



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

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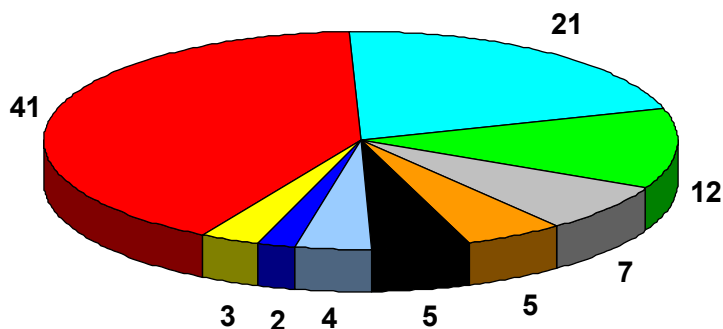
Prevalence of ESRD in children per 1000000 population in Russia (2000-2006)



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

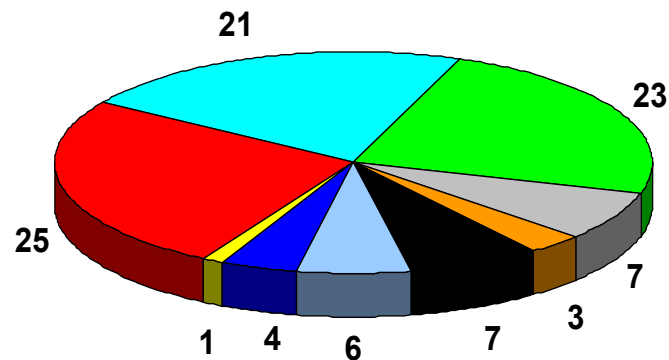
(2006 г.)

CRF, 1493 children



- Obstruct. uropathies
- Hypoplasia/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/age | Fanconi | Ocular involvement | GFR | Cysteamine | 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

| | | | | |
|--------------------|--|--|--|---|
| 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