

Supplementary Figure Legends.

Supplementary Figure 1. The 75-g oral glucose tolerance test (OGTT).

Plasma glucose (mg/dL), insulin ($\mu\text{L}/\text{mL}$), and insulin ($\mu\text{L}/\text{mL}$) to glucose (mg/dL) ratio of the proband, his father, his dizygotic twin sister, and his mother during the 75-g OGTT.

Red line: proband (mutation-positive); blue line: father (mutation-positive); black line: dizygotic sister (mutation-negative); gray line: mother (mutation-negative).

Supplementary Figure 2. The 75-g oral glucose tolerance test (OGTT) with glucose-dependent insulinotropic polypeptide (GIP) and other incretin hormones.

Plasma glucose (mg/dL), GIP (pmol/L), insulin ($\mu\text{L}/\text{mL}$), GLP-1 (pmol/L), and glucagon (pmol/L) of the mutation-positive individuals (proband and his father), and the mutation-negative control (proband's dizygotic twin sister) during the 75-g OGTT. For incretin measurements, the proband underwent another OGTT with a short duration (measured time: 0, 30, 60, 120 min); The proband's glucose values differed from those in Figure 1 and Supplementary figure 1. Incretin values were not available for his mother.

Red line: proband (mutation-positive); blue line: father (mutation-positive); black line: dizygotic sister (mutation-negative).

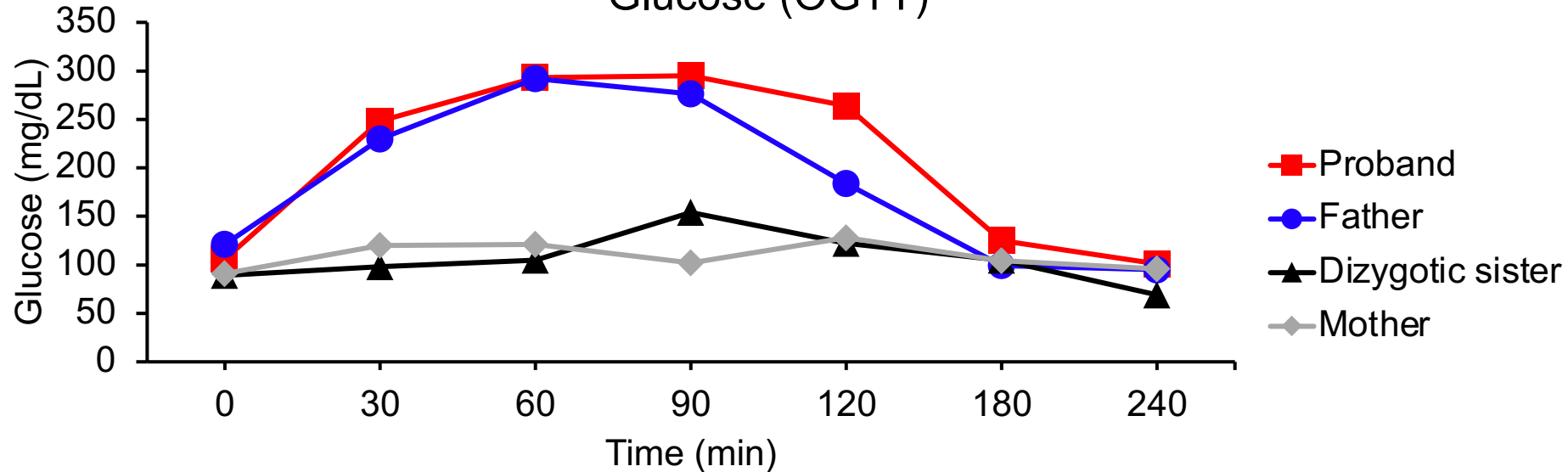
GIP = Glucose-dependent insulinotropic polypeptide, GLP-1 = Glucagon-like peptide-1.

Supplementary Figure 3. Sanger sequencing of *PDX1* in the proband, his father, his dizygotic twin sister, and his mother.

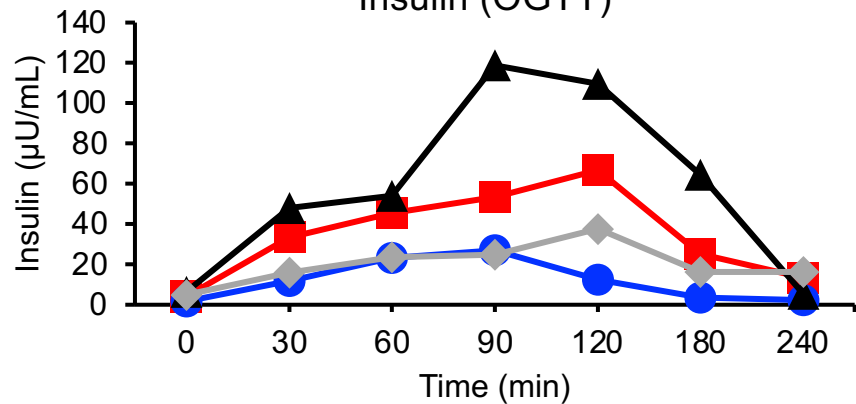
The mutation (c.218delT) is shown with a red arrowhead, and the corresponding wild-type allele is indicated by a black arrowhead.

