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Cushing Syndrome: Establishing a Timely Diagnosis

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A 12-year-old Hispanic female presents to the local emergency room with seizures and severe hypertension. Over the past year, she gained 15 kilograms, with no changes in her appetite or level of physical activity, yet she stopped growing in height. Her BMI increased from 21.9 to 29 kg/m². She was recently diagnosed with type 2 diabetes mellitus and was treated with metformin and diet modification. Her mother noticed that she was more impatient and irritable in the last 4–5 months. Significant findings on her physical exam included severe acne (face, chest, and back); acanthosis nigricans; hirsutism; wide purple striae on legs, abdomen, trunk; facial plethora; fat pads in supraclavicular, temporal, and dorsocervical areas; scattered petechiae on arms and shins; proximal muscle weakness; tinea versicolor; candida albicans; Tanner V pubic hair and Tanner II breast development. Her blood pressure remained > 90% for age-height, even with treatment with three antihypertensive medications.

Alarmed by the severity of her hypertension along with concomitant development of rapid onset weight gain, diabetes, and numerous physical changes in a young girl, the emergency room doctor suspected Cushing syndrome and referred the child to a pediatric endocrinologist. Following is a brief overview of Cushing Syndrome and the integral role of the pediatric nurse in the care of these children.

Background

Cushing syndrome (CS) is a multisystem disorder resulting from the prolonged exposure to excess glucocorticoids (cortisol). CS is characterized by truncal obesity, impaired linear growth, skin changes (acne, acanthosis nigricans, hirsutism, facial plethora, easy bruising, wide purple striae), delayed sexual development, amenorrhea, hypertension, glucose

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Links to Educational Resources

- Adrenal Insufficiency (http://www.cc.nih.gov/ccc/patient_education/pepubs/mngadrins.pdf)
- National Institute of Child Health and Human Development (<http://www.nichd.nih.gov/health/topics/cushing/Pages/default.aspx>)
- Cushing Support and Research Foundation (<http://csrf.net/understanding-cushings/pediatric-cushings/>)
- The Magic Foundation (www.MagicFoundation.org)

intolerance, headaches, osteopenia or osteoporosis, and fatigue. The most common presentation in children is stalled linear growth with persistent weight gain (Magiakou et al., 1994) (Figure 1). Parents who become concerned about their child's weight gain commonly find websites that describe the symptoms of CS, which frequently overlap with other causes of weight gain. Most cases of CS result from the exogenous administration of glucocorticoids (GC).

CS in children is rare. An estimated 10 to 15 of every million people are affected per year, and only about ten percent of these new cases occur in children (Stratakis, 2012). For prepubertal children, adrenal tumors (e.g., adenoma, carcinoma, or bilateral hyperplasia) are a more common cause of CS. In older children, pituitary adenomas are more common (Magiakou et al., 1994).

Evaluation

Symptoms of CS may be insidious. Weight gain is the most common presenting symptom, and lack of concomitant height gain is typically seen in children who develop CS. Pubertal children may present with virilization and adolescent females with concerns about menstrual irregularity (Batista, Riar, Keil, & Stratakis, 2007; Keil & Stratakis, 2013). A careful review of growth data is imperative in the evaluation of any child or adolescent presenting with a concern about weight gain and growth delay. It is important to review the medication history to ascertain any steroid use, including topical steroid compounds, which could affect test results. It is also important to recognize that over the counter and complementary medications may contain steroids. Common illnesses, such as asthma and Crohn's disease, may be treated with steroids, which could lead to symptoms of CS.

Upon suspicion of CS, it is important to document hypercortisolism. The gold standard screening test for hypercortisolism is 24-hour urinary free cortisol excretion (corrected for body surface area). However, a 24-hour urine collection is often difficult to obtain correctly in an outpatient setting, particularly in the pediatric population. Due to the circadian nature of cortisol production, random measurements are not helpful in the diagnosis, but diurnal salivary cortisol measurements may be useful in the outpatient setting (Batista et al., 2007; Keil, 2012; Stratakis, 2012). Once hypercortisolism is documented the child should be referred to an endocrinologist or specialty center with expertise in CS diagnosis. Of course, any child or adolescent with unexplained growth delay with or without weight gain warrants an endocrine evaluation.

Treatment

Surgical resection is the recommended treatment for benign adrenal tumor(s) and ectopic adrenocorticotrophic hormone-producing tumors (Magiakou et al., 1994).

Nursing Considerations

After successful surgical excision of adrenal or pituitary adenoma, there is a period of adrenal insufficiency while the hypothalamic-pituitary-adrenal axis recovers function. During this period, replacement with glucocorticoid (GC) (physiologic dose of 10-12

mg/m²/day) and “sick-day” stress dosing should be maintained. Patients who undergo bilateral adrenalectomy require lifetime replacement with GC and mineralocorticoid (fludrocortisone), as well as stress dosing for acute illness, trauma, or surgical procedures (Lodish, Dunn, Sinaii, Keil, & Stratakis, 2012). (See Links to Resources below).

The nurse has a crucial role in the initial and ongoing education of affected children, parents, and caregivers about the signs and symptoms of adrenal insufficiency, stress dosing with acute illness or trauma or surgery (“sick day rules”), intramuscular injection of hydrocortisone, and communication with school and other health care providers (i.e., medical alert) (VanRyzin, 2013). Challenges in caring for the child or adolescent treated for CS include optimizing growth and pubertal development, normalizing body composition, and promoting psychological health and cognitive maturation. Normalization of body composition in children and adolescents post-cure of CS may not occur; therefore, strategies to improve diet and exercise regimens are important to discuss with the child and family (Keil, 2013).

It is well established that CS is debilitating physically, mentally, and emotionally (Dorn et al., 1995). A prospective study of quality of life in children with CS reported that despite improvement from pre- to 1-year post-cure, residual impairment persisted in several quality of life measures (Keil et al., 2009). It is important for the pediatric nurse to assess whether the child is engaged in age-appropriate activities and identify areas of concern (physical, social, cognitive, emotional) and refer to resources as appropriate (Keil et al., 2009).

Conclusion

Pediatric nurses and other healthcare providers should have a high index of suspicion of possible CS in children who present with obesity, growth retardation, skin problems, history of fractures, and behavior changes. Although CS is rare, it can have long-term deleterious effects on the body and brain; therefore, early diagnosis and treatment is important. The nurse is an essential team player in the identification, evaluation, treatment, and follow-up of children affected by CS. This includes obtaining accurate measurements, plotting on growth charts, assisting with the accurate specimen collection, and assessing appropriate implementation of “sick day rules.” The pediatric nurse has an integral role in providing support and education; future research is needed to enhance quality of life and improve patient outcomes.

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