

RUSSEL-SILVER SYNDROME: A RARE OBSTETRIC CHALLENGE

L McQuillan ¹, M Andrews ², M Morosan ³, B Laverick ²

¹ Department of Obstetrics & Gynaecology, Norfolk and Norwich University Hospital Foundation Trust. ² Maternal Medicine Centre, Norfolk and Norwich University Hospital Foundation Trust. ³ Department of Anaesthetics, Norfolk and Norwich University Hospital Foundation Trust.



INTRODUCTION

Russell Silver Syndrome (RSS) is a rare genetic condition, characterised by asymmetrical intrauterine growth restriction, triangular face shape and feeding difficulties. Patients are usually diagnosed in utero or early childhood and require growth hormone therapy to reach their growth potential. This report considers a case of pregnancy in a patient with Russell Silver syndrome, where there was sub-optimal response to growth hormone.

CASE REPORT

A 20 year old female with a severe form of Russell Silver Syndrome presented in the first trimester of a spontaneously conceived pregnancy. She had a booking weight of 30kg and height of 128cm. A history of minimal response to growth hormone in childhood was noted and her skeletal structure was smaller than predicted. Other medical history included hypoglycaemia, poor dentition, cleft palate, malignant hyperthermia and significant musculoskeletal pain.

An echocardiogram was performed to rule out dilated cardiomyopathy, which is associated with RSS.

The third trimester was complicated by raised blood pressure compared to baseline, with proteinuria. Growth scans showed normal growth trajectory with an estimated fetal weight of 1889g at 35 weeks gestation.

Initially, she was keen to aim for a vaginal birth but opted for elective caesarean following counselling regarding the uncertainty of pelvic dimensions and the merits of avoiding emergency birth. A caesarean section was carried out without complication at 36 weeks gestation with a 55th centile baby.



