

Journal of
Health Psychology

Experiences of stigma over the lifetime of people with xeroderma pigmentosum: A qualitative interview study in the UK

Journal:	<i>Journal of Health Psychology</i>
Manuscript ID	JHP-17-0168.R1
Manuscript Type:	Article
Keywords:	STIGMA, skin condition, CHRONIC ILLNESS, QUALITATIVE METHODS, BELIEFS
Abstract:	This study explored experiences of stigma in 25 adults with xeroderma pigmentosum. Analysis of semi-structured interviews revealed the changing nature of stigma over the lifetime. Bullying occurred in childhood, whereas adults were questioned about both their photoprotection and skin damage, often resulting in internalised feelings of stigma. Resilience and rejection of feelings of stigma increased with age and experiences of stigma differed across cultures. Findings indicate a need to develop social skills training to help people reject feelings of stigma. Future research should explore perspectives of families, friends and formal institutions and their potential to cause or reduce feelings of stigma.

SCHOLARONE™
Manuscripts

•

Experiences of stigma over the lifetime of people with xeroderma pigmentosum: A qualitative interview study in the UK

Abstract

This study explored experiences of stigma in 25 adults with xeroderma pigmentosum. Analysis of semi-structured interviews revealed the changing nature of stigma over the lifetime. Bullying occurred in childhood, whereas adults were questioned about both their photoprotection and skin damage, often resulting in internalised feelings of stigma. Resilience and rejection of feelings of stigma increased with age and experiences of stigma differed across cultures. Findings indicate a need to develop social skills training to help people reject feelings of stigma. Future research should explore perspectives of families, friends and formal institutions and their potential to cause or reduce feelings of stigma.

Keywords

stigma, chronic illness, dermatology, adjustment, appearance

Introduction

Xeroderma pigmentosum (XP) is a rare genetic skin condition in which DNA damage from ultraviolet radiation (UVR) in daylight cannot be effectively repaired leading to pigmentary changes as a result of UVR exposure. XP is a heterogeneous condition and around half exhibit severe sunburn reactions (Sethi et al., 2013). There are increased risks of cancer, with an estimated 10,000 fold increase in the incidence of non-melanoma skin cancer and a 2000 fold increase in melanoma skin cancer before the age of 20 years and approximately 24% of people with XP develop neurodegeneration (Bradford et al., 2011).

People with XP are advised to minimise their exposure to UVR to reduce their chances of developing skin cancer, ocular problems and pigmentary changes. Personal photoprotective measures involve regularly applying sunscreen and covering up with long sleeves, trousers, gloves, glasses and hats with UVR protective face visors and film on windows also recommended.

This strict photoprotection regimen and the visible changes associated with XP have the potential to result in stigmatisation, based on the notion of unacceptable difference of a person or group of people (Goffman, 1963). Health-related stigma can involve both felt and enacted stigma. Felt health-related stigma is an internalised sense of shame relating to the condition or the fear of negative responses by others. Enacted stigma is the experience of being discriminated against or treated differently by others (Scambler, 1989; Jacoby, 1994) . Both can be equally damaging and felt stigma is likely to lead to concealment of the condition to avoid enacted stigma (Scambler, 2009).

There are numerous reasons why stigma may be an issue within XP. Stigma has been reported in other visible skin conditions, for example people with psoriasis thought other people perceived them as dirty and unhygienic (Bewley et al., 2014). However not all people with XP exhibit visible

1
2
3 pigmentary changes. For these participants the distinguishing photoprotection measures required
4
5 may make their condition more visible and therefore could lead to stigmatisation. XP is a rare
6
7 condition (around 2.3 per 1,000,000 live births in Western Europe; Kleiger et al., 2008) and so there
8
9 is little knowledge about it in the general public. This could make disclosure difficult as it would
10
11 require more explanation, and misunderstandings about the causes and severity could lead to
12
13 further stigmatisation.
14
15

16 Health-related stigma can have a significant detrimental effect on individuals, both psychosocially
17
18 and clinically. Stigma has been linked with reduced self-esteem in conditions such as chronic pain
19
20 (Holloway et al., 2007). Stigma can also lead to rational non-adherence whereby people consider the
21
22 costs of following stigmatising treatment regimens and choose not to adhere. Berger et al. (2011)
23
24 found some people with COPD used their inhaler less in public because of fears of stigmatisation.
25
26 Adherence to photoprotection measures is crucial in XP. For those who do not have
27
28 neurodegeneration a normal life expectancy is possible as long as photoprotection measures are
29
30 used. Without any photoprotection life expectancy is severely reduced.
31
32

33
34 To date there is no published research examining stigma in XP, nor any published qualitative study of
35
36 adults with XP. The aim of this study is to explore the range of experiences of stigma in adults with
37
38 XP and the strategies they use to reduce stigma and increase normalization.
39
40
41
42

43 **Method**

44 *Participants and recruitment*

45
46
47
48 Participants were recruited through the National XP Specialist Service at St Thomas' Hospital,
49
50 London. Eligibility criteria were: Adults aged 16 or over, with a laboratory proven diagnosis of XP
51
52 (reduced unscheduled DNA repair) without neurodegeneration, with adequate English to participate
53
54 and who had not opted out of participating in research. Children and those with neurodegeneration
55
56
57

1
2
3 were the focus of separate studies. Thirty-eight clinic attenders were identified as satisfying
4 eligibility criteria by a consultant dermatologist and contacted by the research nurse using letters,
5 phone calls and talking to patients in clinic. Of these six declined to participate (4 male, 2 female)
6 with reasons of lack of time and not wanting to think about XP and seven could not be contacted.
7
8
9 Twenty five interviews were therefore undertaken. Participants were not offered any incentives for
10
11
12
13 taking part.

