Russell-Silver Syndrome

- **Growth**
  - Low-birth-weight, short birth length
  - Small for gestational age
  - Normal mental development

- **Physical features**
  - Large fontanels; pseudohydrocephalic appearance
  - Triangular face (petite face and relatively large head)
  - Forehead nevus flammeus
  - Blue sclera
  - Asymmetry of arms and/or legs
  - Fifth finger clinodactyly
  - Precocious sexual development

- **Gene**
  - Methylation abnormalities in IGF2
Loss of DNA methylation in the telomere imprinting domain can cause RSS, due to loss of expression of IGF2.
Newborn with SGA birth and relatively large head and proportionate short stature.
Typical growth curve and average height of an affected girl. Affected babies usually are SGA with 1.2-2.4 kg. birth weight. There is proportionate short stature but normal head circumference (relative macrocephaly).
Proportionate short stature and leg length discrepancy of the infant’s right leg. In-curved little fingers (clinodactyly) in affected boy.