

Cystinosis – Swallowing problems



Session plan

- Expert patient discussion with Mr Jonathan Terry



- Is the incidence reducing?

Questions

- How old were you when you first noticed swallowing issues?
- What symptoms did you start with
- How have those symptoms evolved over time?
- How did these problems affect you day to day?

- What is your understanding of the causes of the swallowing problems?

- What tests did you have _ useful or not?

- What treatments have you tried and how do you rate them?

- **Swallowing Dysfunction in Nephropathic Cystinosis**
- Barbara C. Sonies, Ph.D., Evan F. Ekman, B.A., Hans C. Andersson, M.D., Megan D. Adamson, M.D., Stephen G. Kaler, D.M.D., M.D., Thomas C. Markello, M.D., Ph.D., and William A. Gahl, M.D., Ph.D.
- N ENGL J MED 1990; 323:565-570 [August 30, 1990](#)

- **Swallowing dysfunction in 101 patients with nephropathic cystinosis: benefit of long-term cysteamine therapy.**
- [Sonies BC, Almajid P, Kleta R, Bernardini I, Gahl WA.](#)
- [Medicine \(Baltimore\).](#) 2005
May;84(3):137-46.

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Original Article

**Nephrology
Dialysis
Transplantation**

Follow-up and treatment of adults with cystinosis in the Netherlands

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Table 1. Clinical characteristics of adult cystinosis patients

Patient no.	Age	Sex	Height (cm)	Age at start of dialysis	Age at renal transplants	Most recent creatinine value ($\mu\text{mol/l}$)	Professional status	Visual acuity	Thyroid function	Glycaemia	Neurological and muscle dysfunction
1	19	F	161	18	—	600	health care studies	decreased	normal	normal	no
2	21	F	160.1	10	12/14	160	shop assistant	normal	normal	normal	sw/dys
3	21	M	158.5	12	14	100	low professional studies	normal	normal	*	no
4	23	F	150	11	12	79	secretary	*	normal	normal	no
5	24	M	150	7	7/9/19	58	does not work	decreased	decreased	**IDDM type 1	epilepsy
6	25	M	168.8	17	19	655	does not work	normal	decreased	*	no
7	29	M	163	13	16	800	driver	normal	*	normal	no
8	30	M	175.0	not started	—	571	mechanic	*	normal	normal	no
9	33	F	154.7	11	22	96	nurse	decreased	*	normal	no
10	36	M	166	22	19/25	120	does not work	decreased	decreased	normal	sw/dys, dyspnoea

sw/dys. swallow dysfunction; *not controlled/unknown; **IDDM, insulin-dependent diabetes mellitus.

Table 2. Cysteamine treatment of adult cystinosis patients

Patient no.	Age at start cysteamine (years)	Duration of cysteamine (years)	Daily cysteamine (mg/kg)	Cysteamine daily doses (times per day)	Cystine measurements (per year)	Last leukocyte cystine content (nmol/mg protein)	Cysteamine eye drops
1	8	11	60	2	2	0.45	yes
2	11	10	40	2	2	0.35	yes
3	3	18	70	4	2	0.11	yes
4	14	9	40	4	1	0.83	yes
5	*	–	–	–	**	–	no
6	*	–	–	–	2	0.23	yes
7	*	–	–	–	**	–	no
8	17	13	40	6	1	0.072	no
9	15	18	55	3	1	0.12	yes
10	25	11	40	4	2	3.00	no

*not treated; **not measured.