14 15 16 *Procedure*

17
18 Interviews took place in participants' homes in a private room, with the exception of one phone
19
20 interview for logistical reasons. Two researchers carried out the one-to-one interviews. Neither had
21 XP but one has an occasional stutter that she disclosed. The interviewers were accompanied by the
22
23 research nurse who obtained informed consent, but was not present for the interview.
24
25
26

27
28 Interviews were based on a topic guide designed for a larger study with the aim of exploring
29
30 experiences of XP and adherence to photoprotection. Initial questions focussing on the participant's
31
32 personal story were used to build a rapport, followed by questions about psychosocial meanings of
33
34 XP, photoprotection and perceptions of risk (Supp Table 1). The topic guide directed the
35
36 conversation but allowed flexibility with interviews led by participants' experiences and responses.
37
38 Interviewers also prompted and probed as necessary to gain a clearer and more detailed
39
40 understanding. Following the interview, field notes documented a description of the participant and
41
42 reflections on the process and content of the interview.
43
44

45
46 The study received a favourable ethical opinion from Camden and Kings Cross Research Ethics
47
48 Committee 15/LO/1395.

49 50 *Analysis*

51
52
53 Interviews were audio-recorded with permission. They were transcribed verbatim, checked for
54
55 accuracy and entered into NVivo 10 software. All transcripts were read by three team members. A
56
57

coding framework was developed on the basis of team discussions and was added to and refined as new codes arose. Initially duplicate coding of the transcript data was undertaken. Following this one person (RA) completed the coding with the other two discussing and checking samples of coding on an ongoing basis.

Analysis involved both retrieval of coded segments and re reading individual transcripts to identify patterns and relationships for individuals and to compare across cases.

Findings

Characteristics of participants

The 25 participants were aged 16-63 years (mean=37.64 yrs, SD= 5.53). Seventeen were male and eight female, reflecting the preponderance of males in the adult clinic caseload (26 male and 17 female). Nine exhibited severe sunburn reactions ('burners') and 16 did not ('non-burners'). Twelve participants had a history of skin cancer. The sample included participants from a range of ethnicities were European (mainly white British), eight of Pakistani ethnicity and eight mainly of Middle Eastern, Indian or Bangladeshi background, reflecting the relatively high prevalence of XP in Pakistan and the Middle East (Fassihi, 2013).

Childhood experiences of stigma

Photoprotection began in childhood for 16 of the 25 participants of whom ten were formally diagnosed with XP as a child. underwent a strict photoprotection regime. The remaining six were 'burners' who although not diagnosed as children, avoided sunlight to prevent severe sunburn. As children, these participants knew they needed to protect themselves from sunlight but did not understand why. The six who experienced severe sunburn burners knew as children but were not diagnosed until adulthood, knew they needed to protect themselves from sunlight but not why this

1
2
3 was the case for them and not for others. Rather than describing specific worries about XP, accounts
4
5 of childhood indicated that a need for protection from UVR led to a general feeling of being different
6
7 to their friends. Whilst many had close friends as children, the things they did to protect from UVR
8
9 isolated them from others and drew unwanted attention. Participant 6 who burned from the age of
10
11 three described his experience:

12
13
14 everybody would go outside and the teacher would say you have to stay behind and put
15
16 cream on. She used to put cream on me, the teacher did. That's one thing that I remember.

17
18 The kids would look at me like what's going on there? (Participant 6, male, 20yrs)

19
20
21 Bullying from other children at school was common. People were teased about their freckles and
22
23 sunscreen, and one participant described having his hat stolen and kicked around. People also
24
25 reported receiving comments and stares from strangers, often adults, which added to feelings of
26
27 difference. Participant 12 who wore a UVR protective visor as a child described how reactions to this
28
29 from strangers affected him:

30
31
32 sometimes I did feel like crying and stuff like that, but other times I just put my head down
33
34 and walked on (Participant 12, male, 21yrs)

35 36 37 *Impact of diagnosis*

38
39
40 Childhood experiences of stigmatisation and feelings of difference became easier to cope with
41
42 following increased age and being given an official XP diagnosis of XP. As participants understood
43
44 their condition more clearly they found it easier to handle questions and could explain why their skin
45
46 was different. Participant 15 recalled:

