Supplemental files submitted with *Knaup et al.*

Table of contents:

family #	no. of affected individuals (SNaPshot confirmed)	no. of affected generations	haplotype analyses
A-29 (1*)	>50 (27)	6	yes
A-30 (2*)	9 (7)	4	yes
A-49 (5*)	13 (4)	4	
A-33 (10*)	7 (2)	3	
A-05	2 (2)	3	
A-11	3(3)	3	yes
A-12	4(1)	3	
A-14	4(2)	3	
A-21	5 (2)	4	
A-23	4 (1)	3	
A-24	3 (1)	2	
A-36	2 (1)	2	
A-37	2 (1)	2	
A-38	>10 (3)	>2	
A-43	4 (2)	3	
A-47	17(9)	3	
A-50	7(4)	4	

Supplementary Table 1: List of ADTKD-*MUC1* families

17 families with at least two affected successive generations were evaluated for ADTKD-*MUC1* via SNaPshot minisequencing. Numbers provided are in many cases derived from patient history taking, where the affected status appeared very likely. Number of affected family members confirmed by SNaPshot minisequencing are given in parenthesis. In selected families haplotype reconstruction was performed by microsatellite analysis. *indicates the nomenclature of renamed families as cited in (Ekici *et al*.[10]).

Supplementary Table 2: List of antibodies (Ab) applied in our study

	company (clone)	host	dilution IB	dilution IHC/IF
Primary Ab:				
11ß HSD	Millipore (AB1296), Darmstadt, Germany	sheep		1:200
alpha Tubulin	GeneTex (GT114), Irvine, USA	mouse	1:10000	
Megalin	Acris (DM3613P), Herford, Germany	mouse		1:5000
Aquaporin-2	Sigma (SAB5200110), Darmstadt, Germany	rabbit		1:1000
MUC1 WT (VNTR)	Cell Signaling (VU4H5), Danvers, USA	mouse	1:1000	1:100/1:100
MUC1 WT (C-term)	Abcam (ab80952), Cambridge, UK	Armenian hamster		1:1000/1:100
MUC1-fs (pAb1-4)	n.a.	rabbit	1:5000	1:5000/1:1000
THP/UMOD	MP Cappel (55140), Cambridge, UK	goat		1:1000/1:50
β-Actin	Sigma (clone AC-15, 5441), Darmstadt, Germany	mouse	1:10000	
Secondary Ab:		**target/host**		
swine Anti-rabbit HRP	Dako (P0447), Santa Clara, USA	rabbit/swine	1:2000	
goat Anti-mouse HRP	Dako (P0448), Santa Clara, USA	mouse/goat	1:2000	
rb Anti-sheep IgG biotinylated	VectorLabs (BA-6000), Burlingame, USA	sheep/rabbit		1:500
rb Anti-goat biotinylated	VectorLabs (BA-5000), Burlingame, USA	goat/rabbit		1:50
Armenian hamster biotinylated	Jackson ImmunoResearch (127-065-160), West Grove, USA	armenian hamster/goat		1:1000
Alexa Fluor 594, Molecular Probes	Thermo Fisher Scientific (A28175), Darmstadt, Germany	mouse/goat		1:500
Alexa Fluor 488, Molecular Probes	Thermo Fisher Scientific (A11055)), Darmstadt, Germany	goat/donkey		1:500
Alexa Fluor 488, Molecular Probes	Thermo Fisher Scientific (A11070)), Darmstadt, Germany	rabbit/goat		1:500
Alexa Fluor 647	Abcam (ab173004), Cambridge, UK	Armenian hamster/goat		1:200

Dilutions for immunoblotting (IB), Immunohistochemistry (IHC) and Immunofluorescence (IF) as indicated.

Supplementary Table 3: List of microsatellites used in haplotype reconstruction and their respective primer sequences

