescribed Vitamin A to our patient. The upper limit of dose of Vitamin A in pediatric patients (1–8 years) is 17,500–35,000 IU. Hypervitaminosis A occurs when the consumption is more than or equal to 100,000 IU for months. Our patient was referred to Department of Pediatrics and Ophthalmology. Thereafter, he was prescribed Vitamin A at a dose of 25,000 IU. The patient is under stringent follow-up every 2 weeks, for systemic check-up including fundoscopy and signs of irritability. For management of pain and discomfort due to the palmoplantar keratoderma, patient was advised limitation of walking and standing, use of soft shoes, control of body weight, and use of appropriate clothing. The patient is under periodic follow-up. Mechanism of manipulating gene expression is now approved which in future, may help to suppress or correct the genetic defect in the chromosomes. The only effective treatment though is nail surgery with radical excision of the nail bed and matrix and grafting at the site but patient being very young, this line of treatment was not accepted by the parents.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

**Financial support and sponsorship**

Nil.

**Conflicts of interest**

There are no conflicts of interest.

What is new?

Full blown cases of pachyonychia congenita are rarely found and our case is unique because it features all the classical manifestations of this rare disease. Very few cases have been reported from India.

References

10. Handa S, Kumar B. Pachyonychia congenita type 1. Indian J Dermatol Venereol Leprol 1994;60:228-.
Pachyonychia Congenita: A Spectrum of KRT6a Mutations in Australian Patients


Abstract

Background: Pachyonychia congenita (PC) is a rare inherited disorder of keratinization characterised by hypertrophic nail dystrophy, painful palmoplantar blisters, cysts, follicular hyperkeratosis and oral leukokeratosis. It is associated with mutations in five differentiation-specific keratin genes, KRT6A, KRT6B, KRT6C, KRT16, or KRT17.

Objectives: Living with Pachyonychia Congenita can be isolating. The aim of this paper is to document a single patient’s experience within a national context.

Method: We report the case of a 2 year old female with an atypical presentation of PC due to a mutation in KRT6A with severely hypertrophic follicular keratoses, skin fragility, relative sparing of nail hypertrophy on one hand and failure to thrive in early infancy. In collaboration with the International Pachyonychia Congenita Research Registry (IPCRR), a database search was performed using Australian residency and KRT6A mutation as inclusion criteria. The IPCRR database was also searched for a matching KRT6A mutation. Six Australian patients were identified in addition to one patient with an identical mutation residing in the United States. The detailed standardized patient questionnaire data was manually collated and analysed.

Results: Fingernail hypertrophy and oral leukokeratosis were the most common features. There was no recording of asymmetric distribution in any other Australian patient. Trouble nursing as an infant and follicular hyperkeratosis also occurred in the American patient, however they did not have asymmetric distribution and the oral leukokeratosis appeared later in life.

Conclusion: This case has unique features. Sharing information can assist patients navigating life with this condition.
Pachyonychia congenita (PC) is a rare inherited disorder of keratinization that affects 5,000 to 10,000 people worldwide (1). It is associated with a mutation in one of five keratin genes (KRT6A, KRT6B, KRT6C, KRT16, KRT17), each defined by a constellation of clinical features. Although historically divided into two types, classification is now based on these genetic subtypes. The International Pachyonychia Congenita Research Registry (IPCRR) has identified more than 100 mutations (2). Characteristic manifestations of PC consist of hypertrophic nail dystrophy, painful palmoplantar keratoderma, cysts, follicular hyperkeratosis, and oral leukokeratosis (2). Painful palmoplantar keratoderma is the most debilitating clinical feature (1).

We report the case of a 2-year-old girl with an unusual presentation of PC due to a mutation in KRT6A. The features of this case are compared with those of other Australian cases of PC with KRT6A defects reported to the IPCRR.

CASE REPORT

The patient was born to Pakistani parents who were first cousins. She presented at 7 weeks with abnormal fingernails and white spots on her buccal mucosa. She had poor feeding from birth and was not gaining weight (failure to thrive). The parents noticed nail changes at 10 days and the leukokeratotic plaques, initially thought to be secondary to candida infection, at 3 weeks. Her parents and three older sisters were unaffected.

Initial examination identified hypertrophic dystrophy of her fingernails and toenails, markedly more pronounced on the right hand and foot, markedly more pronounced on the right hand and foot. Leukokeratotic plaques were identified on the tongue and right buccal mucosa. The nail hypertrophy was unilateral, with the left hand completely spared and showing only mild dystrophy of the fourth, fifth, and lateral part of the second fingernails. Similarly, the left foot was relatively spared, with mild dystrophy of only the distal part of the third and fourth toenails. This distribution is difficult to appreciate in the clinical photographs available.

Hyperkeratotic papules appeared over her thighs (Fig. 3) and trunk (Fig. 4). Hyperkeratotic papules developed on the knee extensor surfaces when she started crawling at 5 months old and were exacerbated by warm weather. Painful bullae over the soles of her feet developed upon commencement of walking at 12 months. The bullae were noted to have spread over her buttocks and thighs a few months later. This also flared during warm weather and resolved with the change of seasons at 18 months. Leukokeratosis of her mucous membranes fluctuated with time. Angular cheilitis (Staphylococcus aureus–positive culture) was treated with a 5-day course of topical antibiotic (neomycin and gramicidin); subsequent recurrences resolved with shorter 3-day courses of antibiotic.

