Experiences of adolescents living with Silver-Russell syndrome

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ABSTRACT
Objective The psychosocial impact of growing up with Silver-Russell syndrome (SRS), characterised by growth failure and short stature in adulthood, has been explored in adults; however, there are no accounts of contemporary lived experience in adolescents. Such data could inform current healthcare guidance and transition to adult services. We aimed to explore the lived experience of adolescents with SRS.

Design/setting/patients In-depth, semi-structured interviews were conducted between January 2015 and October 2016 with a sample of eight adolescents aged 13–18 (five girls) with genetically confirmed SRS from the UK. Qualitative interviews were transcribed and coded to identify similarities and differences using thematic analysis; codes were then grouped to form overarching themes.

Results We identified four themes from the interview data: (1) the psychosocial challenges of feeling and looking different; (2) pain, disability and fatigue; (3) anticipated stigma; and (4) building resilience and acceptance. Despite adolescents accepting SRS in their lives, they described ongoing psychosocial challenges and anticipated greater problems to come, such as stigma from prospective employers.

Conclusions Adolescents with SRS may experience psychosocial difficulties from as young as 10 years old related to feeling and looking different; pain, disability and fatigue; anticipated stigma; and future challenges around employment. We discuss these findings in relation to recommendations for the care of adolescents with SRS to prepare them for adult life.

INTRODUCTION
Silver-Russell syndrome (SRS) is a rare genetic condition characterised by slow growth in utero leading to poor postnatal growth, short stature in adulthood, triangular facial appearance, asymmetry, feeding difficulties,1 low muscle mass and poor muscle function.2 A recent study estimated SRS incidence as 1:15 866—higher than previous estimates3—with males and females equally affected.1 Two molecular abnormalities cause the majority (60%) of SRS cases,1 but a significant minority of patients have no known molecular cause.1 Management of SRS during childhood focuses on optimising final height through nutrition and growth hormone (GH) therapy.

Our recent qualitative research showed that adults living with SRS dealt with psychosocial challenges through childhood and adolescence; they had appearance-related concerns beyond their height and experienced difficulties in forming romantic relationships.6 Women reported experiencing pain and disability, impacting on employment and familial relationships. As part of our broader programme of research,6 7 we interviewed adolescents (aged 13–18 years) to explore their lived experience of SRS.

What is already known on this topic?
► The clinical management of Silver-Russell syndrome (SRS) during childhood has a major focus on optimising height.
► Psychosocial evaluation is not routinely offered to children and adolescents in clinics.
► Appearance-related concerns result in psychological distress for some individuals living with SRS.

What this study adds?
► Aspects of the lived experience, such as pain, disability, feeling different, anticipated stigma and resilience, are similar in adolescents and adults living with SRS.
► Pain can be a symptom for girls with SRS, which is currently undocumented in existing literature. The causes and management of pain requires further investigation.
► Health professionals need to offer psychosocial support to adolescents as they prepare to discharge them to adult services.
### Considerations when interviewing adolescents

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<tr>
<td>Adolescents may be used to a more educational environment, where they may be expected to give a ‘right’ answer</td>
<td>Source: Morse, J.M., <em>Ethics in action: ethical principles for doing qualitative health research</em>. Qualitative Health Research, 2007. 37: p. 1003–1005.</td>
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<td>Qualitative research methods may pose a risk to participants as interviews often involve revealing intimate details about themselves and their lives, which may become an issue for more vulnerable participants who are young or have learning difficulties</td>
<td>Source: Duncan, R.E., et al., <em>Is my mum going to hear this? Methodological and ethical challenges in qualitative health research with young people</em>. Soc Sci Med, 2009. 69(11): p. 1691-9.</td>
</tr>
<tr>
<td>Adolescents have less life experience so may not be as well equipped to anticipate and deal with an interview scenario, they may be less assertive and may lack confidence</td>
<td>Source: O’Reilly, M. and N. Parker, <em>Doing Mental Health Research with Children and Adolescents: A Guide to Qualitative Methods</em>. 2014, London: Sage.</td>
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**Figure 1** Considerations when interviewing adolescents.

