

Format: Abstract

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available at [SciFO.org](https://scifido.org)[Rev Med Chil.](#) 2018 Jan;146(1):111-115. doi: 10.4067/s0034-98872018000100111.**[Nephropatic cystinosis: report of one case].**

[Article in Spanish]

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Nephropatic cystinosis (NC) is a rare disease associated with pathogenic variants in the CTNS gene, with a common variant that consists of a 57kb-deletion involving CTNS. Patients with NC that are treated with cysteamine improve their life quality and expectancy. We report a 12-month-old girl with a poor growth rate since the 4th month of life. She was admitted to the Hospital with acute kidney injury, severe dehydration and metabolic acidosis. She was treated with volume restorative and bicarbonate. Proximal tubulopathy and Fanconi's syndrome was diagnosed. Medical treatment improved renal function that was stabilized in stage 4 chronic kidney disease (CKD). Since infantile NC was suspected, CTNS genetic analysis was considered. Genomic DNA was isolated from peripheral blood to perform PCR for exons 3-12 in CTNS gene and for the specific 57kb-deletion PCR. Afterwards, variant segregation analysis was performed in the familiar trio. The genetic analysis showed that the patient was homozygous for the common 57kb-deletion encompassing CTNS that had been inherited from her asymptomatic heterozygous parents. The molecular confirmation allowed genetic counselling for parents and facilitated the access to cysteamine. Oral treatment with cysteamine resulted in improvement of renal function to CKD stage 3. After 16 months of treatment the patient shows metabolic stability and mild recovery of height. Ophthalmologic follow-up detected ocular cystine crystals 12 months after diagnosis, starting cysteamine drops.

PMID: 29806685 DOI: [10.4067/s0034-98872018000100111](https://doi.org/10.4067/s0034-98872018000100111)[Indexed for MEDLINE] [Free full text](#)Publication type, MeSH terms, Substances, Supplementary concept LinkOut - more resources