Treatment of the nails involved tar cream and keratolytics, including 10% to 40% urea cream and
3% salicylic acid in aqueous cream, in combination with a mechanical paring device and a diamond-tipped grinder. Aluminium chloride hexahydrate 20% was applied to blistered areas daily for 4 weeks. A short course of betamethasone valerate 0.02% and hydrocortisone 1% was prescribed to relieve the irritated hyperkeratotic regions over the trunk and thighs, but her parents did not start this treatment. Systemic therapy with acitretin was offered, but her parents elected to wait until their daughter was old enough to decide for herself.

Sequencing of the known pachyonychia congenita genes identified a heterozygous missense variant in \textit{KRT6A} (c.520T>A; p.Phe174Ile) \cite{3}. This variant was predicted to be pathogenic using in silico analysis (p > 0.99) \cite{4}. Other disease-causing missense mutations in this position have previously been reported (p.Phe174Val, p.Phe174Ser, p.Phe174Cys) \cite{5-8}. The

\textbf{Figure 2.} Hypertrophic toenails: (A) 2 months old, (B) 18 months old, (C) 19 months old, and (D) 2 years and 2 months old.
IPCRR reports that this variant has been seen in one other individual in the United States (8), as mentioned in a previous review (3).

**METHOD**

The IPCRR collected data using their detailed standardized patient questionnaire. The IPCRR performed the database search using Australian residency and *KRT6A* mutation as inclusion criteria, which revealed seven individuals with the *KRT6A* mutation registered in Australia, including our patient. One of the seven patients was excluded because of incomplete data (multiple unanswered survey questions). The remaining six patients had incomplete data for no more than one survey question. The IPCRR worldwide database was also searched for a matching *KRT6A* mutation, which identified the aforementioned patient residing in the United States who shares the same mutation as the patient described in this case. The questionnaire data were then used to analyze the spectrum of clinical presentations in all known individuals with the *KRT6A* mutation in Australia in addition to the patient with the same mutation from the United States.
RESULTS

The six Australian patients were from four different families and shared four different mutations (8). Table 1 presents the clinical features that these patients reported. Fingernail hypertrophy and oral leukokeratosis were the most common features, recorded by all six patients. In five of the six patients, these changes were documented to have occurred before 1 year of age. Follicular hyperkeratosis occurred in four of the six patients. Hoarseness of voice was reported in half. There was no recording of asymmetric distribution in any of the other Australian patients. Trouble nursing as an infant was reported in one other patient, but data for this survey question were missing for three patients.

Columns 3 and 4 of Table 1 contrast our patient with the patient from the United States. Follicular hyperkeratosis was a shared clinical feature, but the American patient also had pilosebaceous cysts. The American patient did not have asymmetric distribution, and oral leukokeratosis appeared later in life. Both of these patients reported trouble feeding in infancy.

One section of the IPCRR questionnaire focused on factors found to exacerbate and relieve symptoms. The six Australian patients unanimously reported that rest, reduced mobility, cool weather, and winter relieved their symptoms. Half of the patients described their symptoms as at their best in the morning, and two-thirds reported that wearing cotton socks helped to maintain their feet in optimal condition. Factors collectively acknowledged to contribute to deterioration in symptoms included hot weather, walking, and hot or sweaty feet.

DISCUSSION

PC is a rare and complex genetic disorder. The IPCRR shows that, as of October 2014, there were 593 genetically confirmed cases of PC, 13 of these in Australia (2). Inclusive of our patient, 7 of the 13 documented cases in Australia have a mutation in KRT6A, with the remainder consisting of KRT16 and KRT17 mutations (2).

The hallmark features of PC due to a KRT6A mutation include early onset and extensive nail disease, in addition to substantial disease in locations other than the palms and soles, namely oral leukokeratosis, cysts (pilosebaceous and steatocystoma), follicular hyperkeratosis, and hoarseness of voice (8). This is confirmed in the IPCRR data from the six Australians with PC due to a KRT6A mutation (Table 1). The unique aspects of our case include

