

# Lived experience of Silver-Russell syndrome: implications for management during childhood and into adulthood

Lisa Marie Ballard,<sup>1</sup> Elizabeth Jenkinson,<sup>2</sup> Christopher D Byrne,<sup>3,4</sup> Jenny C Child,<sup>5</sup> Justin Huw Davies,<sup>6</sup> Hazel Inskip,<sup>7,8</sup> Oluwakemi Lokulo-Sodipe,<sup>9,10</sup> Deborah J G Mackay,<sup>11</sup> Emma L Wakeling,<sup>12</sup> I Karen Temple,<sup>10,13</sup> Angela Fenwick<sup>14</sup>

For numbered affiliations see end of article.

## Correspondence to

Dr Lisa Marie Ballard, Faculty of Medicine, Clinical Ethics and Law, University of Southampton, Southampton General Hospital, Southampton SO166YD, UK; l.ballard@soton.ac.uk

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## ABSTRACT

**Objective** There is limited information on the psychosocial impact of growing up with Silver-Russell syndrome (SRS), characterised by slow growth in utero leading to short stature in adulthood. Such information could aid families in making difficult treatment decisions and guide management strategies for health professionals. We aimed to explore the lived experience of people with SRS across the lifespan.

**Design/setting/patients** In-depth, semi-structured interviews were conducted between January 2015 and October 2016 with a sample of 15 adults (six women) with genetically confirmed SRS from the UK. Qualitative interviews were transcribed and coded to identify similarities and differences: codes were then grouped to form overarching themes.

**Results** Four themes were identified from participant accounts: (1) appearance-related concerns extending beyond height; (2) strategies to deal with real and perceived threats; (3) women's experiences of pain, disability and feeling older than their years; and (4) feeling overlooked in romantic relationships. These themes show that other factors, beyond short stature, affect patient well-being and indicate a mismatch between patient need and healthcare provision.

**Conclusions** Challenges in SRS during childhood and adolescence were central to the psychosocial impact of SRS, and were not limited to height. These challenges, as well as symptoms such as pain and fatigue for women, have not previously been documented. To help individuals with SRS develop strategies to manage psychosocial issues, we recommend clinicians incorporate psychological services as an integral part of multidisciplinary teams managing individuals with SRS during childhood, adolescence and adulthood.

## INTRODUCTION

Silver-Russell syndrome (SRS) is a multisystem disorder affecting 1:30 000–100 000 children, characterised by growth failure that begins prenatally and typically leads to short stature in adulthood. Known genetic causes include errors of genomic imprinting at chromosome 11p15 and maternal uniparental disomy of chromosome 7, but 40% of clinically diagnosed cases do not demonstrate any genetic anomaly.<sup>1</sup>

Children with SRS require multidisciplinary care for problems including severe feeding difficulties, hypoglycaemia, body asymmetry<sup>2</sup> and poor muscle

## What is already known on this topic?

- ▶ The management of Silver-Russell syndrome (SRS) during childhood has a major focus on optimising height during childhood and final height.
- ▶ Psychosocial evaluation is not routinely offered to children and young people in clinics.
- ▶ Appearance-related concerns result in psychological distress for some individuals.

## What this study adds?

- ▶ Individuals with SRS have significant psychosocial concerns throughout their lives especially during childhood and adolescence.
- ▶ Appearance-related concerns were not just related to height and may lead to difficulties forming romantic relationships for young people with SRS.
- ▶ Pain and disability affected female participants in particular, which impacted on family relationships and employment.

function.<sup>3</sup> Management during infancy usually focuses on feeding difficulties, prevention of hypoglycaemia and the avoidance of nutritional insufficiency. In older children, a key management goal has been to increase height during childhood and final height prognosis by optimising nutrition and administering recombinant growth hormone (GH). Individuals with SRS are typically discharged to primary care when at final height.

In short stature conditions, there has been a focus on the influence of height on psychosocial outcomes, but interpretation of findings may be complicated by the influence of associated problems. Studies of the lived experience of Turner syndrome<sup>4 5</sup> and achondroplasia,<sup>6 7</sup> both associated with short stature, showed adverse psychosocial issues in children and adults, including lower quality of life, depression, low self-esteem, difficulty forming relationships and social isolation. Of interest, height was not associated with quality of life scores in young women with Turner syndrome,<sup>8</sup> and a systematic review showed that GH treatment may not ameliorate psychosocial problems.<sup>9 10</sup>

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To date, psychosocial outcomes have not been studied in SRS. Childhood management focuses on height, but there are no long-term data on the outcome of this management strategy, or on the challenges faced by adults and their perceptions of health priorities. Such information will be useful to paediatricians, healthcare professionals and parents to inform management priorities for children and young people with SRS. Obtaining these data, however, is challenging given the rarity of SRS and the difficulty in identifying adults with SRS.

