More Than Meets the Eye: Insight into Cyclical Cushing Syndrome
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INTRODUCTION

Cushing Syndrome (CS) is rarely seen in children, and even less often seen in infancy. Less than 15 out of every million people are affected by CS every year, and only 10 percent of those new cases are in children. There are three main sources of hypercortisolism: the adrenal glands, the pituitary gland, and ectopic. The Clinical Center at the National Institutes of Health (NIH) is an international referral center for rare endocrine disorders. Many screening tools such as certain imaging scans and serial lab tests were developed at the NIH. We identified this topic as an educational need in the pediatric nursing population based on the rarity of the diagnosis and the experience that this family shared with us.

BACKGROUND

In February of 2003, S.E., a 4-year old female presented to our institution with a history of cyclical Cushing syndrome (CS) from birth. During the periods of hypercortisolism the parents noted such changes as rapid weight gain, moon facies, decreased energy and changes in behavior. Medical evaluation indicated that the patient also had elevated cortisol levels and blood pressures during these periods. Despite several outside evaluations, no definitive diagnosis was made, which increased the family’s anxiety and frustration.

At the time of admission, S.E. was asymptomatic, and presented as a normally developing child on no medications. Her height and weight were within the 50th percentile. The family was asked to return when the symptoms reappeared. Within one month S.E. returned with a dramatically different physical presentation including acne, plethora, distended abdomen, irritability and increased appetite. Her weight had increased to the 97th percentile.

DIFFERENTIAL DIAGNOSES

- McCune-Albright Syndrome
- Pituitary tumor
- PPNAD (pigmented polynodular adrenal disease)
- Menochromeic syndrome by proxy
- Prader-Willi Syndrome

TESTING DONE AT THE NIH

- Bone Age (chronological age 4 years, bone age 5 years)
- CT of adrenals – bilateral minimal nodularity
- Ultrasound of pelvis and thyroid – normal
- Echocardiogram of heart – normal
- Elevated 24 hour Urine Free Cortisol (UFC)
- Nodular adrenals by CT
- Failure to suppress serum cortisol to 8 mg dexa methasone (dose adjusted for weight)
- CT of adrenals – bilateral minimal nodularity
- MR1 chroidal abnormalities/polyploidy – normal
- Ultrasound of pelvis and thyroid – normal
- CRH, ACTH, 8mg Dexamethasone suppression test
- Various testing for thyroid, liver function and electrolytes

FINDINGS

The Clinical Center evaluation confirmed the diagnosis of Cushing syndrome as a result of:

- Abnormal diurnal cortisol levels
- Elevated 24 Hour Urine Free Cortisol (UFC)
- Failure to suppress serum cortisol to 8 mg dexamethasone (dose adjusted for weight)
- The diagnosis of micronodular adrenal disease was confirmed by the paradoxical response to the Liddle’s test and low plasma ACTH

S.E. underwent a bilateral adrenalectomy, which was performed without complications.

BACKGROUND

Located in the hypothalamus are several collections of cells that produce and release CRH. CRH stimulates the pituitary gland, which produces ACTH.

ACTH stimulates the synthesis and release of cortisol from the adrenal cortex.

Cortisol decreases the production of CRH and ACTH by a negative feedback mechanism.

S.E. underwent a bilateral adrenalectomy, which was performed without complications.

A Closer Look: S.E.’s Story

CONCLUSION/RECOMMENDATIONS

When the family arrived at our institution they exhibited anxiety regarding the previous denial of their concerns, lack of differential diagnosis, and the suggested diagnosis of Munchausen’s. The nursing staff served in a pivotal role in offering emotional support by listening to their concerns, advocating, providing information and directing them to resources.

FOLLOW-UP/EVALUATION

- Emotional support for the family during testing
- Providing continuity of care throughout their admissions
- Assisted the family in coping with chronic illness
- Discharge teaching
- IM injection teaching
- Sick Day Rules related to adrenal insufficiency

NURSING RESPONSIBILITIES

- Emotional support for the family during testing
- Providing continuity of care throughout their admissions
- Assisted the family in coping with chronic illness
- Discharge teaching
- IM injection teaching
- Sick Day Rules related to adrenal insufficiency
- Information regarding medic alert bracelet

Chart 1: Diurnal Cortisols

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Chart 2: Liddle’s Test

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<th>Urine 11OHS</th>
<th>Urine 18OHS</th>
<th>Urine 21OHS</th>
<th>Urine Creatinine</th>
<th>17OHS/11OHS</th>
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<tr>
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</tr>
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S.E.'s Story

October 9, 2003

S.E.'s 5th Birthday

S.E.’s 4th Birthday

S.E.’s 3rd Birthday

S.E.’s 2nd Birthday

S.E.’s 1st Birthday

S.E. 1/29/00

S.E. 11/20/02

S.E. 4/25/02

S.E. 5/30/99

S.E. 4/18/99

S.E.’s Road to Recovery

S.E. Fast Facts

S.E. Facts

S.E. Profile