Supplementary Materials

A Partial Phenotype of adFNDI related to the Signal Peptide c.55G>A Variant of the AVP gene

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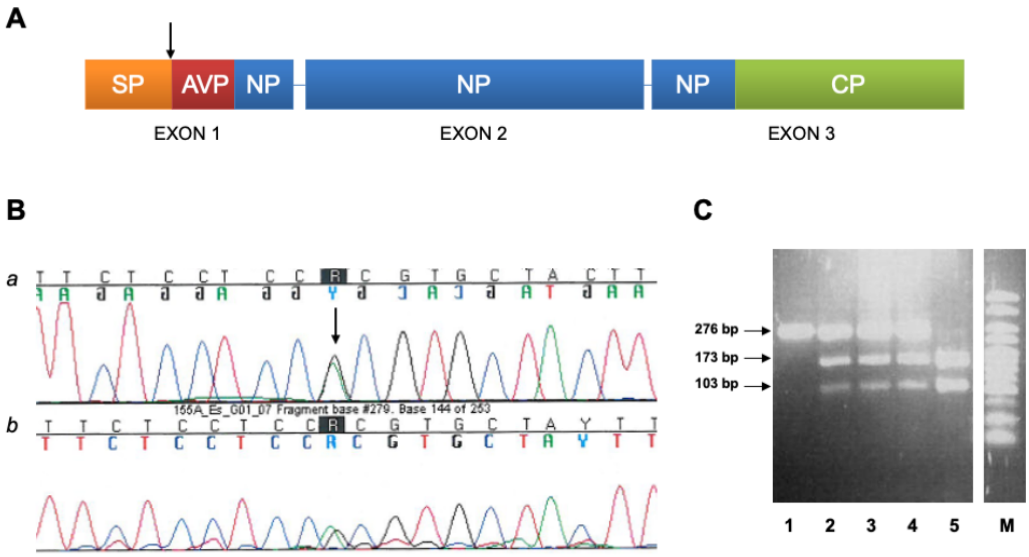

Supplementary Figure. A) Schematic diagram of the coding regions of the *AVP* gene (modified from [Siggard et al, 1999]); SP: signal peptide; AVP: arginine vasopressin; NP: neurophysin II; CP: copeptin **B)** Sequencing chromatograms of the PCR-amplified exon 1 of the *AVP* gene (a, sense; b, antisense) from the index patient (subject III-1); the arrow indicates the c.55G>A (g.279G>A) mutation predicting a p.Ala19Thr transition in the last amino-acid residue of SP; **C)** Agarose gel electrophoresis of the DNA fragments generated by BstUI restriction endonuclease cleavage of PCR products. In the affected subjects, the presence of both normal (173 and 103 bp) fragments and mutant PCR products which remain uncleaved (276 bp) indicates that the c.55G>A mutation is heterozygous. 1: uncleaved PCR products; 2: subject III-1; 3: subject II-2; 4 subject III-2; 5: unaffected control subject; M: DNA marker (Hyperladder V ™, Bioline, Meridian Bioscience).