### TABLE 1. Clinical Features of Individuals with Pachyonychia Congenita Due to a KRT6A Mutation

<table>
<thead>
<tr>
<th>Clinical feature</th>
<th>Australian patients with feature, n (out of a possible 6)</th>
<th>This case</th>
<th>US patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asymmetrical distribution of nail hypertrophy</td>
<td>1 (this case)</td>
<td>Relative sparing of right hand</td>
<td>All nails affected</td>
</tr>
<tr>
<td>Type of mutation</td>
<td></td>
<td>Spontaneous (F174I)</td>
<td>Spontaneous (F174I)</td>
</tr>
<tr>
<td>Familial</td>
<td>3</td>
<td>Birth to 1 year</td>
<td>Birth to 1 year</td>
</tr>
<tr>
<td>Spontaneous</td>
<td>3</td>
<td>10–14 years old</td>
<td>10–14 years old</td>
</tr>
<tr>
<td>Onset of nail hypertrophy</td>
<td></td>
<td>Relative sparing of right hand</td>
<td>Symmetrical distribution</td>
</tr>
<tr>
<td>Birth to 1 year</td>
<td>5</td>
<td>Birth to 1 year</td>
<td>Birth to 1 year</td>
</tr>
<tr>
<td>1–4 years old</td>
<td>1</td>
<td>10–14 years old</td>
<td>10–14 years old</td>
</tr>
<tr>
<td>Asymmetrical skin findings</td>
<td>1</td>
<td>No difficulty</td>
<td>No difficulty</td>
</tr>
<tr>
<td>Leukokeratosis</td>
<td>6</td>
<td>Present</td>
<td>Present</td>
</tr>
<tr>
<td>Leukokeratosis onset</td>
<td></td>
<td>Birth to 1 year</td>
<td>Birth to 1 year</td>
</tr>
<tr>
<td>Unknown (question not answered)</td>
<td>3</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Birth to 1 year</td>
<td>3</td>
<td></td>
<td></td>
</tr>
<tr>
<td>10–14 years old</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Difficulty nursing or sucking as an infant</td>
<td></td>
<td>Difficulty</td>
<td>Difficulty</td>
</tr>
<tr>
<td>Difficulty</td>
<td>2</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Unknown (question not answered)</td>
<td>3</td>
<td></td>
<td></td>
</tr>
<tr>
<td>No difficulty</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Skin manifestations</td>
<td></td>
<td>Follicular hyperkeratosis</td>
<td>Follicular hyperkeratosis and pilosebaceous cysts</td>
</tr>
<tr>
<td>Unknown (question not answered)</td>
<td>2</td>
<td></td>
<td></td>
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<tr>
<td>Follicular hyperkeratosis</td>
<td>2</td>
<td></td>
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<tr>
<td>Follicular hyperkeratosis and pilosebaceous cysts</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Follicular hyperkeratosis, pilosebaceous cysts</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hoarseness of voice</td>
<td>3</td>
<td>Absent</td>
<td>Absent</td>
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<tr>
<td>Ear pain</td>
<td>0</td>
<td>Absent</td>
<td>Present</td>
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relative sparing of fingernail hypertrophy on one hand, severe follicular hyperkeratosis, and blistering involving the proximal limbs. In the Australian cohort, there are no other reports of nail hypertrophy relatively sparing one hand, although one patient described skin calluses affecting the right hand only. New dominant mutations may arise postzygotically, resulting in somatic mosaicism and asymmetric clinical features. This could be investigated by testing a sample of DNA taken from unaffected tissue, but this was not possible in this case. Unaffected tissue might be expected to show no mutation, whereas minimally affected tissue may contain a smaller proportion of cells with the mutation.

Follicular hyperkeratosis occurs in 80% of individuals with PC due to a \textit{KRT6A} mutation and is typically located in areas susceptible to friction (1). All Australian patients reported follicular hyperkeratosis (Table 1). Worldwide, IPCRR data show that in individuals with a \textit{KRT6A} mutation, rates of plantar keratoderma exceed those of palmar keratoderma (2). Because blisters tend to arise in areas of keratoderma, they too are more frequently observed in the plantar region. As evident in this case, the presentation of blisters on the proximal limbs is unique, with no preceding accounts in the literature.

Feeding difficulty and failure to thrive have been reported in two other patients, including the U.S. patient. A possible explanation for this is that leukokeratosis is painful, although the data from the US patient do not support this theory, because the feeding difficulty in infancy predated the appearance of leukokeratosis at 10 to 14 years of age (Table 1). We wonder if failure to thrive is an underreported feature of early infancy in individuals with PC.

As reflected in the Australian data, \textit{KRT6A} is the most frequently mutated gene, followed by \textit{KRT16}, \textit{KRT17}, \textit{KRT6B}, and \textit{KRT6C} (8). Our case represents one of more than 45% of cases of PC that arise spontaneously in patients with no family history (new mutations) (1). The history of consanguinity proved to be irrelevant in this case, as this was a new mutation (autosomal dominant) rather than a recessive disorder. Because atypical features of PC were present, we considered the possibility of a second disorder related to consanguinity, but there were no other clinical features to suggest a specific second diagnosis. The possibility of modifying factors related to consanguinity cannot be excluded.

REFERENCES

ADVOCATE AND PEER COACH SKILL BUILDING:

GROWING OUR PC VOCABULARY
1. **Rare**—Comments, definitions, thoughts from PC Advocates 2016

- Not common, as far as diseases or conditions are concerned, affecting a very small (relatively speaking) number patients, usually making research dollars and research talent difficult to obtain.
- It is something not many people know about. So with PC being ultra-rare, basically NO one knows about it. Educating others is on-going and is part of my regular life, everywhere I go.
- PC is classified as an ultra rare disease affecting just over 300 people in the US.
- Very, very few people globally have a condition.
- PC is an ultra rare disorder; to be an “orphan” disease in the USA there has to be approx 200,000 cases, yet PC has approximately 1,600 cases worldwide.
- Defined by the government as a disease that impacts 200,000 or fewer people. PC is ultra-ultra-ultra rare at 500-700.
- An inadequate word to describe Pachyonychia Congenita. There are only about 400 genetically confirmed cases in the U.S. This is ULTRA rare.

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*Additional comments from PC Project*

Rare Disease is another word for Orphan Disease. In the USA, this is defined as a disease that affects fewer than 200,000 people. However, in other countries there are other definitions of rare. There are about 7,000 rare disorders, and about 30 million people in USA are living with a rare disease. This equates to 1 in 10 (10% of US population)! About 80% of rare disease patients are affected by about 350 different rare diseases (from Global Genes).

**STATISTICS**—Check the PC Data tab on www.pachyonychia.org for monthly updates and please be careful when citing statistics from the IPCRR (registry).

1654 in the IPCRR. These individuals do not all have PC.
892 in the IPCRR have completed their forms, consultations, testing. All do not have PC.
699 in the IPCRR are genetically confirmed with PC worldwide.
311 in the IPCRR are genetically confirmed with PC and live in the USA.

