



3rd-CNE-International-Cystinosis-Conference-2022

Bring good treatment for cystinosis patients around the globe

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Treatment of cystinosis patients in Turkey: Key Points

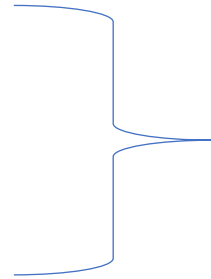
- *Diagnosis and follow-up is not a problem with the educated paediatricians and paediatric nephrologists*
- *Patients mostly seen by paediatric nephrologist; small percentage followed by the doctors of metabolic diseases*
- *Multidisciplinary approach*
- *Every 2–3-month follow-up*
- *Close monitoring the physical and lab*
- *LCL 2-3 times a year*
- *Good communication with the families*
- *Encouraging the family association to connect*
- *Education activities with family association*
- *Problem in transition to adult nephrology remains*

- Specific treatment of cystinosis
 - Cystine-depleting treatment - Cystagon
 - Eye drops - Cystadrops
- Treatment of Fanconi Syndrome

Management of renal Fanconi syndrome

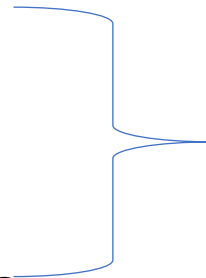
- Free access to water
- Nutritional support

- K Citrate
- K Chloride
- Na bicarbonate



Electrolyte supplementation

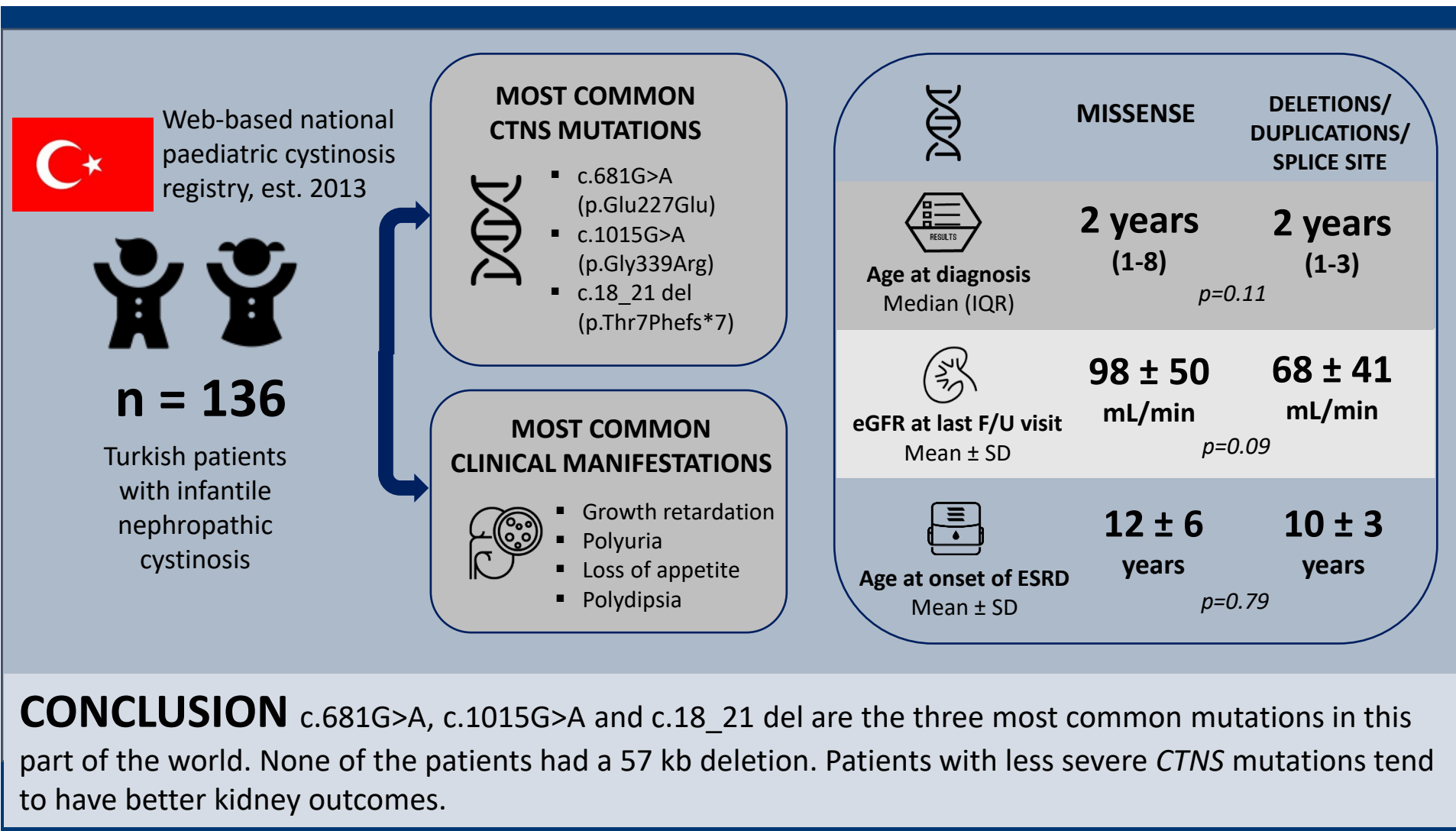
- Phosphate supplementation
- Vit D supplementation
- Alpha-calcidol
- Indomethacin for severe polyuria



