



Child
Growth
Foundation



Silver-Russell Syndrome

Registered Charity: 1172807



About Us

The Child Growth Foundation (CGF) is a UK charity dedicated to supporting families living with rare child growth conditions. We provide information and support to those directly affected, their parents and the healthcare professionals who will work with them.

We also raise awareness of growth conditions among the general public and health professionals and we fund research to increase medical understanding of these conditions.

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Introduction

Silver-Russell syndrome (SRS) was first described by Dr Silver in 1953 and Dr Russell in 1954. At first it was thought that they were describing two separate conditions; it took nearly 20 years for doctors to realise that they had seen different aspects of the same condition. The disorder is usually called Russell-Silver syndrome in the United States and Silver-Russell syndrome in Europe.

SRS is a rare disorder characterised by intrauterine growth retardation (IUGR), poor growth after birth, a relatively large head size, a triangular facial appearance, a prominent forehead (looking from the side of the face), body asymmetry and significant feeding difficulties. The wide spectrum of findings varies both in frequency and severity from one affected individual to another. The majority of individuals with SRS are of normal intelligence, but motor and/or speech delay in early childhood is common.

SRS is genetically heterogeneous, meaning that different genetic abnormalities are known to cause the disorder. Abnormalities involving chromosomes 7 or 11 have been found in up to 60% of SRS patients. However, in approximately 40% of patients with a clinical diagnosis of SRS the underlying cause is still not known.

Consensus guidelines were published in 2017 which give recommendations regarding the investigation, diagnosis and management of individuals with SRS. The management of children with SRS often requires the involvement of many different health professionals.



How Common is SRS?

SRS occurs in all populations and affects males and females in equal numbers. In the past, many infants with IUGR and relatively large head circumference were incorrectly diagnosed with SRS. Because of the difficulty in diagnosis, other cases may go undiagnosed or be misdiagnosed, making it difficult to determine the true frequency of the disorder in the general population. However, recent data suggests that around 1 in 15,000 children will have Silver-Russell syndrome.

What are the Features of SRS?

The features of SRS vary greatly from one individual to another. Some are mildly affected; others may have more serious complications. The wide range of potential features can affect many different parts of the body. It is important to note that each affected individual will have only some of the symptoms listed below. Affected individuals/parents should talk to their medical team about their specific case, associated symptoms and management. With appropriate medical care, most individuals with SRS will live full, productive lives.



Elena aged 3 years



Most likely features:

- Poor growth both before and after birth
- Feeding difficulties
- Body asymmetry
- Relatively large head size at (and after) birth
- Prominent/protruding forehead

