



# Pachyonychia Congenita Project

*Fighting for a cure. Connecting & helping patients. Empowering research.*

## Understanding PC Project

### Training Session 2

# Pachyonychia Congenita Project

**USA = 501(c)3 Public Charity [2003]**

**Europe = Scottish Charitable Incorporated Org SCIO [2016]**

International PC  
Research Registry  
**IPCRR**

Patient Registry  
Genetic Testing

## **Mission Statement**

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# Discovering PAIN in Pachyonychia Congenita

## International PC Research Registry IPCRR

IPCRR - PC Project  
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### QUESTIONNAIRE

**21 pages  
+Images**

Thank you for being part of the IPCRR and for completing this questionnaire. Through the registry, information will be gathered so that physicians and other scientists can more effectively study PC and actively seek a cure for this disease.

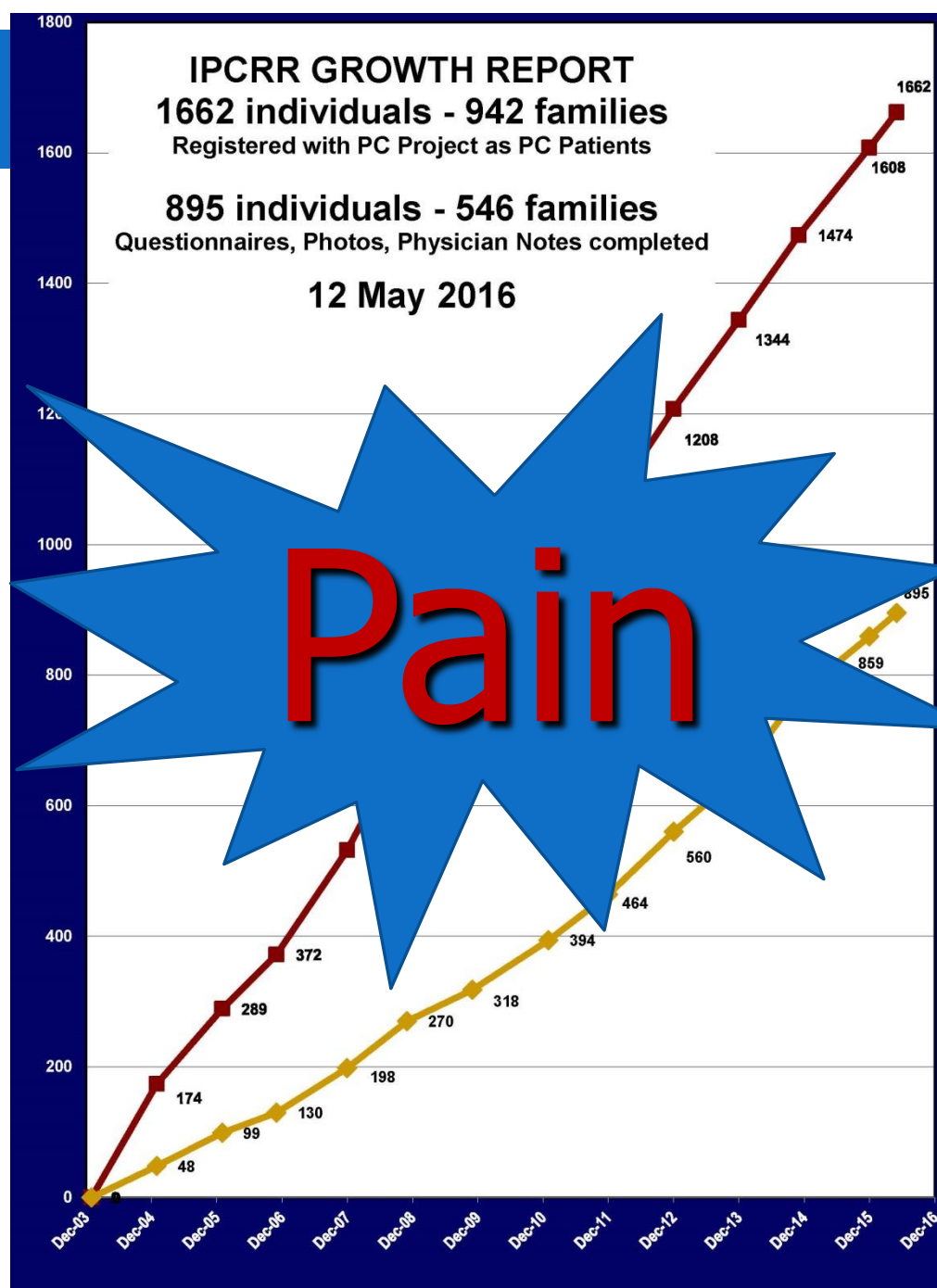
We believe it will take about one hour to complete the questionnaire. You may finish it all at once, or start and stop as you wish. **You may skip any question which you do not wish to answer or which is unclear to you.** You will have an opportunity to discuss your responses with a physician in your telephone consultation.