(QoL) and prior GH treatment had no impact on QoL in adulthood.

We set out to explore and evaluate the lived experiences of adolescents living with SRS to inform clinical management up to adolescence and beyond. Qualitative methods for data collection and analysis allowed for rich data to be generated and examined to gain in-depth understanding of these experiences.

### METHOD

#### Public and patient involvement

A member of the Child Growth Foundation—a UK charity that supports people affected by child growth conditions—who had personal experience of a growth condition, was also part of the research team. This charity member was involved in the development of the funding application, attended project steering group meetings, helped develop patient information leaflets and consent forms, advised on recruitment and analysis and was involved in the dissemination of project findings.

#### Identification of adolescents with SRS

Participants were recruited from a larger study investigating the health consequences of SRS in young people and from a patient support group. Research and development approval was secured from University Hospital Southampton National Health Service Foundation Trust (study sponsor) and, for the 23 UK Genetics Centres, via the National Institute for Health Research UK Rare Genetic Disease Research Consortium Agreement (‘Musketeers’ memorandum’). Individuals were identified across the UK through: (1) study information disseminated by the Child Growth Foundation, UK; (2) prior recruitment to an existing national study ‘Imprinting Disorders: finding out why’; (3) review of positive molecular genetics tests at the Wessex Regional Genetics Laboratory; and (4) contact with regional genetic and paediatric endocrine centres at existing genetics research sites.

Using the first two approaches, participants were sent information by post and invited to contact the study team or were contacted by telephone if they had previously given permission. For the other two approaches, the patient’s overseeing clinician

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**Figure 2** Source of recruitment to qualitative study. *Recruitment of 374 participants was from a larger study. Recruitment to the current study focused on those aged 18-years or younger.*
was contacted and asked to post study information to their patient.

### Study interviews

Interviews were chosen as they allowed the researcher to focus on drawing out individual experiences; explore specific concerns or issues; be flexible when wording questions and add questions about relevant topics that arose. 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. The interview schedule addressed the impact of SRS on key aspects of a participant’s life (education, work, friends, family and intimate relationships). This was piloted with people living with a chronic health condition, before commencing recruitment to our adult lived experience study. Piloting the schedule allowed us to assess how appropriate the questions were, if they were in an order that made sense and to practice asking the questions. We made several adjustments based on interviewee feedback, for example, taking out questions that were difficult to answer, were repetitive or needed reordering. Each interview took 30–60 mins and was audio recorded. Interviews were conducted by a trained female health psychologist (LMB) experienced in discussing sensitive health issues with patients. We gauged parents’ thoughts about the interview and, although this may have placed parents in a ‘gatekeeper’ position, none declined an interview on their child’s behalf. Parents and adolescents had the option of having a parent/trusted person present for the interview, and two participants were interviewed with parents present (see figure 1 for more considerations). All participants were provided with an information sheet and the opportunity to discuss the research and ask questions. Written consent/assent was gained from adolescents and their parents. Participants were reassured that they could discontinue the interview at any time and that it would be audio recorded, but this could be stopped at their request.

### Analysis of study interviews

Interviews were transcribed and analysed thematically using the Braun and Clarke method. Codes were generated from the first few interviews and then refined as subsequent interviews were analysed. Codes were then grouped to create categories and developed into four overarching themes. To ensure rigour in 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. Qualitative data were managed with NVivo software (QSR International, V.11.3.2 (1888) for Mac). To triangulate our findings, we presented them to adolescents living with SRS and their families. Feedback from this reassured us that our conceptual work reflected participant’s lived experiences.

### RESULTS

#### Study participants

Eight participants (five girls) aged 13–18 years were interviewed (see figure 2 and table 1 for details). Six participants were recruited from a broader study (of nine adolescents, six agreed to participate and three were uncontactable) and two from a patient support group. The number of participants declined is not known as adolescents were asked to approach the researcher if they were interested in taking part. Three participants had a loss of methylation at 11p15, four had maternal uniparental disomy of chromosome 7; one was genetically confirmed with the specific diagnosis not revealed to the researcher; six had received GH treatment, one had not and for one this was unknown.