*There are definitely additional PC cases not yet in the IPCRR - but under any definition, it is clear PC is rare.*
ADVOCATE AND PEER COACH SKILL BUILDING
GROWING OUR PC VOCABULARY

2. **PC Advocate/Patient Advocate**—Comments, definitions, thoughts from PC Advocates 2016

- A cheerleader, champion to educate others whether they are doctors, scientists or other patients not only about PC but to also raise money for research.

- A person who advocates/represents PC Project and individuals with PC. Advocacy activities might include: patient rights, matters of privacy and/or confidentiality, patient representation, awareness building, support and education of patients, and their caregivers.

- A representative for all who suffer with a condition and advocate to create awareness and help find a treatment or cure.

- A person who represents PC project and patients with PC to educate and raise awareness for PC. An advocate is trained and knowledgeable in PC and the goals/mission of PC project.

- Someone who goes to bat for people or an organization. Could be a spokesperson, facilitator, champion, public relations specialist for anything PC related.

- One who can speak on behalf of a patient to promote patient’s interests, and support patient throughout the process.

- A person who stands and works for those with the disease. A spokesperson, bodyguard, voice for PC.

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**Additional comments from PC Project**

We want PC Advocates to be the leaders for PC Project and to differentiate a PC Advocate from just an ‘advocate’ or even a ‘patient advocate’ (representing a patient) to an active participant in engaging and building and educating the PC community. Fundraising, education and awareness are important parts in the role of PC Advocates. A group of educated, informed, involved PC Advocates will greatly extend our reach.

Peer Coaches will help us connect with individual patients to unite the community as the number of individuals in the registry continues to grow about 2 each week from all over the world.

It is important that we have a consistent fact-based message about PC and PC Project. Our numbers are small, but a united and committed group can accomplish a great deal to provide hope, support and real progress toward our goals.
<table>
<thead>
<tr>
<th><strong>Definitions</strong></th>
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<tr>
<td><strong>PC Advocate</strong> <em>(public)</em></td>
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<tr>
<td><strong>Represent PC Project</strong></td>
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<td>• Tell your story</td>
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<tr>
<td>• Understand PC</td>
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<td>• Phenotype</td>
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<td>• Impact and QoL</td>
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<td>• Understand PC Project</td>
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<td>• Understand IPCRR</td>
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<td>• Understand IPCC</td>
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| **Peer Coach** *(patients)* |
| • 6 week training |
| • Understand PC |
|   • What it is/is not |
|   • Phenotype |
|   • Genotype |
| • Help patients |
|   • Listen to them |
|   • Non-medical |
|   • Support |
|   • Build patient skills |
ADVOCATE AND PEER COACH SKILL BUILDING

GROWING OUR PC VOCABULARY

3. PC Project—Comments, definitions, thoughts from PC Advocates 2016

- An absolutely unique organization unlike any other than I have ever seen, organizing world-class researchers, educating and supporting patients, and providing resources to aid education, treatment, and providing hope and encouragement to patients who previously would have nowhere to turn.

- The amazing nonprofit organization comprised of physicians and advocates working toward the goal of finding treatments or a cure for PC. Learn more on our website at www.pachyonychia.org.

- The organization that represents and provides support to patients with PC. PC project works with researchers and physicians to advance care and work toward a cure for PC.

- PC Project connects patients, researchers and physicians throughout the world in a united effort to help those with Pachyonychia Congenita.

- A blessing of hope and practical knowledge and help for PC patients. Also able to make things happen in the medical and scientific arena on behalf of PC research, care, treatments and ultimately a cure. I think of PC Project staff as miracle workers.

- Was founded in 2004 to educate, collaborate and reach out to physicians, scientists, patients and their families to assist in diagnosis, treatment (cure/relief) and research efforts.

- A nonprofit organization that is dedicated to the research of Pachyonychia Congenita in hopes of a cure or a method to provide relief to the PC patient. PC Project is also dedicated to patient support, and it provides a registry to confirm and keep records of patients.

Additional comments from PC Project

It’s all about love!

Without you, the PC patients in the IPCRR, supportive family members, and the physicians and scientists in the IPCC, PC Project would not be able to provide any hope, any help, any progress.

PC Project is people and everyone is needed in our community!

THANK YOU!

There are many types of ‘non-profit’ organizations. Officially, PC Project is a 501(c)(3) public charity in the USA and a registered charity in the UK.
4. IPCC—Comments, definitions, thoughts from PC Advocates 2016

- Consortium of scientists and physicians interested in collaborative efforts to develop and deliver effective treatments for PC who are trying to find me a cure or some imminent relief. There are approximately 150 PC leaders to collaborate and research clinical trials to find a cure and/or relief.

- A group of experts – clinical and research doctors -- who are working as a team on Pachyonychia and related diseases.

- The group of scientists and physicians investigating and researching PC.

- It’s an exclusive club for doctors who study PC! The International PC Consortium, made up of scientists and doctors who collaborate to develop and deliver effective treatments for PC.

- A group of interested physicians and researchers whose goal is to learn more about PC with a goal of helping patients better deal with this condition.

- The International Pachyonchia Congenita Consortium consists of elite scientists and physicians who have made rapid progress over the past decade including finding the specific DNA responsible for causing PC. Dozens of peer reviewed articles about PC have been published by this group, and multiple studies have been performed.

- A consortium of scientists and doctors willing and able to collaborate in finding effective treatments and ultimately a cure for PC. I am humbled and grateful for them.