#	Microsatellite ID	Genetic Position (cM)	Physical Position (bp in hg19)	Size (bp)	Forward Primer (5'-3')	ReversePrimer (5'-3')
1	ATA42G12	139.02	107,075,628	178-196	AGCTAGGCACTTGTTGATGG	ATGTTGGTCCACATGTACCC
2	D1S534	151.88	119,678,203	196-216	AGCACATAGCAGGCACTAGC	CGATTGTGCCACTACACAGT
3	D1S305	159.32	154,281,903	156-176	CCAGNCTCGGTATGTTTTTACTA	CTGAAACCTCTGTCCAAGCC
4	D1S2714	161.05	155,115,519	172-182	ATGAATTGCTTGAGCCCA	AGCTATCCTCCCACCTCAGA
5	D1S1153	161.05	155,268,703	270-328	CAGACGAGACCCTAGAGAG	GGATTATAGGCAAGAGCCAC
6	D1S2777	161.05	155,460,360	224-274	GCACCACGGAACTCCAGTAT	CACCACTGTGCCCAGCTAAT
7	D1S303	161.05	155,637,498	181-191	CGACAAGAGCGAAACTCCAT	GCTTCCCAGAGGCTAGGATT
8	D1S1595	161.05	155,688,364	265-293	ATGGTATGAACCTGGAGGTG	GGCAGATAAAAGGACTGCAA
9	D1S2721	161.05	156,153,449	201-247	TTGCTCGGCCAGAGTCT	ACGCATCACACCTGGCTAGT
10	D1S1679	170.84	162,367,764	144-172	GCCATCAAGAAAACTAGTACTGC	ACCATGGTACTCAGCAGTGC
11	D1S1677	175.62	163,559,700	184-212	AGTCAGCTTGATTGACCCAG	CTTAGTGTGACAGGAAGGACG
12	D1S1589	192.05	174,261,084	199-217	TACTCAGGAGGCAGAGATGG	CTGCTTTGGGTTTCACTTGT

Microsatellites 1-4 are proximal and 5-12 are distal of *MUC1*.

Supplementary Table 4: List of microsatellites based genotypes

ID	ATA42G12	D1S534	D1S305	D1S2714	D1S1153	D1S2777	D1S303	D1S1595	D1S2721	D1S1679	D1S1677	D1S1589
A-11_II-2	184	208	172	180	324	266	183	290	232	148	208	200
	187	202	172	180	315	266	187	282	242	156	212	210
A-11_III-1	184	212	168	180	324	266	183	274	242	164	192	201
	187	202	172	180	0	266	187	282	242	155	212	210
A-11_III-2	0	0	172	180	297	268	187	282	236	164	200	200
	184	0	172	180	324	266	183	290	232	148	208	200
A-11_III-3	181	214	172	180	297	268	187	282	236	164	200	200
	184	208	172	180	324	266	183	290	232	148	208	200
A-11_III-4	184	212	168	180	324	266	183	274	0	164	0	200
	187	0	172	180	0	266	187	282	242	156	212	210
A-11_III-5	181	214	170	178	294	266	183	282	238	164	200	204
	184	208	172	180	297	268	187	282	242	168	200	212
A-29_IV-6	184	212	160	174	312	272	181	276	246	160	200	208
	187	204	162	180	297	268	181	282	234	148	212	200
A-29_IV-7	175	206	168	146	294	266	181	276	242	156	196	200
	187	204	162	142	318	268	189	286	240	168	204	216
A-29_IV-8	175	206	168	146	0	266	181	276	242	156	196	200
	184	200	172	142	0	268	189	286	234	168	200	200
A-29_IV-9	181	212	164	178	321	260	185	264	234	160	204	204
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_IV-11	181	212	164	178	321	260	185	264	234	160	204	204
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_IV-13	181	216	174	172	312	268	181	282	240	160	204	200
	184	200	162	180	318	268	185	282	234	172	200	210
A-29_IV-15	184	216	162	178	324	266	185	274	234	148	196	200
	187	200	172	174	318	266	197	276	242	152	200	200

A-29_IV-17	184	200	172	174	318	266	197	276	242	148	192	200
	187	206	172	182	315	266	185	286	234	168	196	210
A-29_IV-18	0	0	168	178	312	266	185	286	228	148	192	200
	187	206	172	182	315	266	185	286	234	168	196	210
A-29_IV-19	181	204	168	178	312	266	185	286	228	0	192	200
	187	206	172	182	315	266	185	286	234	168	196	210
A-29_V-1	184	206	158	142	288	268	185	272	234	172	204	196
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_V-3	184	206	158	142	288	268	185	272	234	172	204	196
	187	212	164	142	310	272	185	290	242	152	196	212
A-29_V-4	184	206	158	142	288	268	185	272	234	172	204	196
	187	212	164	142	310	272	185	290	242	152	196	212
A-29_V-6	187	212	174	180	288	268	185	272	246	152	196	200
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_V-8	187	212	174	180	288	268	185	272	246	152	196	200
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_V-9	187	200	172	174	318	266	197	276	242	152	200	200
	187	212	174	180	288	268	185	272	246	152	196	200
A-29_V-11	189	208	162	180	312	266	185	288	240	160	196	212
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_V-13	187	204	162	180	297	268	181	282	234	148	212	200
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_V-14	184	212	160	174	312	272	181	276	246	160	200	208
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_V-15	184	200	162	180	318	266	193	278	232	156	204	200
	181	212	164	178	321	260	185	264	234	160	204	204
A-29_V-16	181	212	164	178	321	260	185	264	234	160	204	204
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_V-17	187	200	172	174	318	266	197	276	242	152	200	200
	189	213	163	178	303	269	185	284	240	156	204	204
A-29_V-	187	200	172	174	318	266	197	276	242	152	200	200

