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CASE SERIES

Rare Case-Nephropathic Cystinosis

S. N. Sivaraj*, G. Durai Arasan and S. Lakshmi

Department of Paediatrics, Madras Medical College, Chennai – 600003, Tamil Nadu, India;
drnasasivaraj@gmail.com

Abstract

Nephropathic cystinosis is a rare autosomal recessive lysosomal storage disorder characterised by defective cystine transport across lysosomal membranes, leading to multiorgan involvement. We report a case of a 5-year-old male child presenting with growth failure, polyuria, rickets, and dyselectrolytemia. Diagnostic evaluation revealed Fanconi-like features and bilateral hydronephrosis. Genetic analysis was suggestive of nephropathic cystinosis. Early initiation of cysteamine therapy is crucial to delay progression to end-stage renal disease.

Keywords: Fanconi Syndrome, Lysosomal Storage Disorder, Nephropathic Cystinosis, Polyuria, Rickets

1. Introduction

Nephropathic cystinosis is a rare autosomal recessive lysosomal storage disorder caused by mutations in the CTNS gene located on chromosome 17p13, leading to defective cystine efflux and intralysosomal crystal accumulation[1]. The global incidence is approximately 1 in 100,000–200,000 live births, with higher prevalence in consanguineous populations[2]. Based on age of onset and severity, three clinical variants are recognised: infantile nephropathic (classic), juvenile (intermediate), and adult ocular (benign) forms. The infantile form is the most severe, presenting during the first year of life with renal Fanconi syndrome, polyuria, dehydration, growth retardation, rickets, and metabolic acidosis. Progressive cystine deposition affects multiple organs—especially kidneys, eyes, thyroid, pancreas, and muscles—leading to chronic renal failure and systemic complications if untreated[3]. Early diagnosis through biochemical and genetic testing, along with cysteamine therapy and supportive management, can delay progression to end-stage renal disease and improve long-term survival[4].

2. Aim and Objectives

To report and discuss a rare case of nephropathic cystinosis in a pediatric patient, highlighting the clinical features, diagnostic challenges, and management approach.

3. Review of Literature

Epidemiology: Rare autosomal recessive disorder, higher prevalence in consanguineous populations.
Clinical Features: Polyuria, polydipsia, growth retardation, rickets, photophobia, and renal Fanconi syndrome.
Complications: Progressive renal failure, hypothyroidism, ocular involvement, and systemic complications.
Treatment: Oral cysteamine is the mainstay of therapy to reduce cystine accumulation. Supportive measures include electrolyte correction and management of complications. Prognosis: Untreated patients develop ESRD by the first decade of life.

4. Materials and Methods

This case report was based on clinical examination, laboratory investigations, imaging, and genetic analysis. Investigations included:

Blood tests: CBC, electrolytes, renal function tests.
Urine analysis: pH, , calciuria as shown in table 1.
Imaging: Ultrasound abdomen, X-ray for rickets as shown in figure 2.

*Author for correspondence

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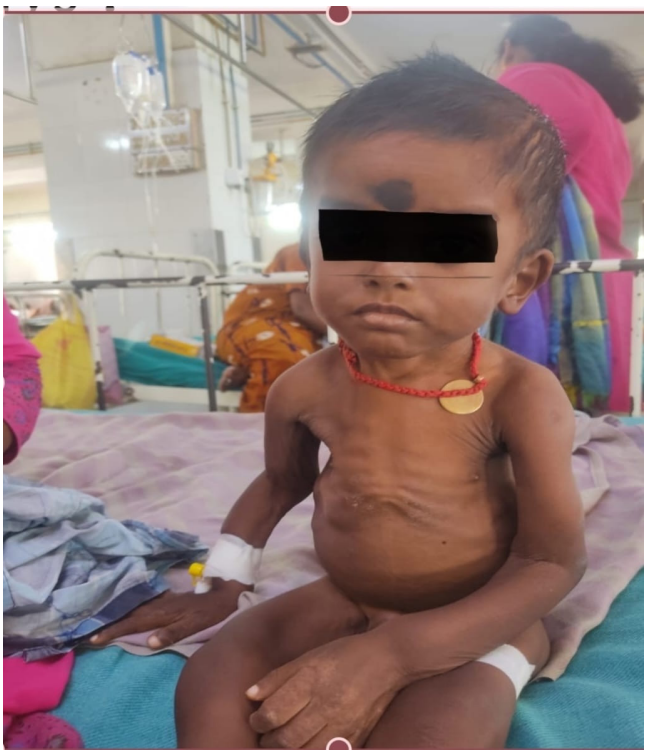
Table 1. Investigation

