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BeyondtheKidneys: Unmasking Urological Complications in Pediatric Nephrogenic Diabetes Insipidus – A Case Report.

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Abstract

Nephrogenic diabetes insipidus (NDI) is a rare disorder of water metabolism characterized by thekidney's profound inability to concentrate urine, even in the presence of normal or elevatedantidiuretic hormone (ADH) [2]. This tubular insensitivity results in the excessive production of diluteurine (polyuria) and compensatory excessive fluid intake (polydipsia). Clinically, NDI presents ininfancy with poor feeding, irritability, fever, and episodes of severe dehydration [1]. Because thesymptoms mimic common childhood illnesses, diagnostic delays are frequent and can result insevere structural urological complications due to chronic high urine output [3]. Diagnosis isconfirmed via a water deprivation test followed by desmopressin administration, which fails toincrease urine osmolality in NDI but does so in Central Diabetes Insipidus [6]. Managing such casesrequires a multidisciplinary approach. Here we present a 10-year-old boy diagnosed with NDI afterpresentingwithmassivebilateralhydronephrosisandatrabeculatedbladder,whowassubsequentlymanaged with targeted pharmacotherapy.

Keywords: Nephrogenic Diabetes Insipidus, Polyuria, Hydronephrosis, Water Deprivation Test, BladderTrabeculation, Pediatric Nephrology

Introduction

Nephrogenic diabetes insipidus (NDI) in the pediatric population is a rare but clinically critical disorder. Itis characterized by the failure of the distal nephron to respond appropriately to arginine vasopressin(AVP),despitenormalorelevatedsystemicsecretion[2,6].Thisdefectdisruptsnormalfluidhomeostasis,resultinginthecontinuousexcretionoflargevolumesofdiluteurineanddrivingcompensatorypolydipsia.If unrecognized, the condition can precipitate severe dehydration and life-threatening electrolyteimbalances [1,3].

Congenital NDI accounts for the vast majority of pediatric presentations [3,4]. The condition is mostfrequently driven by X-linked inheritance patterns caused by mutations in the AVPR2 gene, whichencodes the vasopressin V2 receptor. Less commonly, it results from autosomal recessive or dominantmutations in the AQP2 gene, which encodes the aquaporin-2 water channel [4]. Regardless of themutation, the consequence is an impaired ability of the renal collecting ducts to reabsorb water.

Earlyclinicalsignsininfants—suchaspoorweightgain,vomiting,unexplainedfevers,andirritability—arenotoriously non-specific, often delaying accurate diagnosis [1,3]. Early identification is essential toprevent long-term morbidities such as

growth retardation, neurodevelopmental delays, and functionalabnormalities like secondary bladder dysfunction from chronic volume overload [3]. While acquired formscanoccursecondarytomedications(e.g.,lithium)orobstructiveuropathy[5],congenitaletiologyremainsprimary in children, particularly in males with a family history of unexplained hypernatremia [4].

Case Report

Clinical Presentation

A 10-year-old male child presented to the pediatric outpatient clinic with a long-standing history ofextremepolydipsiaandcontinuouspolyuria,accompaniedbyasensationofincompletevoiding[6].Uponphysicalexamination,thechildhadavisiblyandpalpablydistendedurinarybladder,whichextended midway between the umbilicus and the pelvic brim.

Diagnostic Workup

An initial ultrasound of the kidneys, ureters, and bladder (USG KUB) revealed gross bilateralhydronephrosis and hydroureter. The scan also confirmed a significantly overdistended urinary bladderwith a post-void residual volume of 254 cc, strongly indicating chronic bladder outlet obstruction [3].

To further evaluate the urological architecture, a micturating cystourethrogram (MCU) and diagnosticcystoscopywereperformed.Theseimagingmodalitiesdemonstrateda"treetip"bladdercharacterizedbyirregular walls, with no evidence of vesicoureteral reflux (VUR) or posterior urethral valves (PUV). Directcystoscopyrevealedpronouncedtrabeculationsandahypertrophiedverumontanum.Urodynamicstudies were also completed to assess bladder pressure and capacity.