18	184	200	162	180	318	268	185	282	234	172	200	210
A-29_V-19	184	200	162	180	318	268	185	282	234	172	200	210
	184	200	172	174	318	266	197	276	242	152	200	200
A-29_V-20	184	200	172	174	318	266	197	276	242	148	192	200
	187	206	166	178	314	266	193	284	232	148	196	210
A-29_V-21	181	200	172	184	318	266	193	276	242	148	192	200
	187	206	172	182	315	266	185	286	234	168	196	210
A-29_VI-1	184	212	169	180	321	262	189	284	234	176	204	204
	187	200	172	174	318	266	197	276	242	152	204	196
A-29_VI-2	184	212	162	142	324	266	185	286	234	164	204	204
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_VI-3	184	212	162	142	324	266	185	286	234	164	204	204
	187	200	172	174	318	266	197	276	242	152	200	200
A-29_VI-4	184	206	158	142	288	268	185	172	234	172	204	196
	186	210	172	180	312	272	191	288	242	156	200	204
A-29_VI-5	187	200	172	174	318	266	197	276	242	152	200	200
	187	203	173	180	293	267	185	296	234	160	192	200
A-29_VI-6	184	206	164	180	318	266	197	284	242	156	192	212
	187	212	174	180	288	268	185	272	246	152	196	200
A-29_VI-7	187	212	174	180	288	268	185	272	246	152	196	200
	184	206	164	180	318	266	197	284	242	156	192	212
A-29_VI-8	187	200	172	174	318	266	197	276	242	156	196	200
	187	213	175	178	324	266	193	288	244	160	204	200
A-30_III-1	181	202	174	178	318	266	192	274	244	155	200	210
	184	206	164	176	318	268	184	286	238	143	208	196
A-30_III-2	184	206	158	178	0	268	184	278	234	155	196	200
	187	200	168	178	330	266	184	290	242	167	200	200
A-30_III-4	184	200	172	178	309	266	184	290	236	155	196	200
	187	200	168	178	330	266	184	290	242	167	200	200
A-30_III-5	184	198	172	176	312	266	184	282	242	151	200	200
	181	198	164	180	297	268	180	290	238	155	200	204

A-30_III-6	184	200	172	178	0	266	184	290	236	147	196	200
	187	200	168	178	0	266	184	290	242	167	200	200
A-30_III-7	184	206	158	178	0	268	184	278	234	0	196	200
	187	200	168	178	0	266	184	290	242	167	200	200
A-30_IV-1	184	206	164	176	318	268	184	286	238	143	208	196
	187	200	168	178	0	266	184	290	242	167	200	200
A-30_IV-2	184	200	172	178	309	266	184	290	236	155	196	200
	187	200	168	178	330	266	184	290	242	167	200	200
A-30_IV-3	184	198	172	176	312	266	184	282	242	151	0	200
	187	200	168	178	0	266	184	290	242	167	200	200
A-30_IV-4	184	206	158	178	320	268	184	0	234	0	0	200
	181	208	162	180	326	262	180	0	233	167	204	204

All genotypes used in haplotype reconstruction for members of families A-11, A-29 and A-30. The first column show the merged family and individual IDs separated with an underscore as indicated in the pedigrees in figure 2. Columns 2-13 show the genotypes as allele fragment length (bp) of respective microsatellites.