Figure 1: Typical features of Russell Silver Syndrome

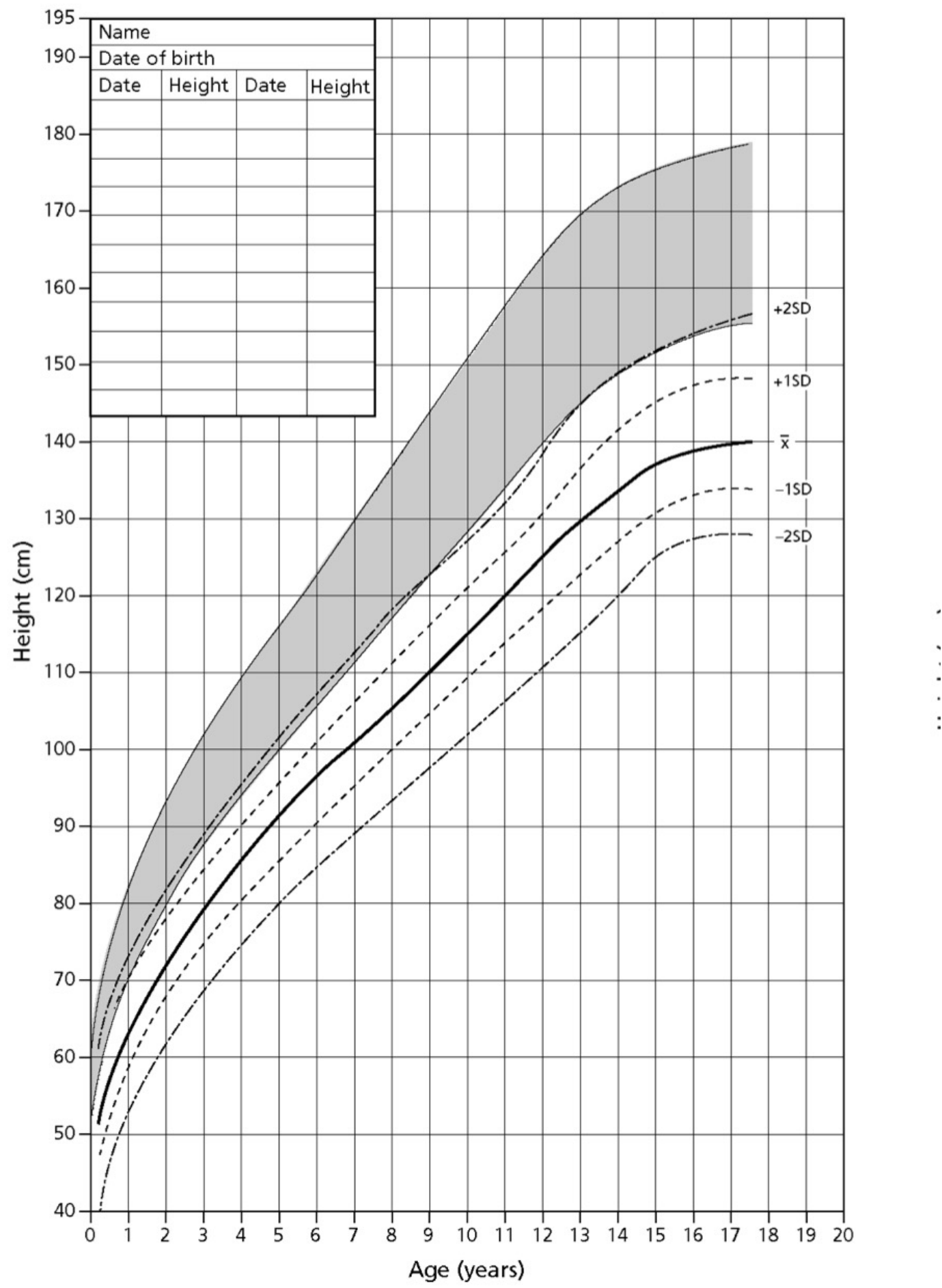


Figure 2: Height centiles for girls with Silver–Russell syndrome. The gray-shaded area indicates normal girls ± 2 standard deviations (SD). ³

DISCUSSION

Pregnancy in RSS presents a challenge to the MDT. This case highlights some of these challenges. This case required multiple collaborative maternal medicine MDT meetings to ensure safe antenatal care, anaesthetic and birth.

RSS is associated with metabolic disorder causing hypoglycaemia which becomes more pronounced with increased fetal demand. Hypoglycaemia can be unpredictable. Prolonged fasting prior to caesarean could be catastrophic and in this case dextrose infusion was given pre op.

Anaesthetic considerations included the use of paediatric equipment and spinal needles to accommodate narrow, shallow interspinal spaces. Avoidance of anaesthetic agents implicated in malignant hyperthermia and accurate prescription of weight adjusted doses of medication and fluid.

Surgical considerations included a comparison to paediatric surgery in terms of circulating blood volume (estimated 3000ml) and surgical instruments and retractors available. Cell salvage, uterotonics and tranexamic acid were used proactively. Blood loss was estimated to be 380ml.

Attention was given to the availability of height appropriate equipment on such as height adjustable beds and cots to enable safe recovery and care of the baby on the postnatal ward.

REFERENCES

¹ Nord: national organisation for rare disorders. 2020. Russell-Silver Syndrome. [Online]. [26 January 2025]. Available from: <https://rarediseases.org/rare-diseases/russell-silver-syndrome/>
² Yakoreva, M., Kahre, T., Žordania, R., Reinson, K., Teek, R., Tillmann, V., Peet, A., Öiglane-Shlik, E., Pajusalu, S., Murumets, Ü. and Vals, M.A., 2019. A retrospective analysis of the prevalence of imprinting disorders in Estonia from 1998 to 2016. *European Journal of Human Genetics*, 27(11), pp.1649-1658.
³ Brook, C.G.D., Clayton, P.E. and Brown, R.S. (2009) *Brook's clinical pediatric endocrinology*. Chichester, UK ; Hoboken, NJ: Wiley-Blackwell.