WEB TABLE I CLINICAL AND BIOCHEMICAL CHARACTERISTICS OF PATIENTS WITH DENT DISEASE

	Pt. 1	Pt. 2#	Pt. 3	Pt. 4	Pt. 5	Pt. 6*	Pt. 7*	Pt. 8	Pt. 9	Pt. 10	Pt. 11	Pt. 12	Pt. 13	Pt. 14	Pt. 15	Pt. 16	Pt. 17	Pt.18
Age at onset, yr	8	0.3	2	4	1	2	2	6.5	1	4	1	1	0.8	2	0.3	1.5	0.5	6
Age at diagnosis, yr	14	5.3	4	8	5	4	4.5	9.5	14	12	10	8	10.8	12	1.8	4	1.5	8
Height SDS	-2.8	-3.8	-3.8	-3.6	-2.6	-3.5	-6.7	-2.8	-1.9	-5.3	-8.4	-7.6	-8.4	-4.8	-3.0	-2.3	-3.2	-3.24
Onset to diagnosis, yr	6	5	2	4	4	2	2.5	3	13	8	9	7	10	10	1.5	2.5	1	2
Polyuria, polydipsia	+	+	+	+	+	+	+	+	-	-	+	+	+	+	+	+	+	+
Salt preference	+	+	+	+	+	+	+	-	-	-	-	-	-	+	-	-	+	-
Rickets	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Night blindness	+	+	+	+	-	+	+	-	+	+	+	+	-	+	-	-	-	+
Creatinine, mg/dL	0.8	0.6	0.4	0.6	0.74	0.5	0.4	1.06	0.8	0.6	1.3	0.5	0.3	0.9	0.8	0.4	0.4	0.5
eGFR, ml/min/1.73 m^2	76	64	78	62	61	100	87	49	75	66	28	69	126	55	49	98	82	76
Potassium, mEq/L	2.8	3.1	3.9	3.1	3.3	3.2	2.8	2.8	3.7	2.5	3.2	3.1	4.3	3.3	3.5	4.0	3.7	3.3
Calcium, mg/dL	8.7	9.9	9.0	10	10.8	8.6	9.7	9.3	9.4	9.3	8.8	7.8	8.8	8.3	8.9	9.4	8.9	9.7
Phosphate, mg/dL	3.0	2.9	2.0	2.9	2.6	2.8	2.6	4.3	2.4	2.2	2.3	2.7	2.5	2.9	2.8	2.6	2.5	2.5
TmP/GFR, mg/dL	1.1	2.5	1.9	1.9	2.2	1.5	1.7	3.6	2.6	1.3	1.2	1.1	1.5	2.5	NA	2.1	1.5	1.4
Alkaline phosphatase, IU/L	1509	1747	3516	1060	1764	2023	1525	926	3841	2278	3465	5317	1893	2257	1757	1041	537	1897
pH	7.42	7.37	7.43	7.40	7.39	7.38	7.43	7.54	7.42	7.40	7.26	7.36	7.37	7.34	7.31	7.39	7.32	7.33
Bicarbonate, mEq/L	25.6	22.7	20.6	24	21.3	22	21.6	20	22.2	21	15	20	22	21	19.8	19.3	15.4	20.8
25 hydroxyvitamin D, ng/mL	42.8	12	NA	NA	53.5	NA	7	37	35.9	23	42	35.8	NA	NA	24.3	123	19.6	NA
PTH, pg/mL	63.4	24.2	68	72	50.2	NA	29.3	16.6	23.3	15	422	116	NA	NA	373	53.2	170	NA
24-hr urine protein, mg	3400	560	1400	560	1100	2000	1200	1600	1860	1100	640	554	860	840	1230	520	1340	1870
24-hr calcium, mg/kg	3.8	16.6	10.2	20	8	8.2	13	10	9.4	5.3	7	6.8	8.8	5	3.9	13.5	4.6	4
â2-microglobulin, µg/L	57156	NA	53000	32340	88000	68000	68900	99199	72300	39948	78000	89900	64000	129967	19463	28700	38889	40000
Aminoaciduria	+	+	-	-	+	+	+	+	+	-	+	+	+	+	NA	+	+	NA
Follow up duration, yr	2.7	5.8	10	NA	1.8	19	2	1.2	5.4	0.3	0.4	0.4	0.5	1.3	8	4.8	20.6	6.25
Height SDS at follow up	-2.2	-4.2	-3.2	NA	-1.9	-6.2	-5.34	-4.3	-1.5	-5.3	-8.4	-7.6	-8.4	-4.6	-3.9	-1.9	-3.0	-4.64
eGFR follow up, m/min/1.73 m^2	58	96	80	NA	67	52	120	56	78	66	28	57	95	50	36	101	66	107
delta eGFR, ml/min/1.73 m^2/yr	6.7	-5.5	-0.2	NA	-3.3	2.5	-19	-5.8	0.56	-	-	-	-	3.84	1.62	-0.62	0.78	-5.0
24-hr calcium, mg/kg	4.6	17.6	10.6	NA	7.37	9.0	4.7	7.43	10	-	6.8	6.6	9.0	5.5	3.78	7.2	5.0	2