Mutant = Mutated sequence, Wild = Wild-type sequence.

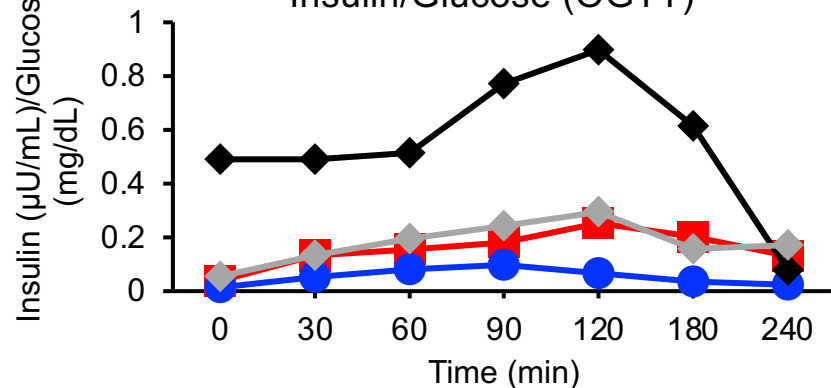
Glucose (OGTT)

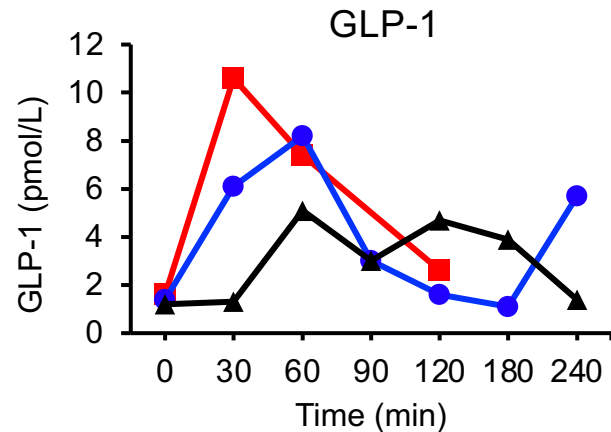
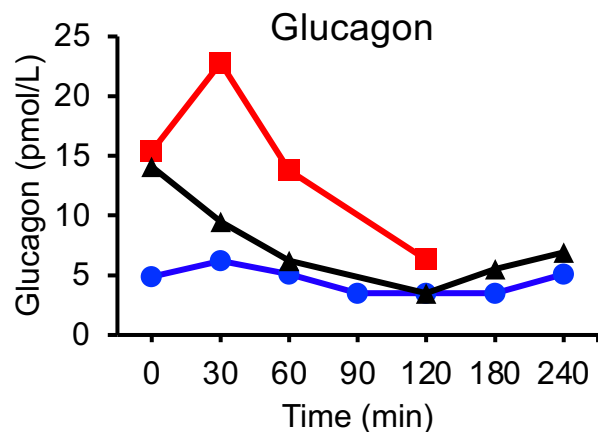
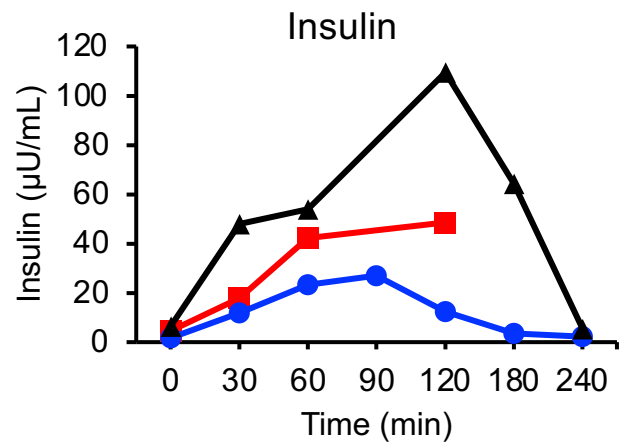
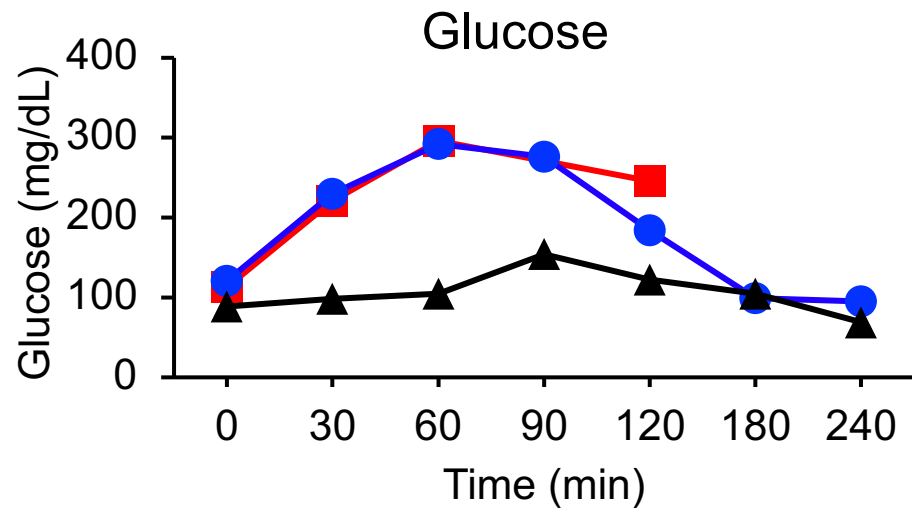
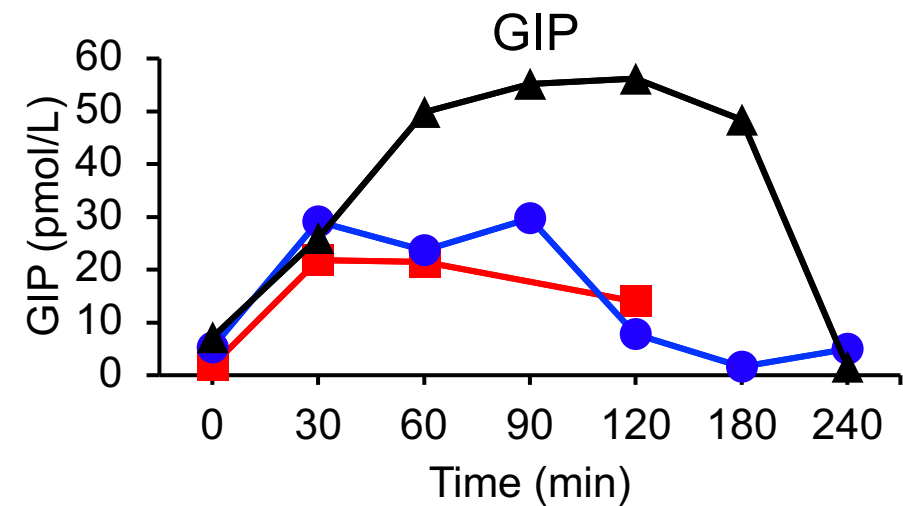


