

# MAGIC NZ

Supporting Children with Growth Disorders  
and their Families



[www.magicnz.org.nz](http://www.magicnz.org.nz)

## *Russell-Silver Syndrome*

### Introduction

In 1953 and 1954, Drs Silver and Russell independently described groups of small-for-gestational-age [SGA] children whose pregnancies had been complicated by intrauterine growth retardation [IUGR]. Their common findings were short stature without catch-up growth, normal head size for age, a distinctive triangular face, low-set ears and incurving fifth fingers. These two groups of patients are now considered to have had variations of the same disorder that we now call Russell-Silver Syndrome [RSS] in the U.S. and Silver-Russell Syndrome [SRS] in Europe.

One interesting and important aspect of the Russell-Silver syndrome is its variation in phenotype. In this context, a phenotype is all the physical characteristics and abnormalities found in an individual patient that are attributed specifically to RSS. Some individuals with RSS have many traits, while others have very few.

### RSS versus SGA: How is RSS Diagnosed?

The diagnosis of RSS is still a judgment call on a physician's part because there is no definitive laboratory test that can answer yes or no in a specific case. Doctors generally base their diagnosis on characteristic, clinical findings that make up the RSS phenotype.

Distinguishing an RSS child from a non-RSS/SGA child is not necessarily straightforward. Reading the information in this brochure and referring to the separate "Small-for-Gestational Age" brochure from The MAGIC Foundation will give you a better understanding of RSS and SGA.

### Characteristics Considered to Distinguish RSS Children From Other SGA Children:

- body asymmetry - LARGE side is "normal" side
- inadequate catch-up growth in first 2 years
- persistently low weight-for-height
- lack of interest in eating
- lack of muscle mass and/or poor muscle tone
- broad forehead
- large head size for body size
- hypoplastic (underdeveloped) chin & midface
- downturned corners of mouth & thin upper lip
- high-arched palate
- small, crowded teeth
- low-set, posteriorly rotated and/or prominent ears
- unusually high-pitched voice in early years
- clinodactyly (inward curving) of the 5th finger
- syndactyly (webbing) of the 2nd and 3rd toes
- hypospadias - abnormal opening of the penis
- cryptorchidism - undescended testicles
- café-au-lait (coffee-with-milk) birth marks
- dimples in the posterior shoulders and hips
- narrow, flat feet
- scoliosis - curved spine, associated with spinal asymmetry and accentuated by a short leg

### Characteristics of SGA Patients in General That Are Seen More Often in RSS Patients:

- fasting hypoglycemia & mild metabolic acidosis
- generalized intestinal movement abnormalities:
- esophageal reflux resulting in movement of food up from stomach into food tube
- delayed stomach emptying resulting in vomiting or frequent spitting up
- slow movement of the small intestine &/or large intestine (constipation)
- blue sclera (bluish tinge in white of eye)
- late closure of the anterior fontanel (soft spot)
- frequent ear infections or chronic fluid in ears
- congenital absence of the second premolars
- delay of gross and fine motor development
- delay of speech and oral motor development
- kidney abnormalities
- delayed bone age early, later fast advancement
- early pubic hair and underarm odor (adrenarche)
- early puberty or rarely true precocious puberty
- classical or neurosecretory growth hormone deficiency
- ADD and specific learning disabilities

## What Should I Do If I Think My SGA Child Has RSS?

- Have your child's diagnosis confirmed by an endocrinologist who is familiar with RSS-SGA patients.
- Make sure your child is measured carefully & frequently. KEEP YOUR OWN RECORDS.
- Find a paediatrician who is willing to learn from experts about RSS-SGA children and who will coordinate care and opinions with consulting specialists.
- Get adequate calories into your child. Insufficient nutrition & low blood sugar damage the developing brain and compound the growth failure.
- Take necessary measures to prevent hypoglycaemia in young RSS children. Pay special attention to the night when everyone is asleep, anytime your child is ill or not eating normally, and when your child is unusually active or stressed.

