



Transition: Silver-Russell Syndrome

Guidance and support for
moving into adult services

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Let's talk about your health and moving from paediatric to adult services: Silver-Russell syndrome

When you reach 16 years old things change with your healthcare in the NHS.

You might have been seeing the doctors and nursing team for years and suddenly there's a decision to be made about your care - you should be part of this conversation.

Some people with Silver-Russell syndrome (SRS) will move into adult services with an adult specialist depending upon their health needs. Others are discharged from their consultants and have no regular medical follow up.

Transition planning to move out of paediatric services should start by school year nine and is usually led by health and social care practitioners, with the full involvement of families/carers and young people.

The National Institute for Health and Care Excellence (NICE) has published a quality standard for transition from children's to adults' services. The standard and statements focus on planning transitions and having a coordinated approach across all services involved in providing care to the young person.

You can read more about this here: www.nice.org.uk/guidance/qs140 or by scanning the QR code below:



The main professional responsible for your immediate health and wellbeing after leaving paediatric care should be your GP.

You need to make sure you have registered with a GP local to where you are living. You might be thinking you don't need to see a doctor regularly and that's totally fine, but you might feel differently and that's ok too.

If you are feeling uncertain about when to see a doctor, whether that's your GP or hospital specialist, speak to a family member or trusted friend who will support you to access the right care.

If you are still unsure, or if there is anything you would like to discuss further with our nursing team, you can contact the CGF Support Line* by:

Emailing: support@childgrowthfoundation.org

Calling: 020 8995 0257

Visiting: www.childgrowthfoundation.org/supportline

By scanning the QR code below:



**By contacting the CGF Support Line you are providing consent for the CGF to collect, process and store your data to provide you with the information or services you are contacting the CGF about. Read the CGF's [Support Line Privacy Statement](#) for full details.*

Now you are entering adulthood, there may be occasions when you are asked to explain your health needs and SRS to someone. This could be part of an assessment for PIP benefits, or with a colleague, friend or employer.

Personal information which YOU CHOOSE to share with others is completely your decision. The CGF have written these explanations as a tool to help you have those conversations.

Explaining Silver-Russell syndrome

Silver-Russell syndrome (SRS), also known as Russell-Silver syndrome (RSS), is a rare genetic condition causing growth problems.

Genetic testing provides an explanation in many, but not all, individuals with SRS.

SRS may not always be diagnosed on genetic testing and you may find that you have been given the diagnosis based on clinical symptoms.

This means that the healthcare professionals have identified that you have a number of features that are seen in people who have SRS.

These may include low birth weight, poor growth through childhood, a large head size, triangular shape face (larger forehead and narrow chin) and feeding difficulties.

Whether you have a genetic or clinical diagnosis, it is worth asking your parents / carers for a copy of the letter confirming the diagnosis (and also a copy of the genetics report if this is available) so that you can keep it to show people who may need to see it.

If you / they no longer have a copy of the letter, you can ask your GP or the paediatrician your care was previously under to give you a copy that should still be in your records.

How SRS might affect you

You can use the space below to record the problems you may have had in the past and may still experience now.

The list is just a prompt of things to think about, so you are prepared to explain as much or as little as you wish, when asked.

You may find that you have experienced some, all or none of these.

You can ask your parents or another adult to help you complete it, or you may feel you want to do this on your own.

| Part of SRS | How it has affected me in the past | How it affects me now |
|---|------------------------------------|-----------------------|
| Height/growth | | |
| Eating/ dietary needs | | |
| Low blood sugar levels (hypoglycaemia) | | |
| Body asymmetry/ joint pain/ mobility | | |
| Learning difficulties | | |
| Other (i.e. emotional health and wellbeing) | | |

SRS into adulthood

Due to SRS, there are things that may be an issue as you get older and healthcare professionals, such as your GP, should be made aware that you may need monitoring more closely due to the increased risk. It may be issues with:

- Weight management
- High blood pressure
- Raised cholesterol (a fatty substance in your blood that at high levels can increase the risk of heart problems)
- Joint pain
- Insulin resistance that can lead to higher blood sugar levels and potentially type 2 diabetes

The GP will need to see you every three to five years for calculation of BMI (body mass index), blood pressure, fasting lipids (to measure cholesterol), fasting blood glucose and HbA1c (to check for high blood glucose levels).

You should see the GP before this if you experience any symptoms of being more tired than usual, drinking more than usual because you feel thirsty or any spells of dizziness.

Whilst your GP will be familiar with carrying out these health care checks, they are unlikely to have expertise in SRS and many won't have even heard of your condition.

