POLISH PATIENTS WITH SILVER-RUSSELL SYNDROME (SRS): CHARACTERISTIC OF GROWTH PARAMETERS ACCORDING TO (EPI)MUTATION

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Introduction

Children with SRS are characterized by intrauterine and postnatal growth retardation, feeding difficulties, relative macrocephaly, triangular face and body and/or face asymmetry. SRS is a rare heterogeneous congenital imprinting disorder associated with loss of methylation in *H19/IGF2*:IG-DMR at chromosome 11p15.5 (11p15 LOM) or maternal uniparental disomy of chromosome 7 (upd(7)mat).

Patients

95 SRS patients:79 with 11p15 LOM (83,2%) – 42 boys and 37 girls and 16 with upd(7)mat (16,8%) – 10 boys and 6 girls, at age from 0,1 to 18,9 year, without growth hormone therapy, were diagnosed and followed. One patient was measured 10 times on average. Body weight, length/height were measured and BMI was calculated. The right and left side of the body were measured to diagnose body asymmetry. Growth parameters were standardized and expressed as SDS scores.

Results

Table 1. The differences between boys and girls according to the (epi)mutation type – descriptive statistics.

feature	11p15 LOM boys						upd(7)mat boys						
	N[1]	mean	SD	median	min	max	N[1]	mean	SD	median	min	max	p[2]
age (y)	538	4,6	4,5	2,9	0,0	18,7	95	3,9	3,8	2,8	0,2	18,0	ns
weight (kg)	521	12,5	9,6	9,5	2,2	71,7	94	11,4	9,5	9,0	2,9	59,7	ns
weight (SDS)	521	-3,9	1,8	-3,6	-9,8	0,9	94	-4,2	1,3	-4,2	-6,9	-1,1	0,005
height (cm)	450	96,2	28,2	92,1	46,0	166,5	85	89,3	22,7	85,5	51,0	159,0	0,04
height (SDS)	450	-3,2	1,4	-2,9	-8,1	0,7	85	-3,5	0,8	-3,5	-5,5	-1,8	0,0003
BMI (kg/m²)	433	13,5	2	13,5	9,5	25,9	84	13,5	1,9	13,0	10,7	23,6	ns
BMI (SDS)	433	-2,7	1,9	-2,4	-8,6	1,2	84	-2,4	1,2	-2,5	-5,6	0,6	ns
feature	**11p15 LOM girls**						**upd(7)mat girls**						
	N[1]	mean	SD	median	min	max	N[1]	mean	SD	median	min	max	p[2]
age (y)	503	4,3	4,2	3,0	0,1	18,9	80	4,8	3,9	3,9	0,1	14,8	ns
weight (kg)	492	12,6	11	9,2	2,0	64,5	80	10,9	7,6	9,1	2,0	35,1	ns
weight (SDS)	492	-3,2	1,8	-3,1	-9,9	1,2	80	-4,9	1,3	-4,9	-7,5	-2,4	<0,00001
height (cm)	394	92,8	26,3	90,0	45,0	159,6	70	90,8	22,9	88,5	47,5	140,2	ns
height (SDS)	394	-3,0	1	-3,0	-6,0	-0,4	70	-4,2	0,7	-4,2	-6,2	-2,8	<0,00001
BMI (kg/m²)	383	14,4	3,5	13,8	8,9	30,0	70	13,2	1,4	12,8	11,2	17,9	0,009
BMI (SDS)	383	-1,8	2	-1,7	-8,5	2,2	70	-2,3	1,0	-2,4	-4,3	-0,8	0,004

1 – number of measurements, 2 – Mann Whitney U test, ns - nonsignificant

Fig. 1. Body weight, height and BMI (SDS) – the differences according to (epi)mutation type in boys and girls group.

