

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 children¹. 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, moonfacies, 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 moonfacies, plethora, distended abdomen, irritability and increased appetite. Her weight had increased to the 97th percentile.

References:
Kell, M. (2004). Cushing's syndrome in children. Cushing's Support & Research Foundation.
http://csrf.net/AboutCushing_Pedia.htm

DIFFERENTIAL DIAGNOSES

- 👉 McCune-Albright Syndrome
- 👉 Pituitary tumor
- 👉 PPNAD (pigmented nodular adrenal disease)
- 👉 Munchausen's syndrome by proxy
- 👉 Prader-Willi Syndrome

February 24, 2003



April 1, 2003



TESTING DONE AT THE NIH

- 👉 Bone Age (chronological age 4 years, bone age 5 years)
- 👉 MRI chest/abdomen/pelvis/pituitary – normal
- 👉 Ultrasound of pelvis and thyroid – normal
- 👉 Echocardiogram of heart – normal
- 👉 DEXA Radius/Femur/Spine L1-L4 – significant osteopenia
- 👉 CT of adrenals – bilateral minimal nodularity
- 👉 Diurnal cortisols (See Chart 1)
- 👉 24 HR urine testing and Liddle Test (See Chart 2)
- 👉 CRH, ACTH, 8mg Dexamethasone suppression test
- 👉 Various testing for thyroid, liver function and electrolytes

Chart 1: Diurnal Cortisols

Date	PM Diurnal Cortisol	AM Diurnal Cortisol
Normal Values	< 5.0 ug/dl	< 20.0 ug/dl
2/18/03	7.5 ug/dl	11.2 ug/dl
4/02/03	31.45 ug/dl	29.85 ug/dl

PM Diurnals averaged from draws at 11:30 pm and 12 MN
AM Diurnals averaged from draws at 7:30 am and 8 am

Chart 2: Liddle's Test

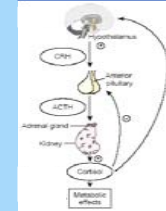
	Urine Creatinine (g/24 hr)	FPORS (mg/24hr)	Urine Free Cortisol (ug/3hr)	Volume of Urine (ml)
Day 1	0.24	1.6	6.6	344
Day 2	0.26	1.6	11.4	492
Day 3	0.23	1.6	9.9	365
Day 4	0.23	1.5	12.1	225
Day 5	0.20	2.8	25	269
Day 6	0.22	5.4	49.2	324
Day 7	0.21	4.8	53.8	225

The Liddle's test, or long dexamethasone suppression test, is used to evaluate adrenal gland function. Normal responses to Liddle's test a patient without CS suppresses on both low and high dose dexamethasone, a patient with Cushing's disease suppresses on high dose, and in a patient with adrenal or ectopic involvement usually no suppression is detected. The response for this patient was characteristic of bilateral micronodular adrenal disease.

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)
 - Nodular adrenals by CT
 - 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.

HPA Axis



- 👉 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.

FOLLOW-UP/EVALUATION

At the 6 month follow up visit the patient's weight had returned to the 50th percentile with no signs of central obesity. Routine endocrine follow-up is indicated to monitor whether this diagnosis represents an isolated finding or if it is part of Carney's Complex (an inherited, autosomal dominant multiple neoplasia syndrome).

October 9, 2003



NURSING RESPONSIBILITIES

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

CONCLUSION/RECOMMENDATIONS

When the family arrived at our institution they exhibited anxiety regarding the previous dismissal 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.

A Closer Look: S.E.'s Story



4/16/99



5/30/99



1/23/00



4/23/01



6/2/01



9/10/02



3/2/02



4/25/02



11/20/02



11/23/02



S.E.'s 4th Birthday



S.E.'s 5th Birthday

