

Pregnancy Out comes in Women with Silver Russell Syndrome

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Introduction:

Silver Russell Syndrome or Russell Silver Syndrome (SRS) is characterized by growth restriction, short stature, phenotypical features (protruding forehead, triangular face, clinodactyly, micrognathia, etc.) and body asymmetry. It has a prevalence of 1:30000 – 1:100000. At least 4 out of 6 from the Netchine–Harbison clinical scoring (NH-CS) system is required to diagnose this condition clinically.

Molecular abnormalities of several chromosomes have been identified to cause SRS. People with this condition will have metabolic abnormalities like diabetes and hypoglycaemia, early adrenarche and musculoskeletal disorders. They also have learning difficulties, growth and neuro-development delay and high- pitched voice.

A very little information exists in the literature about adult life of this group and their obstetric outcomes.

Case Report:

A 27-year-old G₂P₀ woman with SRS was presented for tertiary level obstetric care. Previously, she had a first trimester miscarriage.

She is a product of a consanguineous marriage and had an affected sister. She met the NH-CS system criteria for SRS and had additional phenotypical features of SRS. Her height was 124 cm. BMI was 12 kg/m².

Her cognitive development and cardiac assessment were normal. She had type two diabetes mellitus without end organ involvement. She did not have gynaecological concerns. She conceived spontaneously.

Pregnancy was managed with close monitoring with multidisciplinary assessment. Except the fetal growth which was on the 10th centile, her pregnancy remained uncomplicated, including a satisfactory glycaemic control with metformin. However physical difficulties due to the gravid uterus were significant. Mobility, eating and sleep became difficult for her after 20 weeks.

She had multiple hospital admissions due to lower abdominal pain. At 32 weeks she presented with preterm labour and received corticosteroids.

As she had regular progressive contractions without a descent of fetal head or cervical dilatation, a decision for abdominal delivery was made at 34 weeks and 2 days. Due to the extremely narrow pelvis, cephalopelvic disproportion was suspected.

General anaesthesia was given due to the risk of high spinal due to short height. There was a significant drop in the haemoglobin (120 g/L to 70 g/L) even though the blood loss during the lower segment caesarean section was only 400ml. She was started on a therapeutic dose of iron. Mother was given psychiatrists support and special help for lactation. Her recovery was uneventful. She handled the baby normally with the support of her parents.

Neonate had a low birth weight (2.014 kg, 3rd centile) but a normal length (46cm) and OFC (30.5cm). Dysmorphic features were absent.

Discussion:

- Four pregnancies in women with SRS have been reported in the literature. All of them had live births by Caesarean sections.
- Three patients had preterm deliveries (at 29, 34 and 36 weeks).
- None of the offspring had features of SRS, but they had SGA (n=4), cleft lip and palate (n=1) and one infant died due to pneumonia. GDM was occurred in one case.
- Due to short stature pregnancy is associated with significant physical difficulties in mobility, sleep, etc.

Conclusions:

- Successful pregnancy outcomes can be achieved with patients with SRS with careful monitoring and timely interventions.

Reference: Wakeling EL, Brioude F, Lokulo-Sodipe O, O’Connell SM, Salem J, Blik J, Canton AP, Chrzanowska KH, Davies JH, Dias RP, Dubern B, Elbracht M, Giabicani E, Grimberg A, Grønskov K, Hokken-Koelega AC, Jorge AA, Kagami M, Linglart A, Maghnie M, Mohnike K, Monk D, Moore GE, Murray PG, Ogata T, Petit IO, Russo S, Said E, Toumba M, Tümer Z, Binder G, Eggermann T, Harbison MD, Temple IK, Mackay DJ, Netchine I. Diagnosis and management of Silver-Russell syndrome: first international consensus statement. Nat Rev Endocrinol. 2017;13:105–24.