47
48
49 I called it everything: allergy to the sun. Crazy names came out, like anything. Just
50
51 something to call it, it really puts you in a group and makes it easier. When people ask you
52
53 what's that mean, you say go Google it. Couldn't do that before, right? (Participant 15,

54
55
56 male, 34yrs)

1
2
3 Whilst receiving the diagnosis did not remove all stigma, it made it easier to manage and helped
4 participants feel less different within themselves by providing a sense of group identity. Participant
5
6
7 4, diagnosed at 31 years, was initially reluctant to get tested feeling he was already protecting
8
9 effectively but described how eventually visiting the XP clinic changed the way he saw himself:

10
11
12 I was just gobsmacked in a sense where, when I was at [the XP clinic], I'd seen other patients
13
14 there as well and obviously they've got different skin to myself. I thought I was, myself, my
15
16 sister, and my cousin were the only one probably that has it. I didn't really think of anyone
17
18 else in the world [...] I didn't feel different (Participant 4, male, 36yrs)

19
20
21 For the nine participants who did not have signs of XP as children, issues of stigma did not arise until
22
23 their diagnosis as adults. They were happy to have had a normal childhood without being separated
24
25 from peers or experiencing bullying with the diagnosis bringing the potential for stigmatisation that
26
27 they had not experienced before. Participant 9 explained:

28
29
30 the fact that I had to take care of myself so intensely made me feel like a sick person, which I
31
32 hadn't felt before (Participant 9, male, 38yrs)

33
34
35 However not all participants diagnosed as adults felt this way with the five diagnosed over the age of
36
37 40 years being less impacted. This group was more confident in how they saw themselves and
38
39 learning they had XP did not damage their self-concept. One participant who was diagnosed with XP
40
41 in her forties described how she found it easier to adapt to photo-protection:

42
43
44 because older, you're kind of stuck in your ways all these times. As I said, I don't feel,
45
46 besides I just want to slow down a bit, I don't think or feel differently (Participant 25, female,
47
48 55yrs)

51 *Adulthood*

52
53
54 *Experiences of stigma.* During adulthood, overt negative reactions were less frequent, but comments
55
56 and questioning about skin damage and photoprotection continued reflecting the continuing

1
2
3 requirement to engage in photoprotection which made the condition more visible and highlighted
4 participants' difference. The four participants who currently wore a visor spoke about people who
5 "take the mickey a bit" and experiences of comments, pointing and staring. Remarks and mocking
6
7 about other photoprotection measures were rare, but there were numerous accounts of
8
9 participants being asked why they were wearing thick clothes in summer and three participants (1
10
11 Caucasian and 2 Asian males) were stopped by police, suspicious that they were wearing hoodies in
12
13 summer.
14
15
16

17
18 In addition to distinctive photoprotection, increasingly visible skin damage from UVR exposure and
19
20 surgery scars further distinguished participants. These concerns occurred across all ethnicities, but
21
22 the negative social meanings and impact of skin damage was a particular issue for Asian participants.
23
24 Freckles are rare in Asian skin and drew more attention. As adults, although non-Asian participants
25
26 had concerns about visible skin damage, they rarely experienced enacted stigma. However Asian
27
28 participants described numerous experiences of direct comments and questioning about skin
29
30 damage from strangers, friends and family, making them feel different to others in the Asian
31
32 community. Participant 6, a young Pakistani man, avoided spending time with other Asian people
33
34 because of this and described the negative interactions he had encountered:
35
36

37
38 all I would hear is ah, are your freckles ever going to go away? How the hell are you, a
39
40 brown guy with freckles? They don't understand that. They associate freckles with
41
42 Caucasian people. They, I guess they try to make me feel like an outsider (Participant 6,
43
44 male, 20yrs)
45
46

47 Three participants reported that people assumed they would want to get rid of their freckles and
48
49 made suggestions of remedies to lighten their skin. This was frustrating because it implied blame, as
50
51 if they would do something about it if they wanted to:
52
53
54
55
56
57

1
2
3 people used to say oh, there's this cream. If you keep wearing it, they will fade. Your
4
5 freckles would fade, and I used to get angry with that, thinking what's wrong with me having
6
7 freckles? I don't want them to fade and how can freckles fade? [...] I'm not just being weird
8
9 and I'm not looking after myself; it's because I have a condition (Participant 2, female, 35yrs)

10
11
12 *Rejecting feelings of stigma.* Despite these common experiences, 13 of the 25 participants were
13
14 categorised as 'resilient' in the face of comments and questions. Although degrees of resilience may
15
16 have varied these were participants who may have experienced negative effects of stigma but had
17
18 learned or were learning to reject this and saw this as the other person's problem, not theirs. What
19
20 other people thought was therefore not as much a central part of their descriptions of experiences
21
22 of XP as it was for others. Some also felt stronger from overcoming the challenges of XP, and one
23
24 participant described his uniqueness as a positive:

25
26 I don't have a problem with XP. I have little or no stigma about it, because actually it's
27
28 interesting. I'm an interesting patient. It's almost reversed, almost pride (Participant 20,
29
30 male, 55yrs)

31
32 These participants compared themselves to others and saw that "*everyone is different*" and so could
33
34 normalise XP. Some had little visible difference from XP making this normalization easier, whereas
35
36 for others support from family and friends helped them to adapt and make positive social
37
38 comparisons. One participant described how a particularly severe operation helped her accept that
39
40 she will always look different and that "*people have to just accept that*".