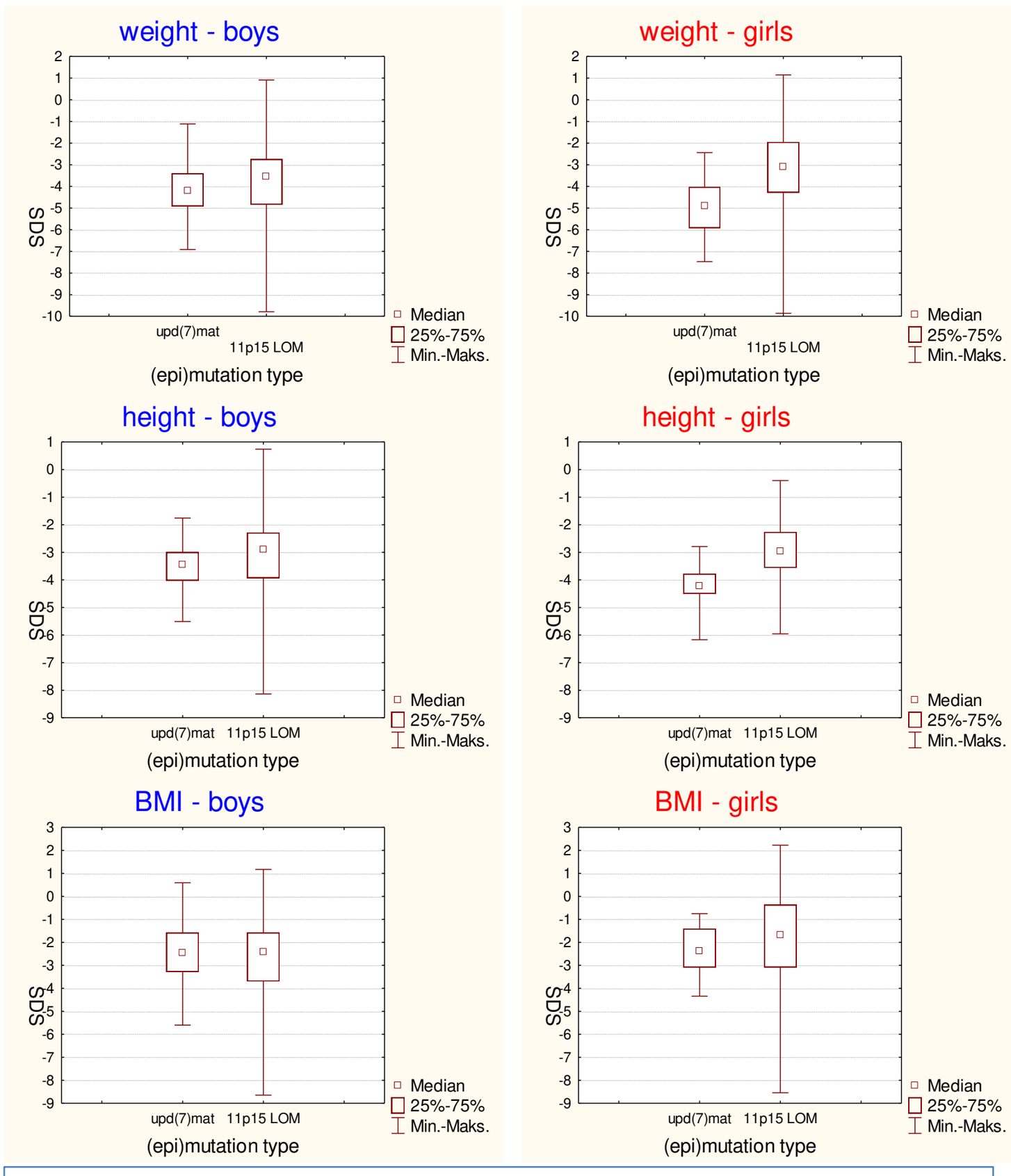


Fig. 2. A 4,5 year old boy with 11p15 LOM and body asymmetry.