We undertook a qualitative research study using semi-structured in-depth interviews, with the aim to characterise the lifelong experiences of people with SRS. Qualitative research explores the meaning and complexity of a phenomenon that cannot be adequately explained numerically; a qualitative methodology is best placed to understand what it is like to live with a particular condition or to have experienced a certain phenomenon.<sup>11–13</sup> In this study, qualitative methods were chosen to add the patient voice to the objective and clinical literature, providing a fuller picture of the lived experience of SRS, and the effect of childhood management strategies on adult outcomes.

## METHODS

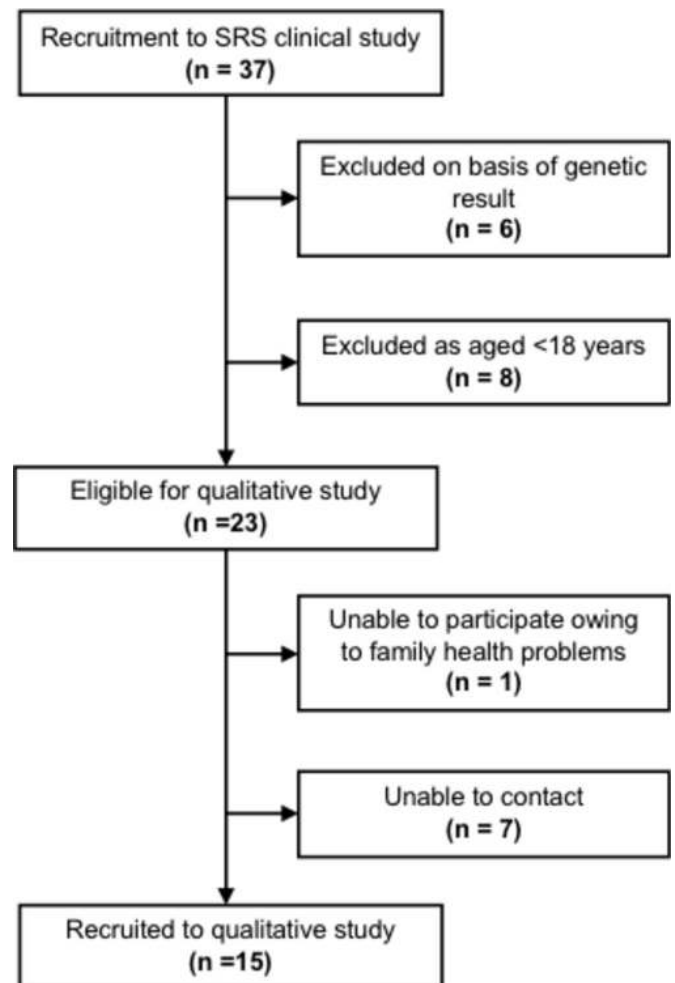
### Identification of adults with SRS

This study formed one limb of a broader study investigating the adult health consequences of SRS. Research and development approval was first gained at University Hospital Southampton NHS Foundation Trust (study sponsor) and then for the 23 UK Genetics Centres via the NIHR UK Rare Genetic Disease Research Consortium Agreement ('Musketeers' memorandum'). Individuals were identified across the UK using different approaches: (1) study information was disseminated through a patient support group: the Child Growth Foundation, UK; (2) individuals with SRS who had been recruited to an existing national study 'Imprinting Disorders: finding out why' and had expressed an interest in further research; (3) positive molecular genetics tests at the Wessex Regional Genetics Laboratory were reviewed; (4) regional genetic centres; and (5) paediatric endocrine centres at existing genetics research sites were contacted.

Using the first two approaches, participants were sent study information by post and invited to contact the study team or were contacted by telephone if they had previously given permission. For the latter three approaches, the individual's overseeing clinician was contacted and provided with study information to post to their patient.

