GENETIC REPORT

Patient data

Name: John

Apellidos: Doe

Gender: Male

Fecha Nacimiento: 12/04/2005

Fecha Entrada: 05/09/2020

RESULTS

Glycogen Storage Diseases Types Ia, III, VI, and HI via G6PC, PYGL, and AGL Gene Sequencing: POSITIVE.

Heterozygous in G6PC for c. 229 T>C and c.820 G>A

This patient is heterozygous in exon 1 of the G6PC gene for a missense mutation defined as c.229 T>C, which is predicted to result in amino acid substitution (p.Trp77Arg).

INTERPRETATIONS

This mutation has been documented to be CAUSATIVE for Glycogen Storage Disease Type Ia (von Gierke disease) (Chevalier-Porst et al. J. Med. Genet. 33:358-360,1996).

This patient is also heterozygous in exon 5 of the G6PC gene for a missense mutation defined as c. 820 G>A, which is predicted to result in the amino acid substitution p.Ala274Thr. To our knowledge, this particular nucleotide substitution has not been previously reported in the literature or in databases.

The p.Ala274 residue is highly conserved across G6PC proteins examined in 10 different mammalian and non-mammalian species, including human, mouse, rat, fly, chimp, nematode and yeast proteins; a glycine can be found at this position in zebrafish. The program PolyPhen-2 (Adzhubei et al. Nat. Methods 7:248-249, 2010) predicts the p.Ala274Thr substitution to be "benign"and the SIFT software program (Ng and Henikoff Genome Res 11:863-874, 2001) predicts this amino acid change to be "tolerated". Despite these findings, the use of computer programs is not equivalent to functional proof, and these results should thus be interpreted in context of clinical findings, family history and other laboratory data.