



Thyroid Function, Lipid Profile and Carbohydrate Metabolism Parameters in Patients with Alstrom Syndrome

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Introduction

Alstrom syndrome is an autosomal recessive genetic disorder with mutation in the ALMS 2p12-13 gene and its characteristic features are: pigmented retinopathy, deafness, growth deficiency, obesity, metabolic syndrome, diabetes, thyroid dysfunction, nephropathy and cardiomyopathy.

Aim of the study

Evaluation of anthropometric parameters, thyroid function, carbohydrate metabolism and lipid profile in five patients with diagnosed Alstrom syndrome, hospitalized in the Department of Pediatrics, Diabetology and Endocrinology in Gdańsk, Poland.

Methods

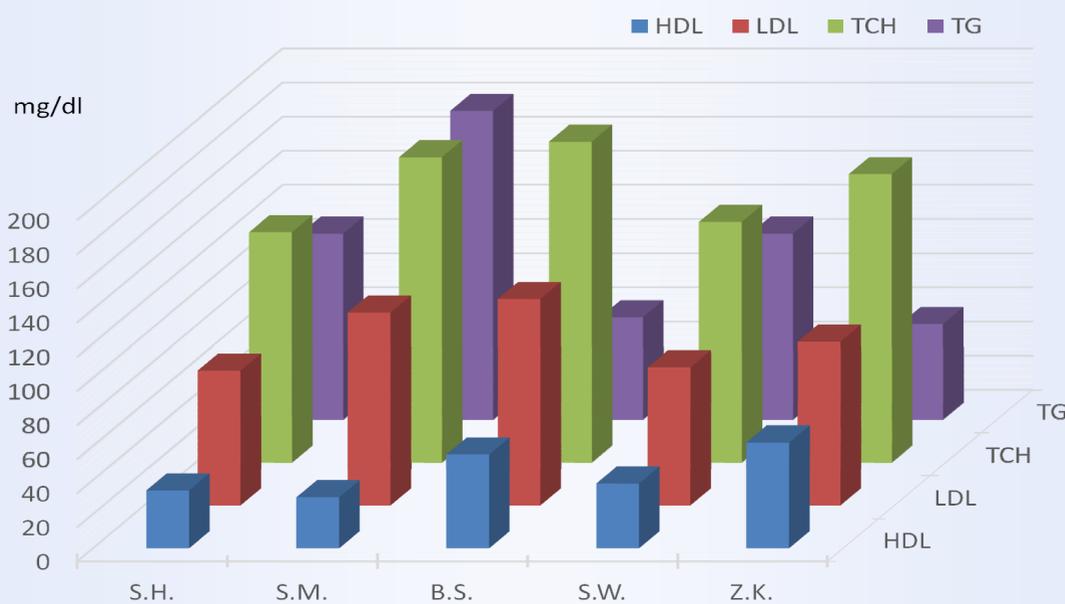
The height, body weight, waist circumference, body mass index (BMI), blood pressure, puberty stage in Tanner scale and laboratory tests such as TSH, fT4, thyroid antibodies, lipid profile, diabetes antibodies, oral glucose tolerance test (OGTT) and thyroid and abdominal ultrasound were performed.

Patients' characteristics

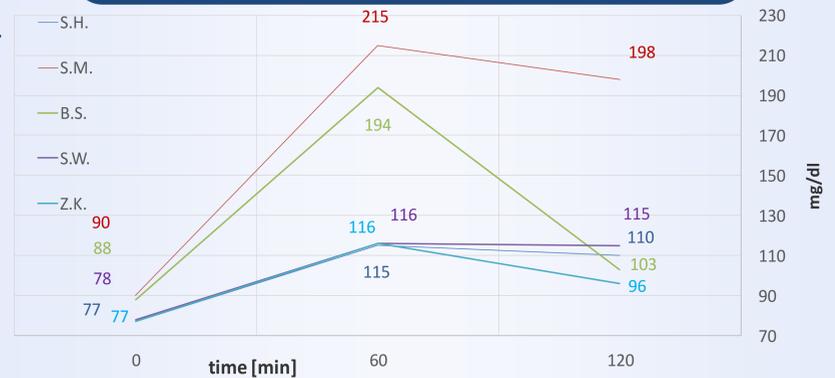
	S.H.	S.M.	B.S.	S.W.	Z.K.
sex	male	male	male	female	male
Age at the test time	17 1/12	14 3/12	14 3/12	7 2/12	4 9/12
Height	158,5cm (<3pc)	152,5cm (<3pc)	159cm (10-25pc)	135cm (90-95pc)	118cm (95pc)
BMI	23 (75-90pc)	24 (90pc)	24,5 (90-95pc)	28,5 (>95pc)	18 (90-95pc)
WHtR	0,48	0,49	0,51	0,6	0,5
Tanner stage	Tanner 3 (G3, P5, testicles 6ml)	Tanner 3 (G3, P4, testicles 8ml)	Tanner 3 (G3, P4, testicles 6ml)	Tanner 2 (Th2, P3)	Tanner 1 (cryptorchidism, hipoplastic genitals)

Blood pressure adjusted to age, sex and height in normal range in all patients

Lipid profile



OGTT - glucose



OGTT - insulin



Thyroid function

- Hypothyroidism was diagnosed in 2 patients, treated with L-thyroxine.
- thyroid antibodies were negative in all patients.
- Thyroid USG revealed normal echogenicity and echostructure; in 2 patients with hypothyroidism cysts were observe.

HbA1c and antibodies

- HbA1c in normal range in all patients
- Positive anti-GAD antibodies in 1 patient with IGT (198mg% after 2 hours of OGTT); later after 6 months diabetes type 1 was diagnosed and insulin was introduced

NAFLD

- Abdominal USG revealed liver steatosis and hepatomegaly in 1 patient (B.S.)
- ALT was in normal range in all patients

Conclusions

- In patients with Alstrom syndrome obesity is very common and it may lead to complications such as IGT, diabetes, lipid disorders and fatty liver disease.
- Patients with Alstrom syndrome could also acquire hypothyroidism and it is important to monitor their thyroid function as well.
- Early screening for metabolic disorders may advance the treatment and improve quality of life in those patients.

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