**INTRODUCTION**
Russell-Silver syndrome (RSS) is characterised by intrauterine growth retardation accompanied by postnatal growth deficiency. Affected individuals typically have proportionately short stature, normal head circumference, typical facial features (broad prominent forehead with small triangular face and small narrow chin), fifth finger clinodactyly and limb length asymmetry that may result from hemihypotrophy with diminished growth of the affected side. Children with RSS may also be at risk for developmental delay (both motor and cognitive) and learning disabilities. It is estimated that the prevalence of RSS is 1 in 100,000.

RSS can be inherited in an autosomal dominant or recessive manner. It is a genetically heterogeneous condition involving 3 distinct loci on chromosomes 7 and 11. Maternal duplication or paternal hypomethylation of $H19$ and $IGF2$, within the imprinting center region 1 (ICR1) at 11p15.5, has been found in up to 64% of RSS patients. Maternal uniparental disomy (UPD), interstitial deletions and submicroscopic duplications of chromosome 7 have been shown to result in RSS, with maternal UPD accounting for approximately 7-10% of cases. The underlying mechanism in approximately 30% of RSS cases is unknown.

Confirming a diagnosis of RSS is important as:
- Management of the syndrome requires that differential diagnoses are excluded
- Determining the disease mechanism enables recurrence risks to be given to the family
  - UPD and interstitial deletions are usually *de novo* events; therefore the recurrence risk is low (unless there is a predisposing parental chromosomal rearrangement).

**TESTING AND REFERRALS**
Diagnostic testing:
- referrals of clinically affected patients are accepted from Clinical Genetics and Paediatrics
- referrals from other specialties are accepted based on details of clinical symptoms
Prenatal referrals:
- only accepted from Clinical Genetics and, where possible, arranged in advance

**STRATEGY AND TECHNICAL INFORMATION**
- Methylation-sensitive multiplex ligation probe amplification (MS-MLPA) analysis of ICR1 at 11p15.5 ($H19$-$IGF2$)
- Microsatellite analysis for UPD7 is undertaken following a normal $H19$-$IGF2$ result
  - parental samples are required at this stage and will be requested prior to analysis

**TARGET REPORTING TIMES**
Diagnostic testing (MS-MLPA/UPD7 microsatellite analysis): 10 days
Prenatal testing (UPD7 - includes maternal contamination check): 3 days

*N.B. Details are correct for the date of printing only – last updated 02/09/2015*