There are six sections in the Questionnaire.

|  |         |
|--|---------|
| Section 1. IDENTIFYING INFORMATION .....       | Page 2  |
| Section 2. DIAGNOSIS .....                     | Page 4  |
| Section 3. FAMILY HISTORY .....                | Page 6  |
| Section 4. PHYSICAL FINDINGS .....             | Page 7  |
| Section 5. TREATMENT AND CARE EVALUATION ..... | Page 17 |
| Section 6. PATIENT PHOTOGRAPHS .....           | Page 22 |

## International PC Research Registry 2004-2016

Discovered  
PAIN is the  
most common  
feature of PC



# International PC Research Registry IPCRR

## Genetic Testing

PC Project has obtained genetic testing for more than 1000 patients through the IPCRR. Tests cost approximately \$2000-\$2500.

Factual data is now available to researchers on those patients with confirmed PC.

IPCRR data available at [www.pachyonychia.org](http://www.pachyonychia.org)

| K6a          | Individuals | Families |
|--------------|-------------|----------|
| A463P        | 6           | 3        |
| c.541-2A>T   | 1           | 1        |
| c.1460-1G>C  | 1           | 1        |
| c.541-1G>C   | 9           | 3        |
| c.541-1G>T   | 1           | 1        |
| c.541-2A>C   | 2           | 2        |
| D432_E470dup | 1           | 1        |
| E163K        | 6           | 4        |
| E461K        | 3           | 2        |
| E461Q        | 2           | 1        |
| E461X        | 1           | 1        |
| E472D        | 4           | 2        |
| E472K        | 15          | 9        |
| E473GfsX91   | 1           | 1        |
| F174C        | 1           | 1        |
| F174I        | 2           | 2        |
| F174S        | 12          | 11       |
| F177del      | 2           | 2        |
| G461K        | 1           | 1        |
| G471_E472del | 1           | 1        |
| H67N         | 3           | 1        |
| H67S         | 1           | 1        |
| H78N         | 2           | 2        |
| H62F         | 1           | 1        |
| H62N         | 6           | 3        |
| K6a          | 1           | 1        |
| L170F        | 5           | 4        |
| L170P        | 1           | 1        |
| L170R        | 2           | 1        |
| L468P        | 7           | 5        |
| L469P        | 10          | 8        |
| L469R        | 4           | 3        |
| N171D        | 2           | 2        |
| N171H        | 1           | 1        |
| N171K        | 16          | 11       |
| N171S        | 20          | 9        |
| N171T        | 4           | 1        |
| N171Y        | 3           | 3        |
| N172del      | 70          | 46       |
| Q435X        | 2           | 1        |
| R164P        | 3           | 1        |
| R466P        | 2           | 2        |
| S176P        | 2           | 1        |
| S505QfsX50   | 1           | 1        |
| T464P        | 1           | 1        |
| V181_Q186del | 25          | 6        |
| Y465C        | 2           | 1        |
| Y465H        | 2           | 2        |
| Y465InsH     | 2           | 1        |
| Y465S        | 2           | 1        |
| Y468H        | 1           | 1        |
|              | 276         | 173      |