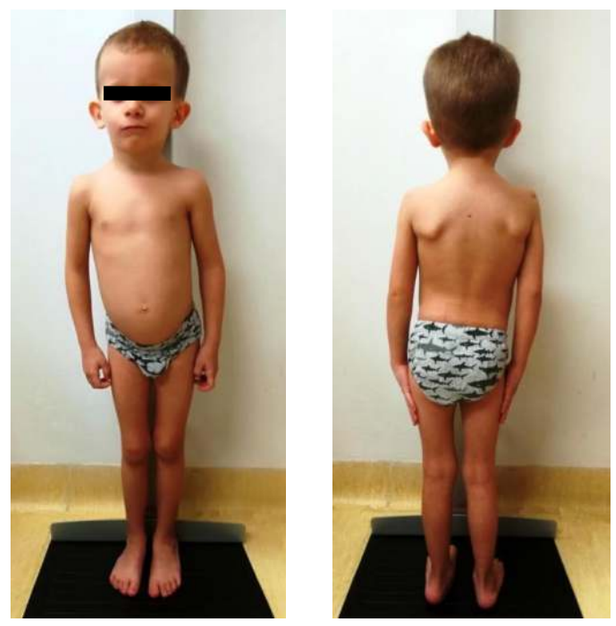


Fig. 3. The frequency of body asymmetry in SRS patients according to (epi)mutation type (percentages).

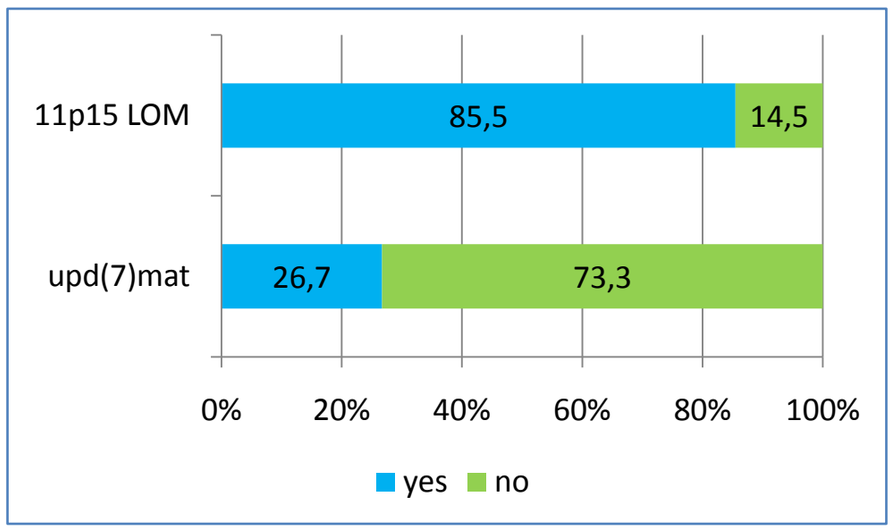
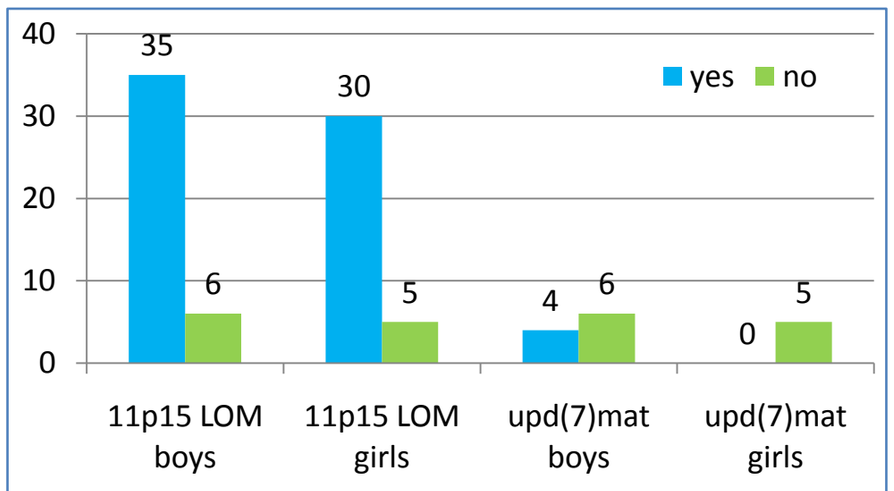


Fig. 4. The frequency of body asymmetry in SRS patients according to (epi)mutation type and sex (number of patients).



Conclusions

1. Growth parameters such as body length and weight were significantly lower in boys and girls with upd(7)mat comparing 11p15 LOM group.
2. Body asymmetry is more characteristic for children with 11p15 LOM .

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