**Additional comments from PC Project**
There are now 207 individuals in the IPCC which is open to all who have an interest in providing clinical or research services related to PC.

**IPCC STEERING COMMITTEE**
Philip David Gard, MBChB (retired)  
C. David Hansen, MD, Univ of Utah, Salt Lake City, UT, USA  
Roger L. Kaspar, PhD, TransDerm, Inc., Santa Cruz, CA, USA  
W. H. Irwin McLean, DSc, FRSE, University of Dundee, Scotland  
Edel O’Toole, MD, PhD, Queen Mary Univ, London, England  
Frances J.D. Smith, PhD, PC Project, Dundee, Scotland  
Eli Sprecher, MD, PhD, Tel Aviv Sourasky Medical Center Israel  
Maurice A.M. van Steensel, MD, PhD, Univ of Dundee, Scotland

**MEDICAL AND SCIENTIFIC ADVISORY BOARD (MSAB)**
All members of the IPCC Steering Committee, plus
Sherri J. Bale, PhD, GeneDx, Inc., Gaithersburg, MD, USA  
Pierre A. Coulombe, PhD, Johns Hopkins University, Baltimore, MD, USA  
Alain A. Hovnanian, MD, PhD, University Paris Descartes Sorboone, France  
Peter R. Hull, MD, PhD, FFDERM, FRCP, Royal University Hospital, Canada  
E. Birgitte Lane, PhD, FRSE, FMedSci, Singapore  
John A. McGrath, MD, PhD, King's College, London, England  
Leonard M. Milstone, MD, Yale School of Medicine, New Haven, CT, USA  
Amy S. Paller, MD, Northwestern University, Chicago, IL, USA  
Michael Polydefkis, MD, Johns Hopkins Univ, Baltimore, MD, USA  
Laure Rittié, PhD, University of Michigan, Ann Arbor, MI, USA  
Dennis R. Roop, PhD, University of Colorado, Aurora, CO, USA  
Jean Y. Tang, MD, PhD, Stanford Dermatology, Redwood City, CA, USA  
Joyce M. Teng, MD, PhD, Stanford University, Palo Alto, CA, USA

Thank you!
5. IPCRR—Comments, definitions, thoughts from PC Advocates 2016

- The International PC Research Registry that PC Project maintains. This registry keeps track of the PC patients that have been genetically confirmed. PC Project pays for new candidates to be tested for PC. This registry is helpful for patient life history as well. Having the registry makes it easier to obtain funding such as grants. PC Project is fortunate to have such a tool, but it takes time and money to keep it up.

- The patient registry which collects data about PC. Critical to the success of the IPCC so they can conduct research, clinical trials, and hopefully someday treatment or cure.

- A patient registry which collects PC data to establish what is really true about PC (especially to help scientists and doctors).

- A compilation of patients with PC, detailed information regarding the degree of involvement, treatments attempted, etc., all with the goal of compiling information to improve the understanding treatment of PC.

- The International PC patient registry for patients with genetically confirmed PC. The registry helps collect data and information on patients with PC which can be used to assist with research.

- It’s our own private club where you must have PC to get in! The purpose of the IPCRR is to register people with PC all with an eye toward growing the knowledge, profile and possibility of finding a cure for PC.

- Consortium of patients to help the physicians and scientists in their research to find me a cure or imminent relief. To raise money for studies, clinical trials, etc. There are approximately 1,600 individuals identified with PC worldwide and approximately 900 families.

Additional comments from PC Project

NOTE: The IPCRR includes a large number of patients who do not have PC. Through the IPCRR registry protocol we help patients obtain genetically-confirmed diagnoses. We have learned important diagnostic differences from many disorders that are mistakenly diagnosed or identified as PC. (See the correct statistics on the ‘Rare’ definition page.)

The IPCRR has been selected as one of the first participants in the NIH Global Rare Disease Registry (GRDR) and IPCRR de-identified data is now available online.
6. **Genetic/Genetics**—Comments, definitions, thoughts from PC Advocates 2016

- Pachyonychia Congenita (PC) is a genetic disorder. This means that a mutation or change in a gene causes the condition. In PC the mutation is found in any of five keratin genes KRT6A, KRT6B, KRT6C, KRT16 or KRT17.
- The study of inherited conditions understanding of genes so that treatment options may become more available.
- Inherited characteristics dictated by DNA.
- Having to do with the DNA that makes up who we are. In the case of us with PC, we have a mutation in our DNA (in a keratin gene) which unfortunately causes the expression of PC characteristics.
- 5 keratin genes that cause a hereditary or spontaneous mutation. PC is an autosomal disorder that has a 50/50 chance of being passed on.
- PC is a genetic disease caused by a mutation of one of 5-keratin genes. PC is a dominate genetic disease so a mutation in a single keratin gene will cause the symptoms of PC. A person with PC has a 50% chance of passing the gene on to their children. Approximately 40% of PC cases are caused by a spontaneous mutation.
- Of the genes. PC is a genetic disorder because the problem is in the makeup of certain keratin genes. This problem exists throughout the entire life of a patient from birth to death.

**Additional comments from PC Project**

Genetics is a major field of study of genes, genetic variation, and heredity in living organisms. Many of the 7,000 identified rare diseases are genetic diseases (caused by a mutation in one or more genes.) We are fortunate that the genes that cause PC have been identified. Many rare diseases do not yet have the genetic cause identified.

Genetics is much more than only PC. A very interesting, educational and user-friendly website is http://learn.genetics.utah.edu/ [NOTE: The information on PC needs to be updated!]