41
42 Concerns about difference were less salient in the eight participants aged 45+. These participants
43
44 were more confident and so were less likely to worry about what other people thought. Those who
45
46 were diagnosed young had worried about what others might think or say in the past, but their
47
48 perspective had changed and they could now reject feelings of stigma. Participant 21 frequently
49
50 spoke about feeling sorry for younger people with XP. He was asked if he worried about what other
51
52 people thought about him:

1
2
3 it's a wrong question, because I'm 62. If you ask somebody, like 25, 20, it would be a
4
5 different answer. It's never bothered me. Maybe it has when I was young (Participant 21,
6
7 male, 62yrs)
8
9

10 Whilst adherence to photoprotection measures was not always high in these participants, they had
11
12 decided on a level of protection they were comfortable with and so did not worry about other
13
14 people's reactions. Whereas many younger participants were reluctant to act differently and draw
15
16 attention to themselves, one 55 year old described how others' opinions about his photoprotection
17
18 did not concern him:
19

20
21 I don't feel any anxiety about who I'm with or what I'm doing [...] If I'm with somebody and
22
23 they want to walk on the sunny side of the street I'll say no, I don't want to. I want to walk
24
25 on that side. So I don't think it affects my, I don't think my behaviour is affected by other
26
27 people (Participant 20, male, 55yrs)
28
29

30 There were also five younger participants who managed to reject feelings of stigma. They had dealt
31
32 with this since childhood and over time changed the way they responded. Participant 5 was teased
33
34 about her freckles throughout childhood but described how she stopped worrying about these
35
36 comments:
37

38
39 I used to think oh, why did I have freckles? If I didn't have them, it would've been better off
40
41 because no one would've said anything to me. But as I grew up and I started to think so
42
43 what, I've got freckles. Why should I listen to other people? They're just making me feel
44
45 bad about me (Participant 5, female, 26yrs)
46
47
48

49 However others who were also bullied as children were less successful in rejecting feelings of stigma.
50
51 In particular, five men aged 21-37 years reacted angrily to those who asked questions or made
52
53 negative comments and sometimes confronted them. This indicated that they did still care about
54
55 and were affected by what people thought or said. These participants were conflicted about how to
56
57

1
2
3 respond and this often depended on their mood at the time. Participant 12 would quietly ignore
4
5 looks and comments from others as a child, but as a young man he became angrier. He explained the
6
7 problems of responding in this way:

8
9
10 I just look back, straight, whatever. Like what are you looking at or something. Or if you
11
12 want to look, come on, let's look then. [...] In that moment it helps, but then you just think
13
14 through it after, I'm being looked at differently. Sometimes that just brings out the side in
15
16 yourself that you don't really, it's not really you (Participant 12, [male](#), 21yrs)

17
18
19 *Felt stigma.* The 12 participants who did not successfully reject negative responses from others were
20
21 more likely to internalise stigma and felt different even in the absence of comments from others.
22
23 Comparisons with others often led to the feeling that they were not 'normal'. This was not always a
24
25 result of worries about what other people thought but rather a self-stigma in the belief that they
26
27 were fundamentally different because of XP. Participant 7 described how XP could dominate his
28
29 thoughts and he would question why he couldn't be like everyone else:

30
31
32 why have I got XP and then why am I suffering from this and why can't I be normal, why
33
34 can't I go out. All of these questions, which are normal questions if you, I clearly mind
35
36 anyway. Because it's not normal to suffer from XP (Participant 7, [male](#), 27yrs)

37
38
39 Accounts of feeling different in adulthood were common and related to both skin damage and
40
41 photo-protection. Ten participants described persistent unwelcome feelings of difference because of
42
43 visible skin damage. Their main concern was that this damage would mark them out as different and
44
45 affect the way people saw them. Participants described questioning themselves during social
46
47 interactions and assuming that people were looking at their skin. When having surgery, the resulting
48
49 scars meant having "*another thing to hide*" which was more worrying than the pain and discomfort.
50
51 One participant was determined to keep her condition from her work colleagues to avoid being
52
53 treated differently, and so having facial surgery was particularly difficult for her:
54
55

1
2
3 my shoulder, they've done that; that was fine. They did my leg, that was fine. Not fine-fine,
4
5 but in comparison to having it on my face because again, on your face people can see it

6
7 (Participant 24, female, 28yrs)
8
9

10 For some it was frustrating to see skin changes, but this did not have a significant effect on their
11 confidence. However for others, appearance dominated their self-concept and reduced their
12 confidence. Participant 2 was bullied for her freckling as a child and described how the feeling of
13
14 needing to blend in and not being good enough continued into adulthood. She explained how her
15
16 visible difference skin damage affected how she saw herself:
17
18