Insulin (OGTT)



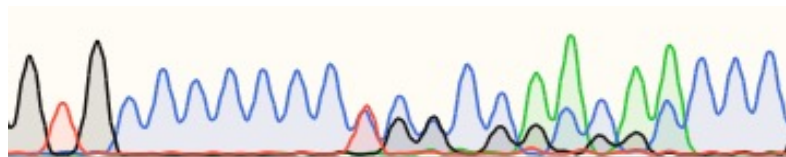
Insulin/Glucose (OGTT)





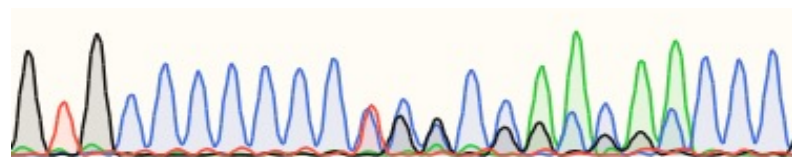
Proband

Mutant G T G C C C C C C C C C C G C C G A C G A C C C C
 Wild G T G C C C C C C C C C T C G C C G A C G A C C C C



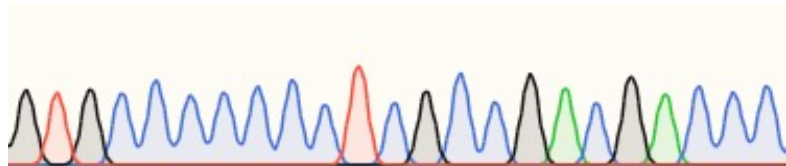
Father

Mutant G T G C C C C C C C C C C G C C G A C G A C C C C
 Wild G T G C C C C C C C C C T C G C C G A C G A C C C C



Dizygotic Sister

Wild G T G C C C C C C C C C T C G C C G A C G A C C C C
 Wild G T G C C C C C C C C C T C G C C G A C G A C C C C



Mother

Wild G T G C C C C C C C C C T C G C C G A C G A C C C C
 Wild G T G C C C C C C C C C T C G C C G A C G A C C C C

