Supplementary material

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Table S1. Clinical characteristics and genotypes of Polish patients with infantile (F1-F12.2) and juvenile (F13, F14) nephropathic cystinosis.

Patient ID	**Gender**	**Date of birth, yr**	**Age at first Symptoms, yrs.**	**First symptoms**	**Age of clinical Diagnosis, yrs**	**Cysteamine treatment**	**CKD FU**	**Extrarenal NC symptoms**	**Supplemental information**	***CTNS* genetype (if applicable)** Novel mutations marked in bold
F1	F	1981	1	failure to thrive, MR, rickets, P/P, FS, HC, ↓eGFR	1.5	irregular systemic since 18. yr., intermittent topical, incidental WBC ½ cystine assessment	progressive CKD (ESKD at 9 yrs.), PD, LDKTx at 13 yrs., 1.KT loss at 22 yrs., HD, DDKTx at 23 yrs., at 36 yrs. still functioning 2.KT (eGFR- 45 ml/min/1.73 m^2)	ocular, pHT, short stature (final growth 140 cm), DM, distal myopathy, dysphagia, CNS involvement	death at 36 yrs. by myocardial infarction	c.18_c.21delGACT (p.Thr7Phefs*7);?

F2	M	1983	1	failure to thrive, MR, P/P, recV, iFS, ↓eGFR	30	regular systemic since 30 yr., topical since 29. yr., incidental WBC ½ cystine assessment	progressive CKD (ESKD at 6 yrs.), preemptive LDKTx, 1.KT loss at 28 yrs., HD, DDKTx at 31 yrs., at 38 yrs. still functioning 2.KT (eGFR- 42 ml/min/1.73 m^2)	ocular (i.a. unilat. keratoplasty), pHT, short stature (final growth 160 cm), CNS involvement, distal myopathy	late diagnosis at the age of 30 yrs., initially treated as CTIN of unknown etiology	**c.141-1 G>A** (p.?); c.225+5_225+6delGTinsCC
F3	M	1984	0.8	failure to thrive, MR, rickets, FS, ↓eGFR	4	irregular systemic since 6. yr., intermittent topical, incidental WBC ½ cystine assessment	progressive CKD (ESKD at 11 yrs.), PD, HD, DDKTx at 15.5 yrs., at 38 yrs. still functioning 1.KT (eGFR- 83 ml/min/1.73 m^2)	ocular (i.a. bilat. keratoplasty), pHT, short stature (final growth 144 cm), DM	short GH therapy	suspected homozygous Ex3 del
F4	F	1986	1.5	failure to thrive, MR, P/P, iFS,	2.5	irregular systemic since 3. yr., intermittent	progressive CKD (ESKD at 8 yrs.), PD, LDKTx at 8.5 yrs.,	ocular (i.a. unilat. keratoplasty, unilat. blindness), pHT, short	at 29 yrs. successful pregnancy, delivery of a healthy child at 33. GW	c.681+1G>A (p.?); c.18_c.21delGACT (p.Thr7Phefs*7)

				↑rcECHO, ↓eGFR		topical, incidental WBC ½ cystine assessment	1.KT loss at 31 yrs., HD	stature (final growth 139 cm), DM, dysphagia, CNS involvement, distal myopathy		
F5	M	1989	0.9	failure to thrive, MR, rickets, P/P, recV, HC, FS, ↑rcECHO	8	shortly systemic between 8.5 and 10 yrs., WBC ½ cystine assessment not performed	progressive CKD (ESKD at 9 yrs.), HD, DDKTx at 10.5 yrs., at 32 yrs. still functioning 1.KT (eGFR- 92 ml/min/1.73 m^2)	ocular, pHT, short stature (final growth 155 cm)		c.681+1G>A (p.?); ?
F6	M	1990	0.8	failure to thrive, MR, rickets, P/P, recV, FS	2	irregular systemic since 4. yr., intermittent topical, incidental WBC ½ cystine assessment	progressive CKD (ESKD at 15 yrs.), PD, LDKTx at 16 yrs., at 31 yrs. still functioning 1.KT (eGFR- 89 ml/min/1.73 m^2)	ocular, pHT, short stature (final growth 155 cm)		no causative variants
F7	F	1991	0.8	failure to	2	irregular systemic	progressive CKD	ocular, subnormal	short GH therapy, at 27	not performed