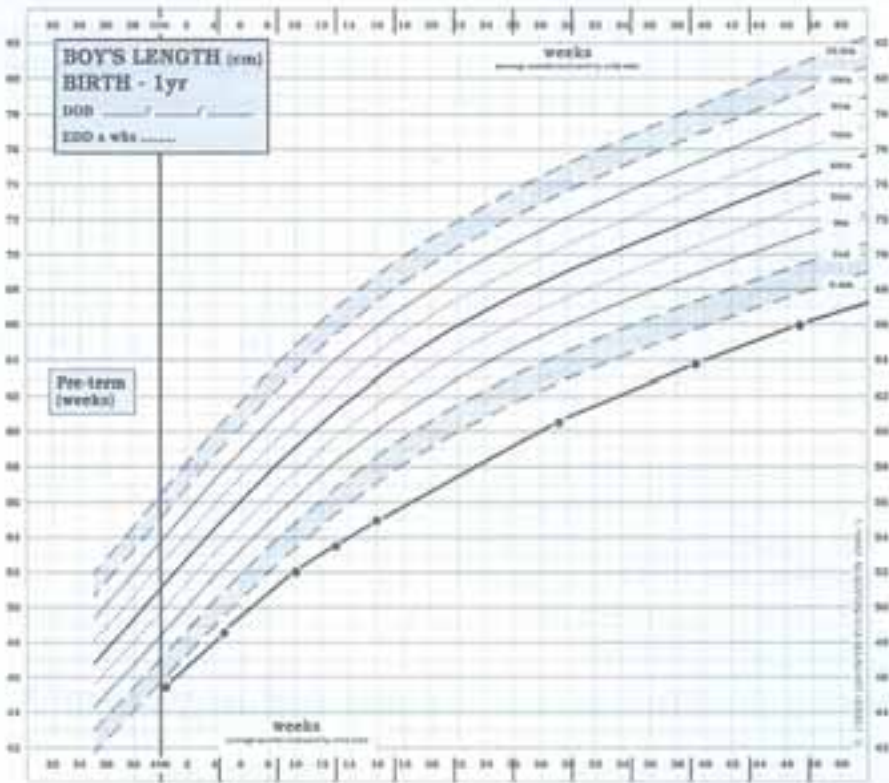
Growth and puberty

Almost all infants with SRS have a birth weight and length below the 2nd centile (<-2SD) even at full term. After birth, weight often continues to fall farther away from the normal range. Parents often report poor appetite (some children never cry for food) and struggle to get a SRS child to gain weight. Growth velocity for length/height continues to be slower than normal throughout infancy and childhood, with no 'catch-up' growth.

Bone age

Most SRS children have a delayed bone age in early childhood. However, the delayed bone age of SRS children is not associated with a late growing period (as seen in children with constitutional growth delay). Instead, SRS children typically experience a rapid acceleration of their bone age, often around age 8-9 years, and their bone age then becomes advanced.

There is only limited information regarding final height individuals with SRS who have not received growth hormone (GH) treatment, but in one study this was reported as approximately 151 cm in males and 140 cm in Asymmetry



Typical growth chart for a boy with SRS in the first year of life. Birth length is just below 0.4th centile. Growth velocity is slightly slower than normal with no 'catch-up' growth.

Asymmetry

In many children with SRS, all or part of one side of the body is smaller than the other (asymmetry). This results from the underdevelopment of one side of the body (hemihypotrophy). The extent and severity of



asymmetry is extremely variable. In most cases, asymmetry is found in just leg and/or arm length but, in some cases, one entire side of the body is affected. Children may experience difficulties with balance and walking as a result. Although, in most cases, asymmetry is apparent at birth, it may not become evident until later during childhood. Asymmetry may also improve with age. Treatment with growth hormone does not appear to increase the severity of asymmetry.

Craniofacial features

Characteristic craniofacial features are commonly seen in affected children. They are particularly noticeable in infancy and early childhood and become less obvious with age.

The most common finding is a large head size compared with the rest of the body (called head sparing/ relative macrocephaly). The head circumference is almost always far higher on the growth curve than either weight or length. This feature is almost invariably present from birth. This, along with the tendency for the jaw to be small (micrognathia), gives rise to the typical triangular facial shape seen in children with SRS. As a result, children with SRS may be investigated for hydrocephalus (a build-up of fluid surrounding the brain), though this is not associated with the condition.



Tristan aged 6 years



Another common facial feature is an abnormally prominent/protruding forehead, where the forehead protrudes out when the face is viewed from the side.

Other craniofacial features associated with SRS are less common, but can include:

- delayed closure of the 'soft spot' (anterior fontanelle) on the top of the head;
- low-set, posteriorly rotated ears;
- a small mouth with downturned corners;
- a high, narrow roof of the mouth (palate)

A variety of dental abnormalities have been reported including missing and/or small teeth and dental crowding.

Feeding difficulties

Gastrointestinal problems are common in children with SRS. These can include inflammation of the tube that carries food from the mouth to the stomach (oesophagitis), backflow of the contents of the stomach or small intestines into the oesophagus (gastroesophageal reflux), delayed gastric emptying (where ingested food takes longer than normal to digest causing the child to feel full) and failure to gain weight or grow at the expected rate for age and sex (failure to thrive/faltering growth). Some children with SRS simply never have a sensation of hunger during early childhood, while others may develop an aversion to food.

