Prevalence of endocrine complications in Duchenne muscular dystrophy

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Background: Duchenne muscular dystrophy (DMD) is caused by mutations in the dystrophin gene and results in a progressive muscular damage and degeneration. **Endocrine complications result from decreased energy expenditure, immobility and glucocorticoid (GC) treatment.** Due to the multidisciplinary management and emerging genetic and molecular therapies longer survival is expected and there is an increasing emphasis on the quality of life in DMD.

Aim: To determine prevalence of selected endocrine complications in a national cohort of boys with DMD in regard to treatment with GC.

Methods: 29 boys with DMD (age (mean+/-SD) 11.5+/-5.4 years) were studied at annual multidisciplinary visit. Levels of IGF-1, IGF-BP3, TSH, free T4, glucose, insulin and vitamin D3 were determined in the morning following an overnight fast. Bone mineral density and body composition were determined by DEXA. Data were expressed as mean+/-SD, groups were compared by T-test, correlations were made by Pearson’s coefficient. P-values < 0.05 were considered statistically significant.

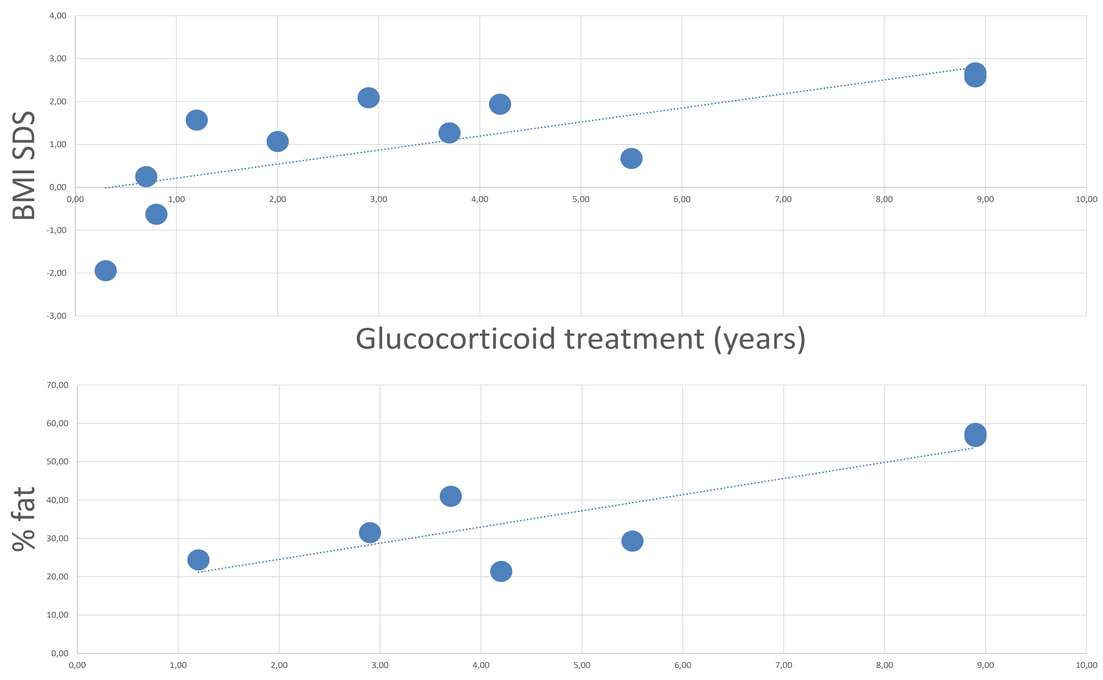
Results:

- 8/29 subject were **short stature.**
- 7/29 subjects were **obese**, 3/29 were **underweight.**
- **HOMA-IR** was increased in 8/29 and none presented with impaired glucose tolerance or diabetes.
- **No cases of acute adrenal insufficiency were reported.**
- **Lumbar spine-BMD was decreased** in 8/29, vitamin D3 levels were decreased (< 50 nmol/L) in 10/29 mean of the cohort, 24/29 were receiving vitamin D3 supplements and in one boy antiresorptive therapy.

Table 1. GC treated and non-treated subjects didn’t differ regarding frequency of evaluated endocrine complication.

	No-GC	GC
N	16/29	13/29
Age (years)	12.3	10.5
GC therapy duration (years)	NA	3.6
BH-SDS	-1.0	-.5
BW-SDS	-.19	.54
BMI-SDS	.52	1.05
HOMA-IR	3.2	3.2
Lumbar BMD (Z-score)	-.8	-1.3
WB Fat (%)	40	39
WB Lean (%)	56	60
D3	59	49

Figure 1. GC treatment duration correlated positively with BMI-SDS and % fat mass (r=0,722, p=.012 and r=0.84, p=.018, respectively.



Conclusions:

1. **Short stature, under and overweight and prediabetes** are common however not obligatory endocrine complications of DMD.
2. **GC treatment duration is an important risk factor for obesity and metabolic complications in DMD.**

References

1. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. Lancet Neurol. 2018 Mar;17(3):251-267.
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