### Study interviews

Interviews were chosen to collect data as they allow the researcher to: focus on drawing out individual experiences; explore specific concerns or issues; be flexible about the wording of questions; and add questions about relevant topics that arise.<sup>13</sup> As SRS is a rare condition and individuals may be known through support groups, robust confidentiality measures were instituted, including numerical participant identification and the use of age ranges (rather than specific ages). The interview schedule was constructed to address the impact of SRS on key stages in a participant's life (education, work, friends, family, intimate relationships). Each interview took between 60 and 90 minutes and was audio recorded. A trained female researcher (LMB) experienced in discussing sensitive health issues with patients in a healthcare setting conducted the interviews. Although LMB met some participants at a clinical appointment prior to interview, it was made clear in the patient information sheet and in verbal discussions that the study was to explore what it is like to live



**Figure 1** Source of recruitment to qualitative study. SRS, Silver-Russell syndrome.

with SRS. The study team had no preconceived ideas as to the nature of experiences participants would report.

### Analysis of study interviews and thematic identification

Interviews were transcribed and analysed thematically using the well-established Braun and Clarke method.<sup>11</sup> Tentative codes were generated from the initial transcripts and interviews were analysed as they were conducted, allowing the codes to be refined. Codes were grouped to create categories, and were developed into four overarching themes. To ensure rigour in relation to developing themes, coding and categories were independently reviewed by three researchers (LMB, AF and EJ). Tracy's criteria were used to enhance the validity and overall quality of the study.<sup>14</sup> NVIVO software (QSR International, V.11.3.2 (1888) for Mac) was used to manage the qualitative data.

## RESULTS

### Study participants

Thirty-seven adults with a molecular diagnosis of SRS were identified for the broader study, of whom 15 (six female) agreed to participate in a study interview [figure 1](#). Of the 15 participants: 14 had a loss of methylation at 11p15 and one had maternal uniparental disomy of chromosome 7. The mean final height SD score (SDS) of this group was  $-2.68$  (men  $-2.61$  SDS; women  $-2.78$  SDS) ([table 1](#)). Nine participants received GH (three female). No significant differences between participants from

**Table 1** Characteristics of study participants

| Participant number (sex) | Age   | Genetic classification | Height | Height SDS | Childhood GH treatment |
|--------------------------|-------|------------------------|--------|------------|------------------------|
| P01 (male)               | 31–40 | 11p15LOM               | 153.23 | −3.66      | Yes                    |
| P02 (male)               | 31–40 | 11p15LOM               | 171.25 | −1.02      | No                     |
| P03 (male)               | 31–40 | 11p15LOM               | 150.3  | −4.09      | Yes                    |
| P04 (male)               | 31–40 | 11p15LOM               | 156.85 | −3.13      | No                     |
| P05 (male)               | 21–30 | 11p15LOM               | 171.95 | −0.92      | Yes                    |
| P06 (male)               | 31–40 | 11p15LOM               | 171.9  | −0.92      | Yes                    |
| P07 (male)               | 31–40 | 11p15LOM               | 168.05 | −1.49      | Yes                    |
| P08 (male)               | 31–40 | 11p15LOM               | 146.07 | −4.71      | Yes                    |
| P09 (male)               | 61–70 | 11p15LOM               | 153.9  | −3.56      | No                     |
| P10 (female)             | 31–40 | 11p15LOM               | 149.53 | −2.37      | No                     |
| P11 (female)             | 21–30 | Upd(7)mat              | 160.87 | −0.49      | Yes                    |
| P12 (female)             | 31–40 | 11p15LOM               | 130.57 | −5.52      | No                     |
| P13 (female)             | 21–30 | 11p15LOM               | 147.7  | −2.67      | Yes                    |
| P14 (female)             | 41–50 | 11p15LOM               | 143.45 | −3.38      | No                     |
| P15 (female)             | 51–60 | 11p15LOM               | 150.1  | −2.28      | Yes                    |

GH, growth hormone; SDS, SD score.

the broader study who did and did not participate in the qualitative study in terms of height, weight, asymmetry and molecular diagnosis were found.

### Findings

Four themes were identified from participant accounts: (1) ‘it’s not just all about height’: appearance-related concerns extending beyond height; (2) resilience: strategies to deal with real and perceived threats; (3) ‘I feel like I’m an old lady’: women’s experiences of pain, disability and feeling older than their years; and (4) ‘mayor of the friend zone’: feeling overlooked in romantic relationships.