Hypoglycaemia

Infants and children with SRS are at increased risk of hypoglycaemia (recurrent episodes of low blood sugar levels). This is likely to be due to their lack of subcutaneous fat, poor appetite and large head size. This is



most likely to happen when a child does not eat for an extended period of time. Symptoms associated with hypoglycaemia include weakness, hunger, dizziness, sweating and/or headaches. However, studies have found that infants with SRS can have night-time hypoglycaemia with little or no physical symptoms. Excessive sweating is common, particularly at night. Sometimes this can be a sign of hypoglycaemia, but not all SRS children who sweat a lot have low blood sugar levels.

Developmental milestones, learning and behaviour

Motor development skills may be delayed due to low muscle tone (hypotonia) and relatively large head size, especially in infancy and toddlerhood. Delay in speech development is also common, particularly in those patients with maternal uniparental disomy of chromosome 7 (mUPD7) (see 'What causes SRS?', below). Early intervention (physio, occupational and/or speech therapy) is important. Parents should ask their paediatrician for more information and refer as and if appropriate.



Taylor aged 3 years

The majority of children with SRS have normal intelligence. However, there is evidence for differences in frequency of learning and/or behavioural problems between the different genetic subtypes of SRS, with greater risk for children with maternal uniparental disomy of chromosome 7(mUPD7).



Genitourinary abnormalities

A variety of abnormalities affecting the organs of the reproduction and urinary systems have been reported, including failure of one or both testes to descend into the scrotum (cryptorchidism), abnormal placement of the urinary opening on the underside of the penis (hypospadias) and under-development of the uterus and upper part of the vagina (Rokitansky syndrome). Structural kidney (renal) abnormalities may also occur. Genitourinary abnormalities are also found at increased frequency in children who are born small-for-gestational-age and who do not have SRS.

Additional features

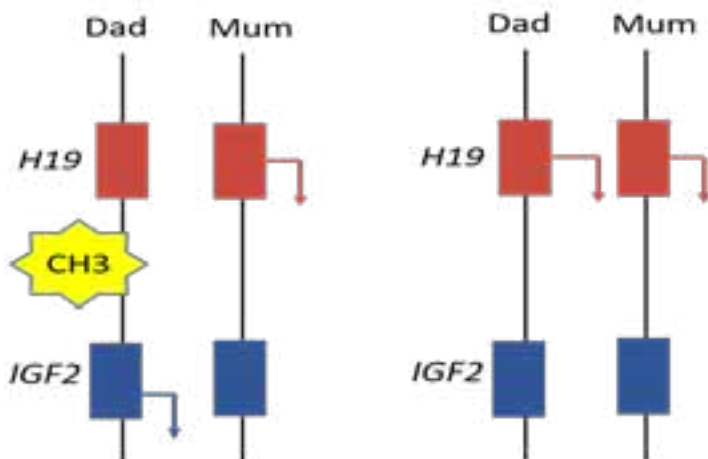
Other features have been described in the medical literature with varying frequency. These include:

- Curvature of the spine (scoliosis)
- Hip dislocation
- Short and in-curving 5th fingers (clinodactyly)
- Fingers that are fixed in a bent position (camptodactyly)
- Webbing of the second and third toes (syndactyly)
- Structural heart or kidney defects
- Cleft palate (an opening in the roof of the mouth).

Congenital anomalies are more common in children with loss of methylation on chromosome 11p15 (see 'What causes SRS?', P12).



Chromosome region 11p15 11p15 Loss of Methylation in SRS



On left: ICR1 in chromosome region 11 p15 contains two imprinted genes. H19 (in red) is expressed from the copy inherited from mum. IGF2 (in blue), a growth promoting gene, is expressed from the copy inherited from dad. Arrows represent gene expression. Methylation (CH3; in yellow) of dad's chromosome causes H19 to be switched off and IGF2 to be switched on. Mum's copy is not methylated, which gives the opposite pattern of gene expression.