SUPPLEMENTARY METHODS

Cell culture and reagents. HeLa cells were purchased from the German Collection of Microorganisms and Cell Cultures (DSMZ, Braunschweig, Germany). The cells were cultivated in DMEM, 1.0 g glucose/L, 10% fetal calf serum, 2 mmol/L L-glutamine, 100 U penicillin and 100 µg streptomycin/ml. Medium and penicillin/streptomycin were supplied by PAN-Biotech (Aidenbach, Germany), fetal calf serum (standard "Gold") by PAA Laboratories (Coelbe, Germany). If not stated otherwise, all reagents were purchased from Sigma-Aldrich (Taufkirchen, Germany). Human urinary primary tubular cells (hUPTs) were generated and cultivated as described by *Zhou et al*.[33]

Cloning of MUC1 cDNA´s. The cDNA coding for the human MUC1 gene including 22 tandem repeats (MUC1/22TR) was a kind gift from Prof. Dr. Olivera Finn (University of Pittsburgh) and is described elsewhere[34]. For the generation of the MUC1/22TR-fs plasmids, a PCR-Fragment generated by the following primers: 5´-TAA TAC GAC TCA CTA TAG GG -3´ and 5´- GG GGC TGA GGT TGA CAT CGT GGG CTG -3´ was amplified using the MUC1/22TR cDNA as a template. Primer 1 binds to the T7 promoter within the backbone of pcDNA3, the second primer contains one additional basepair and covers the restriction site *BbvCI* in order to generate a frameshift directly upstream of the VNTR. The amplified PCR fragment was digested with the restriction enzymes *HindIII* and *BbvCI* and cloned into the original MUC1/22TR cDNA. An internal Flag-Tag (DYKDDDDK) followed by the restriction site *SacII* was fused directly to the end of exon 2 in analog to the method of Burdick and colleagues[35] into the MUC1/22/TR-wt and -fs plasmid by PCR-mutagenesis using the following primers: 5´- TAC ATC AAT GGG CGT GGA TA -3´, 5´- A GGC CGG GGC TGG CTT GTT GTC -3´, 5´- CTT GTC GTC GTC ATC CTT GTA ATC AGC ATT CTT CTC AGT AGA -3´ and 5´- GAT GAC GAC GAC AAG CCG CGG GCT GTG AGT ATG ACC AGC AGC -3´ and the restriction sites *HindIII* and *BbvCI*. For cloning, PCR fragments were purified using NucleoSpin Extract II from Macherey-Nagel (Düren, Germany). All plasmids were sequenced by standard procedueres at GATC Biotech (Konstanz, Germany).

Generation of antibodies. Four different rabbits were immunized with a synthetic peptide against the following amino acid sequence: NH_2-CHLGPGHQAGPGLHRPPSPR-$CONH_2$, corresponding to the VNTR of the frameshift MUC1 protein (MUC1-fs). All animals were immunized at day 1, 60 and 90. For testing, serum was taken 60, 90 or 120 days after first injection. To obtain MUC1-fs antibodies, all sera of day 120 were affinity purified with a column coupled with the synthetic MUC1-fs VNTR peptide. All steps were carried out commercially by Pineda–antibody-service (Pineda, Berlin, Germany).

Human samples. Kidney biopsies were collected retrospectively. The study was approved by the local ethics committee (protocol no.4103 and 181_15 Bc).

Transfection and Immunoblotting. HeLa cells were transfected either with equal amounts of empty vector (pcDNA3), wildtype human MUC1 cDNA (MUC1/22TR, 22 repeats of VNTR) or frameshift human MUC1 cDNA (MUC1/22TR-FS) using jetPEI transfection reagent (Polyplus-transfection, Illkirch, France) according to the manufacturer's instruction. 24 hours after transfection, cells were washed twice with PBS and homogenized into extraction buffer (8 M urea, 10% glycerol, 1% SDS, 10 mM TrisHCl pH 6.8, protease inhibitor complete™ (Roche, Mannheim, Germany)). Protein concentration was measured according to the manufacturer´s manual using the DC Protein Assay (BioRad, California, USA). Equal amounts of protein were separated by SDS PAGE, transferred to PVDF membranes (Millipore, Bedford, MA, USA) and stained with antibodies against the wildtype MUC1 VNTR (MUC1 (VU4H5) mAb; no. 4538; Cell Signaling, Danvers, MA, USA; dilution 1:1000) or one of four antibodies (dilution 1:5000) generated to detect the MUC1-fs protein (pAb1-4-fs). IVTT extracts were generated following manufacturers description (TNT, T7 Quick Coupled Transcription/Translation System, Promega, Wisconsin, USA) using linearized plasmids for MUC1/22TR or MUC1/22TR-FS). For antibody testing, pure serum of immunized rabbits was used 1:20.000. As loading control, β-actin was stained using the monoclonal antibody ß-actin (dilution: 1:10.000, Sigma Aldrich, St. Louis, USA no. A5441). Signals were visualized by the ECL system from GE Healthcare (Munich, Germany). For detailed antibody information, see supplementary table 2.

siRNA knockdown in hUPTs (human urinary primary tubular cells). hUPTs were seeded 24 hours prior to transfection for siRNA experiments and knockdown was performed as reported previously by *Warnecke et al*.[36]. Following siRNAs were used: siMUC1_5 Cat. No.: SI00162988, siMUC1_7 Cat. No: SI02780673, siMUC1_10 Cat. No.: SI04949826 and siMUC1_11 Cat. No.: SI04949833 (all Qiagen, Hilden, Germany).