19
20
21 I just want to be normal. I know I keep saying normal, and I am normal, but like I said when
22
23 you see other women, and you just think why can't I? You know, blemishes. Why am I so
24
25 different? I look odd. I just look odd (Participant 2, female, 35yrs)
26
27

28 Participants also spoke about feeling different because of their photoprotection. They worried that
29
30 they couldn't participate with others outside, and when they did they had to act "weird" by
31
32 reapplying sunscreen or dodging the sun. This was a particular issue for younger participants who
33
34 were more likely to be forming new relationships and avoiding time outside during daylight hours
35
36 limited these opportunities. Participant 12 did not experience enacted stigma at university, but
37
38 described how XP makes him feel different to other students:
39
40

41
42 everyone's always socialising and everything like that. So today, sunny days they'll always
43
44 be out on campus and everything and just relaxing in the sun. So obviously I can't do that.

45
46 So it's just about fitting in (Participant 12, male, 21yrs)
47
48

49 *Support and stigma-free spaces.* ~~Whilst feelings of difference were common in those participants~~
50 ~~who did not reject negative responses from others, there were situations where felt stigma was~~
51 ~~reduced.~~ Positive responses from those close to participants often reduced felt stigma for those who
52
53 did not reject negative responses from others and created safe, stigma-free environments. Family
54
55
56
57
58

1
2
3 and friends were described as providing emotional support but more importance was placed on
4 their role in normalising XP by understanding and incorporating photo-protection into normal life.

5
6
7 For example, participant 22 was highly adherent to photoprotection, planning all of his activities
8 around this and also described how his friends would do the same, without making it feel like an
9 imposition:
10
11

12
13
14 they didn't even think a second thing about it. It's like we don't go out until it's sunset.

15
16 That's it. Or do you want to come here? Things they are organising, like dinners for their
17 birthdays. It's like right, we'll go out at 9:00 and then you can. It's not like oh, then you can
18 come. It's like we'll go at this time. They don't need to say why or... I think that's just good
19
20
21

22 friends (Participant 22, male, 26yrs)
23
24

25 Another source of normalization was formal institutions such as schools, workplaces and hospitals.
26 For example having window film installed at schools and workplaces meant UVR protection did not
27 need to be worn inside. However four participants described unsupportive workplaces. Some felt
28 they had missed out on employment opportunities because of their XP and employers did not
29 always act on requests for adaptations to reduce UVR exposure. Participant 18 was reluctant to ask
30 for time off work for appointments relating to XP:
31
32
33
34
35
36
37

38 they're just money-making strategies coming through and whatnot. They really don't care.

39 And even when you tell them you've got to drop to a hospital appointment, they're like ah
40
41
42

43 (Participant 18, 31yrs)
44
45

46 The XP clinic was an important formal resource providing both emotional support and practical
47 solutions such as cards to help explain XP to others and tinted sunscreen that is less noticeable than
48 standard sunscreen as it blends in more easily. Participant 2 described feeling like an outsider
49 throughout her interview, but explained that she didn't feel this way at the XP clinic:
50
51
52
53
54
55
56
57

1
2
3 There are other people, so you're not, you are different, but you're not so different. So it
4 brings, it felt nice, and obviously the XP team, they obviously deal with XP so you don't feel
5 ashamed, different. You don't feel that I can't say this and I mustn't say that (Participant 2,
6
7
8
9 female, 35yrs)

10
11
12 *Managing stigma and normalization*

13
14 *Concealing visible difference.* The participants described wanting to reduce visible skin damage and
15 fears about future skin changes. The use of clothing, make-up or specialist camouflage to cover skin
16 damage was described by seven participants and improved confidence in social interactions.
17 However four of these participants had internalised stigma and for them concealing their difference
18 was a necessity. They would not go out without make-up or camouflage because of worries about
19 their appearance and so these routines added to the everyday burden of XP.
20
21
22
23
24
25
26
27

28 As well as covering skin damage, all participants were keen to minimise the visible difference of
29 photo-protection. This sometimes meant utilising practical adaptations like tinted sunscreen, but
30 often led to reductions in protection. For some participants who struggled to reject feelings of
31 stigma, the use of this strategy was inconsistent due to uncertainty about how much protection to
32 use. Photoprotection can be stigmatising, but it allows people with XP to participate with others
33 without worrying about UVR exposure and can help prevent visible skin damage. There was
34 therefore a conflict between avoiding visible damage and the associated stigma, avoiding attention
35 from wearing photoprotection and increasing participation with others. Participant 22 who wore a
36 visor which regularly received negative attention described conflicting concerns:
37
38
39
40
41
42
43
44
45
46

47 it's just looking different. Then at the back of it, so knock-on effect, you're worried about the
48 skin changes it'll make if you don't wear all that. Then there's particularly what women will
49 think of you (Participant 22, male, 26yrs)
50
51
52
53
54
55
56
57
58
59
60