#### ‘It’s not just all about height’: appearance-related concerns extending beyond height

At interview, 93% (n=14) of participants were employed, 80% (n=12) had a romantic partner (46% (n=7) were married), 40% (n=6) had their own families, and most appeared content with themselves and their lives: ‘I think in my thirties it has been a much more comfortable acceptance of this is who I am and kind of take it or leave it’ (P02 male). Many participants reported times when they had felt unhappy with the way they looked (table 2) and used negative words, such as ‘horrific’, ‘unattractive’, ‘ugly’ and ‘hate’ to describe how they felt about themselves. They reported concerns regarding how their appearance affected their psychological well-being, which included height but this was not the only concern raised—some participants stated that being short and asymmetrical was worse than only being short; others said that ‘it was not

**Table 2** Examples of appearance-related concerns

| Participant | Quote   |
|-------------|---|
| P09 male    | ‘Short and ugly’  |
| P03 male    | ‘Wonky mouth’—asymmetrical face                                 |
| P04 male    | ‘Unattractiveness of overweight’ ‘bulbous’—asymmetry and weight |
| P13 female  | ‘Small, but not in proportion’                                  |
| P14 female  | ‘My teeth, that’s what’s held me back’                          |
| P15 female  | ‘Angular face and hanging teeth’                                |
| P12 female  | ‘Physically unusual’  |

**Table 3** Examples of characteristics participants reported they would like to change

| Participant | What has had the biggest impact   |
|-------------|-----------------------------------|
| P03 male    | Speech (due to cleft lip/palette) |
| P04 male    | Asymmetry                         |
| P14 female  | Asymmetry                         |
| P10 female  | Asymmetry/scars on legs           |
| P12 female  | Weight                            |
| P06 male    | Weight                            |
| P05 male    | Restricted leisure activities     |
| P09 male    | Height                            |
| P13 female  | Height                            |
| P15 female  | Pain                              |

just about height at all’. Only two participants stated their height had impacted on their lives the most (table 3).

#### Resilience: strategies to deal with real and perceived threats

Participants described adversities (table 4), which they perceived to be related to their diagnosis of SRS, such as: feeling different from others; discrimination; infertility; bullying; comments and staring from strangers; and negative experiences with health-care. Strategies used by participants in response to adversity were classed as coping or adaptive strategies to reduce stress<sup>15</sup> or cultivated resilience, for example, capitalising on other characteristics (such as humour and/or intelligence); accessing social/group support; and comparing themselves with others perceived to be less fortunate than themselves. Maladaptive ways of dealing with adversity that may have increased stress and/or psychological distress (anxiety and depression) were also identified. This included social avoidance; comparing themselves with others perceived to be more fortunate than themselves (often siblings); and not disclosing their diagnosis of SRS to others, reducing the likelihood of social support (table 5).

#### ‘I feel like I’m an old lady’: women’s experiences of pain, disability and feeling older than their years

Female participants, especially those over 30 years, tended to depict their experience of SRS as one of pain and disability, although a few shared experiences of pain and the impact during adolescence. Some had painful joints and fatigue such that they used wheelchairs, were in receipt of disability living allowance, and some were considering modifications to their homes (stair lift). They described feeling generally ‘unwell’ and unusually tired compared with those around them. Participants felt these issues adversely affected their employment status as: they were either not employed; had to reduce their working hours as they could no longer manage; or felt they could not disclose how much their pain or disability affected them from fear of being dismissed. Participants were also concerned about the impact of their condition on their families as they had to care for them or that they were unable to attend social events due to illness or pain table 6. Only one male participant discussed experiencing pain in his neck and lower back. He attributed this to a discrepancy in leg length, which did not appear to limit his activities.

#### ‘Mayor of the friend zone’: feeling overlooked in romantic relationships

While friendships had flourished for most participants through childhood and adolescence, this was less evident for romantic relationships. One participant described being ‘mayor of the friend zone’, which has been defined as ‘an ostensibly platonic relationship