- **Autosomal** refers to the fact that the genes causing PC are not related to the genes that determine sex.
- **Dominant** means only one copy of the gene has the mutation for the disease signs to show.
- **Recessive** means that both copies of the gene have the mutation for the disease signs to show.
ADVOCATE AND PEER COACH SKILL BUILDING
GROWING OUR PC VOCABULARY

7. **Skin**—Comments, definitions, thoughts from PC Advocates 2016

- Organ often taken much for granted until conditions adversely affect it, and in the case of PC to the point where it can govern one’s life and activities.
- Ouch! Hurts me! Constant excruciating debilitating pain from my skin, my calluses, blisters, etc. on my hands and feet that makes me feel like such a FREAK!
- PC skin is hard, hurtful, and can be removed.
- Painful in certain areas for me and for other PC patients, especially on the bottoms of my feet. Although it affects each of us with PC differently, our skin is a challenge to deal with every single day.
- The stuff that covers bones, organs, muscles to form a body.
- Skin is made up of keratin. In a person with PC one of 5 (not 4) keratin genes have a mutation. The mutation of the keratin gene causes the skin (keratin) to be weaker. The body produces more keratin to compensate. This leads to thickened skin, ie callouses, blisters. This causes pain and limitations for people with PC.
- Where PC is located, and a PC Patient does not have the luxury of normal skin in the affected areas. Some patients have blisters and calluses on their feet and hands. Other patients have cysts all over their bodies.

**Additional comments from PC Project**

Quick! What's the body's biggest organ?

You might be surprised to find out it's the skin, which you might not think of as an organ. No matter how you think of it, your skin is very important. It covers and protects everything inside your body. Without skin, people's muscles, bones, and organs would be hanging out all over the place. Skin holds everything together. It also: protects our bodies; helps keep our bodies at just the right temperature; allows us to have the sense of touch. From a fun and educational website: [kidshealth.org/en/kids/skin.html](http://kidshealth.org/en/kids/skin.html)

There are many types of skin both inside and outside the body. For example, the skin on your palms and soles is different than the skin on your back or the skin that covers organs inside your body.
ADVOCATE AND PEER COACH SKILL BUILDING
GROWING OUR PC VOCABULARY

8. "Dr." — Comments, definitions, thoughts from PC Advocates 2016

- PC patients often need to educate the Dr that they see about the facts of PC. PC is ultra rare and many physicians have never seen or heard of PC. It is important for a PC patient to have a physician willing to listen and learn about PC. The PC patient needs a physician that will work with them as a team.

- It is also my job not to let that doctor do any useless and/or painful procedure because of ignorance or take any medications that I know from PC Project data would be a waste of my time.

- A person who most likely doesn't know anything about PC. That is why each patient needs to steer their doctors to the PC website.

- A healthcare professional with training in a specific field.

- Someone who obtains extensive training in medicine and who dedicates their life to trying to help people afflicted with whatever condition bothers them.

- My partner in crime who studies, researches, and ultimately finds the cure for PC.

- Someone who doesn’t need to initially know much about PC but who is willing to learn. Also, he or she very much needs to listen to me and trust that I know what I’m talking about. I am in charge of helping that doctor understand PC and how it affects me and my children with PC.

- HELPS ME!!! My doctors provide maintenance and relief to help me try to live a normal life. But, please doctors find me a cure!

Additional comments from PC Project
It is important for patients to really understand who they are interacting with when someone is called ‘doctor.’

- MD (Medical Doctor/Physician) may be a clinician or a scientist
- PhD (Scientist)
- DPM (Podiatrist)
- PA (Physician Assistant)
- A person who holds a doctorate—he was made a Doctor of Divinity (Minister)

NOTE: PC is a skin disease with pain as a main component. However, only recently did we learn that dermatologists do not normally treat pain or prescribe pain medications. Pain medications are more often prescribed by General Practioners, Internists, Pain Specialists (neurologists, anesthesiologists, etc.) treat pain.
ADVOCATE AND PEER COACH SKILL BUILDING
GROWING OUR PC VOCABULARY


- PC patients need to work together to form a patient voice. Each person and family affected by PC needs to work to spread awareness and educate others.
- ME!
- The collective experience of PCers shared with researchers, physicians, and others who suffer with PC.
- My voice.
- What I hope to be whenever I am asked about PC. I represent not only me, but all PC Patients and PC Project. As a voice, I need to be able to clearly and concisely share my story and the broader PC story in hopes that others can better understand PC and be motivated to help.
- What we patient advocates are intended to be. We will speak for patients at meetings and fund raisers.
- On an individual basis, the efforts of a patient to convey their medical concerns to a person or organization that can help them. Collectively, a group of patients or an organization representing them who function as a patient PC Advocate.

Additional comments from PC Project
This term has become very popular in the last ten years. There are now multiple organizations formed around this phrase. There is a new agency PCOR (Patient Centered Outcome Research) funded by Congress, but this is not focused on rare disease and our applications have not been successful due to the low impact.

None of the ‘patient voice’ momentum has or will make a difference for PC or for getting approval for PC clinical trials or raise funds for PC research…

_unless_ we have one unified ‘PC Advocate’ message from the PC community.

