Renal Glycosuria in Two Siblings

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Renal glycosuria denotes the renal tubular abnormality in individuals who excrete a variable amount of glucose in the urine at normal levels of blood glucose. The age at which the disorder is first detected varies, but in the majority of patients it is in the second decade(1). Though in one instance it was discovered in a 4 weeks old patient(2), reporting in pediatric age is less frequent. Even with the use of titration method for renal filtered glucose, different observers reached opposite conclusions.

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about the possible pattern of inheritance, both autosomal dominant and recessive trait being interpreted(1). Two siblings diagnosed as renal glycosuria as per the criteria laid by Marble and Ferguson(3) are described.

Case Report

A 3-year-boy was admitted with complaints of abdominal pain of 15 days duration. There was no history of polyuria, polyphagia and polydipsia or dysuria. He was anemic and systemic examination did not show any positive findings. His blood counts were within normal limits. Urine was persistently positive for reducing substance in all the samples taken in a day. It was confirmed to be glucose by dipstick method and osazone preparation. Blood sugar after overnight fast was 83 mg/dl and GTT was normal. Blood urea was 25 mg/dl and serum creatinine 0.8 mg/dl. Urine culture did not grow any organisms and ultrasonography of kidney was normal. Urinary excretion of bicarbonates, phosphates and aminoacids were within normal limits and also the acid-base studies in the blood. Urine sugar was persistently positive on follow up of 2 years. A 13-year-old sister and 10-year-old brother were screened and found to be normal. However, a 7-year-old sister had asymptomatic persistent glycosuria with normal fasting blood sugar and GTT. Other investigations were also normal. They were born to non-consanguineous parents. Both parents when screened for glycosuria were negative. There was no family history of Diabetes mellitus.

Discussion

The criteria(3) used for the diagnosis of renal glycosuria are (i) constant glucosuria
even in the fasting state, (ii) lack of significant influence of the amount of dietary carbohydrate, (iii) identification of glucose as the urinary sugar and, (iv) evidence that the patient metabolizes carbohydrate normally. If the above criteria are rigidly adhered, the condition is not common(1). Only 94 cases have observed among 50,000 cases of mellituria seen at the Joslin clinic. The condition is usually discovered in the investigation of an accidentally discovered asymptomatic glycosuria(2). Both the siblings of the present report were discovered like wise and confirmed as per the criteria.

Glucose concentration of the glomerular filtrate is determined by the glucose concentration of the plasma. As the concentration of glucose increases in the plasma, the concentration of glucose presented to the proximal tubules also increases till an amount greater than proximal tubule can reabsorb. This value is called maximum tubular reabsorption capacity for glucose (TmG). If all the nephrons are normal, glucose is reabsorbed completely until TmG is reached. This is plotted as a curve and depending upon the degree of splay of the curve from the normal, two types A and B of renal glycosurias are described. Type A is characterized by a low TmG and Type B an exaggerated splay and a normal TmG(4). Recently, a third, Type O renal glycosuria has also been described(5).

As the disease manifests with varying TmG in siblings and parents, the genetics is complicated and is explained by autosomal recessive trait and autosomal dominant mutation, siblings being homozygous for mutant gene and doubly heterozygous for two mutant genes. Various family descriptions and their genetics is well described in the monograms(1,4).

Due to the limitations in diagnostic facilities, the inheritance of the present family was not established but appears to be autosomal recessive trait with homozygous mutant genes in the affected sibling not expressing in the heterozygous parents. One such family has been reported in the literature(4).

Patients with renal glycosuria are asymptomatic, no treatment is indicated. It is important not to confuse the condition with diabetes mellitus(4). There is disagreement among experts as to whether renal glycosuria progresses over time to diabetes mellitus. Diabetes has been present in the families of patients with renal glycosuria and patients followed up to 70 years have been found to excrete glucose significantly without developing diabetes mellitus(3). A short term follow up of 2 years did not show diabetes in the present cases nor in the siblings or parents.

With the increasing awareness of diabetes mellitus in children even in the tropics, a case of glycosuria must be thoroughly investigated for diabetes mellitus and chronic renal disease before labelling as renal glycosuria which is rarely encountered in children.

REFERENCES


3. Marble A, Dan Ferguson B. Diagnosis and Classification of diabetes mellitus and the non diabetic meliturias. In: Joslin’s Diabetes Mellitus, 12th edn, Eds
Case Reports

Case 1: An 11-year-old boy presented with swelling in front of neck for 2 months and dysphagia for 2 weeks. He was clinically euthyroid. There was mild thyromegaly and a 2 × 2 cm firm non-tender nodule in the right lobe, moving side to side and with deglutition. A RAI (radioactive iodine) scan showed a cold nodule in the right lobe and aspiration cytology revealed only a few follicular cells. In view of the short history of dysphagia and a cold thyroid scan, thyroid malignancy could not be ruled out. A right hemithyroidectomy was done which histologically proved to be a benign thyroid adenoma.

Case 2: An 11-year-old girl presented with swelling in the front of neck for one month. She was clinically euthyroid.

The overlying skin on the thyroid was indurated, a solitary nodule 4 × 2 cm in size was present in the right lobe of the gland, firm, tender moving side to side with deglutition. There was no significant lymphadenopathy. Thyroid scan revealed a cold nodule in the right lobe and on aspiration cytology only a few follicular cells were found. In view of the clinical findings and the report of thyroid scan a provisional diagnosis of thyroid malignancy was made and the gland was explored. The tissues around the right lobe were indurated and on dividing the strap muscles whole of the right lobe was found to be replaced by thick pus. Right lobectomy was done and the histopathology showed evidence of chronic non-specific abscess with mild hyperplasia.

Discussion

Rare solitary thyroid nodules continue to present problems in diagnosis(4). It is