### **Prevent hypoglycaemia by:**

- feeding frequently during the day & night
- keeping snacks with you at all time
- feeding through gastrostomy tube
- making prior arrangements with your doctor and local ER to start IV glucose if feeding is impossible
- having urine ketone sticks at home

Seek appropriate consultation for recurrent ear infections, hypospadias, undescended testicles, leg length discrepancies, etc. *But remember:*

- Only emergency surgery should be done until the child is gaining weight well.
- A young SGA child should NEVER be fasted or kept NPO for more than 4 hours for ANY reason without glucose-running IV.
- For surgery, IV glucose should be given during the procedure and continued in the recovery room.

## Why Does My Child Have RSS?

It is not your fault! You could not have done anything to prevent it! RSS occurs through complicated genetic mechanisms and could never be caused by you.

## What Can I Expect Regarding My Child's Cognitive Abilities?

An infant with RSS is generally born with normal intelligence. Learning disabilities and Attention Deficit Disorder (ADD) appear to be increased in incidence in RSS. Autism and similar disorders like pervasive developmental disorder (PDD) may also be increased. It is unclear whether these problems just appear to be increased in RSS, are innate to RSS or are acquired through early malnutrition and hypoglycaemia, both of which are preventable.

## What Treatments are Available for RSS?

For RSS and non-RSS/SGA patients, the prospect for a normal life with a normal adult height is closer than ever before. Please contact us if you have questions about treatment options for your child, have difficulty arranging medical care for your child or are interested in learning more about ongoing research in the field. Since the treatment for RSS children and non-RSS/SGA children who have failed to achieve catch-up growth is similar, the information listed below is the nearly the same as the information provided in the "Small-for-Gestational Age" brochure from The MAGIC Foundation.

## How Can I Help my RSS Child Grow?

Simplistically, there are two aspects of the RSS child's growth -- weight and height. Particular attention should be paid to weight gain during the first 2 to 3 years of life, as many children born RSS may struggle to gain weight (although ironically, some children born RSS may gain excessive weight during childhood). As previously discussed, a paediatric GI doctor can help diagnose and treat, or rule out, any GI medical issues that may impede an RSS child's ability to gain weight. Reflux (often silent) and delayed gastric emptying appear to be the most common of these GI problems. A nutritionist can also help provide advice on simple additives and other means of adding calories to a child's diet. There are also medications like the antihistamine "cyproheptadine" (brand name: Periactin) which can act to increase an RSS child's appetite. Please contact MAGIC for more in-depth literature on this topic.

The second growth aspect is length/height. Inadequate caloric intake may contribute to growth failure. If optimum caloric intake can be attained, some "catch-up" in weight and height growth curves may occur. If a child is still significantly short after this period, and the child is 2 to 3 years old or older, the family may consider growth hormone therapy (GHT) in order to increase the child's height growth velocity.

It is important to note two things. First, research has not found any means except growth hormone therapy to stimulate catch-up growth, maintain a normal height during childhood, and increase an SGA child's adult height (the majority of RSS children are born SGA and appear to respond to GHT in a similar way). The United States F.D.A. approved growth hormone therapy as "...long-term treatment of children who were born SGA and who have not achieved catch-up growth by the age of 2." Second, most SGA children are not GH deficient according to standard testing measurements. Studies are ongoing, but it appears that many of these short SGA children do not respond normally to growth hormone and thus require more growth hormone than the typical child. Instead of being growth hormone "deficient", experts now consider SGA children to be growth hormone "insufficient."

## Growth Hormone Therapy

In many developed countries, growth hormone therapy is the recommended and available treatment for SGA children who fail to achieve catch-up growth by age two. However, many RSS/SGA children do not meet the entry criteria to receive publicly funded GHT in NZ. This does not mean that GHT is not the appropriate treatment for your child. Growth hormone therapy is the only treatment currently available to increase the RSS/SGA child's height. Not all parents choose GHT for their child. We at MAGIC will support you in whatever choice you make for your child. When GHT is medically indicated, MAGIC NZ would like the entry criteria to give parents the right to accept or decline treatment as a medical, not a financial, decision.

