

Supplementary Table 3. Pathogenicity prediction of rare variants of HNF1 β identified in this study

Position	Protein change	SIFT	Mutationtaster	CADD	Polyphen-2	ACMG evidence	ACMG ^a
36104563	E105K	Tolerated	Disease causing	28.1	Damaging	PS3PM2PP3	Likely pathogenic
36047297	G454R	Damaging	Polymorphism	12.5	Damaging	PS3PM2PP3	Likely pathogenic

RefSeq:NM_001304286.2.

HNF1 β , hepatocyte nuclear factor 1-beta; SIFT, Sorting Intolerant From Tolerant (<http://cadd.gs.washington.edu>); CADD, Combined Annotation Dependent Depletion (<http://cadd.gs.washington.edu>); ACMG, American College of Medical Genetics.

^aThe classification of rare variants identified in this study according to the standards and guidelines recommended by the ACMG.