She was diagnosed with primary adrenal insufficiency and was started on hydrocortisone: 5mg/Kg every 6 hours on the first day, which was then gradually reduced until attaining the maintenance dose (15 mg/m²/day divided in 3 doses). She was also started on fludrocortisone (0.1 mg/day). The antibodies against adrenal cortex were positive, confirming the diagnosis of Addison’s disease. The patient had no history of chronic mucocutaneous candidiasis and her thyroid function, parathyroid hormone and calcium levels were all normal; antimicrosomal antibodies were negative. Antibodies against intrinsic factor, anti-thyroid, anti-transglutaminase and anti-endomysium were all negative.

**Genetic analysis**

For the genetic analysis, DNA of the patient was obtained from peripheral leukocytes by chemagic DNA Blood100 Kit (Perkin Elmer Chemagen). All the AIRE coding region as well as intron exon boundaries were subjected to PCR amplification and direct sequencing using the BigDye terminator kit and were run in the 3730 xl DNA Analyzer (Applied Biosystems, Foster City, CA U.S.A.).

This analysis showed two heterozygous variants: NM_000383.2:C.1411C>T (p. Arg471Cys) in exon 12 (Figure 2) and IVS9+6G>A in intron 9 of the AIRE gene. No other mutation was found.

**DISCUSSION**

In the present case report, we describe a young patient with a diagnosis of type 1 diabetes who was...