

# Interdisciplinary Cystinosis Clinic Rosenheim – A Treatment Model for Rare Multiorgan Diseases

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and the German Cystinosis Patient Support Group

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## Background

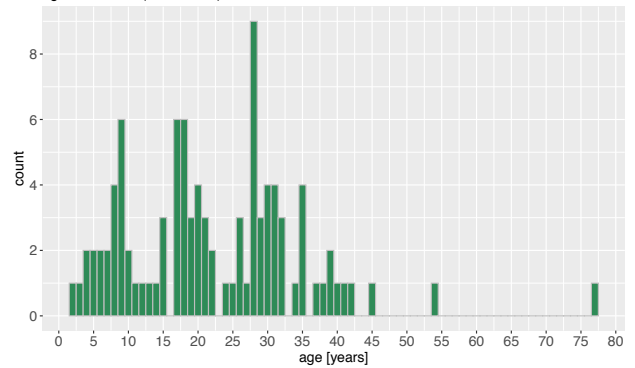
Nephropathic cystinosis is a rare, genetic lysosomal storage disease characterized by the abnormal accumulation of the amino acid cystine in nearly all organs and tissues. The organs most affected are the kidneys, the endocrine organs, the muscles, and the eyes.

In Germany, approximately 130 individuals have cystinosis. Before treatment became available, children with nephropathic cystinosis died at an early age. After the introduction of therapy with cysteamine bitartrate in the early 1980s, cystinosis was transformed from a lethal disorder into a chronic disease, which affects multiple organs.

The rate of survival and the extent of organ damage directly correlate with time of diagnosis and adherence to cysteamine depleting therapy. A major side effect of therapy is halitosis, a bad odor, leading to reduced adherence.

For patients with infantile nephropathic cystinosis transfer from pediatric to adult care is a special challenge with respect to the characteristics of the disease being a rare and a multiorgan disease. In addition there are still unresolved open questions such as long-term outcome.

age distribution (31.03.2020)



## Clinical data at time of presentation

born before 1997	n = 94	born after 1997
n = 41	Patient population	n = 53
m = 23 f = 18	Gender	m = 28 f = 25
<b>Age distribution [years]</b>		
<b>(at time of presentation at the clinic)</b>		
26.40 (16.90 - 39.01)	mean min. - max.	8.30 (0.04 - 17.31)
<b>Age at diagnosis [month]</b>		
21.14 (4.00 - 72.00)	mean min. - max.	19.35 (0.00 - 96.00)
<b>Cysteamine doses [mg/kgKG/d]</b>		
25.96 0.00-57.10	mean min. - max.	46.99 0.00-81.20

## Materials & Methode:

In order to improve outpatient care for people with cystinosis, a model case for patient treatment of rare diseases in the form of a comprehensive interdisciplinary clinic was established through a collaboration between the Cystinosis Patient Support Group and the Rosenheim Children's Hospital. The primary objectives were to build a long-term multidisciplinary team that would address all organs involved and to create a transition model for these patients.

### Outpatient clinic:

- A multidisciplinary team of experts at a single, dedicated venue in Rosenheim
- For children and adults to secure transition
- A "one-stop approach", 10 hours course, 13 different specialties/specialists
- Clinic takes place 5 times a year, over the course of two days
- Additional care close to home - usually once a year

## Adherence to cysteamine therapy

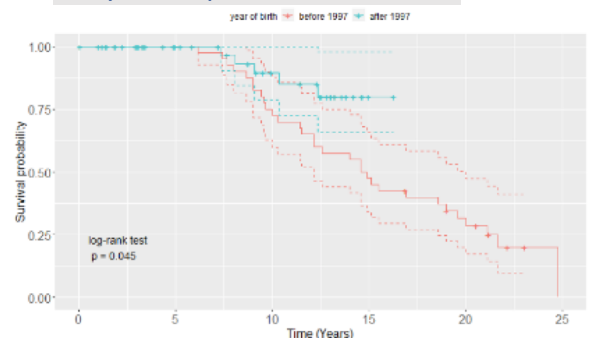
	born < 1997 n = 41		born > 97 n = 53
	22	Good adherence	50
	2	Questionable adherence	2
	17	No adherence	1

## Results

### Clinic

- very well accepted
- 10/2012 - 06/2019
- 378 patient contacts  
156 adults  
222 children  
97 individual patients
- - Permanent team
- - Different stages of disease
- - Long-term follow-up
- - Large number of patients
- - Specialized physicians in each field
- - Early diagnosis and interventions
- - Bundling of medical findings
- - Enhance patient communication
- Adult patients were regularly seen by nephrologists, without addressing specific extrarenal complications of Cystinosis.
- Patients born before 1997 showed an impaired renal outcome and low adherence.
- Halitosis was the main reason for adult patients to stop therapy.
- Reintroduction of therapy was possible in adult patients: full dose n=5, partial dose n=8.

## Kidney-Function: patients born and after 1997



## Conclusion

The interdisciplinary clinic was very well received by both children and adults, which underlines the importance of a multidisciplinary approach for this very rare multiorgan disease. Our approach is a model case also for other rare diseases.