A Rare Reason of Type 2 Diabetes: Alström Syndrome

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Objectives: Alström Syndrome (ALMS), occurred due to mutations in ALMS1 gene, is a rare X-linked recessive disorder. 700 cases have been reported so far. Main clinical findings are rod cone dystrophy, neurosensorial deafness, obesity and type 2 diabetes. Hypogonadism, hypothryroidism, growth hormone deficiency, hypertriglyceridemia, cognitive dysfunction, cardiomyopathy, renal, hepatic and pulmonary disorders could also be seen. Herein, we report a patient with ALMS who experienced both type 2 diabetes and hepatic disorder.

Methods: A 15-year-old boy was referred to Pediatric Endocrinology outpatient department because of hyperglycemia. He was born at 40 weeks after a normal pregnancy as a son of consanguineous family and his birth weight was 3600 g. It was learned that he encountered visual problems when he was 1 and moderate deafness when he was 6 years old. The patient presented with truncal obesity, achantosis nigricans, moderate deafness, visual problems and nystagmus. His height was 163 cm (SDS: -1.03) and weight was 76.8 kg (SDS: 1.45). Fasting blood glycemia was 255 mg/dl, insülin 86.5 mikroIU/mL, ALT 113 U/L, AST 75 U/L and triglyceride level was 238 mg/dl. Abdominal ultrasound revealed grade 1 steatohepatitis. Rod cone dystrophy was detected.

Results: Due to Type 2 Diabetes accompanied with rod cone dystrophy, deafness and high transaminase levels, ALMS was considered. Homozygote c.10975C>T mutation was detected in ALMS1 gene. At first, life style changes were advised as the treatment of type 2 diabetes. However hyperglycemia continued. Because of high transaminase levels glargine insülin was initiated instead of metformin.

Conclusions: First choice in treatment of type 2 diabetes are life-style changes and metformin. Metformin is a hepatotoxic drug. We thought that if there is a suspicion of hepatic pathology metformin should not be considered as a first line therapy.