**Abstract**

**Background:** Pancreatic agenesis is a rare condition in medical history. A recent case emerged that completely lacked a pancreas. To study the patient’s genotype, we used sequencing and PCR to identify mutations in the *IPF1*gene.

**Results:** PCR ampflication of the patient’s genomic DNA showeda deletion of a single cytosine in codon 63 of exon 1. We used four different sequencing primers to reveal the point deletion on both DNA strands suggesting homozygosity. We prepared expression plasmids containing the mutation to determine whether the point deletion results into termination after 59 additional nucleotides and transfected them into Cos-1 cells. A western blot analysis showed that the expression of these plasmids in Cos-1 cells expressed truncated proteins with 16kD weight which is significantly lower than the expression of wildtype IPF1 proteins (42-43kD) in Cos-1 cells.

**Conclusions:** We showed that one cytosine deletion in codon 63 caused a frameshift mutation for the 59 genes downstream leading to pancreatic agenesis in homozygous cases, emphasizing the critical role of the *IPF1* gene in normal pancreatic development.