A Cohort of 23 Individuals with Cystinosis: Up to 30 Years of Follow-up

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Background
Since cysteamine was introduced as a treatment for cystinosis in the 1970s the outcomes for individuals with cystinosis have markedly improved. This presentation explores the outcomes of 23 patients seen at a single

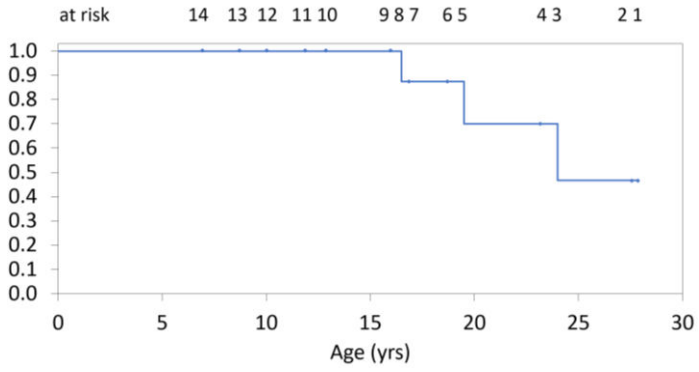
Results
Of 23 patients, 9 transferred into the program, 5 as adults. Three adolescent presentations[1] were diagnosed at 10 (9 - 11) years, those with infantile cystinosis at 1 (0.1 - 2.7) years old. Follow-up was for 8.6 (1.3 - 27.6) years. Four are sib-pairs with the diagnosis of the younger made either soon after an initial adolescent presentation or by an elevated WBC cystine level just after birth. Three patients are no longer followed (one died and two moved). The remaining 20 are 22.3 (5.7 - 55.4) years old.

Four patients required treatment for hypothyroidism, two of whom presented with adolescent cystinosis. Only two developed diabetes both with adolescent cystinosis. For all patients seen 10 (43%) reached kidney failure at 16.3 (12.2 - 25.7) years of age and received a kidney transplant with one transplant still functioning after 40 years.

When last seen the 14 patients followed from diagnosis were 17.1 (6.9 - 31.2) years old. Mixed leukocyte WBC cystine levels at diagnosis were 1.41 (0.85 – 3.82) nmol 1/2cystine/mg protein.
The average WBC cystine (nmol 1/2cystine/mg protein) on treatment by age were:

	Overall	<10 years	10 to 20 years	>20 years
Median	0.25	0.26	0.24	0.12
Range	0.15 – 0.42	0.14 – 0.51	0.11 – 0.33	0.11 – 0.42
avg #/patient	68	34	30	26
#patients	14	14	11	5

None of these 14 patients have hypothyroidism or diabetes. Their height SDS is -1.1 (3.62 - 0.39). Three had kidney failure at age 19.5 (16.5 - 24) years. Survival analysis shows a median age of 24 years for kidney failure. One patient with maintained native kidney function has delivered two healthy babies with breast milk containing minimal cysteamine[2]. One patient has received an autologous hematopoietic stem cell gene therapy transplant and is not taking cysteamine.



Conclusions
For patients seen from diagnosis, with the use of growth hormone in a majority, the median HSDS was higher at -1.1 than in in a large European cohort[3]. The average WBC cystine levels between 10 - 20 years of age were not higher than those <10 years of age and median kidney survival longer at 24 years than 19.5 years in a large European 1990s cohort[3].
For infantile cystinosis, with low WBC cystine levels, non renal sequelae appear unlikely to occur before 30 years of age.

1) Midgley J et al Natural history of adolescent-onset cystinosis Ped Nephrol 26:1335-7 (2011)
2) Chan L et al Pregnancy and Breastfeeding in Nephropathic Cystinosis With Native Kidneys Kid Int Reports 7:1716–1719 (2022)
3) Emma F et al An international cohort study spanning five decades assessed outcomes of nephropathic cystinosis Kidney Int 100:1112–1123 (2021)
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