INTRODUCTION

There is little information on metabolic profiles and body composition in children with Silver-Russell syndrome (SRS).

AIM

To evaluate anthropometrics, glucose and lipid profiles; total body less head (TBLH) and lumbar spine (L1-L4) bone mineral density (BMD) and body composition in patients with SRS.

METHOD

31 SRS patients [16 subjects with 11p15 loss of methylation (11pLOM) and 15 subjects with maternal uniparental disomy of chromosome 7 (mUPD7); mean age 7.4±4.3 years], and non-SRS subjects [34 small for gestational age (SGA), 13.4±2.7 years, and 44 appropriate for gestational age (AGA), 6.9±1.4 years] were enrolled.

All patients underwent a cross-sectional evaluation for anthropometrics, biochemical glucose [fasting blood insulin and glucose (FBG), Homeostatic Model Assessment for Insulin Resistance standard deviation (HOMAIR-SD)] and lipid (triglycerides and cholesterol) profiles; at the same time TBLH and L1-L4 BMD Z-score, TB fat mass percentage (FM %) and limbs/TB fat ratio were measured by Dual-energy X-ray absorptiometry (DXA).

RESULTS

- Height SD in SRS (-2.1±1.2 SD) was significantly lower compared to AGA (0.02±1.3; p 0.001), but similar to SGA (-1.06±1.1 SD). Body Mass Index (BMI) was significantly lower in SRS (-1.6±1.2 SD) than in SGA (-0.7±1.2; p 0.04) and AGA (-0.1±1; p 0.003) groups and it was lower in SGA than in AGA (p 0.04) (Table 1).

- SRS children had higher fasting insulin (10.5±7 µU/mL) and HOMA-IR SD (0.72±1.8) than AGA (4.2±3.9 µU/mL; 0.7±1.3; p<0.001). In particular, children with 11p1LOM had a significantly higher FBG (92.5±7.5 mg/dL) than AGA (85±7 mg/dL; p 0.018). Similar results were found in SGA group, that showed higher insulin (14.5±9.9 µU/mL; p 0.0001), FBG (91±13 mg/dL; p 0.003) levels and HOMA-IR SD (1.3±1.7; p<0.0001) than AGA. Glucose profile was not dissimilar in SRS and SGA. SRS (77.9±40.6 mg/dL; p 0.002) and SGA (76.2±34 mg/dL; p 0.001) groups showed higher triglycerides levels than AGA (53.5±21.9 mg/dL) (Table 1).

- A significantly higher FM was found in SRS (27.5±6.8%) than AGA (22.9±9.5%; p 0.03); furthermore, a higher lower limbs/total body fat ratio was found in AGA (0.44±0.04) in 3S (0.4±0.05; p 0.0002) and in SGA (0.39±0.07; p 0.0003). TBLH and L1-L4 BMD Z-score were significantly lower in SRS (-1.3±0.6, p 0.0001 and -0.85±1.0, p 0.01, respectively) and SGA groups (-1.7±2.8, p 0.0001 and -1.01±0.9, p 0.009) compared to AGA group (0.1±1.1 and -0.26±1.2, respectively) (Table 1).

CONCLUSIONS

- BMI, glucose and lipid profiles as well as body composition assessments are mandatory in children with SRS.

- 11p15LOM seems to be at higher risk of glucose dysregulation.

REFERENCES


ACKNOWLEDGEMENTS

Italian Association of Silver Russell Syndrome (ASIRS Onlus)
Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genova - a Department of Excellence.

CONTACT INFORMATION

Prof. Mohamad Maghnie. e-mail: mohamadmaghnie@gaslini.org
Dr. Giuseppa Patti e-mail: giuseppapatti@gaslini.org