Diabetes Mellitus and Glucose Metabolism

DIABETES COMPLICATIONS II

Embarrassed to Eat: Two Cases of Gustatory Hyperhidrosis

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Introduction

Diabetic Gustatory Hyperhidrosis is characterized by profuse sweating with eating and may be a manifestation of Diabetic autonomic dysfunction. Most patients have evidence of other microvascular complications including nephropathy, retinopathy, peripheral neuropathy and other signs of autonomic neuropathy.

We present 2 cases of gustatory hyperhidrosis associated with longstanding poorly controlled type 1 diabetes.

Case 1: 49 year old Male with past medical history of longstanding type 1 diabetes with poor control, complicated with diabetic retinopathy, polyneuropathy, albuminuria presented to endocrine clinic for management of diabetes. His hemoglobin A1c was 10.8%. He was on basal-bolus Insulin at home. However, he admitted to missing most doses of prandial Insulin. On further questioning, he mentioned having episodes of profuse head and neck sweating while eating any type of food. He attributed these episodes to "low blood sugars" without checking and therefore tried to avoid Insulin. However, he continued having these episodes. He was diagnosed with Diabetic gustatory hyperhidrosis and started on topical Aluminum hexahydrate.

Case 2: 34 year old Female with past medical history of long-standing DM type 1 complicated with poly- neuropathy, autonomic dysfunction, nephropathy, Retinopathy, chronic kidney disease stage III presented for follow up of her diabetes. Her hemoglobin A1c was 9.8%. She was on basal-bolus Insulin at home and reported good compliance. Given her extensive polyneuropathy, she was questioned about hyperhidrosis. She reported having profuse facial and neck sweating with eating all types of food which led to increased embarrassment while eating in public. She was diagnosed with diabetic gustatory hyperhidrosis and started on topical aluminum hexahydrate, with plans for Botox if symptoms persisted.

Discussion

Diabetic Gustatory Hyperhidrosis is an under- recognized condition and may be misdiagnosed as hypoglycemia, anxiety, gastroparesis or other conditions. This gustatory sweating is a source of severe distress and embarrassment for patients and can have serious emotional, social and professional implications. Associated symptoms may also be mistaken for hypoglycemia and in turn lead to nonadherence with Insulin and other diabetic medications causing suboptimal glycemic control.

Topical anti-perspirants like Aluminum Chloride hexahydrate are often used as first line therapy. Second line treatment options include glycopyrrolate, Oxybutynin and Botulinum toxin.

Conclusion

Most patients are reluctant to mention these symptoms to health care providers and diligent history taking with specific questions in high risk patients may help in early identification and management of this condition.

Early identification and management can also help promote overall confidence, quality of life and better glycemic control.

Pediatric Endocrinology

SEXUAL AND GENDER DEVELOPMENT IN THE PEDIATRIC POPULATION

Novel Genes Involved in Sex Differentiation Identified by Whole-Exome Sequencing in a Cohort of Children with Disorders of Sex Development

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Background: Disorders of sex development (DSD) are classified as a congenital discrepancy between external genitalia, gonadal and chromosomal sex. Despite extensive laboratory and imaging investigations, the etiology of DSD is unknown in more than 50% of patients and the diagnosis is often delayed to the second decade of life. Our objective was to evaluate the etiology of DSD by whole-exome sequencing (WES) in children in whom hormonal and candidate gene approaches had not identified the etiology. **Methods**: Nine patients diagnosed with DSD (eight 46,XY and one 46,XX) were enrolled. Patients underwent hormonal evaluation, including ACTH, GnRH and hCG tests. Candidate genes were sequenced in accordance with the hormonal results. WES was performed for the probands and their parents. **Results:** The eight 46,XY patients presented with micropenis, cryptorchidism and hypospadias at birth and the 46,XX patient with fusion of the labia majora. In six of the nine patients (66%), a pathogenic mutation was identified by WES that explained the phenotype: four known DSD-causing genes—*POR*, *CHD7*, *HSD17B3* and *WT1*—and two novel genes—*BMP4* and *RFXP2*. In three patients, variants of unknown significance were found. An 11-y-old boy had a novel *de-novo* mutation in *BMP4*. In humans, mutations in this gene, encoding bone morphogenetic protein 4, are associated with autosomal dominant microphthalmia. BMP4 is expressed in the urethral epithelium and has a role in the development of external genitalia and the pituitary. This is the first report of a *BMP4* mutation in a child with DSD. A 12-y-old boy had a mutation in *RFXP2*, encoding insulin-like 3 hormone receptor, which has been previously reported in adult males with cryptorchidism. This is the first case of an *RFXP2* mutation in a child with DSD. **Conclusions:** WES has a crucial role in early diagnosis of the etiology of DSD, making extensive endocrine testing unnecessary, and has important implications for sex of rearing decisions.