### Table 2 Quotes linked to the text regarding the psychosocial challenges of living with Silver-Russell syndrome (SRS)

**Table 2**

<table>
<thead>
<tr>
<th>Having a stammer*</th>
<th>It’s affecting me sometimes talking in class, because I don’t really want to, in case people (…) laugh at me because I don’t really want to be trying to start saying one word and so I tend to not really speak that much. And I don’t like it when teachers pick on you, because that just makes me go ‘I just can’t do it’. (P03_F)</th>
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<tr>
<td>Early issues with appearance</td>
<td>Because children are always looking for differences, it doesn’t bother me (having SRS). When I was ten, I’d get really bothered about it. I would think ‘Oh why is it me, why did I have to have it?’ and it was like ‘I’m different, why!’ (Now it’s kind of gone down again). (P02_F)</td>
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<tr>
<td>Feeling left behind</td>
<td>P01_F’s mum: Your friendship went off a little bit, because they were all going off going out to the cinema and parties.</td>
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<td>P01_F: And they didn’t invite me much. Interviewer: How did that feel? P01_F: Sometimes I felt sad about it.</td>
<td></td>
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<tr>
<td>Feel different to others with SRS</td>
<td>I think I felt like they felt they were different to me - they got different - I don’t know how it works, but I did feel like some people were different than other people. I’ve met some people who were just smaller than other people, but then I met other people that had other stuff and all of that. (P05_M)</td>
</tr>
<tr>
<td>Treated differently</td>
<td>It was different in different years, so in Year 7, people used to give me very odd looks and kind of dirty looks, for no other reason than I was just small. People used to treat me a lot differently. The most noticeable time that this happened was I was in a football team for about three years, one girl there refused to acknowledge that I was older than I looked. It’s surprising really. You get very different reactions when you’re a certain height. (P07_F)</td>
</tr>
<tr>
<td>Appearance</td>
<td>It was more when I was younger - it was more noticeable. So, my eyes were a bit further apart when I was younger. They’ve gone more normal now. But they were more spaced apart when I was younger. (P07_F)</td>
</tr>
</tbody>
</table>

*There may not be a specific link between SRS and stammer, but 40% of children with SRS have delayed speech. [37]
Findings

Four themes were identified: (1) the psychosocial challenges of feeling and looking different; (2) pain, disability and fatigue; (3) anticipated stigma; and (4) building resilience and acceptance.

The psychosocial challenges of feeling and looking different: ‘I’m different! Why?’

Participants described feeling different from those around them (table 2), which was exacerbated by how others treated them. For example, participants reported they had previously experienced bullying: ‘I got called names […] small, midget, all that sort of stuff’ (P05_M); however, most said they were not being bullied currently. Participants described feeling and being treated differently from around age 10–12 years, which caused some to change their behaviour. One reported feeling that she must go above and beyond what was expected of her academically, to combat the perception that she may have special needs: ‘She [teacher] just treated me like I was a total idiot’ (P02_F). Another facet of this theme was looking different from peers. Participants discussed wanting to change their ‘spaced apart’ eyes or ‘big forehead’. Two of the girls felt that they would fit in if they were taller describing how being smaller than their peers led to them being treated as someone younger.

Pain, disability and fatigue: ‘Oh, are you disabled or something?’