				thrive, MR, rickets, iFS		since 12 yr., intermittent topical, incidental WBC ½ cystine assessment	(ESKD at 12 yrs.), HD, LDKTx at 12 yrs., 1.KT loss at 18 yrs., HD, LDKTx at 23 yrs., at 31 yrs. still functioning 2.KT (eGFR- 132 ml/min/1.73 m^2)	final growth (155 cm; 3-10 perc.), dysphagia	yrs. successful pregnancy, delivery of a healthy child at 32. GW	
F8	F	1992	1	failure to thrive, MR, rickets, P/P, recV, FS	1.5	irregular systemic since 3. yr., intermittent topical, incidental WBC ½ cystine assessment	progressive CKD (ESKD at 12 yrs.), PD, DDKTx at 13 yrs., 1.KT loss at 28 yrs., PD	ocular (candidate for corneal transplantation), pHT, short stature (final growth 146 cm), DM	short GH therapy	c.18_c.21delGACT (p.Thr7Phefs*7); c.18_c.21delGACT (p.Thr7Phefs*7)
F9	M	1993	1.5	failure to thrive, MR, rickets, FS, ↓eGFR	2	irregular systemic since 3. yr., intermittent topical, incidental WBC ½	progressive CKD (ESKD at 7.5 yrs.), PD, DDKTx at 9.5 yrs., at 24 yrs. still functioning	ocular, pHT, short stature (final growth 153 cm), DM	death at 24 yrs. by cerebral stroke	c.18_c.21delGACT (p.Thr7Phefs*7); c.18_c.21delGACT (p.Thr7Phefs*7)

						cystine assessment	1.KT (eGFR- 63 ml/min/1.73 m^2)			
F10	F	1998	1.3	failure to thrive, MR, rickets, P/P, recV, FS, HC	2	irregular systemic since 3. yr., WBC ½ cystine assessment not performed	progressive CKD (ESKD at 11 yrs.), PD	ocular, subclinical pHT, short stature (< 3. perc.), dysphagia	death at 12 yrs. by choking	not performed
F11	F	2007	0.9	failure to thrive, MR, FS, HC, ↑rcECHO	4	irregular systemic since 4. yr., regular since 10 yr., irregular topical since 4. yr., regular topical since 11. yr., regular WBC ½ cystine assessment since 10. yr.	progressive CKD (ESKD at 14 yrs.), PD	ocular, subclinical pHT, short stature (< 3. perc.)	short rhGH therapy	c.681+1G>A (p.?); c.681+1G>A (p.?)

F12.1	F	2008	1.5	failure to thrive, MR, rickets, P/P, FS, HC	1.5	adequate systemic since 1.5 yr., regular topical since 2. yr,, regular WBC ½ cystine assessment	at 13 yrs. normal eGFR (eGFR- 116 ml/min/1.73 m^2)	ocular, subclinical pHT, normal growth (25-50. perc.)	ulcerative colitis since 11. yr.	57kb-deletion (?_3533579)_(3561452_?)del ;c.314_317delACTC (p.His105Profs*12)
F12.2	F	2011	0.5	iFS, HC	0.5	adequate systemic since 0.6 yr., regular topical, since 1. yr., regular WBC ½ cystine assessment	at 10.5 yrs. normal eGFR (eGFR- 97 ml/min/1.73 m^2)	ocular, subclinical pHT, normal growth (10-25. perc.)		57kb-deletion (?_3533579)_(3561452_?)del ;c.314_317delACTC (p.His105Profs*12
F13	F	1996	2	iP, HC	20	only topical since 24. yr., incidental WBC ½ cystine assessment	slow progressive CKD (at 27 yrs. eGFR-87 ml/min/1.73 m^2),	ocular, normal final growth (161 cm)	initially nutcracker syndrome as a reason of proteinuria was suspected, then after kidney	c.225+5_c.225+6delGT/insCC; c.530A>G (p.N177S)

									biopsy at 13. yr.- FSGS. Ophthalmological evaluation at 20 yr. revealed corneal cystine deposits	
F14	F	2005	9.5	iP, HC, ↓eGFR	12	regular systemic and topical since 16 yr., regular WBC ½ cystine assessment since 15. yr.	slow progressive CKD (at 17 yrs. eGFR-72 ml/min/1.73 m^2)	ocular, normal growth (10-25. percentile)	initially after kidney biopsy at. 10. yr.- CTIN was suspected. Ophthalmological evaluation at 12. yr. revealed corneal cystine deposits	**c.629T>C (p.Leu210Pro)**; **c.462-27_462-3del**

Abbreviations: CKD- chronic kidney disease, CNS – central nervous system, CTIN- chronic tubulointerstitial nephritis, DDKTx- deceased donor kidney transplantation, DM- diabetes mellitus, eGFR- estimated glomerular filtration rate, ESKD- end stage kidney disease, F- female, FS- Fanconi syndrome, FSGS- focal segmental glomerulosclerosis, FU- follow-up, HD- hemodialysis, GH- growth hormone, GW- gestational week, HC- hypercalciuria, iFS- incomplete FS, iP- proteinuria, KTx- kidney transplantation, KT- kidney transplant, LDKTx- living donor kidney transplantation, M- male, PD- peritoneal dialysis, P/P- polyuria/polydipsia, pHT- primary hypothyroidism, ↑rcECHO- hyperechogenicity of renal cortex, recV- recurrent vomiting, WBC- white blood cells