Immunohistochemistry. Paraffin sections (2 µm) were dewaxed in xylene and rehydrated in a series of ethanol washes. For antigen retrieval before staining of MUC1-wt VU4H5 and -fs protein, all slides were cooked in a microwave 20 min in 0.1 M citrate puffer. For antigen retrieval before staining MUC1-wt ab80952, THP and 11ßHSD, slides were cooked for 7 or 5 min in 1xTRS (Target Retrieval Solution, DAKO, Glostrup, Denmark) in a standard pressure cooker. In the case of MUC1-wt ab80952 and THP staining, an additional avidin and biotin blocking was performed (Biotin Blocking System, DAKO). Endogenous peroxidase activity was blocked by incubation for 10 min with Peroxidase Real (DAKO). Slides for the MUC1-wt VU4H5 and -fs protein where blocked with 2.5% Normal Horse Serum (Vector Laboratories) for 20 min, for 11ßHSD 10% Normal Rabbit Serum was used for 30 min and for Muc1-wt ab80952 as well as THP Protein Block (DAKO) was use. All blocking steps where performed at room temperature.
Sections were incubated with primary antibodies (see Supplementary Table 2) diluted in Antibody Diluent (DAKO) overnight at 4°C (MUC1-wt VU4H5, ab80952, MUC1-fs, THP) or at 37°C for 60 min (11ßHSD). In case of THP, 11ßHSD and MUC1-wt ab80952, sections were incubated with biotinylated secondary antibody for 30min at room temperature. Next, slides were incubated with streptavidin/biotinylated alkaline phosphatase (Vectastain Elite ABC Kit, Vector Laboratories, Burlingame, USA) for 30 min. For MUC1-wt VU4H5 and MUC1-fs protein staining, slides were incubated with ImmPRESS Reagent Kit Anti-mouse, -rabbit Ig for 30 min. Finally, AEC+ solution (DAKO) or DAB solution (ImmPACT DAB Peroxidase Substrate Kit, Vector Laboratories) was used as chromogen according to the manufacturer's instructions. For identification of proximal tubules and collecting ducts sections of paraffin-embedded renal biopsies were immunohistochemically stained using antibodies specific for Megalin (mouse monoclonal, Acris, DM3613P, 1:5000) and Aquaporin-2 (rabbit polyclonal, Sigma, SAB5200110, 1:1000). All biopsies were fixed

and stained according to our standard techniques (Ventana BenchMark ULTRA stainer and UltraView DAB IHC Detection Kit, Roche).
All incubations were performed in a humidified chamber. Between incubations, specimens were washed three times in Tris-buffered saline (50 mmol/L Tris-HCl and 136 mmol/L NaCl, pH 7.4). Samples were processed in parallel throughout. Finally, the sections were counterstained with hematoxylin solution according to Mayer (DAKO) and analyzed with a Leica DMRB microscope (Leica, Bensheim, Germany).

Immunofluorescence. Paraffin sections (2 µm) were dewaxed in xylene and rehydrated in a series of ethanol washes. Antigen retrieval was performed by cooking all slides for 10 min (MUC1-wt (VU4H5) and THP) or 20 min (MUC1-fs, MUC1-wt (ab80952) and THP) in 0.1 M citrate puffer using a microwave. Next all slides where blocked with sterile 1% BSA/ PBS. Primary antibodies where diluted in 1% BSA/ PBS and incubated overnight at 4°C (see Supplementary Table 2). Following the slides where incubated for 1.5 hours at room temperature in Alexa Fluor secondary antibodies diluted in 1% BSA/PBS. For nuclear DNA counterstaining, DAPI (Thermo Fisher Scientific, D1306), diluted 1:1000 in PBS, was added for 4 min.
Samples were always processed in parallel and incubated in a humidified chamber. Between incubations, specimens were washed three times in PBS. A Leica DMRB microscope (Leica, Bensheim, Germany) was used for analyzing the slides.

