study aims to identify the pathogenic sites in a core pedigree of Usher syndrome (USH). A core pedigree of USH was analyzed by whole exome sequencing (WES). Mutations were verified by polymerase chain reaction (PCR) amplification and Sanger sequencing. Two pathogenic variations (c.849+2T>C and c.5994G>A) in MYO7A were successfully identified and individually separated from parents. One variant (c.849+2T>C) was nonsense mutation, causing the protein terminated in advance, and the other one (c.5994G>A) located near the boundary of exon could cause aberrant splicing. This study provides a meaningful exploration for identification of clinical core genetic pedigrees [1)].

1)
Jia Y, Li X, Yang D, Xu Y, Guo Y, Li X. Identification of two novel pathogenic compound heterozygous MYO7A mutations in Usher syndrome by whole exome sequencing. Int J Pediatr Otorhinolaryngol. 2018 Jan;104:186-190. doi: 10.1016/j.ijporl.2017.11.020. Epub 2017 Nov 22. PubMed PMID: 29287864.