**Table 4** Adversities—quotes linked to the text regarding resilience: strategies to deal with real and perceived threats

| Childhood: aged 0–10                        |   |
|---|---|
| Negative experiences with healthcare        | <i>I used to go to [hospital], I used to have to stand in this three-sided cubicle with a wooden round floor that used to revolve, with no clothes on, and they used to take photos of me. That has always stuck in my mind. I absolutely hated that, even as young as probably two, three, four years old, I really felt humiliated. (P14 female)</i>  |
| Adolescence and young adulthood: aged 11–21 |   |
| Negative experiences with healthcare        | <i>I remember when I was 15 they suggested to me that I have growth hormone treatment. The growth hormone paediatrician basically they told me that if I don't have it then I would probably not find a relationship and that I would find it very difficult. (P08 male)</i>  |
| Feeling different                           | <i>It was I think only when I got into kind of secondary school that I was the biggest and the tallest in year seven. Because I'd had the growth hormone. It's not very feminine to be the bigger girl and the taller girl, and I developed much earlier than all my friends. I was quite self-conscious in that sort of stage, and that's impacted my confidence. (P11 female)</i>                                     |
| Comments and staring                        | <i>We were having some crazy party and this one girl looked at me and she started freaking out because she was saying 'what's wrong with you, you look different, your left side's bigger' and then she just freaked out because she'd just kind of noticed it for the first time and her reaction that I saw was very disconcerting. (P04 male)</i>  |
| Bullying                                    | <i>As a child [I was] obviously bullied a lot, because your face is angular and different, you can hide your arms and hands, but you can't hide your face. Especially when I hit secondary school, that's when it really got to the point where it's all about what you look like. Secondary school was the hardest I think for me, but it was definitely the early teens that was really, really bad. (P15 female)</i> |
| Infertility                                 | <i>At one time the scan showed I had two ovaries and no womb, and the next showed I only had one ovary and a hypoplastic uterus is what they call it. So, when I found that out that was pretty devastating. (P12 female)</i>   |
| Psychological distress                      | <i>I went through a very low time [aged 13] and I was self-harming, because I physically hated the way that I looked, I hated myself and knew there was nothing that I could do about it to change it, and I couldn't accept the fact that that was who I am. (P13 female)</i>  |
| Discrimination                              | <i>I remember one particular incident, I walked in and the guy in charge saw me and he went to [his colleague] 'Do me a favour. What is he doing here, how can he help?' (P08 male)</i>   |
| Adulthood: >21 (see themes 3 and 4)         |   |

in which one person is romantically interested in the other'.<sup>16</sup> Lack of perceived romantic appeal led participants to feel they were more likely to be treated like friends by those with whom they wanted to have a romantic relationship. Although this theme was identified in the data from both male and female participants, SRS appeared to have a greater impact on male participants' romantic relationships compared with those of their peers, which for some also pervaded into adulthood. The antecedents were difficult to determine, but it is possibly related to participants' feelings of falling short regarding society's perception of quintessentially male appearance/behaviour. This may have resulted in a subsequent lack of confidence and self-esteem. Male participants described times during adolescence and adulthood when their appearance (height and lack of muscle mass) made them feel less masculine, leading to them feeling

'sub-standard'. Female participants described similar experiences of not fitting the 'feminine ideal'. Some (male and female) thought they could not have relationships with people they were attracted to because they would be 'automatically excluded'. They did not have expectations that potential romantic partners would compromise their standards to be with someone who looked like them (table 7).

## DISCUSSION

To our knowledge, this is the first study to explore the psychosocial impact of SRS. Many individuals experienced significant adversity during childhood and adolescence relating to living with SRS, as well as physical and psychosocial issues. However, it should be noted that, at the time of interview, many participants

**Table 5** Quotes regarding coping strategies linked to the text about resilience: strategies to deal with real and perceived threats

| Childhood: aged 0–10                        |   |
|---|---|
| Capitalising on other characteristics       | <i>I had to learn to use my mind pretty quickly, and a sense of humour, as a way to survive. (P02 male)</i>   |
| Non-disclosure                              | <i>That's not something I ever spoke about to anybody [being diagnosed with SRS aged 10] except my Dad, but only again to discuss options and whatever else. (P04 male)</i>   |
| Social avoidance                            | <i>Going to sleepovers was always a nightmare. With the SRS I can't eat late. I will throw up, I used to do it all the time when I was little. It got to the point that I'd say to my mum 'Can you pretend that you've grounded me and that I'm not allowed to go out.' I just didn't want to go out. So, I was lying to my friends just because I was too embarrassed in case something happened. (P13 female)</i> |
| Adolescence and young adulthood: aged 11–21 |   |
| Accessing social support                    | <i>One thing that I think SRS has never held me back on is my social life. (P03 male)</i>   |
| Upward social comparison                    | <i>I realised my brother was a machine, he'd do decathlons and pole vaulting and I always competed with him, or tried to, never beat him, but occasionally when he saw that I was quite emotional then he would let me win and even though in my teens I used to have a mini breakdown about every 3 months and I would cry. (P08 male)</i>   |
| Social avoidance                            | <i>The depressive episode came and then I left home, so just before my 17th birthday I went. The SRS, the shortness, the asymmetry floored me, I couldn't deal with it. The worst thing in hindsight was that I didn't tell anyone. I kept it inside. I kept myself from people as well. (P04 male)</i>   |
| Adulthood: >21                              |   |
| Accessing support groups                    | <i>In about the last three or four years I joined, to find out if other people were having these same problems. My health just got worse and then that's when I thought I could do with knowing more about it because I think I'd just hidden away from it all my life. It helped to see that a few of us were having the same problems. (P10 female)</i>   |
| Downward social comparison                  | <i>You know there are people out there who are handsome, normal, normal height, normal everything, good jobs, whatever. And they're married and they're miserable. (P09 male)</i>   |