On right: In some patients with SRS, loss of methylation on dad's copy causes H19 to be switched on and IGF2 to be switched off (like mum's copy). As a result, there is no expression of IGF2, which in turn results in poor growth.

Approximately 1% of individuals with SRS have been shown to have variants (mutations) in genes in the IGF2 pathway (IGF2, HMGA2, PLAG1) or CDKN1C. Single gene variants are most often seen in rare familial cases of SRS and testing for these is currently not routinely arranged.



What Causes SRS?

In the last few years it has become possible to confirm the clinical diagnosis by genetic testing in around 60% of individuals with SRS. Two main genetic changes (involving chromosome 7 and chromosome 11) are currently known to cause SRS. These are specific to the condition and not seen in children with IUGR and poor postnatal growth but no other features of SRS.

Chromosomes, which are present in the nucleus of human cells, carry the genetic information (genes) for each individual. We normally have 46 chromosomes. Pairs of human chromosomes are numbered from 1 to 22; the sex chromosomes are known as X and Y. Males have one X and one Y chromosome; females have two X chromosomes. Each chromosome has a short arm (“p”) and a long arm (“q”). Chromosomes are further sub-divided into many bands that are numbered. For example, “chromosome 11p15.5” refers to band 15.5 on the short arm of chromosome number 11. The numbered bands specify the location of the thousands of genes that are present on each chromosome.

Everyone has two copies of each gene – one inherited from the father and one inherited from the mother. In most cases, both genes are “turned on” or active. However, some genes are preferentially silenced, or “turned off”, based upon which parent that gene came from. This phenomenon is known as genetic imprinting. Genetic imprinting is controlled by chemical switches through a process called methylation. Accurate genetic imprinting is necessary for normal development. Problems with imprinting have been associated with several disorders, including SRS.



Chromosome 11

Imprinted genes tend to be found clustered or grouped together. Several imprinted genes are found in a cluster on chromosome 11p15.5. This cluster is divided into two functional regions known as imprinting centre regions (ICR1 and ICR2). Researchers have identified several imprinted genes regulated by these imprinting centres. These genes play a critical role in the regulation of fetal growth. Abnormalities in this region have also been shown to cause Beckwith-Wiedemann syndrome, an imprinting disorder which results in overgrowth.

Around 30-60% of cases of SRS have been shown to be due to changes (loss of methylation/ hypomethylation) affecting the ICR1 region on chromosome 11 (11p15 LOM). This, in turn, affects the activity of two genes (maternally expressed H19 and paternally expressed IGF2) which are believed to play a role in the development of SRS. Further research is necessary to learn more about the role of these genes and the complex genetic mechanisms responsible for SRS.



Emily aged 2 years



Georgia aged 2 years

Chromosome 7

Around 5-10% of individuals with SRS have been found to have both copies of chromosome 7 from their mother, rather than one from each parent. This is called maternal uniparental disomy of chromosome 7 (mUPD7). The exact way in which this affects growth and development is not fully understood, though this is likely to be due to increased activity of maternally-expressed gene(s) and/or under-activity of paternally-expressed gene(s) on chromosome 7.



Clinical SRS

The genetics underlying SRS are complex and the specific reasons for the development of the symptoms of this disorder are not fully understood. Currently, genetic testing for the currently known causes of SRS is normal in around 40% of children who have a clinical diagnosis. Research is ongoing to try to identify the underlying cause(s) in this group of children.

Other imprinting disorders

Rarely, other disorders of genomic imprinting can result in clinical features of SRS. Additional testing for these conditions may be considered in children with overlapping features. For example, changes affecting an imprinted region on chromosome 14q32 result in a condition known as Temple syndrome. Children with this condition commonly have IUGR, poor postnatal growth, low muscle tone, delay in development of motor skills and early puberty - all features which can be seen in SRS. Asymmetry is rarely a feature in Temple syndrome.