The perception of feeling different may have been cemented by the experience of pain and disability and others’ reactions to this. Of the strategies for responding to adversity identified in the data (table 3), some may be classed as coping or adaptive strategies, such as capitalising on social support. Participants appreciated opportunities to be with people like them, for example, at school with a person who is also hard of hearing, or at a convention with others who have SRS. While some participants described downward social comparison with others—for example, P02_F feeling ‘quite well off’ compared with other girls with SRS—others described a form of ‘upward comparison’, comparing themselves with people they believed to be more fortunate than themselves. One participant described comparing themselves unfavourably with their unaffected sibling: ‘I used to think it’s not fair, because [they’ve] got nothing wrong with [them] and

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<th>Table 3</th>
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<td>Pain during daily activities</td>
<td>I struggle to walk sometimes, like really struggle. I’ll be fine for about thirty seconds, and then I get a burning sensation going up my legs, and they go, my legs go inwards, so I can’t, and then I can’t walk. (P07_F)</td>
</tr>
<tr>
<td>Adapation due to pain</td>
<td>Well my wrists, I can write short passages but writing longer, I find it really difficult, so I’d use a laptop for it, because it just hurts. (P03_F)</td>
</tr>
<tr>
<td>Comments from others</td>
<td>I mean my feet hurt from just walking home today (…) and like [to] be able to walk long distances, because my feet really hurt after a short amount of time. And PE in particular is really difficult for me. (P02_F)</td>
</tr>
<tr>
<td>Physical disability</td>
<td>It really bothered my dad actually because he was going ‘She’s not disabled, why is she in a wheelchair?’ And I don’t think it actually bothered me, but the fact that people stared did, because it was a real difference, because if you are just walking around-no one stares at you, but I was pushed into the disabled queue, and all the kids were looking at me getting out of the chair, and I just went ‘Oh, I feel a bit on the spot’. (P02_F)</td>
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<tr>
<td>Fatigue</td>
<td>I used to do karate and gymnastics and I used to play football, but I’ve stopped them now (…) because I normally get tired easily. I have to stop all activities. (P08_M)</td>
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For some, the inability to participate fully in physical education was no great loss: ‘They’re all able to like throw things, all the things and I can’t’. But that doesn’t really bother me because I’m not really into sports in the first place (P03_F), but we speculate it is possible that not being able to take part in sports from an early age may have led to a dislike for this subject. Others, who would have liked to be more involved in sport, were fearful of exclusion due to perceived lack of ability.

Anticipated stigma: ‘I hope it doesn’t affect getting a job’

Some participants anticipated future stigmatisation or discrimination because of having SRS (table 4). Some had concerns that symptoms such as pain might make working a challenge; others did not anticipate SRS being a barrier to employment and relationships. Participants did not anticipate SRS having an impact on them dating in the future unless ‘guys don’t like small girls or something. I don’t think so.’ (P02_F). Three participants had started to date, although periods of dating were brief (4 weeks) and three confirmed they had not yet started dating. P06_F and P08_M declined to elaborate, with P06_F saying, ‘I’m not really fussed; like I don’t really bother’ and P08_M said he would rather be single’. P04_M thought that he had no spare time for dating but knew that some of his friends had started.

Building resilience and acceptance: ‘You’re not always going to be perfect’

Of the strategies for responding to adversity identified in the data (table 5), many could be classed as coping or adaptive strategies, such as capitalising on social support. Participants appreciated opportunities to be with people like them, for example, at school with a person who is also hard of hearing, or at a convention with others who have SRS. While some participants described downward social comparison with others—for example, P02_F feeling ‘quite well off’ compared with other girls with SRS—others described a form of ‘upward comparison’, comparing themselves with people they believed to be more fortunate than themselves. One participant described comparing themselves unfavourably with their unaffected sibling: ‘I used to think it’s not fair, because [they’ve] got nothing wrong with [them] and

