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FAMILIAL RENAL GLYCOSURIA - AN UNUSUAL CAUSE OF GLYCOSURIA IN FEMALE CHILD

Sreelatha S	Assistant professor Department of paediatrics, Kurnool medical college, Kurnool
P Ramya*	post graduate Department of paediatrics, Kurnool medical college, Kurnool *Corresponding Author
P Sandeep	post graduate Department of paediatrics, Kurnool medical college, Kurnool
L Gomathy	post graduate Department of paediatrics, Kurnool medical college, Kurnool

ABSTRACT Glycosuria can occur in the setting of global dysfunction of the proximal tubule known as fanconi syndrome. Glycosuria in this instance accompanies the excessive urinary secretion of amino acids, phosphate, and other solutes that are typically absorbed in the proximal tubule .The occurrence of glycosuria in the absence of both generalized proximal tubular dysfunction and hyperglycemia is called renal glycosuria and recognized as inherited disorder and is called familial renal glycosuria. Here we present a case of 7 year old female child diagnosed incidentally as familial renal glycosuria who came for cervical lymphadenopathy associated with grade 1 tonsillitis

KEYWORDS :

INTRODUCTION

Renal glycosuria is a benign disorder characterized by presence of large amounts of glucose in urine with normal blood glucose levels. There is no other defect of proximal tubular function. Normally the filtered glucose is completely reabsorbed by proximal tubule until tubular maximum is exceeded. Mutations in SLC5A2 [SGLT -2 CODING GENE] underlie most if not all cases of FRG. Even in its infrequent severe forms characterized by complete absence of tubular glucose reabsorption clinical manifestations are rare. No treatment is necessary

CASE REPORT

A 7 year old female child brought to opd with complaints of swellings in the both side of neck of size 2*2 cm with intermittent episodes of fever admitted in ward by assuming to be cervical lymphadenopathy. Later the mother repeatedly complaining of thick sticky urine for the past 1 year for which she consulted many hospitals but were undiagnosed and referred to GGH knl .

At initial days of admission work up regarding cervical lymphadenopathy done. On usg neck enlarged matted lymph nodes of size 3*2 cm given on right 1a level and multiple discrete enlarged lymph nodes noted in anterior cervical region. FNAC, ESR, MONTOUX, SPUTUM CBNAAT done to rule out TB lymphadenitis. All investigations not suggestive of TB lymphadenitis

Simultaneously workup regarding thick sticky urine done. A thorough clinical examination was done and child height and weight plotted on growth charts which appropriate for age .complete urine examination was sent showed glucose +++ but was negative for all other substances like nitrites , leucocyte esterase, urobilinogen ,blood and protein and importantly negative for ketones .Repeated further to confirm the glucose in urine In view of excluding type 1 diabetes mellitus FBS and PPBS and glycosylated hemoglobin were done values are 90 and 135 mg/dl and 5.7 respectively .Repeated again for confirmation but results were same .

No History regarding polyuria , polyphagia , wasting , and episodes of breathlessness , failure to thrive, muscle dystonia and ocular defect Developmental history normal Family history suggestive second degree consanguineous product with similar complaints for the sibling who was healthy , developmentally normal child for which above investigations done outside and were normal in spite of thick sticky urine

- General examination normal
- Vitals normal
- Anthropometry appropriate for age
- Other lab investigations done to rule out fanconi syndrome and are as follows
- Blood urea 23 mg/dl
- Sr creatinine 0.5mg/dl

Liver function test total bilirubin 0.9 mg/dl

- SGOT - 33 SGPT - 12 ALP- 173
- Serum calcium 10.4 mg/dl
- Serum electrolytes sodium 143 mmol/L
- Potassium 4.1mmol/L
- Chlorides 101mmol/L
- Urinary electrolytes as follows
- Sodium 47 mmol/gm creatinine
- Potassium 8.8 mmol/gm creatinine
- Chloride 43 mmol/gm creatinine
- Urine calcium 9 mg/dl
- Urine creatinine 39 mg/dl
- All of them are in normal limits
- The fractional excretion of sodium was calculated and was normal
- Urine was analyzed for reducing substances and was clinitest strongly positive
- Both parents were tested for glycosuria which was absent on CUE and dipstick method
- The diagnosis of renal glycosuria made by exclusion