1
2
3 *Managing disclosure.* Differing approaches to disclosure of the XP diagnosis were found within the
4 sample. Some described limiting stigma by avoiding disclosure wherever possible. Their close family
5 knew and some told employers if they felt it was necessary, but they did not discuss it openly. They
6 argued that they “*don’t really think it’s any of their business*” and that people wouldn’t understand.
7
8 Not telling others meant they would be treated the same as everyone else, but required effort to
9 conceal their XP. Another smaller group were more open, but struggled to decide who to tell and
10 how much information to give.
11
12
13
14
15
16
17

18 These approaches were mostly adopted by participants who had internalised stigma, although three
19 participants who rejected feelings of stigma were also selective in their disclosure. They did not hide
20 their XP, but brushed off questions with simplistic explanations of being ‘allergic’ or ‘sensitive’ to sun
21 unless they felt close to the person.
22
23
24
25
26
27

28 There was a group of twelve other participants who were open about their XP. These participants
29 saw openness as the best strategy to reduce potential stigma, as they believed that if people
30 understood more about XP they would not judge the way they look or the steps they take to protect
31 themselves. One participant explained that his friends knowing about his XP means they don’t
32 question his behaviour:
33
34
35
36
37

38 because most of my friends know about my skin condition, they just brush it off like yeah,
39 that’s fine. As long as you’re safe, we’re happy to chill out and let you put [suncream] on
40
41
42

43 (Participant 19, male, 18yrs)
44
45

46 Some were proactive in telling people about XP to increase awareness, but mostly these participants
47 did not give much thought to whether to disclose their XP. Whilst they did not specifically tell
48 people, they were happy to discuss it if asked. This was generally a successful approach which had
49 the benefit of not drawing unnecessary attention to themselves but also avoided the burden of
50 hiding their condition.
51
52
53
54
55
56
57

Discussion

This is the first study to explore experiences of stigma among people with XP and showed feelings of unacceptable difference to be widespread. Most studies of stigma focus on a single point in time, whereas this study identified changes in experiences of stigma from childhood, through diagnosis and adulthood, and compared responses to stigma across younger and older participants.

Participants' accounts revealed that the nature of stigmatisation changed significantly over their lifetime. Childhood was the time of the most direct enacted stigma including accounts of bullying. In adulthood it was mostly questioning rather than direct comments, and felt stigma relating to concerns about being different was more salient. Whilst studies interviewing children with other visible conditions have found these types of childhood-specific stigma issues (e.g. Bogart, 2015), the current study also accessed pre-diagnosis accounts of stigma in childhood and how this changed following diagnosis. For those who had experienced adverse reactions to UVR and needed to photoprotect from childhood a diagnosis of XP reduced stigma by giving an explanation for their difference and a group identity. Conversely for those diagnosed under the age of 40 who had not previously experienced adverse impacts of XP, diagnosis generally brought the potential for a stigma they had not previously experienced. Early diagnosis and support at this time are however key to both reduce skin cancers and also to provide formal assistance to young adults, families and schools to help them understand XP and create safe, stigma-free environments.

As adults, distinctive skin lesions and appearances associated with XP combined with the unique photoprotection measures required, resulted in multiple experiences of stigma. Whilst visible skin changes are the main source of stigma in most skin conditions, our findings showed that in XP, photoprotection measures were often the principale l-sign of difference to themselves and others. This often led to 'rational' non-adherence, consistent with previous research in chronic hand eczema where patients were embarrassed to use protective measures such as gloves in public (Mollerup et

1
2
3 al., 2013). Compared to non-adherence in other skin conditions the consequences can be much
4
5 more severe in XP due to the high skin cancer risk. Additionally, non-adherence to photoprotection
6
7 can conflict with other attempts to reduce stigma by using photoprotection to increase UVR-safe
8
9 participation with others and prevent further visible skin changes. The relationship between
10
11 photoprotection adherence and stigma is therefore complex and varied within this heterogeneous
12
13 group.
14
15

16 ~~It was clear throughout the findings that increasing age reduced feelings of difference.~~ The
17
18 qualitative approach of the current study gave insights into current experiences of middle-aged and
19
20 older people with XP and identified how increased age reduced stigma self-consciousness, adding
21
22 further depth to previous quantitative findings of reductions in stigma with age in conditions such as
23
24 HIV (Emlet et al., 2015). Whilst participants of different ages in our sample rejected stigma, this was
25
26 more common in those over 45 years old who also appeared more comfortable with their level of
27
28 photo-protection and confident in their self-concept. These participants were more likely to engage
29
30 in positive reappraisal of their situations, a coping strategy which has been found to support
31
32 resilience and positive adjustment in people with visible differences (Egan et al., 2011).
33
34
35

36 In addition to age differences in experiences of stigma, the current study revealed significant ethnic
37
38 differences. Asian participants experienced more enacted stigma than non-Asians and were regularly
39
40 made to feel like outsiders in their communities because of their freckling. Cultural factors that
41
42 contribute to stigma were also found in a study interviewing South Asian women with vitiligo
43
44 (Thompson et al., 2010). The current study adds to this literature, and by exploring cultural
45
46 differences within the wider context of stigma, insights into experiences of stigma common across
47
48 different cultures were not lost.
49
50