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<td>Worries about pain affecting employment prospects</td>
<td>I hope that doesn’t affect getting a job because of my wrists - my wrists do hurt, writing and that, and I just wonder. Probably being a veterinarian, maybe. I hope to have a good future. Hope it doesn’t really affect me. (P03_F)</td>
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<td>Anticipated stigma</td>
<td>I also hope that because you see stuff like people giving jobs to taller people because they think they’re more able and stuff. I just hope people don’t judge me because of that. I would tell them that, I think that’s the main thing, if I tell them that I had this thing [SRS], that they’d say ‘Oh, she’s weird, she has a genetic condition’. (P02_F)</td>
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<td>Worries about height affecting employment prospects</td>
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<td>Independence for those with MatUPD7</td>
<td>I hope to start making my own doctor’s appointments. Because I will be living in a house before very long [and] working in a phone shop. (P08_M)</td>
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<td>Dating</td>
<td>No, I don’t actually really have any relationships of that kind, I’ve only got friends. I think some of my [friends] have had boyfriends and girlfriends, but I don’t really hear much about them. (P04_M)</td>
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Previous research on patients with Turner’s syndrome likely to have a significant psychosocial impact on adults living the conclusion that childhood and adolescent challenges are studies suggest likewise; psychosocial evaluation and support by et al.106:1195–1201. doi:10.1136/archdischild-2020-321376 Arch Dis Child 2021;106:1195–1201. doi:10.1136/archdischild-2020-321376

These findings parallel those of our adult study6 and reinforce example, appearance, hearing problems and having a stammer. experiencing bullying associated with SRS when younger, for treated differently, often as early as age 10 years. Some reported experiences of adolescents living with SRS. While resilience was described, most participants described psychosocial challenges and anticipated more to come. For example, stigma owing to their height, having a genetic condition and feeling different from their peers. Most of the girls in the sample reported experiencing pain and discussed how physical disability played a part in their lives.

Participants described feeling different from peers, and being treated differently, often as early as age 10 years. Some reported experiencing bullying associated with SRS when younger, for example, appearance, hearing problems and having a stammer. These findings parallel those of our adult study6 and reinforce the conclusion that childhood and adolescent challenges are likely to have a significant psychosocial impact on adults living with SRS.6 Previous research on patients with Turner’s syndrome suggested that QoL outcomes could be improved by addressing concerns about height by age 6–12 years at the latest.16 17 Our studies suggest likewise; psychosocial evaluation and support by around age 10 years would assist during adolescence and in the transition to secondary school.

Participants reported coping with feeling different by comparing themselves with others less fortunate than themselves, capitalising on social support from friends and family and accessing support groups. These skills, which foster resilience, are important to positive adjustment when feeling or looking different.18 Accessing support groups can be particularly beneficial; and some participants were members of online support groups. Participants may have gained social capital by forming friendships19. Downward comparison can be made with those in a similar situation, increasing self-esteem20 and upward comparison can increase feelings of hope and motivation;21 simply being with others who are similar may give people some respite from feeling different.20 Accessing the support offered by friends and family can also reduce stress22 and help cope with the challenges that living with a chronic condition can raise.23

Most girls in the study reported pain and disability, largely absent in accounts from boys; fatigue was discussed by a minority of all participants. This novel finding is of interest and complements our previous data in adults with SRS where we found that adult women’s experience of SRS was one of pain, fatigue and disability, a phenomenon little documented in existing literature.6 Although approximately a third of adolescents in Europe experience fatigue,24–26 it may be more significant in individuals with SRS because of their tendency to low lean mass.6 Our findings require verification in other cohorts and further study regarding the causative mechanisms of pain and disability among girls/women living with SRS.

Participants reported being treated differently due to their appearance and wanting to change the way they looked in order to fit in. These findings again echo those from our adult

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<td>Downward comparison</td>
<td>I haven’t got Russell Silver Syndrome as bad as some of the other people in there (Child Growth Foundation Convention). (P06, F)</td>
</tr>
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<td>Social support – Child Growth Foundation Annual Convention</td>
<td>Because you can talk about it more. Because sometimes when you’re talking to your friends about it, they’re always like ‘Oh no’, and they’re not really interested because probably it’s just my friend who has something. But the people there, they have it too, and you can share experiences with them. (P02, F)</td>
</tr>
<tr>
<td>Social support – friends like me</td>
<td>But one of my friends, who I’ve known before Secondary is in my Year, and she’s also hard of hearing, so that’s nice for me, because it’s someone who I can relate with. (P03, F)</td>
</tr>
<tr>
<td>Social support – friends</td>
<td>I think I’ve got quite a few friends as well. (P04, M)</td>
</tr>
<tr>
<td>Social support – family</td>
<td>My mum and dad have been there for me all the time, through my injections and all through the growth and all that. I think it might have impacted on them, the injections, because I don’t think they liked me taking injections. But I feel like it’s impacted them in a good way, because they’ve seen all the changes that have happened to me, and they’re quite proud of me. (P05, M)</td>
</tr>
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DISCUSSION
To our knowledge, this is the first study exploring contemporary experiences of adolescents living with SRS. While resilience was described, most participants described psychosocial challenges and anticipated more to come. For example, stigma owing to their height, having a genetic condition and feeling different from their peers. Most of the girls in the sample reported experiencing pain and discussed how physical disability played a part in their lives.