Around 25-30% of children with SRS due to 11p15 LOM also have LOM at other imprinting regions on ICR2 and/or other chromosomes. This is known as multi-locus imprinting disturbance (MLID). The clinical significance of this finding is not yet well understood and testing for MLID is currently only available on a research basis in the UK.

How is SRS Diagnosed?

The diagnosis of SRS is based on clinical findings. Because many of the symptoms are nonspecific, making a diagnosis of SRS remains difficult. Consensus guidelines for investigation and diagnosis of SRS have recently been published, based on the Netchine-Harbison clinical scoring system for SRS (see next page).



Netchine-Harbison clinical scoring system

Clinical diagnosis is considered if patient scores at least 4 out of 6 from the following criteria:

Testing for known genetic causes of SRS (chromosome 7 and 11) can confirm the clinical diagnosis in up to 60% of individuals. Knowing the underlying genetic cause can also help guide treatment as some problems are more common in association with abnormalities of chromosome 7 or 11.



James aged 5 years

It is important to remember that a positive SRS molecular test result confirms a SRS clinical diagnosis, but a negative test result does not exclude a clinical diagnosis.

Clinical Criteria	Definition
SGA (birth weight and/or birth length)	≤ -2 SDS for gestational age
Postnatal growth failure	Height at 24 \pm 1 months ≤ -2 SDS or Height ≤ -2 SDS from mid-parental target height
Relative macrocephaly at birth	Head circumference at birth ≥ 1.5 SDS above birth weight and/or length SDS
Protruding forehead	Forehead projecting beyond the facial plane on a side view as a toddler (1-3 years)
Body asymmetry	Leg length discrepancy (LLD) of ≥ 0.5 cm or arm asymmetry or LLD < 0.5 cm with at least two other asymmetrical body parts (one non-face)
Feeding difficulties and/or low BMI	BMI ≤ -2 SDS at 24 months OR current use of a feeding tube or cyproheptadine for appetite stimulation

Abbreviations: BMI, body mass index; SGA, small for gestational age.

Table from Nature Reviews Endocrinology (2017) 13,105–124.



Other diagnoses to consider

Poor growth prior to birth is a feature of many different congenital disorders. Although some of these disorders may have signs and symptoms that are similar to SRS, they usually have other physical features that help differentiate them. For example, relative microcephaly (head size smaller than expected for height and weight) is almost never seen in SRS. Alternative diagnoses should also be considered if there is global developmental delay or significant intellectual disability, no history of feeding difficulties, distinctive facial features (different from those described in SRS), additional congenital anomalies and/or other features atypical for SRS.

3M syndrome is an extremely rare genetic disorder characterised by low birth weight, short stature, distinctive facial features, and subtle skeletal changes. Characteristic facial features include a long, narrow head, an unusually prominent forehead and a triangular-shaped face with an up-turned nose and full lips. Affected individuals may also have additional features including prominent heels and/or increased flexibility (hypermobility) of the joints. Intelligence is normal. 3M syndrome is inherited as an autosomal recessive genetic trait.

Chromosomal instability disorders such as Fanconi anaemia, Bloom syndrome, and Nijmegen breakage syndrome may have similar features to SRS. In individuals with these disorders, the chromosomes within their cells are unstable and break and rearrange easily (chromosome instability), damaging DNA found within cells. In most people, damage to DNA is repaired. However, in affected individuals, breaks and rearrangements occur more often and their bodies are slow or fail to repair the damage, which in turn affects the genetic code. These disorders are commonly associated with IUGR and short stature as well as additional features, including small head size (microcephaly), limb



abnormalities, frequent infections and an abnormal sensitivity of the skin to ultraviolet (UV) light (photosensitivity). It is important to recognise these diagnoses as growth hormone treatment is contraindicated in this group of disorders.

Other rare conditions which may cause similar features to SRS are listed in tables 3 and 4 of the consensus guidelines.