Due to the unrelenting polyuria, routine urinalysis was repeatedly performed, consistently showing aremarkably low specific gravity [2]. Paired osmolality testing was subsequently ordered; urine osmolalitywas profoundly low at 55.2 mOsm/kg, while serum osmolality remained normal, raising strong clinicalsuspicion for diabetes insipidus [6].

A formal water deprivation test was executed to confirm the diagnosis [6]. In a healthy physiologicalresponse, fluid restriction concentrates the urine (>600 mOsm/kg) as serum osmolality rises [2]. In thispatient, urine remained persistently dilute (<400 mOsm/kg) despite dehydration. Following theadministration of exogenous desmopressin, no appreciable increase in urine concentration occurred [6].This failure to respond to desmopressin definitively differentiated the condition from central diabetesinsipidus [3], cementing the diagnosis of Nephrogenic Diabetes Insipidus.

Management And Outcome

Followingtheconfirmeddiagnosis,thepatientwasinitiatedonadual-pharmacotherapyregimen.Hewasprescribed hydrochlorothiazide, a thiazide diuretic that paradoxically reduces urinary output in NDI byinducingmildvolumedepletion,therebyenhancingproximaltubularreabsorptionofsodiumandwater[3].Additionally, oxybutynin chloride was started to manage the bladder dysfunction and trabeculation. Thepatient remains under close follow-up and has shown notable clinical improvement.

Discussion

Thiscasehighlightsaclassicdownstreamconsequenceofundiagnosedorpoorlymanaged NDI:non-obstructive urological dilation [3]. The continuous, massive output of dilute urine eventuallyoverwhelms the functional capacity of the lower urinary tract. This chronic volume overload forces thebladdertostretchandhypertrophy,ultimatelyresultingintrabeculation,secondaryhydroureter,andgrossbilateral hydronephrosis—findings dramatically visible in our 10-year-old patient.

Definitive diagnosis requires a carefully monitored water deprivation test and exogenous desmopressinchallenge [6]. Genetic testing is also becoming increasingly vital, allowing clinicians to precisely identifyAVPR2orAQP2mutations[4],offeraccuratefamilycounseling,andmapoutlong-termprognoses.Whiledefinitive curative gene therapies are not yet clinically available, prompt symptomatic management usingthiazides,non-steroidalanti-inflammatorydrugs(NSAIDs),andadequatehydrationcandrasticallyreducepatient morbidity [3,5].

Conclusion

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Nephrogenicdiabetesinsipidusisararebutimpactfuldisorderdrivenbyrenalinsensitivitytovasopressin[2,6].
This condition predisposes children to severe polyuria, resulting not only in a high risk ofdehydrationandhypernatremia[1]butalsoinprogressive,severeurologicalcomplicationslike hydronephrosis and bladder hypertrophy if left unchecked [3]. Early recognition and aggressive earlymanagement are paramount to prevent irreversible renal and developmental damage [3,4]. Continuedresearch into the molecular basis of AQP2 channels and vasopressin V2 receptors continues to pave theway for future, targeted therapies [4,6].

References

1. Behrman,R.E.,Kliegman,R.M.,&Jenson,H.B.(Eds.).(2004).Nelsontextbookofpediatrics(17thed.).Saunders.
2. Robertson,G.L.(1981).Diabetesinsipidus.EndocrinologyandMetabolismClinicsofNorthAmerica,10(2),365–388.
3. Bockenhauer,D.,&Bichet,D.G.(2015).Pathophysiology,diagnosisandmanagementofnephrogenicdiabetes insipidus. Nature Reviews Nephrology, 11(10), 576–588.https://doi.org/10.1038/nrneph.2015.105
4. Knoers,N.V.A.M.,&Deen,P.M.T.(2001).Molecularandcellulardefectsinnephrogenicdiabetesinsipidus. Pediatric Nephrology, 16(12), 1146–1152. https://doi.org/10.1007/s004670100023
5. Grünfeld,J.P.,&Rossier,B.C.(2009).Lithiumnephrotoxicityrevisited.NatureReviewsNephrology,5(5),270–276. https://doi.org/10.1038/nrneph.2009.37
6. Christ-Crain,M.,etal.(2019).Diabetesinsipidus.NatureReviewsDiseasePrimers,5(1),54.https://doi.org/10.1038/s41572-019-0103-2

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