Participants described feeling different from peers, and being treated differently, often as early as age 10 years. Some reported experiencing bullying associated with SRS when younger, for example, appearance, hearing problems and having a stammer. These findings parallel those of our adult study6 and reinforce the conclusion that childhood and adolescent challenges are likely to have a significant psychosocial impact on adults living with SRS.6 Previous research on patients with Turner’s syndrome suggested that QoL outcomes could be improved by addressing concerns about height by age 6–12 years at the latest.16 17 Our studies suggest likewise; psychosocial evaluation and support by around age 10 years would assist during adolescence and in the transition to secondary school.

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Participants reported being treated differently due to their appearance and wanting to change the way they looked in order to fit in. These findings again echo those from our adult

Figure 3 Recommendations by theme. SRS, Silver-Russell syndrome.

study, where adults reported difficulties in adolescence forming romantic relationships that, for some, persisted into adulthood. However, adolescent participants did not report present or anticipated issues with dating, perhaps some were too young or participants may have hesitated to discuss such sensitive topics with the interviewer, as suggested by one young person who declined to comment on the topic. Previous research has found that having a visible difference does not appear to prevent adolescents from embarking on romantic relationships, but many experienced concern about future relationships or current relationships progressing. Future research should include the exploration of romantic relationships in relation to visible differences for people living with SRS.

Currently, clinical management of SRS focuses on height optimisation, but our data show that appearance-related concerns and psychosocial issues require at least equal attention. This study demonstrates enduring issues for people living with SRS and gives rise to specific recommendations for professionals working with this group (figure 3). Support for children and adolescents should include: increased understanding of psychosocial issues among clinicians, teachers and family members; access to psychological support for appearance-related concerns and management of pain and fatigue; peer support; career advice sensitive to concerns regarding living with a health condition; and guidance on diet and exercise, especially after GH treatment stops, as people with SRS may be at risk of metabolic problems in adulthood. When implementing these recommendations, it is important to highlight that severity of condition or visible difference is not related to psychological distress.

Strengths, limitations and future directions
Our study had several limitations. First, our sample was small and lacked diversity (eg, gender, ethnicity and culture) though this was unavoidable due to the rarity of SRS and difficulties in recruitment of adolescents. However, as our adolescent data and adult reports of adolescence are similar (ie, pain, disability, feeling different, anticipated stigma and resilience), the current study adds weight to our adult findings. Moreover, as it explored a specific, rare and previously undescribed experience, a smaller number of participants was acceptable due to higher information power. Second, we suspect our findings regarding more sensitive topics, such as dating and mental health issues, may not represent the full picture for adolescents living with SRS.

CONCLUSION
Adolescents with SRS may experience psychosocial difficulties related to feeling and looking different; pain, disability and fatigue; anticipated stigma; and future challenges around employment. Unlike conditions such as Turner’s syndrome that have medical issues monitored into adulthood, adolescents living with SRS are discharged from healthcare at a pivotal time when they require the most psychosocial support. Clinicians involved in the care of adolescents with SRS should be aware of the experiences highlighted here to support their patients now and prepare them for the future when discharged from the specialist service. Without such guidance, people living with SRS risk psychosocial problems that could be ameliorated with appropriate prevention and/or treatment interventions.

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