













- correlates with the severity of renal disease[J]. *J Am Soc Nephrol*, 2002, 13: 1230-1237.
- [10] PEI Y, OBAJI J, DUPUIS A, PATERSON A D, MAGISTRONI R, DICKS E, et al. Unified criteria for ultrasonographic diagnosis of ADPKD[J]. *J Am Soc Nephrol*, 2009, 20: 205-212.
- [11] LI W, MA Y, YU S, SUN N, WANG L, CHEN D, et al. The mutation-free embryo for *in vitro* fertilization selected by MALBAC-PGD resulted in a healthy live birth from a family carrying *PKD 1* mutation[J]. *J Assist Reprod Genet*, 2017, 34: 1653-1658.
- [12] ZHANG S, MEI C, ZHANG D, DAI B, TANG B, SUN T, et al. Mutation analysis of autosomal dominant polycystic kidney disease genes in Han Chinese[J/OL]. *Nephron Exp Nephrol*, 2005, 100: e63-e76. doi: 10.1159/000084572.
- [13] YU C, YANG Y, ZOU L, HU Z, LI J, LIU Y, et al. Identification of novel mutations in Chinese Hans with autosomal dominant polycystic kidney disease[J/OL]. *BMC Med Genet*, 2011, 12: 164. doi: 10.1186/1471-2350-12-164.
- [14] CHANG M Y, CHEN H M, JENQ C C, LEE S Y, CHEN Y M, TIAN Y C, et al. Novel *PKD1* and *PKD2* mutations in Taiwanese patients with autosomal dominant polycystic kidney disease[J]. *J Hum Genet*, 2013, 58: 720-727.
- [15] YANG T, MENG Y, WEI X, SHEN J, ZHANG M, QI C, et al. Identification of novel mutations of *PKD1* gene in Chinese patients with autosomal dominant polycystic kidney disease by targeted next-generation sequencing[J]. *Clin Chim Acta*, 2014, 433: 12-19.
- [16] JIN M, XIE Y, CHEN Z, LIAO Y, LI Z, HU P, et al. System analysis of gene mutations and clinical phenotype in Chinese patients with autosomal-dominant polycystic kidney disease[J/OL]. *Sci Rep*, 2016, 6: 35945. doi: 10.1038/srep35945.
- [17] PATEL V, WILLIAMS D, HAJARNIS S, HUNTER R, PONTOGLIO M, SOMLO S, et al. miR-17~92 miRNA cluster promotes kidney cyst growth in polycystic kidney disease[J]. *Proc Natl Acad Sci USA*, 2013, 110: 10765-10770.
- [18] ZHENG W, SHEN F, HU R, ROY B, YANG J, WANG Q, et al. Far upstream element-binding protein 1 binds the 3' untranslated region of *PKD2* and suppresses its translation[J]. *J Am Soc Nephrol*, 2016, 27: 2645-2657.
- [19] AUDRÉZET M P, CORNEC-LE GALL E, CHEN J M, REDON S, QUÉRÉ I, CREFF J, et al. Autosomal dominant polycystic kidney disease: comprehensive mutation analysis of *PKD1* and *PKD2* in 700 unrelated patients[J]. *Hum Mutat*, 2012, 33: 1239-1250.
- [20] NEUMANN H P, JILG C, BACHER J, NABULSI Z, MALINOC A, HUMMEL B, et al. Epidemiology of autosomal-dominant polycystic kidney disease: an in-depth clinical study for south-western Germany[J]. *Nephrol Dial Transplant*, 2013, 28: 1472-1487.
- [21] HOEFELE J, MAYER K, SCHOLZ M, KLEIN H G. Novel *PKD1* and *PKD2* mutations in autosomal dominant polycystic kidney disease (ADPKD)[J]. *Nephrol Dial Transplant*, 2011, 26: 2181-2188.
- [22] OBEIDOVA L, ELISAKOVA V, STEKROVA J, REITEROVA J, MERTA M, TESAR V, et al. Novel mutations of *PKD* genes in the Czech population with autosomal dominant polycystic kidney disease[J/OL]. *BMC Med Genet*, 2014, 15: 41. doi: 10.1186/1471-2350-15-41.
- [23] ROSSETTI S, STRMECKI L, GAMBLE V, BURTON S, SNEDDON V, PERAL B, et al. Mutation analysis of the entire *PKD1* gene: genetic and diagnostic implications[J]. *Am J Hum Genet*, 2001, 68: 46-63.
- [24] KOPTIDES M, MEAN R, DEMETRIOU K, CONSTANTINIDES R, PIERIDES A, HARRIS P C, et al. Screening of the *PKD1* duplicated region reveals multiple single nucleotide polymorphisms and a *de novo* mutation in Hellenic polycystic kidney disease families[J/OL]. *Hum Mutat*, 2000, 16: 176. doi: 10.1002/1098-1004(200008)16:2<176::AID-HUMU11>3.0.CO;2-H.
- [25] PHAKDEEKITCHAROEN B, WATNICK T J, AHN C, WHANG D Y, BURKHART B, GERMINO G G. Thirteen novel mutations of the replicated region of *PKD1* in an Asian population[J]. *Kidney Int*, 2000, 58: 1400-1412.
- [26] VAN DER KLIFT H M, JANSEN A M, VAN DER STEENSTRATEN N, BIK E C, TOPS C M, DEVILEE P, et al. Splicing analysis for exonic and intronic mismatch repair gene variants associated with Lynch syndrome confirms high concordance between minigene assays and patient RNA analyses[J]. *Mol Genet Genomic Med*, 2015, 3: 327-345.