Management Recommendations

Early diagnosis and intervention can help improve growth and ensure that affected children reach their highest potential. Treatment needs to be tailored to each individual child and their particular needs. Management will often require the coordinated efforts of a team of specialists including:

- Local paediatrician
- Paediatric endocrinologist (management of growth and puberty)
- Paediatric gastroenterologist (feeding problems, reflux, etc)
- Dietician
- Orthopaedic surgeon (asymmetry, scoliosis, hip dislocation)
- Orthodontist/ dentist
- Community paediatrician (developmental assessment)
- Speech therapist
- Physiotherapist
- Occupational Therapist
- Psychologist



Parents may also wish to be referred for genetic counselling if they are planning to have further children.

Recommendations for management are described in further detail in the published consensus guidelines:

<http://www.nature.com/nrendo/journal/v13/n2/full/nrendo.2016.138.html>

Growth and puberty

Failure to thrive is very common in children with SRS, due to a combination of feeding difficulties and gastrointestinal problems, such as reflux. In the first 2 years of life the main goal is to ensure adequate intake of calories. This, in turn, will allow growth, avoid malnutrition and help maintain blood sugar levels. In some cases, a feeding tube may be necessary to assist feeding. Initially a nasogastric tube (a thin tube that runs from the nose to the stomach through the oesophagus) may be used. If feeding difficulties are severe and persistent, a gastrostomy tube (inserted directly into the stomach through a small incision in the abdomen wall) may be needed.

It is, however, important not to overfeed a child with SRS (which can occur quickly, especially with feeding tubes). There is evidence that rapid weight gain in early childhood can lead to problems with the body's metabolism and increased risk of high blood pressure and heart disease later in life. Babies born small-for-gestational-age should therefore remain well-nourished but stay lean. It is important to monitor weight-for-height. Increasing the calories of a child with SRS can result in a brief spurt of length/height growth which often then levels off. The child then simply becomes more overweight rather than gaining any further height.



Several studies have now shown that growth hormone (GH) therapy significantly improves childhood growth and final adult height in SRS. SRS patients and non-SRS small-for-gestational-age (SGA) patients gain a similar amount of incremental height on GH therapy (about 7-9cm). Additional benefits of GH treatment are increased appetite, lean body mass and muscle power, which can result in improved motor development and reduced likelihood of hypoglycemia.

In the UK, there is a licence for GH therapy for children who were born SGA who have not displayed adequate catch up growth by the age of 4 years. A low starting dose of GH is recommended and should be adapted to growth velocity. Most negative side effects of GH treatment are transient, meaning they go away after stopping GH. SRS children and non-SRS SGA children have the same risk of experiencing negative side effects, and this risk is quite small. IGF-1 levels (which are routinely measured during GH therapy) are often high in children with SRS, especially in those with 11p15 LOM.



Louise aged 1 year

GH deficiency is uncommon in SRS and GH stimulation testing can have risks as it requires fasting. In addition, children born SGA who are GH deficient do not respond any differently to GH treatment than those who are not GH deficient. GH stimulation testing is therefore not recommended for most SRS children.

Before puberty, children typically enter an early stage of sexual maturation known as adrenarche. In children with SRS, bone age starts to advance at around 7 or 8 years, when they enter adrenarche. This



may happen even earlier, especially if there is a period of rapid weight gain. Children may then enter true ('central') puberty, which accelerates bone age even further. If not diagnosed and treated, this can lead to a reduced final height, even if the child has been treated with GH. From mid-childhood, children with SRS need to be monitored closely by a paediatric endocrinologist to look for early signs of adrenarche and puberty. If necessary and appropriate, puberty can be delayed by using a medicine known as gonadotropin-releasing hormone analogue (GnRHa).

Feeding difficulties

It is important to consider the possibility of underlying gastrointestinal problems and to treat these effectively as early as possible.

Gastroesophageal reflux can result in arching of the back and/or a tendency to bring feeds back up; it can also be "silent", with almost no physical symptoms. Acid reflux can be helped by providing smaller, more frequent meals and upright positioning of babies so gravity can help prevent food from flowing back up into the oesophagus.