	21/5	23/5	25/5
Urea	52	56	35
Creatinine	1.7	1.3	1.0
Sodium	109	110	112
Potassium	2.2	2.9	3.9
Calcium	3.3	4.5	5.4
Phosphorus	3		2

Urine electrolyte	22/5	23/5
Sodium	40	36
Potassium	5.1	5.54
Calcium	1.8	2.6
Creatinine	4.7	4.0
Chloride	25.5	28.6
Ca/cr ratio	0.38	
Anion gap	19.5	



Figures 1. Patient images.

Special tests: TSH, ceruloplasmin.
Genetic analysis: CTNS mutation screening as shown in figure 2.

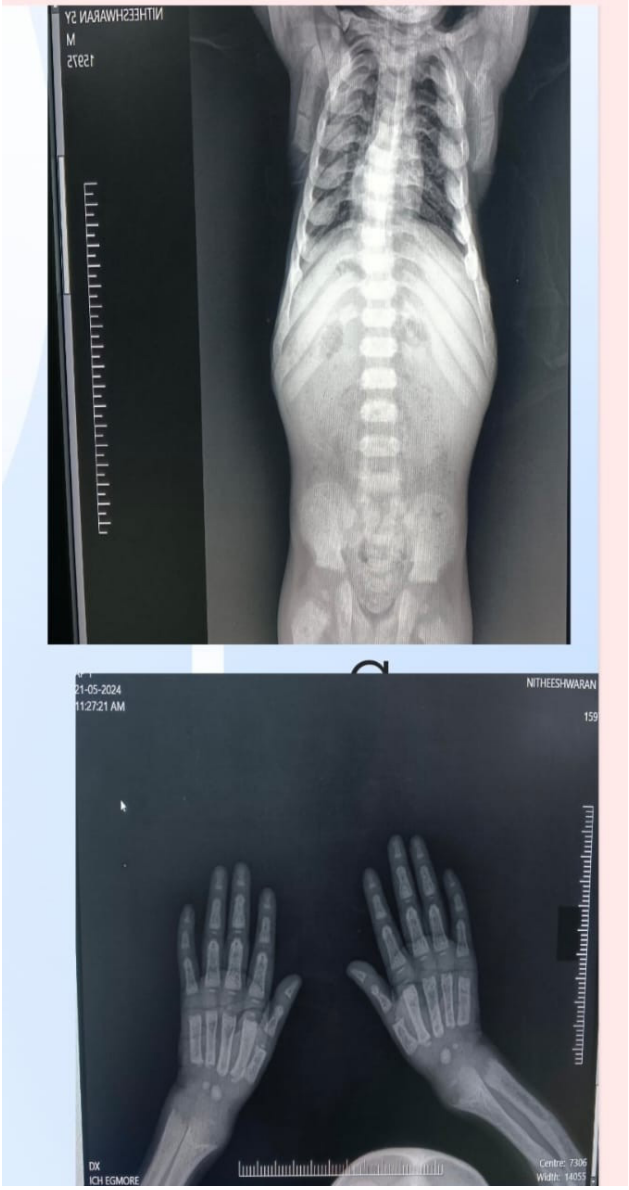
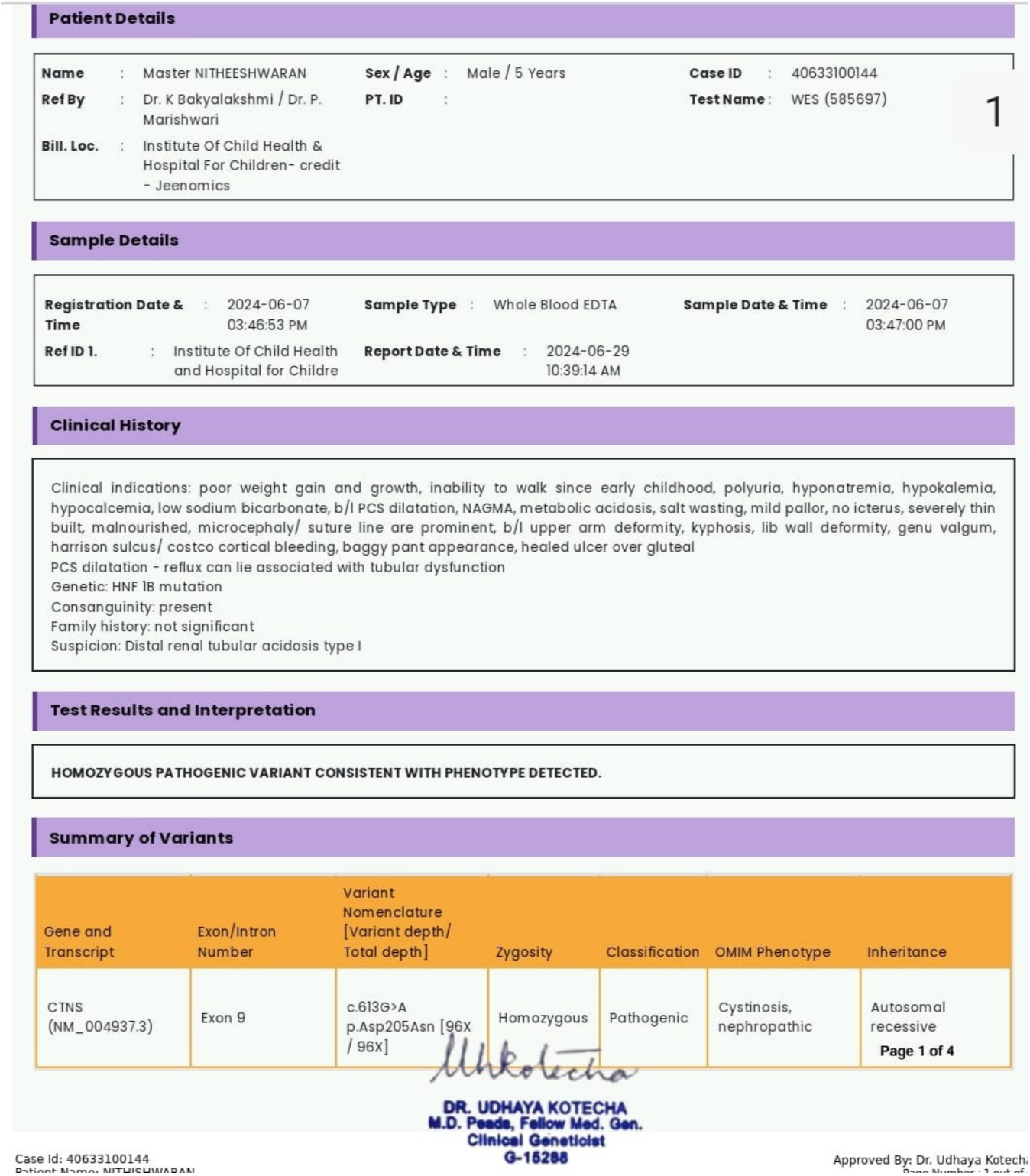


Figure 2. Xrays.