What is our message? What is our ‘ask’? What actions matter? Is our voice strong? Or weak? Who is our audience—funders, researchers, patients, friends, community, family, others…?
10. **Grand Rounds**—Comments, definitions, thoughts from PC Advocates 2016

- A medical meeting or seminar where experts in the field come together to examine and discuss difficult, challenging, or unusual patients.
- Training for doctors, typically an academic setting intended to help doctors learn more about specific medical conditions.
- A tool used for educating medical students and physicians. They are not made to help the patient, but to let students see a rare disease.
- A conference where doctors learn about PC by seeing it for themselves first-hand.
- An opportunity for a variety of PC patients to educate doctors and medical students about PC in one setting.
- A physician meeting or conference where a group of physicians invite patients. The physicians will examine the patient to learn more about PC.
- Patients participating and showing PC in a large setting to educate doctors, residents, researchers, etc. It can be awkward and embarrassing for me as a patient but so important to get the word out and show PC!

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*Additional comments from PC Project*

We need to schedule many more PC Grand Rounds events each with a group of PC patients and a knowledgeable speaker. We hope advocates will seek to interest educators in their areas to experience a Grand Rounds the PC way. (See SAMPLE Grand Rounds handout.)

We need to help patients to ‘just say no’ to any one patient Grand Rounds and especially any Grand Rounds using a single young child. When invited, be ready to connect with PC Project. We will fund a truly beneficial and educational Grand Rounds experience. Any one PC patient does not help the PC story — PC is a syndrome of conditions and understanding and recognizing the bigger picture of PC is an essential part of awareness and education.

Some PC Project Grand Rounds—Stanford/UC Davis/UC SF, Atlantic Derm Society, Tennessee Area Derm Society, Innsbruck/Austria Genodermatosis Training Conference, etc.
ADVOCATE AND PEER COACH SKILL BUILDING:

PC CARE—SHARING BEST TIPS
ADVOCATE AND PEER COACH SKILL BUILDING
PC Care—Sharing Best Tips

Awareness—Even with the same gene/mutation PC affects each individual differently.
Empathy—An Advocate/Peer Coach knows that each person feels ‘their way for care’ is the best.
Education and Resources—The PC website at www.pachyonychia.org is the most reliable source of information on PC and PC care. To keep the website constantly updated and relevant, we need you to both build (share your tips and ideas) and teach others using the available website tools and give us feedback.

On the dropdown list under Living With PC, there is a specific page “Caring For PC” that has information on PC Care with links to the Caring For PC Videos on the right side bar.

Facebook is not the best place to host good PC care information.

The PC Wiki (see next page for sample of PC Wiki) includes a lot of care tips from PCers. We collect this information from patient emails and calls, during patient support meetings and from the Facebook Patient Chat. Facebook is not the best place to host good PC care information.
Shoes and Sandals - Listed Alphabetically

**Abeo**

From *The Walking Company*:

Abeo® biomechanical footwear™ infuses advanced technology into classic shoe designs and is found exclusively at the Walking Company’s 200 plus nationwide stores and thewalkingcompany.com. When you visit any location of The Walking Company, we will utilize advanced scanning technology to digitally analyze your foot in order to help you maximize the benefits of Abeo®’s revolutionary footwear.

K16 patient: “In 2010 I discovered The Walking Store, particularly a shoe they carry referred to as the Abeo Smart System (generally I have used the sandal form of this shoe). This shoe has made a huge difference for me – I actually couldn’t wear the shoes I used to wear before I started wearing this shoe. I just wanted to pass this along and say that this is the most comfortable shoe I have found to date.”

**Bandals**

K16 patient: “Bandals. Fashion flip flops are so comfortable, they have a thick base and are very flexible. The best thing is that you can change the top to match your outfit. I live in Maine in the summer as they really help and being able to change the tops means you only need one pair!”

**Camper**

K16 patient: “Camper shoes and Camper women’s shoes have a spirit and personality of their own and present a diverse range of footwear that combines graphic design with imagination and functionality. To the senses, Camper shoes are unique, original and constantly a surprise. From the iconic Camper Pelotas range, to the socks in retro sneakers, to the asymmetrical Camper Turtles, that are playful and whimsical. Camper men’s shoes and Camper women’s shoes are filled...”
QUESTIONS FOR PC SPEED LEARNING
## QUESTIONS FOR PC SPEED LEARNING

<table>
<thead>
<tr>
<th>Questions Specialists Will Ask</th>
<th>Questions Patients Will Ask</th>
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<tbody>
<tr>
<td>What is the main thing you'd like to change about your PC and why?</td>
<td>How can I best help other specialists (doctor/nurse/researcher) learn about PC?</td>
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<tr>
<td>How does PC affect your life and quality of life?</td>
<td>How did you get interested in PC?</td>
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<tr>
<td>Why is PC Project important to you?</td>
<td>What motivates you to help PC Project and/or PC Patients when there are so many other diseases?</td>
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<td>When were you diagnosed? Are most people diagnosed early in life?</td>
<td>How does the IPCRR (registry) help research?</td>
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<td>What is the hardest thing about having a rare disease?</td>
<td>What is your biggest frustration in your scientific/medical world?</td>
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<td>On a scale of 0 to 10, what is your pain level right now (plantar pain)? How is your pain</td>
<td>Why is research on rare diseases so important?</td>
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<td>affecting you in this meeting today?</td>
<td>What is the most important thing I can do as a PC Advocate to move research forward?</td>
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<td>How could a physician help you most?</td>
<td>Why does it take so long does to get a new drug to market (available to patients?)</td>
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<tr>
<td>Are there any things that lower your pain or increase your pain?</td>
<td>What qualities makes PC Project unique from other organizations?</td>
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<td>What restrictions does PC put on your life?</td>
<td>If a treatment or cure is found for PC, how will patients be able to access such due to the high costs for rare disease treatments?</td>
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<td>How did you first hear/find PC Project?</td>
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OPPORTUNITIES FOR PC ADVOCATES / PEER COACHES
### Sign Up Sheet—PC Advocate Opportunities

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<tr>
<td>Clinical Studies / Clinical Trials</td>
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<td>Fundraising / Awareness Events</td>
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<td>Meetings and Events</td>
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<td>Webinars</td>
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<tr>
<td>Your Personal Participation Score</td>
<td>Please participate as often as possible with PC Project efforts. It makes a real difference!</td>
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CLINICAL STUDIES / CLINICAL TRIALS
To get a drug to market, there are many steps and a long road of discovery and testing. In order to prove effectiveness of a drug, we have to do observational studies to set proper endpoints.