SRS, Silver-Russell syndrome.

**Table 6** Quotes related to 'I feel like I'm an old lady'—women's experiences of pain, disability and feeling older than their years

| Adolescence: 11–21              |   |
|---------------------------------|---|
| Pain                            | <i>If physical education was first thing in the morning I'd have pains in my legs then for the rest of the day and all that night.</i> (P10 female)   |
| Impact of pain on mental health | <i>Probably in my teenage years. I was treated with depression for years, part of that is just being chronically ill for so long. It's not really been the fact that I'm short, it's just being in pain and it being a wearing thing on you for so long.</i> (P12 female)   |
| Adulthood: >21                  |   |
| Pain                            | <i>Now I do get a bit of back pain and hip pain, and I've got curvature of the spine, so I'm going to have. But I can remember being told when I used to go to the hospital that I'd probably be in a wheelchair by the time I was forty, but that ain't never going to happen.</i> (P14 female)  |
| Impact on employment            | <i>I mean you do have to be careful. I'm very, very careful not to take sick leave. I went to [my manager] one day and I said look, I'm really, really struggling, and she said well if you can't do the job you might need to find another one.</i> (P15 female)   |
| Fatigue                         | <i>I can't sort of walk far and I can't do stairs very well and you just generally get sort of fatigued and that, but that's the problem that's causing the most issues.</i> (P12 female)   |
| Limiting daily activities       | <i>And there have been days when actually it's hard to describe, but you actually just can't walk far. It really, really hurts and you've just got to sort of not do much that day.</i> (P15 female)  |
| Impact on others                | <i>My daughters, at twelve and nine, they can cook a meal, they can put the washer on, the eldest can iron. They Hoover up and if I need them to look after me when I'm having a bad day, they do, they're really, really good. They'll do anything for me. So, I'm very lucky in that respect. But then sometimes I think 'Is that fair on them?'</i> (P10 female) |

had successful careers, were in romantic relationships, and some had, or were starting to have, families of their own.

Challenges during childhood and adolescence were central to all four themes. Participants reported starting to feel 'different' around aged 10 and began comparing themselves with others. They experienced bullying, staring and/or comments from strangers; and encountered problems developing romantic relationships during adolescence. Participants described disquiet regarding these issues, which were in addition to concerns about their height, and which for some led to anxiety and depression and persisted into young adulthood and beyond.

The issues experienced by participants in our study are likely to be above and beyond those experienced by young people without a health condition. Other studies have suggested that people with conditions affecting growth (idiopathic short stature, achondroplasia, Turner syndrome, constitutional growth delay) are more likely to experience psychosocial issues (lower

quality of life, depression, low self-esteem, difficulty forming relationships, social isolation) compared with people within the average height range and no diagnosed health condition.<sup>4 6 17 18</sup> Whether this is due to short stature or having a health condition, or both, is not easily understood.