Antibody pre-adsorption assay. Prior to routine immunohistochemistry (see above) the pAb3-fs was incubated on a roller platform with the MUC1-fs peptide (NH_2-CHLGPGHQAGPGLHRPPSPR-$CONH_2$) used for generating the antibodies pAbx-fs, a hSPAG4 (human Sperm Associated Antigen 4) peptide (NH_2-RSAEPGPGEPEGRRARGPSC-$CONH_2$) also consisting of 20 amino acids (used for the generation of hSPAG4 antibody, described in *Knaup et al.*[22]) and “no peptide” (antibody diluent, DAKO) for 2h at room temperature. The peptides were applied at a final concentration of 20µg/ml. Subsequently the antibodies were applied at identical dilutions, as described above.

SUPPLEMENTARY FIGURE LEGENDS

Supplementary Figure 1: Partial co-localization of mucin 1 and THP in kidney tubules. Immunofluorescent staining of healthy control kidney shows partial co-localization of mucin 1 (red), stained with the C-term mucin 1 antibody and THP (green, Tamm Horsfall Protein), an established kidney tubule marker of the TAL (thick ascending limb). Arrows indicate kidney tubules with shared expression of both markers.

Supplementary Figure 2: **Localization of mucin 1 in kidney tubule system.** Immunohistochemical analyses of serial human kidney sections for mucin 1 and distinct markers of the kidney tubule system (megalin, THP (Tamm Horsfall Protein), 11ßHSD (11-ß-hydroxysteroid dehydrogenase) and aquaporin-2. Partial co-localization is visible for mucin 1 and THP (TAL, thick ascending limb) as well as for aquaporin-2 (CD, lower collecting duct), whereas 11ßHSD (CD, collecting duct) nearly completely co-localizes with mucin 1. No spatial overlap can be detected for mucin 1 and megalin (PT, proximal tubule).

Supplementary Figure 3: **Localization of MUC1-fs in kidney tubules.** Immunohistochemical analyses of serial human kidney sections for MUC1-fs and distinct kidney tubule markers (megalin, THP (Tamm Horsfall Protein), 11ßHSD (11-ß-hydroxysteroid dehydrogenase) and aquaporin-2. Partial co-localization is visible for MUC1-fs and THP (TAL, thick ascending limb) as well as for aquaporin-2 (CD, lower collecting duct), wheras 11ßHSD (CD, collecting duct) nearly completely co-localizes with MUC1-fs. No spatial overlap can be detected for MUC1-fs and megalin (PT, proximal tubule).

Supplementary Figure 4: Immunohistochemical validation of MUC1-fs antibodies. The four individual and purified antibodies against MUC1-fs (pAbx-fs) were used in immunohistochemistry on consecutive sections of one patient with ADTKD-*MUC1* (ADTKD-0048). All antibodies produced identical positive signals, with pAb3-fs showing the most intense staining.

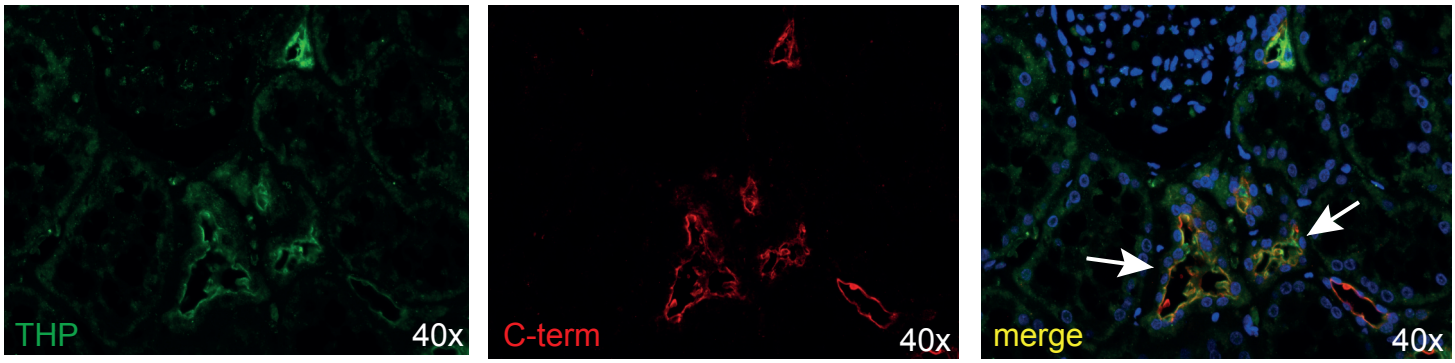
SUPPLEMENTARY REFERENCES

34. Cascio S, Zhang L, FinnOJ. MUC1 Protein Expression in Tumor Cells Regulates Transcription of Proinflammatory Cytokines by Forming a Complex with Nuclear Factor-κB p65 and Binding to Cytokine Promoters. J Biol Chem. 2011 Dec 9;286(49):42248-56.