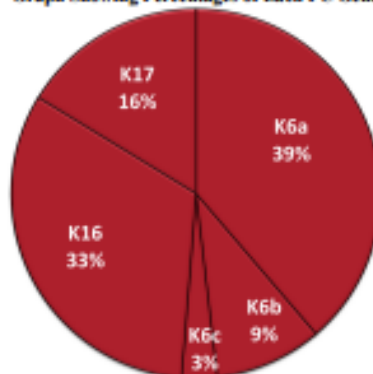
| K16                  | Individuals | Families |
|----------------------|-------------|----------|
| E413K                | 1           | 1        |
| K15X                 | 9           | 1        |
| L124_D126del         | 1           | 1        |
| L124_D126del & K134N | 1           | 1        |
| L124_N125delInsH     | 8           | 1        |
| L124H                | 11          | 4        |
| L124P                | 5           | 4        |
| L124R                | 3           | 2        |
| L126P                | 2           | 2        |
| L126Q                | 1           | 1        |
| L132P                | 29          | 16       |
| L421P                | 1           | 1        |
| M121K                | 4           | 2        |
| M121R                | 3           | 1        |
| M121T                | 9           | 5        |
| M353I                | 1           | 1        |
| N125D                | 6           | 3        |
| N125S                | 45          | 24       |
| R127C                | 55          | 17       |
| R127G                | 3           | 3        |
| R127H                | 1           | 1        |
| R127L                | 3           | 2        |
| R127P                | 6           | 5        |
| R127S                | 7           | 2        |
| R418_R419del         | 4           | 1        |
| R418P                | 4           | 2        |
| S12X                 | 1           | 1        |
| S130del              | 7           | 6        |
|                      | 231         | 111      |

| K6b          | Individuals | Families |
|--------------|-------------|----------|
| E461K        | 4           | 4        |
| E472K        | 40          | 11       |
| G429_A457dup | 1           | 1        |
| H178N        | 2           | 1        |
| L469R        | 1           | 1        |
| N172del      | 19          | 7        |
|              | 67          | 25       |

| K6c         | Individuals | Families |
|-------------|-------------|----------|
| E472K       | 10          | 4        |
| H62-E470del | 5           | 1        |
| L469P       | 2           | 1        |
| N172del     | 5           | 1        |
|             | 22          | 7        |

| K17            | Individuals | Families |
|----------------|-------------|----------|
| A66_L99del     | 1           | 1        |
| E111A          | 2           | 1        |
| L388P          | 7           | 3        |
| L91P           | 1           | 1        |
| L95P           | 4           | 4        |
| L95Q           | 3           | 1        |
| L99_K101del    | 1           | 1        |
| L99P           | 9           | 6        |
| M88K           | 3           | 2        |
| M88R           | 3           | 1        |
| M88T           | 6           | 3        |
| N90_D93delInsI | 3           | 1        |
| N92_L99del     | 1           | 1        |
| N92D           | 6           | 2        |
| N92S           | 49          | 27       |
| R94C           | 8           | 5        |
| R94H           | 1           | 1        |
| R94S           | 2           | 1        |
| S97del         | 2           | 2        |
| Y98D           | 4           | 1        |
|                | 116         | 65       |

Graph Showing Percentages of Each PC Gene



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International PC  
Research Registry  
**IPCRR**

Other services --

Patient Newsletters

Patient Support Meetings



## Mission Statement

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International PC  
Consortium  
**IPCC**

Collaborative research  
Publications  
Clinical Trials

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# IPCC

## Collaborative Research

### 9th Annual IPCC Symposium

Raleigh, NC

Publicize the disorder, Publish "Best Practice" Guidelines, Multi-center Clinical Studies  
Established YouTube Channel  
Published Articles



2012

IPCC 2013: Why are we here?



10th Annual IPCC Symposium  
Park City, UT NIH/NIAAMS-ORDR Grant  
Better understand mechanisms of PC  
Develop better objective endpoints  
IPCC Steering Committee formed  
Orphan drug designation for Topical Sirolimus  
FDA approval for Phase 1b Topical Sirolimus



2013

Publications  
Clinical Trials

### 11th Annual IPCC Symposium

Albuquerque, NM

Gather more basic science data on PC  
IRB approved PC Pain Study and clinical exam  
Section 1 - Paris, France (19 patients)  
Section 2 - Edinburgh, Scotland (36 patients)  
Biopsies from affected/unaffected PC skin collected  
Tape stripping samples collected for PC proteomic study  
Topical Sirolimus Phase 1b trial begins

2014

### 12th Annual IPCC Symposium

Atlanta, GA

Move forward to formal clinical trials  
IRB approved PC Pain Study and clinical exam  
Section 3 - Newark, NJ (47 patients)



Topical Sirolimus Phase 1b trial completed  
Three treatments selected for clinical trials  
Botox, Retinoids (new version), Topical Sirolimus



2015

  
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PROJECT



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