51 We interviewed over half of adults without neurodegeneration on the register of the only XP
52
53 specialist centre in the UK, suggesting a strong basis for research and practice recommendations
54
55 within the UK. A strength of the study was its flexible, qualitative approach which allowed cultural
56
57

1
2
3 differences within the UK that emerged during interviews to be explored. However there may be
4
5 cross-cultural differences in experiences of stigma within the international XP population with
6
7 ~~Prevalence of XP is considerably higher in the Middle East than Western Europe (Fassihi, 2013).~~
8
9 ~~Cultural~~ practices of covering up with clothing and societal preferences for lighter skin tones
10
11 affecting patients' and the public's perceptions of both visible skin changes and steps taken to
12
13 protect from UVR. There is therefore a need for further international research to explore
14
15 experiences of stigma across cultures.
16
17

18
19 A further limitation was that accounts of stigma in childhood depended on participants' recollections
20
21 which may not be entirely accurate. This approach had the benefit of accessing accounts of changing
22
23 experiences of stigma across the lifetime, however specifically interviewing children with XP about
24
25 their current experiences would add valuable insights to this narrative.
26
27

28
29 Our findings suggest a need to develop interventions to address stigma in order to aid adjustment in
30
31 XP. Aiming this at young adults may be particularly valuable as this appears to be a key time when
32
33 people with XP begin to either internalise or reject stigma. Moss (2005) found that a high perceived
34
35 ~~appearance-severity of a in patients with~~ disfiguring conditions on appearance was a predictor of
36
37 poor adjustment. He suggested that this may in part be due to a lack of social skills causing further
38
39 negative reactions from others which are then internalised. Social skills training could therefore be
40
41 beneficial to help people cope with enacted stigma, increase resilience and aid with disclosing their
42
43 condition to others. Our findings supplement previous research which showed that whilst openness
44
45 and disclosure of conditions can at times risk increased stigma, it is often a positive experience that
46
47 increases access to social support (Kent, 2000; Sayles et al., 2007). An online programme consisting
48
49 of social skills training and exposure-based techniques reduced appearance concerns, anxiety and
50
51 depression in people with visible differences (Bessell et al., 2012). Given the geographical spread of
52
53 patients, these sorts of online resources would be appropriate for use in XP.
54
55
56
57
58
59
60

1
2
3 Participants' accounts highlighted the potential for family, friends and formal institutions to cause
4 feelings of stigma as well as their role in reducing stigma by normalizing XP and the associated
5 photoprotection. This suggests that these groups should also be the focus of interventions. Research
6
7 aimed at gaining the perspective of potential sources of stigma and support could inform
8
9 interventions and campaigns to educate people and reduce stigma, rather than putting the emphasis
10
11 wholly on individuals to reject stigma.
12
13
14
15

16 Whilst reducing instances of enacted stigma and helping people reject feelings of stigma should be
17
18 priorities, it is likely that people with XP will still want to reduce the visibility of their
19
20 photoprotection. Any efforts to increase adherence must address feelings of stigma whilst allowing
21
22 patients to make informed decisions about risk and the level of photoprotection they are
23
24 comfortable with.
25
26
27
28
29

30 **References**

- 31
32
33 Berger BE, Kapella MC and Larson JL. (2011) The experience of stigma in chronic obstructive
34
35 pulmonary disease. *Western Journal of Nursing Research* 33: 916-932.
36
37
38 Bessell A, Brough V, Clarke A, et al. (2012) Evaluation of the effectiveness of Face IT, a computer-
39
40 based psychosocial intervention for disfigurement-related distress. *Psychology, Health and*
41
42 *Medicine* 17: 565-577.
43
44
45 Bewley A, Burrage DM, Ersser SJ, et al. (2014) Identifying individual psychosocial and adherence
46
47 support needs in patients with psoriasis: A multinational two-stage qualitative and
48
49 quantitative study. *Journal of the European Academy of Dermatology and Venereology* 28:
50
51 763-770.
52
53
54
55
56
57
58
59
60