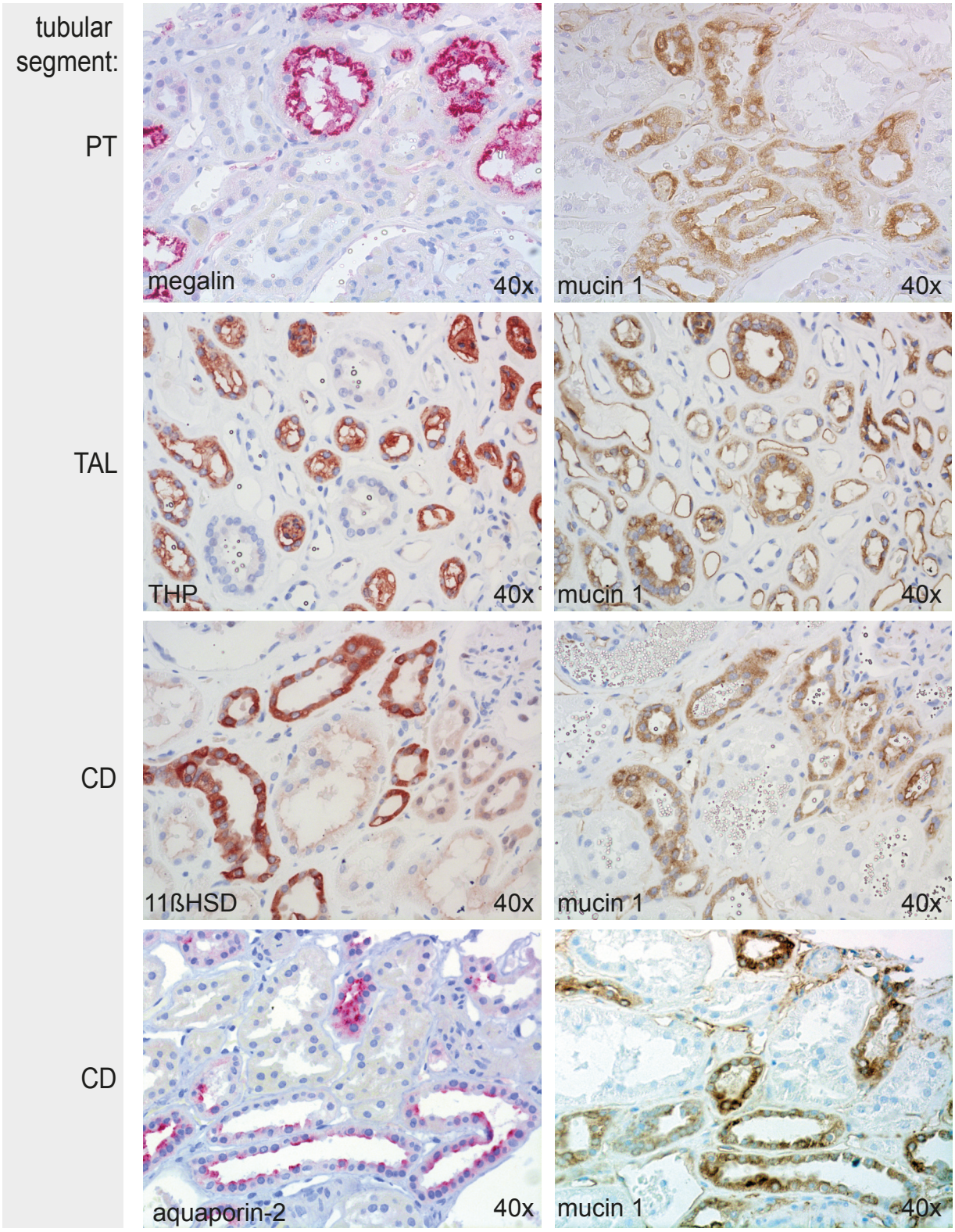
35. Burdick MD, Harris A, Reid CJ, Iwamura T, Hollingsworth MA. Oligosaccharides expressed on MUC1 produced by pancreatic and colon tumor cell lines. J Biol Chem. 1997 Sep 26;272(39):24198-202.