Preventing or treating rickets

- GH treatment for poor growth – It is not available in Turkey for CKD and cystinosis. The indication should be a true GH deficiency it is recently changed
- Carnitine supplementation

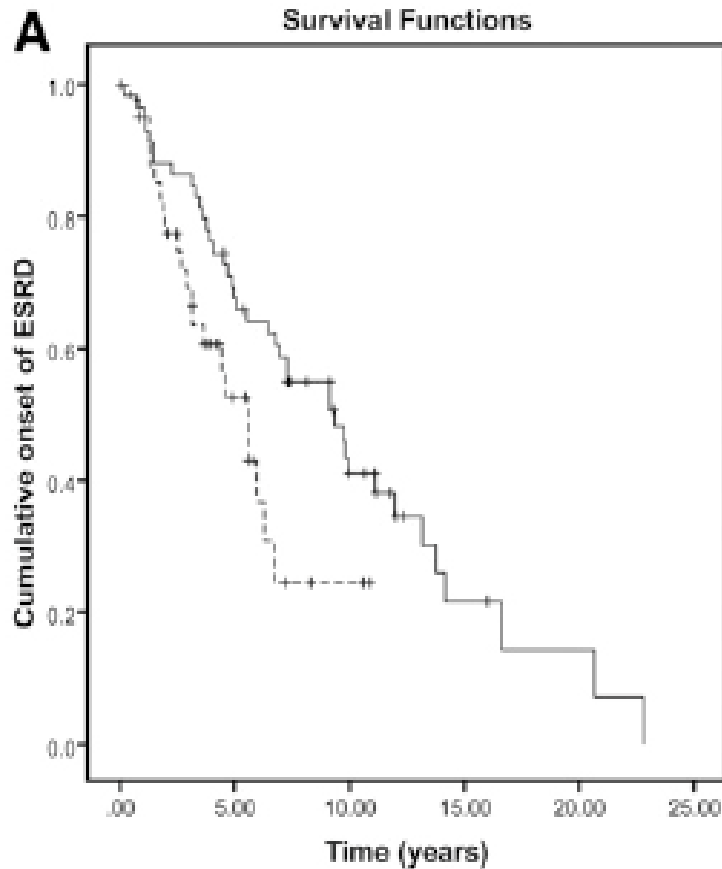
The Clinical and Mutational Spectrum of Turkish Cystinosis Patients



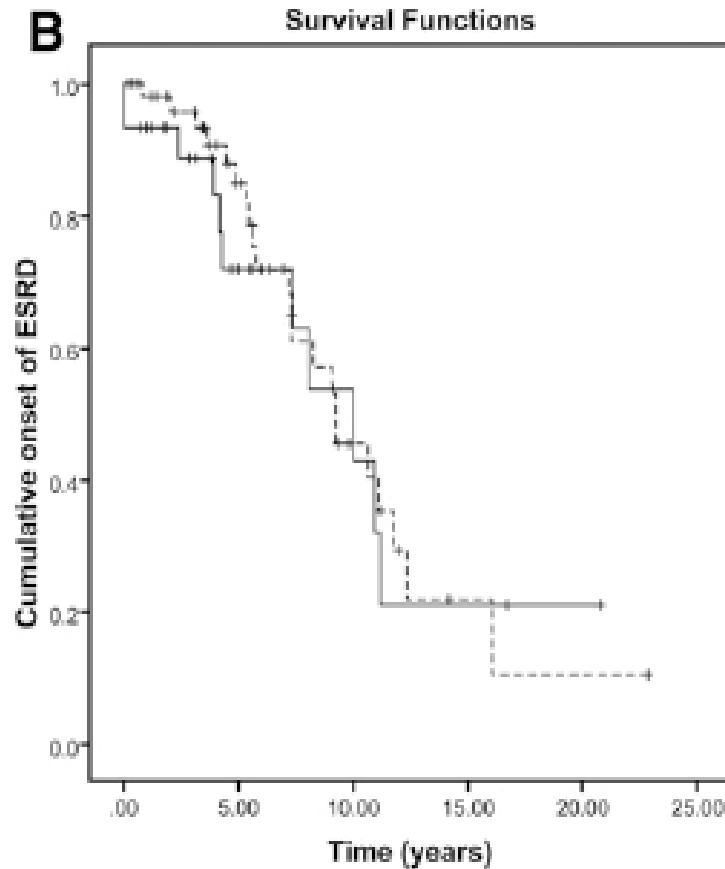
The Clinical and Mutational Spectrum of Turkish Patients with Cystinosis

