

Atypical Cystic Fibrosis Adolescent Case Study (with normal sweat test) Referring with Diabetes Mellitus Symptoms Found to Carry Homozygous R352Q Mutation

Mehmet Keskin¹, Özlem Keskin², Sevgi Bilgiç Eltan², M. Fatih Deveci³
Gaziantep University Medical School, Gaziantep ¹ Pediatric
Endocrinology, ²Pediatric Allergy and Immunology, ³Paediatrics

Introduction: Cystic fibrosis is an autosomal recessive genetic disorder affecting typically the lungs, the pancreas, the gastrointestinal tract and tissues that produce mucus secretion, such as sweat glands. Impaired glucose tolerance and cystic fibrosis-related diabetes are the most common complications of cystic fibrosis. Cystic fibrosis-related diabetes is an other type of diabetes mellitus and carries some of the characteristics of type 1 and type 2 diabetes. We hereby present an atypical cystic fibrosis case who is referred to our center as diabetes mellitus.

Case Presentation: 15 year old, male referred to our center with symptoms of drinking too much water, peeing more often and losing weight. He had reported oily stool since 1 year of age. This has been analysed in other centers, but found to have no specific reason. His parents were first degree relatives/cousins. In his physical examination, his weight was: 45.5 kg (10-25th percentile), height was: 165 cm (75th p), and puberty was inline with Tanner stage 5. In his laboratory test; serum glucose: 497 mg/dl, HbA1c: 12.7%, C-peptide: 1.04 ng/ml (0.9-7.1), pH:7.39, HCO₃:22.7 mmol/l, vitamin A:367 ng/ml (400-1500), vitamin D: 9.1 ng/ml (20<), vitamin E: 2.5 µg/ml (5-16), steotocrit in gaita: negative, sweat test found to be normal. Fecal elastase: 35 µg/dl (<100: exocrine pancreatic insufficiency). Insulin adacık antibodies (-), glutamic acid decarboxilase antibodies (-), MODY gene was negative. With cystic fibrosis prediagnosis, homozygous R352Q mutation has been found on exon 8, in CFTR Full Sequence Analysis. This mutation has already been described in former cystic fibrosis cases.

Results: Cystic fibrosis coupling with diabetes is rarely seen in childhood. Such a case study, having been presented with diabetes symptoms, and found to be diagnosed as atypical cystic fibrosis has not been presented in the literature, so far.

