

## Russell-Silver Syndrome

In 1953 and 1954, respectively, Dr. H. K. Silver and Dr. A. Russell independently described a special group of IUGR (Intrauterine Growth Retardation) children with short stature along with a small triangular face, low-set ears, incurved fifth fingers, and other characteristics. Their independent and varied findings were eventually determined to be a variation of the same syndrome. (A syndrome is a group of physical signs and symptoms which make up a specific disease or disorder.) As a result, the syndrome received its name.

Russell-Silver Syndrome (RSS) is also known as Silver-Russell Syndrome, Silver Syndrome, and Russell Syndrome.

One of the interesting and at the same time unsettling aspects of this syndrome is the very wide variation of its characteristics. Some individuals with RSS have many of the documented traits, while others have very few. Mental retardation is not characteristic of this disorder.

### ***Almost, without exception, every RSS individual has:***

- Low birth weight (Intrauterine Growth Retardation)
- Decreased birth length
- Triangular shaped face (lessens with age)
- Scaphocephaly (long narrow head) at birth
- Normal head size appearing large because of reduced body length and weight
- Postnatal growth retardation
- Poor appetite in early years
- Fifth finger clinodactyly (incurving)

### ***Common characteristics:***

- Hypoglycemia (low blood sugar) in infancy and early childhood (2-3 years)
- Asymmetry (unequal in length or size)
- Late closure of the fontanel (soft spot)
- Broad forehead
- Hypoplastic mandible (small chin)
- Downturned corners of the mouth
- Thin upper lip
- Crowding of teeth
- Microdontia (small teeth)
- Unusual, high-pitched voice (usually disappears in later years)
- Abnormal ears (low-set, small, and/or prominent)
- Syndactyly of the toes (fused or webbed - degree of fusing varies)
- Hypospadias (abnormal location of urethral opening)

- Cryptorchidism (undescended testicles)
- Delayed bone age
- Weak muscle tone
- Developmental delays

***Rarer traits:***

- Hydrocephalus
- Blue sclerae (a blue tinge in the whites of the eye)
- High-arched palate
- Congenital absence of the second premolars
- Frequent ear infections or chronic fluid in the ear (can result in temporary hearing loss)
- Migraine headaches
- Scoliosis (often due to asymmetry)
- Vertebral abnormalities
- Renal abnormalities
- Reflux
- Precocious (earlier than usual) puberty
- Growth hormone deficiency
- Cafe-au-lait spots (flattened, irregularly spaced oval spots which are usually the color of cream and coffee)
- High energy
- ADD
- Passing out spells

***What To Do If You Suspect Russell-Silver Syndrome:***

1. Consult a Geneticist to confirm or rule-out the diagnosis of RSS.  
 2. See a Pediatric Endocrinologist who will evaluate and follow the patient to determine what physical abnormalities need treatment. The rarity of this syndrome makes it difficult to find physicians familiar with RSS and treatment options. Some of the actions taken by the Pediatric Endocrinologist might include:

- Examining the patient's growth curve
- Documenting daily caloric and nutritional intake
- Measuring for asymmetry
- Examining male genitals for hypospadias and/or cryptorchidism
- Taking a bone x-ray to determine the bone age versus chronological age
- Testing for growth hormone deficiency
- Testing for hypoglycemia (A common characteristic of hypoglycemia is excessive sweating on the head & upper trunks)

3. Contact other RSS families. The Endocrinologist often only treats the physical problems. However, the syndrome poses many more concerns for families than can be addressed entirely by physicians. Day to day challenges, such as feeding techniques, finding clothes to fit, learning difficulties, etc., may be made less stressful and eased or eliminated by contacting other families of RSS. Any medical treatments discussed by other RSS families should be discussed with your doctor.

## ***Treatments***

Currently, there are various methods used by physicians and professionals to treat problems resulting from RSS. However, limited knowledge about RSS and the relatively recent use of several of these treatments often makes it difficult to determine their effectiveness and, consequently, recommend them or verify concrete results. It is very important you consult with a qualified professional before agreeing to any of the following:

- **Diet Change** - Hypoglycemia treatment, increase caloric intake, improve growth
- **Periactin** - Antihistamine used as an appetite stimulant
- **Feeding Pump** - Increase caloric intake
- **Recombinant Growth Hormone** - Used to increase growth velocity and final height
- **Gastrostomy** - Increase caloric intake
- **Ear Tubes** - Improve fluid drainage from ears
- **Lupron** - Attempt to delay puberty
- **Speech Therapy**
- **Remedial Reading**
- **Physical Therapy**
- **Occupational Therapy**
- **Shoe Lifts** – Asymmetry
- **Limb Lengthening Surgery** - Leg asymmetry
- **Corrective Surgery** - Various physical abnormalities

## ***Long Term Effects of RSS and Quality of Life***

The diagnosis of RSS does not mean a reduced quality of life. A person with RSS typically lives a normal life. However, their short stature may pose some challenges not encountered by average- sized individuals.

The early years are often the most difficult. For instance, poor eating; developmental delays; weak muscle tone; learning disabilities; asymmetry; and other traits cause concern and require intervention. This will be time-consuming, stressful, and create uncertainty. However, the extra effort and caring will pay off over time. Typically, the triangular-shaped face will lessen; muscle tone will strengthen; motor coordination will develop; appetite will improve; speech will become clear; learning will occur; and RSS infants will become healthy and happy adults.

It is recognized the severity with which an individual is affected by RSS varies. Certain traits can create more difficult obstacles to overcome than others.

## ***Why does my child have RSS?***

There is no known etiology (the study of all factors that may be involved in the development of a disease) of RSS. Most cases of RSS occur sporadically with no previous family history. However, there are rare occurrences where RSS is present in more than one member of a family. Research is continuing in an attempt to locate the source of RSS. There are several findings which indicate a possible location on a specific chromosome. However, there is no confirmation yet.

***It is extremely important you realize nothing you did before or during your pregnancy caused your child to have RSS. Most parents have trouble releasing***

***themselves from guilt. This is common. However, please try to ease your stress by understanding you are not responsible for the existence of RSS in your child.***

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The MAGIC Foundation is a national nonprofit organization created to provide support services for the families of children afflicted with a wide variety of chronic and/or critical disorders, syndromes and diseases that affect a child's growth. Some of the diagnoses are quite common while others are very rare.

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