You need to find a doctor who is willing to help you in this decision process by focusing on the medical aspects of growth hormone therapy. Factors to discuss with your doctor may include potential incremental height for your child (based on the parents' heights), whether your child can benefit from the possible added muscle mass and strength, your child's age, bone age and other health issues of your child. Growth hormone therapy requires a commitment. However, the initial commitment can be for a trial period of at least six months to determine growth response. The injection is a simple subcutaneous (top layers of skin) injection every night. Most children and parents who have chosen this treatment think it has turned out far better and far easier than they expected. If you are interested in talking with other families who have chosen or declined growth hormone therapy, MAGIC can connect you.

## Does a Delayed Bone Age Mean My Child Will Have Catch-Up Growth Later?

NO. Although most if not all RSS/SGA children have a bone age that is relatively delayed compared to their chronological age, studies have shown bone age to be an UNRELIABLE predictor of adult height in SGA children. One possible reason is that many RSS/SGA children experience a rapid acceleration of their bone age just prior to the onset of, and during, puberty. Within a span of just 12-18 months, an RSS/SGA child's previously delayed bone age can quickly surpass his chronological age, negating any "extra growing time" that usually is present with delayed bone age. Growth hormone therapy should not be declined in the expectation that the RSS/SGA child will attain catch up growth due to their delayed bone age.

An RSS/SGA child's bone age should be regularly monitored throughout childhood to see how quickly it is advancing. For the older RSS/SGA child, bone ages are also used to determine if any incremental height remains for the child (as long as the bone plates are open and not fused).

## Other Health Issues Associated with Being Born SGA

Multiple studies, short-term and longitudinal, have found an increased risk of health problems such as insulin resistance, cardiovascular disease, hypertension, obesity, and type-2 diabetes among adults who were born SGA or with low-birth weight. Explanations for these risks vary from intrauterine nutrition to genetic causes. In addition, some correlations have been found between persistent short stature and psychosocial difficulties and/or behavioral problems. Clearly, a great deal of research still needs to be done to narrow down and clarify the exact risks of being born SGA and identify which children are at risk. Until then, children and adults born SGA should be monitored carefully by their physicians in light of these possible risks.

## A Note to Parents

You have learned that your baby or child has RSS/SGA. You may have a lot of confusion or be frightened regarding the well being of your child. As a concerned parent, you probably wish to learn as much as you can about the condition and what you and your health care professional can do to help your child as he/she grows and develops.

**Advocate for Your Child:** Most importantly, be your child's #1 advocate, trust your parental gut instinct and love your beautiful RSS child. Treatment is available to help your child. The appropriate medical care under the proper specialists will greatly improve your child's outcome. The treatment of SGA children's problems should be approached in a systematic and timely fashion. The major problems that require intervention in the various age periods are all different, but most of these problems can be solved or dealt with successfully if you get the help you need.

**Ask Questions:** As you learn about RSS/SGA, you will probably have questions that are specific to your child. Leave no questions unanswered, even if you think the questions are simple or silly. Don't be afraid to ask questions or get a second opinion from another specialist. A greater understanding of this condition will allow you to provide optimal care for your child.

**Network:** Coping with the time-consuming special attention and services necessary to care for an RSS child can be overwhelming, especially if you try to face it alone. Good physicians may have no experience with routine needs of RSS children. Day-to-day challenges such as feeding, formulas, fitting clothes, school issues and peer pressures can be less stressful if you are in contact with other families who "have been there". MAGIC NZ can put you in touch with other families affected by RSS/SGA - people who have experienced and solved problems similar to yours.

For more information visit [www.magicnz.org.nz](http://www.magicnz.org.nz), email [jan@magicnz.org.nz](mailto:jan@magicnz.org.nz)  
or write to MAGIC NZ, PO Box 1493, Wellington.

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