CURRENT EXAMPLES
- Need responses on new nail removal addendum
- Need participation in sweat testing or other opportunities at PSM
- Need blood samples and/or biopsies for additional research study on itch
- Need many to use the pain app over a period of weeks/months to establish a base line
- Need K6a high pain participants in the Activity Tracker and Pain App Study with a normal control (same gender, close in age and location)
FUNDRAISING / AWARENESS EVENTS

We need all 8 PC Advocates to hold a 2016 Awareness Event with family, friends, community. We also need all 8 PC Advocates to post about their plans, their event and to contact and reach out and encourage everyone they know in the PC community to do something in this area. It is really important that we begin to motivate and activate the PC community in fundraising.
MEETINGS AND EVENTS
There may be future meetings, events or grand rounds that we may need PC Advocates to attend as representatives for PC Project. Also, if you hear of a grand round or other meeting/event that PC Project should have representatives at, please let us know.
PC NewsBrief

We would like to have each PC Advocate contribute to the PC NewsBrief. We can do this on a rotation basis—select the month you’d like to do a feature. Select what you know and would like to share. This can also be written in your native language.

One example: We think we could use quite a few short ‘meeting reports’ about your experience at the training meeting. Also, your experience at the IPCC meeting. Other ideas are welcome. We’ll be sure to have your submissions reviewed and edited as needed for correctness.
PC WEBSITE
The PC website is filled with information. In order to be able to direct others to the information is important that PC Advocates are PC website experts. You can help us make a plan on how to use/master the website, the key website features that help most patients, the things we need to improve. Also, an important role for PC Advocate is to let PC Project know about other websites that have information posted about PC as well as sites that need to add a link to the PC Project website. We also hope to eventual have an introductory page in each language for the website.
PEER COACHES
Selecting / Training PC Peer Coaches. We need your assistance as we begin to extend the Peer Coach program.

Peer Coaching. We will continue to work with PC Advocates to offer one-on-one coaching opportunities, to match your skills/interest with new patients.
SOCIAL MEDIA
This is an area where PC Advocates can make a major difference. We need all of your eyes, ears, comments, posts, likes and more. With eight PC Advocates sending the PC message and engaging others on social media, our ’reach’ to identify and help many patients will definitely increase, our research will be stronger and our goals will be more quickly achieved. See hints/guidelines in the Challenges section of this training manual.
Facebook Patient Chat Guidelines

This is a private group and is provided for those in the International PC Research Registry. It is a supportive environment where people affected by PC may connect, exchange information, find understanding and build friendships. We are committed to providing a safe, welcoming community. We encourage your comments, photos, videos, questions and links.

If you are interested in becoming a part of our community, we encourage you to like our Facebook organization page titled Pachyonychia Congenita Project.

1. PC is not one thing – it is a syndrome of conditions
Your experiences with PC may or may not be similar to others. If you are comfortable, please consider sharing your general PC type (PC-K6a, PC-K6b, PC-K6c, PC-K16 or PC-K17) with your comments. If you are not sure of your PC type, you are welcome to contact PC Project for this information.

Please also always remember that there are nearly 100 different mutations so that even within the general PC types, individual symptoms may differ quite a bit and what works for you may or may not work for others.

2. No medical advice
Posts on PC Patient Chat are for educational purposes only and are not for the purpose of rendering medical advice. The information presented is not intended to replace the counsel of your physician. PC Project does not endorse any medications, products, equipment or treatments.

Posts regarding specific drugs or medications or treatments (even ones that are ‘all natural’ or ‘over-the-counter’ medications) are not appropriate for PC Patient Chat. Those participating on PC Patient Chat may have special conditions so that a general recommendation of any drug or medication could be harmful to them. So, please no medical advice on PC Patient Chat. (If you have a need or you have a suggestion, contact PC Project so our Medical Advisory Board can be of help or review. Almost every drug or medication has ‘warnings’ attached that must be included with references to the item).

3. Respect
Please do not engage in abusive behaviour or conversations. We will remove comments that are harassing, aggressive, threatening, harmful, obscene, racially offensive, sexually explicit or otherwise inappropriate.

Please show respect for fellow members and for medical professionals and researchers. We are a team working together to find effective treatments.

PC Patient Chat is for personal use only. Posts that advertise, promote services or products, distribute unsolicited information or distribute information for financial gain will be removed. PC Patient Chat page is offered under general Facebook guidelines and is monitored by PC Project staff. We reserve the right to remove posts that violate the community and our guidelines. We must ban users who violate these guidelines.
WEBINARS

We want to host a series of informational webinars. PC Advocates can help (a) plan the topics (b) help us select the presenters (c) host the webinars which is a key to success (d) give us the wider options of various time zones.