**Pt. 6 and 7 had nephrocalcinosis at initial diagnosis; #Pt. 2 developed nephrocalcinosis on follow up at 6-yr of age; ˆDelta eGFR computed for patients with follow up >1 yr; SDS standard deviation score; eGFR estimated glomerular filtration rate; TmP/GFR tubular maximum for phosphate/GFR; PTH parathormone; NA not available.*

WEB TABLE II RESULTS OF GENETIC TESTING FOR *CLCN5* GENE IN 15 OF 18 PATIENTS. *OCRL1* GENE WAS SEQUENCED IF NO MUTATION WAS FOUND IN *CLCN5*

	*Mutation**	*Location*	*Type of mutation*	*Amino acid change**	*Location of mutation^*	*SIFT[@] score*	*PolyPhen-2[##] (sensitivity; specificity)*
Pt. 1	c.731C>T	Exon 7	Missense	p.Ser244Leu[#]	Helix G	Damaging, 0	Probably damaging; score 0.999 (0.14; 0.99)
Pt. 2	c.1511T>A	Exon 9	Missense	p.Met504Lys[$]	Helix O	Damaging, 0	Probably damaging; score 0.992 (0.70; 0.97)
Pt. 3	c.2186T>G	Exon 12	Nonsense	p.Leu729X[$]	CBS-2 domain	-	-
Pt. 4	c.1511T>A	Exon 9	Missense	p.Met504Lys[$]	Helix O	Damaging, 0	Probably damaging; score 0.992 (0.70; 0.97)
Pt. 5	c.731C>T	Exon 7	Missense	p.Ser244Leu[#]	Helix G	Damaging, 0	Probably damaging; score 0.999 (0.14; 0.99)
Pt. 6	c.174G>C	Exon 3	Missense	p.Trp58Cys[$]	Helix B	Damaging, 0	Probably damaging; score 1.000 (0.00; 1.00)
Pt. 7	c.174G>C	Exon 3	Missense	p.Trp58Cys[$]	Helix B	Damaging, 0	Probably damaging; score 1.000 (0.00; 1.00)
Pt. 8	c.1579G>C	Exon 10	Missense	p.Glu527Gln	Helix P	Damaging, 0	Probably damaging; score 1.000 (0.00; 1.00)
Pt. 9	c.1942C>T	Exon 11	Nonsense	p.Arg648X[#]	Between CBS domains	-	-
Pt. 10	c.169G>C	Exon 3	Missense	p.Gly57Arg	Helix B	Tolerated, 0.08	Probably damaging; score 0.999 (0.14; 0.99)
Pt. 11	c.731C>T	Exon 7	Missense	p.Ser244Leu[#]	Helix G	Damaging, 0	Probably damaging; score 0.999 (0.14; 0.99)
Pt. 12	c.731C>T	Exon 7	Missense	p.Ser244Leu[#]	Helix G	Damaging, 0	Probably damaging; score 0.999 (0.14; 0.99)
Pt. 13	c.1909C>T	Exon 10	Nonsense	p.Arg637X[‡]	CBS-1 domain	-	-
Pt. 14, 15	No mutations identified in *CLCN5* or *OCRL1* genes						

*CBS cystathionine beta synthase. *Numbering according to cDNA sequence (GenBank NM_001282163.1) & protein sequence (GenBank NP_001269092.1) with A of first coding methionine no. 1; [#]Reported by Lloyd [12]; [$]Sethi [9]; [‡]Takemura [13]; ^Predicted topology adopted from Wu [14]; [@]Sorting intolerant from tolerant (SIFT) predicts substitutions with a score <0.05 as damaging and others as tolerated (http://sift.jcvi.org/); [##]PolyPhen-2 score represents the probability that a substitution is damaging; values near 1 are predicted to be deleterious (http://genetics.bwh.harvard.edu/pph2).*