Adrenal

ADRENAL CASE REPORTS I

Long-Term Outcomes of Two Siblings with X-Linked Congenital Adrenal Hypoplasia Due to a Mutation in NR0B1 (DAX1) Gene: Reproductive and Neuropsychiatric Aspects

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Background: X-linked congenital adrenal hypoplasia (CAH) is a rare disease caused by mutations in the *NR0B1* (*DAX-1*) gene. Non-classical manifestations have been described, including late-onset adrenal insufficiency (AI) and gonadotropin-independent precocious puberty (GIPP). We report long-term endocrine and neuropsychiatric outcomes of two siblings with CAH due to mutation in *NR0B1*. ***Case report:*** A 2-yr-old boy was referred due to progressive clinical signs of puberty since 6 months of age. At the age of 3 yr, AI was diagnosed, and the molecular analysis revealed a mutation in the *NR0B1* (p.Cys65Leufs*6). Glucocorticoid replacement resulted in reduced testicular volume and decreased testosterone levels. At 11 yr, cyproterone acetate was indicated due to pubertal progression and bone age advancement. At 17 yr the patient had incomplete sexual development and no pubarche. Testosterone levels declined, despite pubertal levels of basal and GnRH-stimulated gonadotropin levels, indicating partial hypogonadotropic hypogonadism. Adult height was 156 cm (SDS: -2.7) within his target height of 161 cm (SDS: -2.1). This patient also presented a psychiatric diagnosis of mood disorder and attention-deficit/hyperactivity disorder (ADHD), and was under methylphenidate, topiramate and sertraline. Both the patient and his mother had SNP array performed, which excluded contiguous gene syndrome. His younger brother also harbored the same mutation in the *NR0B1,* confirmed shortly after birth. AI was diagnosed with 1 month of age. Cortisone acetate and fludrocortisone were initiated. At 11 months of age, he presented signs of pubertal development with an elevated ACTH and testosterone levels with suppressed gonadotropins, confirming the diagnosis of GIPP. He was treated with cyproterone acetate. At 8 yr, a pubertal response to the GnRH test was detected, and leuprorelin was added. At 9 yr, due to the low growth velocity and advanced bone age, rhGH was started. However, this patient presented a poor compliance and severe obesity (BMI 33 kg/m^2). Treatment for GIPP and secondary CPP was stopped at 10 yr, with bone age of 13.5 yr and height of 151 cm (SDS: - 2.3). The diagnosis of ADHD and autism spectrum disorder was made after neuropsychiatric assessment and the patient received treatment with methylphenidate and sertraline. ***Conclusion:*** Pubertal development of patients with CAH due to *NR0B1* mutations can be heterogeneous. However, the intriguing neuropsychiatric features in two siblings may suggest a role of *NR0B1* in neuropsychological development or other still unknown underlying genetic defect.

Diabetes Mellitus and Glucose Metabolism

CLINICAL AND TRANSLATIONAL GLUCOSE METABOLISM AND DIABETES

Improving Care Transitions: Optimizing Diabetes Medication Reconciliation

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Introduction

Medication reconciliation (MR) is essential in caring for hospitalized patients with diabetes (DM). Inaccuracies prevail, affecting transitions of care. Factors contributing to errors are driven by providers, patients, and systems of practice and increase in transitions out of hospital. A study showed that MR errors occurred in about 38 % of admissions. Common discrepancies among DM subjects were related to DM and cardiovascular drugs. An intervention aiming to reduce MR discrepancies at discharge achieved 70% less errors due to communication between inpatient providers, primary care and patients within 24 hours. Systems-based interventions and multidisciplinary approaches show promise to improve processes. However, a comprehensive assessment of the elements of practice to target and how interventions may be more effective is lacking. Our purpose was to examine aspects of practice among personnel responsible for the MR steps, and within the hospital workflow in order to identify gaps in the process. We intend to recognize factors to be targeted to optimize MR for DM, and to provide a multipronged approach to inform changes in hospital processes.

Methods:

We used quantitative and qualitative methods to investigate errors in the MR process as part of a hospital quality improvement program. We randomly included patients 18 years or older with type 1 or 2 DM evaluated by the endocrine team for 6 months. Chart reviews were conducted to assess type and frequency errors. Interviews of nurses, pharmacists, clinicians, and DM educators were sought to understand unique situations and the roles of health providers in the MR process.

Results

Twenty-two subjects were identified with one or more of the following gaps in their MR pertaining to DM medications: a) missing, b) redundancy, and c) dosing error. Scenarios included ≥2 discrepancies (4 of 22); ≥1 medication inappropriately changed from home regimen (18 of 22); redundantly adding a medication (4 of 22); wrong dose of medication (1 of 22); incomplete prescription for DM supplies (8 of 22).

Conclusion:

We identified deficits and their attribution to professionals and categorized errors in hospital workflows. Observations, providers' insights and literature review enabled an