While the issues experienced during adolescence were particularly problematic, young adults with SRS also experienced significant concerns at a time when specialist care ceased, when participants were facing challenging times and at a stage of life when young people experience the trials and self-doubt commonly experienced during adolescence.<sup>19</sup> Likewise, in adults with cleft lip or palate, new issues and experiences are encountered just as the young person is discharged from paediatric services.<sup>20</sup> Clarke *et al*<sup>21</sup> suggest transition as a good point for appearance-related concerns to be assessed, enabling health professionals (HPs) skilled in dealing with these issues to support individual needs. We would argue that for young people with

**Table 7** Quotes in relation to 'mayor of the friend zone': feeling overlooked in romantic relationships

| Adolescence and young adulthood: aged 11–21                              |  |
|--|--|
| Mayor of the friend zone—male perspective/low self-esteem                | <i>I had pretty low sort of sexual self-esteem all the way through my teenage years. I was very definitely kind of mayor of the friend zone. I'd become very good friends with all of these very beautiful girls [...] I sort of really struggled with forming relationships.</i> (P02 male)   |
| Mayor of the friend zone—female perspective                              | <i>I was quite boy mad actually when I think back. I just didn't have a lot of boyfriends. They all liked me, but just not that way. I was a bit of a clown really. I was one of the lads.</i> (P14 female)  |
| Lack of confidence   | <i>More lack of romantic relationships at the time. I mean when I was younger I probably had at school, I had one girlfriend and that was it. Maybe that was a consequence of it [SRS], potentially getting back to that confidence aspect as well probably did have an effect, no it definitely probably did have an effect at the time I would say, probably right up until or through till university.</i> (P05 male)                         |
| Difficulties were over and above peer's difficulties                     | <i>I remember when I was 13 I liked someone at school, I asked her out every month for a year and she kept on turning me down. It was then I was aware that I was different 'cause other people were in relationships and I wasn't. It became quite an important thing [...] I think I felt sorry for myself. Not having a relationship. I didn't have many later on in life.</i> (P08 male)   |
| Lack of height and muscle/falling short of 'masculine ideal'             | <i>Even at that age [I felt] like the football players or the taller blokes or stronger perhaps did better with girls than those who were perhaps shorter or fatter. I didn't actually have any relationships that I considered to be mature until I actually went to university anyway.</i> (P07 male)  |
| Adulthood: >21   |  |
| Lack of height and muscle/falling short of 'masculine ideal'             | <i>Because of my height, I guess most girls are looking for guys who're a bit taller. And my lack of muscle. Girls are looking for guys that have got a bit more muscle. Not every girl. But generally, if you take a hundred random women, how many of them, if you stick in a guy [...] six feet tall with average build, and you take me, you'd be lucky if one of them picks me over the other [...] and that's just reality.</i> (P03 male) |
| Feeling substandard—male perspective                                     | <i>The late 20s phase with the relationships I think I probably saw myself as being worse, as being sub-standard, being less of a man in a traditional sense and also kind of being in an environment where the idea of what a man was was quite narrowly defined as being kind of you know big and muscular and sort of hairy.</i> (P02 male)   |
| Feeling substandard—female perspective/falling short of 'feminine ideal' | <i>I think if I met someone, with me being my height and having my joint problems, it would take somebody special anyway, because I'm not the tall leggy blonde with blue eyes that a lot of men would want. So, it would take somebody with a special heart anyway to be with somebody who is physically unusual.</i> (P12 female)  |

SRS, Silver-Russell syndrome.

SRS—and possibly other health conditions—psychosocial assessment should take place much earlier than this.

Our findings suggest that for HPs managing children and young people with SRS there needs to be greater appreciation and focus on appearance-related concerns, in addition to height, as well as psychosocial issues. A similar focus on factors other than height has improved psychosocial adjustment in children and young people with idiopathic short stature.<sup>10</sup> It is possible that a management focus on improving height may communicate height as an aesthetic attribute, with an unhealthy focus on appearance that could contribute to appearance-related concerns beyond height. HPs also need to consider adopting a tailored approach to support, due to the gender differences in the reported experiences of individual's SRS.

Regular psychosocial assessments are recommended for individuals with SRS.<sup>1</sup> This is particularly pertinent as objectively rating the severity of an individual's appearance-altering condition by HPs cannot reliably predict who will, or will not, experience psychological distress due to appearance-related concerns.<sup>22</sup> Our findings indicate that a psychologist should be an integral member of a multidisciplinary team managing individuals with SRS during childhood, adolescence and adulthood, to aid in the development of strategies to manage any appearance-related concerns and other psychosocial issues that arise.