5. Results (Including Observations)

Patient details: 5-year-old male, born to consanguineous parents as shown in figure 1, with developmental delay, poor weight gain, and polyuria.
Anthropometry: Weight 6.2 kg (<1st percentile), height 75 cm (<1st percentile), microcephaly.
Lab findings: Severe dyselectrolytemia (hyponatremia, hypokalemia, hypocalcemia, hypophosphatemia), metabolic acidosis, anaemia (Hb 8 g/dL), elevated TSH, and ALP 1097 IU/L. Imaging: *USG Abdomen*:
B/L Hydronephrosis
No Nephrolithiasis/Calcinosis, increased echoes.



Patient Details

Name	: Master NITHEESHWARAN	**Sex / Age**	: Male / 5 Years	**Case ID**	: 40633100144
Ref By	: Dr. K Bakyalakshmi / Dr. P. Marishwari	**PT. ID**	:	**Test Name**	: WES (585697)
Bill. Loc.	: Institute Of Child Health & Hospital For Children- credit - Jeenomics				

1

Sample Details

Registration Date & Time	: 2024-06-07 03:46:53 PM	**Sample Type**	: Whole Blood EDTA	**Sample Date & Time**	: 2024-06-07 03:47:00 PM
Ref ID 1.	: Institute Of Child Health and Hospital for Childre	**Report Date & Time**	: 2024-06-29 10:39:14 AM		

Clinical History

Clinical indications: poor weight gain and growth, inability to walk since early childhood, polyuria, hyponatremia, hypokalemia, hypocalcemia, low sodium bicarbonate, b/l PCS dilatation, NAGMA, metabolic acidosis, salt wasting, mild pallor, no icterus, severely thin built, malnourished, microcephaly/ suture line are prominent, b/l upper arm deformity, kyphosis, lib wall deformity, genu valgum, harrison sulcus/ costco cortical bleeding, baggy pant appearance, healed ulcer over gluteal
PCS dilatation - reflux can lie associated with tubular dysfunction
Genetic: HNF 1B mutation
Consanguinity: present
Family history: not significant
Suspicion: Distal renal tubular acidosis type I

Test Results and Interpretation

HOMOZYGOUS PATHOGENIC VARIANT CONSISTENT WITH PHENOTYPE DETECTED.

Summary of Variants

Gene and Transcript	Exon/Intron Number	Variant Nomenclature [Variant depth/ Total depth]	Zygosity	Classification	OMIM Phenotype	Inheritance
CTNS (NM_004937.3)	Exon 9	c.613G>A p.Asp205Asn [96X / 96X]	Homozygous	Pathogenic	Cystinosis, nephropathic	Autosomal recessive

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DR. UDHAYA KOTECHA
M.D. Peads, Fellow Med. Gen.
Clinical Geneticist
G-15288

Case Id: 40633100144
Approved By: Dr. Udhaya Kotech

Figures 3. Genetic report.

X-Ray: f/s/o rickets, cupping and splaying of wrists, knees.
Course: Treated with IV antibiotics, electrolyte correction, vitamin D supplementation, desmopressin trial, and genetic confirmation of nephropathic cystinosis.

6. Discussion

Nephropathic cystinosis presents with proximal tubular dysfunction leading to Fanconi syndrome and rickets. Early diagnosis is often delayed due to nonspecific features. This case emphasises the importance of considering cystinosis in children with growth failure and renal tubular acidosis. Genetic confirmation and cysteamine therapy remain key to management. Supportive care for electrolyte imbalance and prevention of complications are crucial.

7. Summary and Conclusion

Nephropathic cystinosis is a rare but important cause of growth failure and renal dysfunction in children. Clinical suspicion, early diagnosis, and initiation of cysteamine can significantly improve outcomes. Awareness among clinicians is essential to avoid delays in treatment.

8. References

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