



Alterations in Metabolic profile and Body Composition in children with Silver Russell Syndrome

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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 (-1.96±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±1.1; 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 11p15LOM 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±9µU/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±21.3mg/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) than in SRS (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, 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).

	SRS	SGA	AGA	p-values		
				SRS vs SGA	SRS vs AGA	SGA vs AGA
Height SD	-2.1±1.2	-1.96±1.1	0.02±1.3	0.61	0.001	0.001
BMI	-1.6±1.2	-0.7±1.2	-0.1±1.1	0.04	0.003	0.04
Fasting glucose (mg/dl)	87.1±10.6	91±13	85.6±7	0.16	0.6	0.003
Fasting insulin (µU/ml)	10.5±7	14.5±9	4.2±3.9	0.07	<0.001	0.0001
HOMA-IR SD	0.72±1.8	1.3±1.7	-0.7±1.3	0.25	<0.001	<0.0001
Tryglicerides (mg/dl)	77.9±40.6	76.2±34.6	53.5±21.3	0.95	0.002	0.001
TB fat (%)	27.5±6.8	26.1±8.7	22.9±9.5	0.38	0.03	0.09
Lower limbs / TB fat ratio	0.4±0.05	0.39±0.07	0.44±0.04	0.71	0.0002	0.0003
TBLH z-score	-1.3±0.6	-1.7±2.8	0.1±1.1	0.99	0.0001	0.0001
L1-L4 z-score	-0.85±1	-1.01±0.9	-0.26±1.2	0.70	0.01	0.009

Table 1. Auxological data, glucose and lipid profiles and body composition in SRS versus non-SRS subjects

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.

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ACKNOWLEDGEMENTS

Italian Association of Silver Russell Syndrome (AISRS Onlus)

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