

A

cDNA deletion

...cgcagGAAGCTCGTAAACGGTTTGCACAAAGCTACCATTACATATGATCAGgtgcgttgaat

E A R K R F D K A T I T Y D Q

E A R K

gDNA deletion

aa~~fatgtcaatgtgatgggttaa~~attatgggtttcccagcttgttaccaaatacatat~~ttatgt~~

atgtgcatctatatgaaacagGCACGAGAAAAATATCTTTCATTAAGGAAGAGCACACGAT...

A R E K Y L S L R K S T R

R H E K N I F H *

B

...TTCTTCAAACAGgcaagctttttccttgaaaacaattttcagagcaagaccacaaataact

F F K Q A S F F L K T I F R A R P Q I T

ctggatccaatccgcttg---9th intron---GGATATGAACTACTACATCAAATGGAG..

L D P I R L G Y E L L H Q M E

Supplemental Figure S1

A, The nature of the mutation in *AGDI*. The dashed line and solid line indicate the deleted genomic (gDNA) and cDNA sequences at the junction of exon 6-intron 6. Note that the mRNA has been alternatively spliced because of the deletion resulting in a frame-shift mutation. **B**, Correction for the 9th exon-intron NCBI- annotated *AGDI* gene. The solid line indicates an extension of actual intron sequence in the wild type gene. The amino acid sequence that has to be deleted is indicated by the italicized letters.