36. Warnecke C, Zaborowska Z, Kurreck J, Erdmann VA, Frei U, Wiesener M, et al. Differentiating the functional role of hypoxia-inducible factor (HIF)-1alpha and HIF-2alpha (EPAS-1) by the use of RNA interference: erythropoietin is a HIF-2alpha target gene in Hep3B and Kelly cells. FASEB J. 2004 Sep;18(12):1462-4.

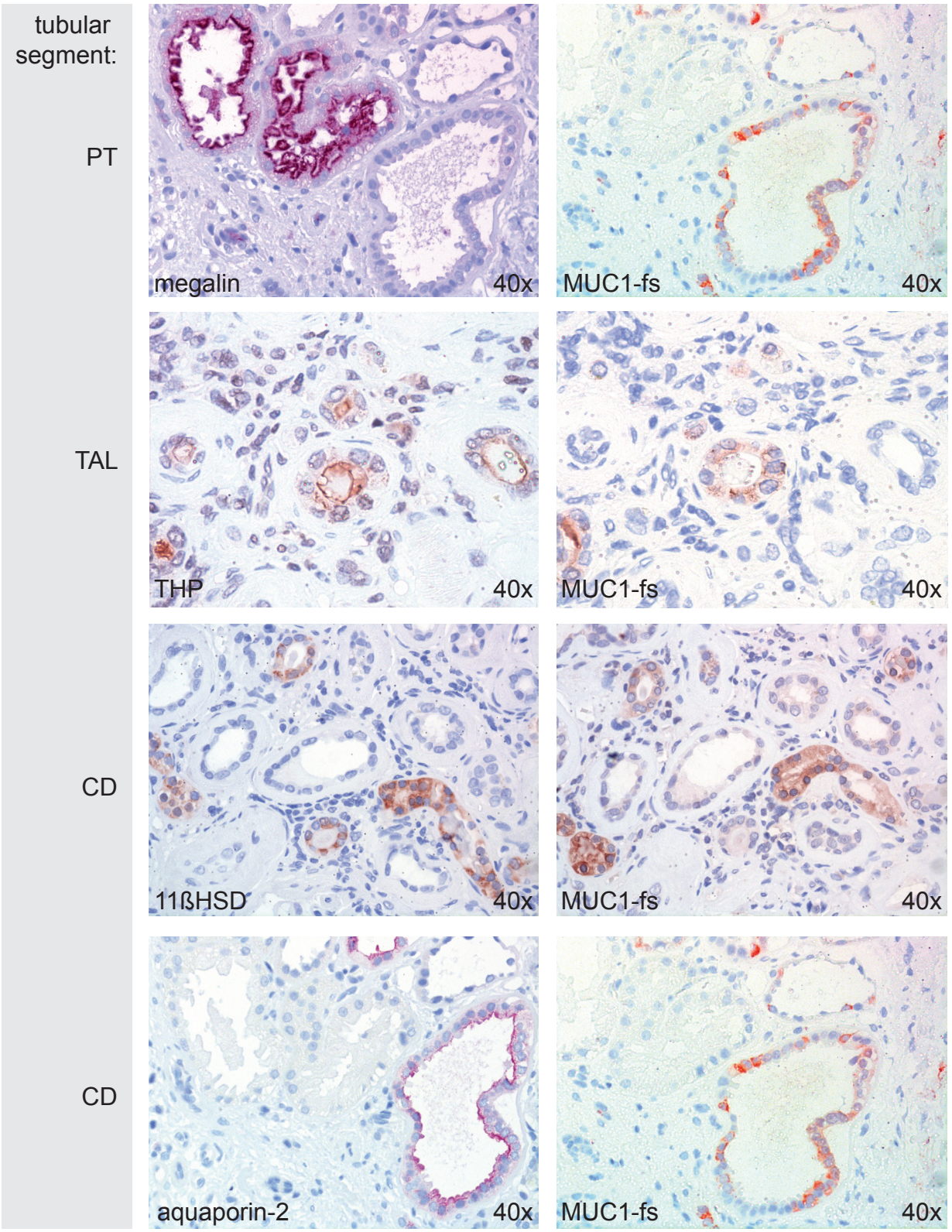
Supplementary Figure 1: Partial co-localization of mucin 1 and THP in kidney tubules



Supplementary Figure 2: Localization of mucin 1 in kidney tubule system



Supplementary Figure 3: Localization of MUC1-fs in kidney tubules



Supplementary Figure 4: Immunohistochemical validation of MUC1-fs antibodies