DISCUSSION

With normal renal tubular function, glucose will only be excreted in urine when blood glucose concentrations are elevated, and the renal tubular re absorptive capacity for glucose is exceeded. Glucose transport in the human kidney and intestine is mediated by sodium-coupled glucose transporters, termed SGLT, and glucose transporters, termed GLUT. Glucose is transported across the brush border into the epithelial cell via SGLT, and across the basolateral aspect out of the epithelial cell by GLUT. The intestine and kidney share high-affinity Na+ /glucose co-transporters (SGLT1), but a low-affinity Na+ /glucose co-transporter (SGLT2) is kidney specific. In the normal nephron, filtered glucose is reabsorbed in the proximal convoluted tubule (PCT), about 90% being reabsorbed in the first segment of the PCT by high- affinity SGLT1, and the remainder in the second and third segments of the PCT by low-affinity SGLT2. Renal glycosuria (also known as benign glycosuria or nondiabetic glycosuria) is a benign, inherited condition in which glucose is excreted in the urine despite normal blood glucose concentrations.3 The condition is asymptomatic and self limiting, and is usually only discovered incidentally. However, glycosuria may be associated with other tubulopathies, such as Fanconis syndrome, cystinosis, Wilson's disease, hereditary tyrosinaemia, and oculocerebrorenal syndrome (Lowe's syndrome). These other tubulopathies, however, are associated with growth failure, muscle dystonia, polyuria, polydipsia, dehydration or ocular defects (glaucoma, cataracts). In renal glycosuria no other renal tubular dysfunction is present.

Benign glycosuria can be of two types: Type A: Classic glycosuria caused by a reduced tubular threshold and maximal reabsorptive rate for glucose. Type B: Normal maximal reabsorptive rate for glucose, but reduced tubular threshold. Plasma glucose, glucose tolerance, insulin and HbA1c are all normal, and all other renal tubular

abnormalities are absent in both types. The molecular mechanism giving rise to benign renal glycosuria has been discovered to be due to a defect in the low-affinity SGLT2 in the proximal convoluted portion of the renal tubules.

Inheritance is autosomal recessive, but because the condition is asymptomatic, a family history is often non-contributory. Suggested investigations for suspected cases of renal glycosuria include blood tests for urea, creatinine and electrolytes, calcium, phosphate, uric acid, glucose, and measurement of the HbA1c. Urine should be sent for urinalysis and microscopy, and measurement of phosphate, and the tubular maximum of phosphate reabsorption should be calculated. In cases where another tubulopathy is suspected on the basis of abnormal laboratory results or clinical findings, a 24-hour urine specimen should be collected and sent for amino acid analysis. Referral to a paediatric nephrologist is then recommended. Renal glycosuria requires no treatment or special dietary restrictions, and the prognosis is excellent. Once diabetes mellitus and other renal tubular disorders are excluded, all that remains is to explain the condition, and reassure the parents.

CONCLUSION

The case illustrates an unusual cause of glycosuria, the commonest cause of which is uncontrolled diabetes mellitus. In young non-obese children, type I diabetes mellitus is the commonest type of diabetes, where the absence of insulin production and secretion from the beta cells in pancreatic islets causes hyperglycaemia and ketosis. Renal glycosuria is a benign autosomal recessive condition caused by a defect in SGLT2 in the PCT of the nephron, which allows renal glycosuria to occur in the absence of a raised serum glucose level. Benign renal glycosuria is not associated with other renal tubular abnormalities. The condition is benign, and asymptomatic, and no treatment or dietary restriction is necessary.

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