- 1
2
3 Bogart KR. (2015) People are all about appearances: A focus group of teenagers with Moebius
4
5 Syndrome. *Journal of Health Psychology* 20: 1579-1588.
6
7
8 Bradford PT, Goldstein AM, Tamura D, et al. (2011) Cancer and neurologic degeneration in
9
10 xeroderma pigmentosum: Long term follow-up characterises the role of DNA repair. *Journal*
11
12 *of Medical Genetics* 48: 168-176.
13
14
15 Egan K, Harcourt D, Rumsey N, et al. (2011) A qualitative study of the experiences of people who
16
17 identify themselves as having adjusted positively to a visible difference. *Journal of Health*
18
19 *Psychology* 16: 739-749.
20
21
22 Emlet CA, Brennan DJ, Brennenstuhl S, et al. (2015) The impact of HIV-related stigma on older and
23
24 younger adults living with HIV disease: Does age matter? *AIDS Care - Psychological and*
25
26 *Socio-Medical Aspects of AIDS/HIV* 27: 520-528.
27
28
29 Fassihi H. (2013) Spotlight on 'xeroderma pigmentosum'. *Photochemical and Photobiological*
30
31 *Sciences* 12: 78-84.
32
33
34
35 Goffman E. (1963) *Stigma: notes on the management of spoiled identity*, Englewood Cliffs, NJ
36
37 Prentice-Hall.
38
39
40 Holloway I, Sofaer-Bennett B and Walker J. (2007) The stigmatisation of people with chronic back
41
42 pain. *Disability and Rehabilitation* 29: 1456-1464.
43
44
45 Jacoby A. (1994) Felt versus enacted stigma: A concept revisited. Evidence from a study of people
46
47 with epilepsy in remission. *Social Science and Medicine* 38: 269-274.
48
49
50 Kent G. (2000) Understanding the experiences of people with disfigurements: An integration of four
51
52 models of social and psychological functioning. *Psychology, Health and Medicine* 5: 117-129.
53
54
55
56
57
58
59
60

1
2
3 Kleijer WJ, Laugel V, Berneburg M, et al. (2008) Incidence of DNA repair deficiency disorders in
4
5 western Europe: Xeroderma pigmentosum, Cockayne syndrome and trichothiodystrophy.
6
7 *DNA Repair* 7: 744-750.
8
9

10 Mollerup A, Johansen JD and Thing LF. (2013) Knowledge, attitudes and behaviour in everyday life
11
12 with chronic hand eczema: A qualitative study. *British Journal of Dermatology* 169: 1056-
13
14 1065.
15
16

17 Moss TP. (2005) The relationships between objective and subjective ratings of disfigurement
18
19 severity, and psychological adjustment. *Body Image* 2: 151-159.
20
21

22 Scambler G. (1989) *Epilepsy*, London: London : Tavistock / Routledge.
23
24

25 Scambler G. (2009) Health-related stigma. *Sociology of Health and Illness* 31: 441-455.
26
27

28 Sethi M, Lehmann AR, Fawcett H, et al. (2013) Patients with xeroderma pigmentosum
29
30 complementation groups C, e and v do not have abnormal sunburn reactions. *British Journal*
31
32 *of Dermatology* 169: 1279-1287.
33
34

35
36 Thompson AR, Clarke SA, Newell RJ, et al. (2010) Vitiligo linked to stigmatization in British South
37
38 Asian women: A qualitative study of the experiences of living with vitiligo. *British Journal of*
39
40 *Dermatology* 163: 481-486.
41
42
43
44
45
46
47
48
49
50
51
52
53
54
55
56
57
58
59
60

Acknowledgements

We most grateful to the 25 participants who contributed to this study and to Lesley Foster who was instrumental in the recruitment of participants and facilitation of the interviews. We also thank the specialist XP team at St Thomas' Hospital and particularly Tanya Henshaw and Sally Turner for helpful discussions. Thanks are also due to the PPI group for their ongoing involvement and to the programme leads, Robert Sarkany and John Weinman,

Funding

This article presents independent research funded by the National Institute for Health Research (NIHR) under its Programme Grants for Applied Research scheme (RP-PG-1212-20009). The views expressed are those of the authors and not necessarily those of the NHS, the NIHR or the Department of Health.

Declaration of Conflicting Interests

None declared

Supplementary Table 1: Extracts of themes and questions from topic guide**1. Participants personal story of their XP (brief introduction)**

Can you tell me when you first became aware of having a skin problem and what happened?
When was this problem first formally diagnosed as XP?

2. Psychosocial meanings of XP**Nature of the burden**

It will be really helpful to understand what are the main ways it effects your activities, such as work/school/leisure activities/relationships? Have you found ways to reduce this impact over time?

Do other people understand/support you – in what ways?

Meanings and emotions

Going back to when XP was diagnosed – what was your understanding of XP at that time? How did you feel about having this condition? How do you feel about it now? Has the way you think about XP changed since you were diagnosed?

How does XP effect how you see yourself more generally, if at all? Do you see yourself in a different way because of your XP?) Has this changed at all over time?

I'm wondering if you feel different to other people who don't have XP or not? How? Has this changed at all over time?

Biggest worries about having XP?

(restrictions on activities, feeling different, fears about reactions of others, experience of social isolation/exclusion, fear of cancer, uncertainty about the future?)

Do you worry about what others think about you because of your XP?(family, friends strangers) Have you had any experiences that have made you feel this way or not
What things do you do when you feel this way?

3. Protection practices

Ask about each type of photoprotection practice (sun cream, visors, clothing to cover up and not going out in daylight and adapting environment with film on windows etc)

What do they do, how important they think it is, what are their experiences and any reasons why you might not do your UV protection?

Assistance and facilitating by others?

Have there been times in the past when UV protection has been harder/easier? Have you changed the different types of things you do over time?

Before we end is there anything else that you'd like to add that is important for me to understand what it is like living with XP?