We have gained insight into what it is like to live with SRS through in-depth interviews, something that has not been achieved for other growth conditions. Previous research, mostly based on survey data, indicates that individuals living with short stature conditions experience a range of psychosocial issues similar to those detailed in this study.<sup>4–6 17 18 23–25</sup> Echoing our study findings more closely, women with Turner syndrome report that height is also not their main concern;<sup>4</sup> they had significantly more appearance and body image concerns than women without Turner syndrome,<sup>20</sup> and experience difficulties when dating.<sup>5 8</sup> More in-depth research is needed to explore these issues in all conditions that affect growth to ensure appropriate support is offered to children and their families. Excessive tiredness and joint pain also requires further research and long-term follow-up to understand it more fully. This may be related to the lack of muscle bulk and function in this syndrome that is often underestimated.<sup>1</sup> It shows itself in childhood by delaying motor milestones but is subsequently not such an issue.

Our study has several limitations. We relied on participants recalling past experiences, which may have changed over time and was skewed towards a higher reporting of memories from adolescence.<sup>26</sup> Adolescence was a crucial time when treatment and regular hospital appointments ceased, so it may be expected that adults recall more experiences from this time. Second, some participants had health conditions that may be part of, or in addition to, SRS (eg, dyslexia, cleft palate) which may lead to poor psychological adjustment.<sup>27</sup> Teasing out which experiences were related to SRS and which were related to other health conditions was not possible, therefore we presented participants' views on what they perceived to be related to SRS. Third, adults recalling their experiences may bear little resemblance to those of current children and young people with SRS who benefit from advancements in managing SRS. However, through speaking to young people and observing clinical appointments with children and their families, we remain confident our findings are pertinent to those actively receiving treatment. Lastly, participants were primarily those who volunteered for the broader study. These may have been a self-selecting group of individuals with complex issues, keen to have a thorough medical examination. Reflecting on the sample of participants we would say this was not the case.

Participants had a range of complex health issues, with some classing themselves as 'unaffected' by SRS. Figure 1 shows that this substudy included all but one of the eligible participants who could be contacted, indicating a low risk of further selection bias.

The strengths of our study include several novel contributions to the field of appearance research. Our study investigated areas that are rarely reported in the literature and have not previously been described in SRS.<sup>28–33</sup> Methodological quality in our study was enhanced by adapting the interview schedule after each interview ensuring a broad range of issues were captured. We also presented our key findings to young people and adults with SRS and their families who had not taken part in our study to triangulate our results. This gave us reassurance that the conceptual work carried out was still rooted in participants' experiences.

## CONCLUSION

The SRS literature to date describes the physical characteristics of SRS with little insight into living with this rare genetic condition. We have identified that individuals with SRS experience significant psychosocial issues during childhood, adolescence and adulthood which extend beyond a concern about height. These findings are relevant to paediatricians who are well positioned to anticipate and address psychosocial issues in children and young people with SRS. Such issues could be ameliorated through early psychosocial intervention; ensuring clinical appointments explore other concerns as well as growth; involving other HPs, such as psychologists and general practitioners; and by developing an adult service for individuals with SRS.

## Author affiliations

<sup>1</sup>Faculty of Medicine, Clinical Ethics and Law, University of Southampton, Southampton, UK

<sup>2</sup>Faculty of Health and Applied Sciences, University of the West of England, Bristol, UK

<sup>3</sup>Nutrition and Metabolism Unit, University of Southampton, Southampton, UK

<sup>4</sup>Southampton National Institute for Health Research, Biomedical Research Centre, University Hospital Southampton NHS Foundation Trust, Southampton, UK

<sup>5</sup>Membership and Communications, Child Growth Foundation, Sutton Coldfield, UK

<sup>6</sup>Department of Endocrinology, University Hospital Southampton NHS Foundation Trust, Southampton Children's Hospital, Southampton, UK

<sup>7</sup>MRC Lifecourse Epidemiology Unit, University of Southampton, Southampton, UK

<sup>8</sup>NIHR Southampton Biomedical Research Centre, University of Southampton and University Hospital Southampton NHS Foundation Trust, Southampton, UK

<sup>9</sup>Faculty of Medicine, Human Development and Health, University of Southampton, Southampton, UK

<sup>10</sup>Wessex Clinical Genetics Service, University Hospital Southampton NHS Foundation Trust, Southampton, UK

<sup>11</sup>Faculty of Medicine, University of Southampton, Southampton, UK

<sup>12</sup>North West Thames Regional Genetics Service, London North West Healthcare NHS Trust, London, UK

<sup>13</sup>Faculty of Medicine, Human Development and Health, University of Southampton, Southampton, UK

<sup>14</sup>Faculty of Medicine, Clinical Ethics and Law, University of Southampton, Southampton, UK

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