Patients in whom treatment of cystinosis began at age <2 years old had later onset of ESRD ($P=0.02$).

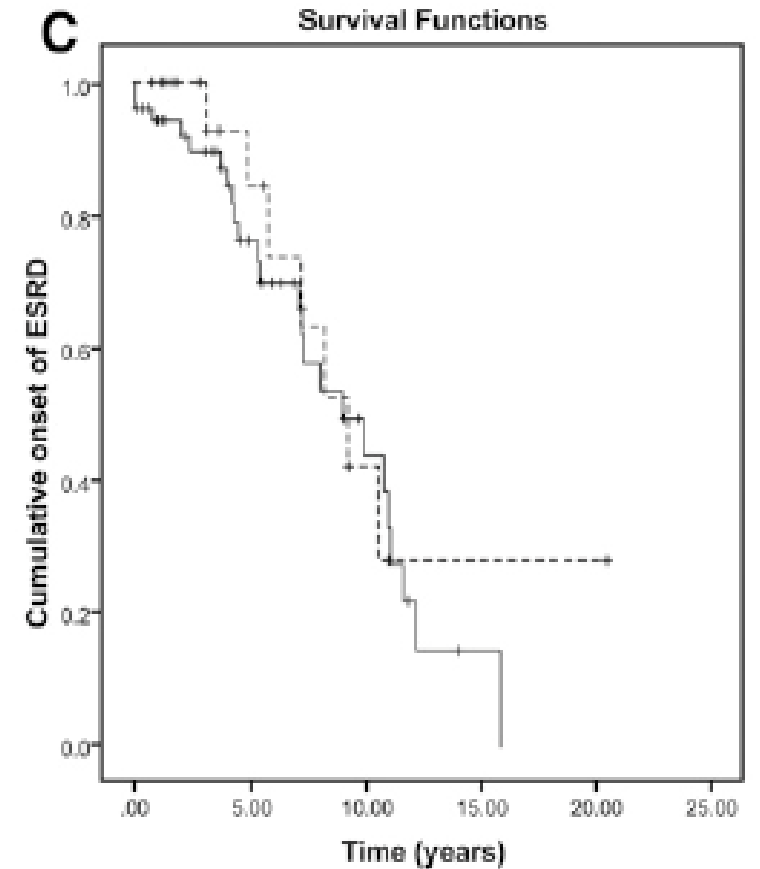
Yılmaz, Aytül Noyan, İsmail Dursun, İbrahim Gökçe, Beltinge Demircioğlu Kiliç, Selçuk Yüksel, and Fatih Ozaltın, on behalf of the contributors of The Journal of Clinical Investigation, 2017. doi: <https://doi.org/10.2215/CJN.00180117>



Cysteamine treatment initiated at age <2 versus >2 years old $p=0.02$



Missense versus deletion/duplication/splice mutations $p=0.79$



Most common mutations versus others $p=0.54$

Renal transplantation in cystinosis

- Most of our patients receiving living-related transplantation with heterozygote parents
- We have seen no recurrence of cystinosis in renal graft
 - graft biopsies: cystine crystals in invading host cells, but not in tubular or glomerular epithelium
- Graft survival is excellent in our hands as well
- Nephrectomy of the native kidneys because of persistent polyuria is rarely required
- We use the same immunosuppressive treatment as in non-cystinosis patients
- We start cysteamine treatment when patient can take oral medicine after transplantation and continues life long

Renal Transplantation



TR36, compound heterozygous for the c.681 G>A (p.E227E) and the (c.1015 G>A; p.G339R), is noncompliant and ESKD at approximately 10 years of age; she was transplanted with her mother's kidney at age 14

TR 6



Diagnosis at age 1, transplanted at age 19



Recently transplanted at age 21



A Father with cystinosis ICSI, intra-cytoplasmic sperm injection



A mother with cystinosis

We are not good at transition

- Rare Disease
- Adults used to see less rare diseases but not now: they should learn rare diseases starts in childhood
- Adult nephrologists very busy with the prevalent diseases
Hypertension and Diabetic Nephropathy



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