The functional consequence of S148L HNF1B mutation as a cause of bilateral renal hypodysplasia with MODY5

**Case Study:** Maturity-onset diabetes of the young (MODY), which is known as monogenic diabetes, refers to autosomal dominant forms of diabetes mellitus involving multiple genes. According to the related genes with a principal role in the pancreatic β-cell development or insulin secretion, MODY is classified into 13 subtypes. The mutation of hepatocyte nuclear factor 1β (HNF1B) results in MODY5 associated with congenital anomalies of the kidney and urinary tract. We conducted the mutational analysis of the HNF1B gene in a child of ESRD with bilateral renal hypodysplasia and MODY and found a heterozygous S148L HNF1B mutation. And we evaluated the functional consequences of HNF1B mutation through in vitro study. It suggests that kidney and pancreas were differently affected in the S148L of HNF1B. The S148L mutant can cause problems with tissue-specificity of their gene expression to affect the binding affinity and specificity of DNA binding.