Medications to help prevent reflux may also be prescribed. In rare cases of severe gastroesophageal reflux, (especially when a gastrostomy tube is being placed), a surgical procedure known as fundoplication may be recommended. During this surgical procedure, the upper curve of the stomach is wrapped around the lower portion of the oesophagus. This procedure strengthens the valve between the oesophagus and stomach and helps prevent acid reflux.

Decreasing the quantity of foods high in fat and providing smaller, more frequent meals can help improve delayed gastric emptying.

Constipation is also common in SRS and can cause a child to feel full so they do not want to eat. A diet rich in fibre and fluids will help.

Medication may also be prescribed to help with this.



Hypoglycaemia

Management of hypoglycaemia includes frequent feeding, dietary supplementation and the use of complex carbohydrates such as cornflour. To avoid low blood sugar levels, children with SRS should never go without food for long periods (even for medical procedures, such as surgery). They should go to A&E for glucose infusion when they are ill and unable to eat food by mouth. If a child with SRS is to have elective surgery, it



Emma and Lyla, both aged 5 years

will be important that the overseeing doctor puts measures in place to monitor and avoid hypoglycaemia. It is helpful for parents to be taught to measure ketones in the urine as an early warning sign, particularly when a child is unwell.

Developmental milestones, learning and behaviour

Some children with SRS, particularly those with mUPD7, may need additional support with development and learning. Early intervention is important to ensure that they reach their potential. Special services that may be beneficial include physiotherapy, occupational therapy, speech and language therapy and other medical, social, and/or vocational services. An individual education plan (IEP) may be developed to support children in school if special services are required; for some children an Education, Health and Care Plan (EHC Plan), may be appropriate.

Speech problems are common (especially in children with mUPD7) and speech and language therapy may be recommended. A hearing check



should also be performed to rule out hearing loss as the cause of speech problems.

A recent study indicates that some individuals with SRS have difficulty with autistic traits and these tend to be more common in individuals with SRS mUPD7. It is important for families and clinicians to be aware of this increased likelihood of Autism Spectrum Disorder (ASD) as, in some cases, a full assessment for ASD may be appropriate. In addition, individuals with SRS mUPD7 may be more likely to have difficulty with learning, compared to their peers so it is important to consider whether additional support with learning and development may be useful. It is important to note that there was variability within each group, indicating that some individuals may have more difficulty with autistic traits or learning than others. Therefore, a referral for additional support or services may be beneficial for some individuals with SRS but not required for others.

However, it is important to stress that there is considerable variability between individuals and only some affected children will need additional help.

Dental problems

Braces and oral surgery may be needed to correct dental problems, such as crowding of the teeth.

Asymmetry

Difficulties can sometimes arise with walking due to limb asymmetry. Any child with significant asymmetry (lower leg length difference over 0.5cm) should be seen by an orthopaedic surgeon. Special braces and shoes may help improve balance and gait. In a small number of cases, surgical intervention may eventually be required; this is usually performed when growth has ceased.



Genitourinary problems

Cryptorchidism (undescended testicles) can sometimes resolve spontaneously, although some boys require surgical treatment. Hypospadias requires surgery, ideally by an experienced paediatric surgeon.

Psychosocial issues

Short stature and other medical issues can lead to problems with self-image in some children, adolescents and adults. Referral for psychosocial support may be beneficial in those experiencing issues with self-image, peer relationships and other social interactions.

SRS in Adulthood

Research about the long-term health of adults with SRS is limited and most adults with SRS are not routinely followed up. Diagnostic scoring systems used to diagnose SRS in childhood are not useful in older people as the clinical features of SRS become less obvious with increasing age. This means that making the diagnosis of SRS in adults is more challenging so if a diagnosis is suspected, early molecular confirmation is recommended.

