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sy680 is a novel allele of pkd-2

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Description:

Summary: a new allele of <u>pkd-2</u> was isolated in a behavioral genetic screen for male mating defects, and found to result in a substitution of Arginine for Glycine in the equivalent of human PKD2 alanine 615.

Article:

The *C. elegans* ortholog of polycystin-2 is encoded by <u>*pkd-2*</u> (Barr et al., 2001). From an EMS screen of a <u>*plg-1*</u>; <u>*him-5*</u> strain for male mating defective mutants and a secondary behavioral screen for defects in discrete steps of male mating behavior, namely response to contact to hermaphrodites and vulval location (described in Schindelman et al., 2006), we identified a new allele of <u>*pkd-2*</u> based on mapping and complementation. <u>*sy680*</u> fails to complement <u>*pkd-2(sy606)*</u> for defects in response to contact with hermaphrodite and vulval location. Here we report the sequence of this allele. PCR amplification and sequencing of <u>*pkd-2*</u> exons indicated that there was a c-->t transition in the transcribed DNA strand (g-->a in the *pkd-2* sense strand; Figure 1A). This change leads to an altered codon, a Glycine to Arginine substitution the PKD-2 protein. This position corresponds to A615 of the human protein (Figure 1B).

Α.	
N2:	FGY
N2:	TGCATTCGCACAGTTTGGATATTTGTGCTT
sy680:	TGCATTCGCACAGTTT <mark>A</mark> GATATTTGTGCTT
sy680:	FRY

B. C. elegans PKD-2 <i>H.</i> sapiens PKD2	TKTGVNRVNSVIENGLTNAPFDDVTSSENSYLNIKACVVFVAWVKVFKFISVNKTMSQLS TSN-VEVLLQFLEDQNTFPNFEHLAYWQIQFNNIAAVTVFFVWIKLFKFINFNRTMSQLS * *: ::*: * *:.:: : .: ** * .***:*******	438 587
C. elegans PKD-2 H. sapiens PKD2	STLTRSAKDIGGFAVMFAVFFFAFAQFGYLCFGTQIADYSNLYNSAFALLRLILGDFNFS TTMSRCAKDLFGFAIMFFIIFLAYAQLAYLVFGTQVDDFSTFQECIFTQFRIILGDINFA :*::*.***: ***:** ::*:*:**:** ****: *:*: :. *: :: :: *:****:**:	498 647
C. elegans PKD-2 H. sapiens PKD2	ALESCNRFFGPAFFIAYVFFVSFILLNMFLAIINDSYVEVKAELARKKDGEGILDWFMNK EIEEANRVLGPIYFTTFVFFMFFILLNMFLAIINDTYSEVKSDLAQQKAEMELSDLIRKG :***.:** :* ::***: *****************	558 707

Figure 1. A. The position of lesion in the <u>pkd-2</u> DNA sequence. B. Location of lesion in an alignment of *C. elegans* and human proteins. Position of substitution shown in red.

References

Barr MM, DeModena J, Braun D, Nguyen CQ, Hall DH, Sternberg PW. The *Caenorhabditis elegans* autosomal dominant polycystic kidney disease gene homologs *lov-1* and *pkd-2* act in the same pathway. Curr Biol. 2001 Sep 4;11(17):1341-6. PubMed PMID: 11553327.



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Schindelman G, Whittaker AJ, Thum JY, Gharib S, Sternberg PW. Initiation of male sperm-transfer behavior in *Caenorhabditis elegans* requires input from the ventral nerve cord. BMC Biol. 2006 Aug 15;4:26. PubMed PMID: 16911797; PubMed Central PMCID: PMC1564418.

Reagents

Strains: PS7518 <u>plg-1</u>(e2001d) III; <u>pkd-2(sy680)</u> IV; <u>him-5(e1490)</u> V PS3400 <u>pkd-2(sy606)</u>

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