Russell-Silver syndrome

**Eponyms:**
- asymmetry-congenital short stature sexual development variations
- RSS

**Inheritance:**
- autosomal dominant
- genetic heterogeneity
- genomic imprinting
- sporadic
- X-linked dominant

**Semeiological Synthesis:**
Cutaneous-facio-skeletal disorder. Low birth weight, normal head circumference, large head (pseudohydrocephalus), skeletal asymmetry, short stature, triangular face, clinodactyly, syndactyly, café-au-lait spots. Occasionally maternal uniparental disomy.

**Group**

<table>
<thead>
<tr>
<th>Sub group</th>
<th>Signs</th>
<th>DERMATOLOGICAL DISORDERS</th>
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<td>pigmentation changes</td>
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<td></td>
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<td>- cutis, café au lait spots</td>
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<td>sebaceous-sudoriparous glands, changes</td>
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<td>- hyperhidrosis, diaphoresis, hyperhidrosis</td>
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<td>subcutaneous changes</td>
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<td>- acromial dimple</td>
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<td>- subcutaneous dimples, skin dimples</td>
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**GENITAL DISORDERS**
- female genitalia, modified not including ambiguity
  - clitoris, hypertrophic, clitoromegaly
- genital dysfunctions
  - puberty, precocious; sexual precocity
- male genitalia, modifications not including ambiguity
  - cryptorchidism
  - hypospadias

**LABORATORY DATA**
- chromosomal assignment
  - chromosome 17q localization
  - chromosome 7p localization
- gene, structural-functional anomalies
  - gene analysis-DNA analysis
  - IGFBP1 insulin-like growth factor-binding protein-1, gene chr.7p14-p12
  - IGFBP3, gene chr. 7p12-13
  - RSS (SRS) Russell-Silver syndrome, gene chr.17q25
  - Russell-Silver syndrome (RSS), gene chr.7p.11.2
- pituitary hormones, modified functions
  - somatotrin (growth hormone-releasing factor, GHRF), low levels
  - plasma nonprotein-organic constituents, anomalies
  - hypoglycemia

**NEOPLASTIC DISEASES**
- cancer, genetic features
  - tumour susceptibility

**OCULAR DISORDERS**
- sclera, changes
  - sclerae blue

**OROCRANIOFACIAL ANOMALIES**
- facies, modified appearance
  - dysmorphic face
  - facial dysmorphism due to cranial changes including microcephaly
- facies, small, triangular, narrow
- proptosis, prominent eyes, exophthalmos, protuberant eyes, protruding eyes
forehead-orbital region, changes
- frontal bossing
lips, modified appearance
- lips, mouth, downturned corners
- lower lip, thin
mandibular changes
- micrognathia, mandibular hypoplasia, small jaw, not including: severe micrognathia, agnathia

OTHERS
inheritance
- inheritance, autosomal dominant
- inheritance, genetic heterogeneity
- inheritance, genomic imprinting
- inheritance, sporadic
- inheritance, X-linked dominant
supergroups
- cutaneous-genito-skeletal disorders
- facio-genito-skeletal-urinary
- facio-skeletal disorders

PRENATAL-NEONATAL MODIFIED DATA
foetal changes
- birth length, decreased; low birth length
- birth weight, low; foetal growth decreased, intrauterine growth retardation, intrauterine growth restriction, IUGR
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques
prenatal diagnosis
- prenatal diagnosis, echographic

SKELETAL DISORDERS
brachydactyly
- brachydactyly, brachyphalangy, in syndromic association
- brachymesophalangy
fingers, modified form, deformity
- clinodactyly
- fingers, broad, clubbing, including spatulate, stubbing, bullous
- Kirner deformity
fontanelles-sutures, changes
- fontanelles, cranial sutures, closure delayed
limb anomalies, limb defects
- asymmetric growth, hemihypertrophy, limbs asymmetry
ossification, changes
- bone age, delayed
skull shape, changes
- plagiocephaly, skull asymmetry
spine, changes
- sacral hypoplasia, agenesis
- vertebra, absence
stature, growth, modified habitus
- growth delayed, failure to thrive, growth retardation
- stature, short, including micromelia, including short limbs
- stature, very short
syndactyly
- syndactyly, in syndromic association
**vertebral changes**
- vertebral anomalies, unspecified type

**UROLOGICAL DISORDERS**
- kidney, malformations
  - kidney, malformation, including horseshoe kidney

**Super group:**
- cutaneous-genito-skeletal disorders
- facio-genito-skeletal urological disorders
- facio-skeletal disorders

**Super aggreg.**
**Aggregations:**
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques

**NEOPLASTIC DISORDERS**
- tumour, susceptibility

**OTHER**
- craniofacial dysmorphism due to cranial changes, including microcephaly
dysmorphic face

**Differential diagnosis:**
- 3400 Bloom syndrome
- 7830 Dubowitz syndrome
- 9855 floating-harbor syndrome
- 11910 hemihypertrophy, idiopathic
- 2455 Jung-Smith syndrome
- 14780 Levi dwarfism
- 16050 megalocornea-mental retardation syndrome
- 16820 MMM syndrome
- 17120 MULIBREY syndrome
- 21600 neonatal progeroid syndrome
- 27509 Partington syndrome
- 20960 Pitt-Rogers-Danks syndrome
- 101 placental chromosome mosaicism
- 6846 short stature-macrocrania-peculiar face syndrome
- 23760 SHORT syndrome
- 28775 triploidy

**Bibliography**
OMIM ID: 180860
- Smith's Recognizable Patterns of Human Malformation. 6th Edition pag.92-94
- Annuario Orphanet-Italia delle Malattie Rare, 2005 pag.262, 743