It is well recognised that being SGA at birth with accelerated gain in weight for length, particularly in early life, increases the risk of metabolic problems in adulthood. A recent study of individuals with SRS aged ≥ 18 years recorded impaired glucose tolerance (predisposing to diabetes) in 25%, high blood pressure in 33% and high cholesterol levels in 52%. Affected individuals can be monitored for metabolic problems in adulthood by their GP who can refer on for specialist advice if needed. In adults with SRS, other health problems have been highlighted (e.g. muscle and joint pains) but there remains uncertainty



as to whether such problems are seen more frequently when compared to adults without SRS.

Overall, however, most adults with SRS will have a



Emily age 14, Megan age 17 and Georgia aged 18 years

normal quality of life, educational attainment and normal fertility.

Very little research has been published about the long-term health of adults with SRS. Most adults with SRS are not routinely followed up, and the small numbers of adults reported have had very few medical problems. However, it is well recognised that being SGA at birth with accelerated gain in weight for length, particularly during early life, increases the risk of metabolic problems in adulthood.

Can this happen again?

In most families, only one child is affected and the chance of parents having another baby with SRS is likely to be very low. Similarly, the chance of an individual with SRS having an affected child themselves is also likely to be very low. However, in rare cases, familial occurrence of SRS has been noted and the risk of recurrence can be as high as 50%. Genetic investigation is important before parents are advised about recurrence risk. Genetic counselling may be of benefit for parents planning another pregnancy and affected individuals who are thinking of starting a family of their own.



Further Support

If you are still unsure, or concerned about anything you have just read, the Child Growth Foundation operates a support service, by telephone or email, that may be able to help further.

You can email the CGF at:

info@childgrowthfoundation.org

Or call us on:



0208 995 0257

Our website has further information that you may find helpful:

childgrowthfoundation.org

We also manage a number of closed patient support groups through Facebook visit our main page to find out more:

facebook.com/childgrowthfoundation/

Overseas Support

Further information and support for people outside of the UK can be found at the following:

- USA - www.magicfoundation.org
- Worldwide - <https://silverrussellsyndrome.org/>
- NORD - rarediseases.org/rare-diseases/russell-silver-syndrome/



References

Wakeling E, Brioude F, Lokulo-Sodipe O, et al. Diagnosis and Management of Silver-Russell syndrome: first international consensus statement. *Nat Rev Endocrinol.* 2017;13105-124.

A PDF version of this consensus paper can be downloaded from:

<https://www.nature.com/nrendo/journal/v13/n2/pdf/nrendo.2016.138.pdf>

A summary of the consensus guidelines, written for affected individuals and their families can be found on the CGF website.

A full list of references regarding the diagnosis and management of SRS can be found at the end of the consensus paper. Abstracts or original articles can be found on the internet in PubMed:

<https://www.ncbi.nlm.nih.gov/pubmed/>

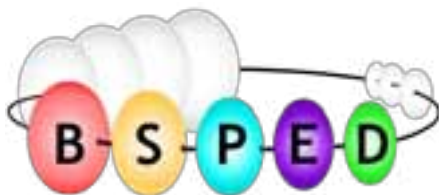
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Endorsement

This guide has been endorsed by the British Society for Paediatric Endocrinology and Diabetes (BSPED). The BSPED aims to improve the care of children and young people with endocrine disorders or diabetes mellitus, by bringing together professionals from a range of disciplines.

www.bsped.org.uk





Further Information

Further Information:

If you have any questions regarding the information contained in this sheet, then please contact:

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Web: <https://childgrowthfoundation.org>

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FUNDING

The Foundation funds research into many aspects of growth conditions such as the causes, effects, treatments and psychological impact. It also offers essential advice and experience to parents of children who have been diagnosed with growth problems. The annual convention provides a great forum for people to get together to discuss problems and solutions with others in a similar position. It also provides a chance to meet and learn from the doctors and professors dealing with child growth in the UK.

The CGF is entirely self-sufficient and is an independent charity. It relies on donations and membership subscriptions to keep going. If you have found this information leaflet helpful, please consider becoming a member and/or making a donation - www.childgrowthfoundation.org.



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