Joint efforts from parents and doctors to promote the care for cystinosis patients in Russia

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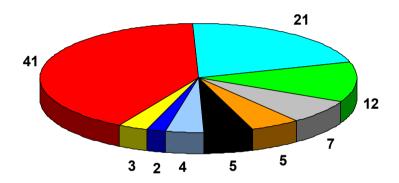
## Prevalence of ESRD in children per 1000000 population in Russia (2000-2006)





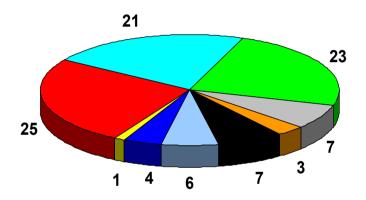
# Causes (%) of pediatric CRF and ESRD on RRT in Russia

(2006 **Г.**) CRF, 1493 children



- **■** Obstruct.uropathies
- Hypoplasy/Dysplasia
- **■** Glomerular diseases
- □ Cystic diseases
- Pyelonephritis/VUR/TIN
- Hereditary nephritis
- HUS
- Systemic/vasculitides
- Other

#### ESRD on RRT, 483 children



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#### Cystinosis: a new challenge

- Lack of knowledge and diagnostic tools resulted in ignorance of the disease
- First case was diagnosed in 2007. Since that the program to search for cystinosis patients was launched in Institute of pediatrics NCZD
- So far no molecular genetic tests available
- Nevertheless, 5 cases were diagnosed during less than 2 years

#### **Patients**

Gender/a ge	Fanconi	Ocular involve ment	GFR	Cystea mine	Tx
m/3	+	+	83	+	
f/2	++	+	68	+	
f/3	++	++	53	+	
m/14	+	+		+	+
f/24	+	+++		+	+

#### Social aspects

- Lack of cysteamine despite all medication for ESRD children must be supplied and funded by government
- No support from social security for those not in ESRD
- No skills and motivation to diagnose cystinosis from medical professionals

### Psychological aspects

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Phases of attitude	"Careful" optimism	Frustration with scaring reality	"Life is going on" – let's do something	There might be the way out if we act jointly
Doctors	We are so smart to diagnose a rare disease	Now how do we manage it without drug and experience?	But the problem has a solution in other areas. And we've a professional community.	We may be a team with other professionals and with the parents
Parents	We have a diagnosis at last. But our disease can't be "cured"?	God! There is no drug on the market here. And it's so expensive!	But we are not alone in that situation and doctors really want to help us	We may be a team with other families and with medical professionals

#### **Activities**

- Working group established based on an Institute of Pediatrics NCZD (4 members)
- Four talks for cystinosis given on a major national conferences for pediatrics and nephrology
- A review article published in Nefrologia I dializ http://www.nephro.ru/magazine/article.php?id=20759
- Parents organized "mini society" to care children with cystinosis
- Letters to Ministry of Health and local governments
- Local TV was used to tell about some of these patients
- Contact to international Cystinosis Society established

#### Results

- The manufacturer expressed an interest for registration process
- A purchase of cysteamine abroad assisted for parents. Compensation secured from local social security funds for 3/5 patients
- Meetings of "Cystinosis mini-society" every 6 months
- Quality of life improved according to questionnaire in 4/5 families
- Awareness of the doctors on sites probably increases

#### Further considerations

- More actively use of mass media
- Further assist for cysteamine registration and import
- Introduction of molecular genetic tests
- Creation of similar "mini-societies" for other diseases patients