'Advice for young people and young adults living in the UK: Silver-Russell syndrome' has been written by SRS specialists and is for you to give to your GP so they have the right information and understand why these checks might be necessary. They are important so any signs of trouble can be picked up and treated early.

Advice for young people and young adults living in the UK: Silver-Russell syndrome

Name:

DOB:

NHS no:

Clinical/ genetic diagnosis (attach genetic test report(s)): Choose an item.

Silver-Russell syndrome (SRS), also known as Russell-Silver syndrome (RSS), is a rare genetic condition which causes growth restriction.

- Characteristic features are most obvious in early childhood:
- Low birth weight
- Continued poor growth
- Prominent forehead
- Relatively large head size at birth
- Body asymmetry
- Feeding difficulties

A genetic cause can be identified in around 60% of patients with a clinical diagnosis of SRS: most commonly loss of methylation on chromosome 11p15 (30-60%) and maternal uniparental disomy (matUPD7; 5-10%).

The condition is very variable in severity. Most children with the condition will be treated by an Endocrinologist with growth hormone until final height. Some will need long-term input from multiple medical specialities.

Research suggests that self-esteem and wellbeing is generally good but some individuals may benefit from referral to a psychologist for counselling.

Many young people and adults with SRS will lead active and healthy lives. This is likely to help avoid some health issues which appear to be more frequent in young people and adults with SRS. Sometimes specific changes to lifestyle or medical therapy may be required. These **health issues** include:

- **Weight management problems** (overweight/obesity)
- **Metabolic syndrome** (including obesity, insulin resistance/ type 2 diabetes, high blood pressure, raised blood lipids such as cholesterol, coronary heart disease, polycystic ovarian syndrome)
- **Dizzy spells** (and other signs suggestive of autonomic dysfunction)
- **Joint pains** (in some, but not all, cases this may be secondary to scoliosis or body asymmetry)

Long-term surveillance

Our knowledge of the health issues experienced in adulthood is still limited and the optimal surveillance strategy is not known.

Currently, in addition to maintaining a healthy lifestyle, we would suggest:

- 3-5 yearly assessments at your GP surgery for:
 - Calculation of body mass index (height and weight)
 - Blood pressure
 - Fasting blood lipids (including cholesterol and triglycerides)
 - Fasting blood glucose and HbA1c level
- Referral by your GP for specialist advice (e.g.: endocrinology/ lipid clinic/ cardiology), as needed
- GP to consider referral to other specialities (e.g.: rheumatology), depending on symptoms

Healthy lifestyle

Between the assessments with the GP as above, healthy lifestyle choices should be prioritised.

NHS advice about making healthy choices, including eating a balanced diet, being a healthy weight, and exercising can be found here: www.nhs.uk/live-well or by scanning the QR code below:



If individuals are concerned about their weight increasing or decreasing outside of a healthy range for them, they should speak to their GP surgery between the 3-5 yearly assessments to access further support.

Family planning

- For most individuals with SRS the chance of having a child with the condition is very low.
- For those with rare copy number or single gene variants, the chance may be up to 50%.
- Review of genetic test results is essential before giving advice to individuals planning to start a family. Referral to the local genetic service can be arranged via your GP.
- Females who fail to start their periods by 16 years of age need investigation for very rare structural uterine anomalies.

International consensus guidelines for clinical diagnosis and management of SRS

Read the full guidelines '*Diagnosis and management of Silver–Russell syndrome: first international consensus statement | Nature Reviews Endocrinology*' here: www.nature.com/articles/nrendo.2016.138 or by scanning the QR code below:



Read the summary for affected individuals and families here: www.childgrowthfoundation.org/srs-consensus-2017 or by scanning the QR code below:



Further information and support

Visit the Child Growth Foundation website here: www.childgrowthfoundation.org or by scanning the QR code below:



Visit The MAGIC Foundation website (including the booklet on 'Transitioning to Adulthood') here: www.magicfoundation.org or by scanning the QR code below:



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Disclaimer

We have taken every care to ensure the accuracy of the information contained in this resource. The information enclosed should not be used as a substitute for the advice from a clinician, GP or other healthcare professional.

Feedback

Your feedback helps us to ensure we are delivering information to the highest standard. If you have any comments or suggestions, please contact us at info@childgrowthfoundation.org or on 020 8798 2139.

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The CGF is an independent charity that relies entirely on the generosity of individuals, groups and organisations to continue our work. If you have found this resource helpful, please consider becoming a member, fundraising for our charity and/or making a donation at www.childgrowthfoundation.org.

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