Diagnostic Delay of Untreated Chronic Diabetes Insipidus and Rapidly Progressive Puberty in a 10-Year-Old

Pranshu Bhardwaj B.S.[1]; Rachel M. Coleman M.D.[2]; Marie L. Rivera-Zengotita M.D.[3]; John H. Rees M.D.[4]; Angelina V. Bernier M.D.[5]

[1]University of Florida College of Medicine, Gainesville, Florida

[2]Department of Pediatrics, University of Florida Health Shands Hospital, Gainesville, FL

[3]Department of Pathology, Immunology and Laboratory Medicine, University of Florida Health College of Medicine, Gainesville, FL

[4]Department of Radiology, Division of Neuroradiology University of Florida College of Medicine, Gainesville, FL;

[5]Department of Pediatric Endocrinology, University of Florida Health Shands Hospital, Gainesville, FL.

INTRODUCTION

A 10-year-old Hispanic male presented with a 5-year history of polydipsia and polyuria. He underwent evaluation in Venezuela, where diabetes insipidus (DI) was reportedly 'ruled out'; however, no head MRI was performed. After two years in the US struggling to acquire insurance, he presented to his pediatrician with worsening symptoms.

CASE PRESENTATION

A head MRI, ordered to evaluate dilute high-volume urine output, revealed a suprasellar mass extending superiorly into the hypothalamus. He was admitted for diagnostic evaluation and met the criteria for DI. Notably, he had an elevated β-Human Chorionic Gonadotropin (β-HCG) level. Biopsy confirmed the diagnosis of a Central nervous system (CNS) germinoma. He was treated with DDAVP and proton therapy with subsequent remission of his tumor.

DISCUSSION

Multiple medical and cultural factors led to various lengthy delays in care and diagnosis. The patient did not present with symptoms more typical of CNS Germinomas such as headaches, nausea, and vomiting. His increased stretched penis length and Tanner staging, which were identified later in his disease course, were contradicted by his pre-pubertal testicular volume and bone age. Poverty in Venezuela, lack of insurance, anxiety regarding COVID-19, and the language barrier also contributed to these delays. To our knowledge, this is the first case report of a pediatric patient presenting with a 5-year history of untreated polyuria and polydipsia due to undiagnosed DI with a β-HCG secreting CNS germinoma, without metastasis. This study highlights a rare presentation of DI and emphasizes the importance of supporting Spanish-speaking families as they navigate our complex healthcare system.