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THE EVALUATION OF THE CHILD WITH SHORT STATURE

WHAT DOES A CHILD NEED TO GROW?

- food (money)
- hormones
- good genes
- a good start (intrauterine factors)
- absence of disease, infection, stress
- love

IMPORTANT GROWTH FACTORS

- prenatal
  - insulin
  - prolactin, IGF-1 and IGF-2
- postnatal
  - growth hormone, IGF-1
  - T3
- puberty
  - gonadal hormones

GROWTH CHARTS

- WHO 2006/2007
- CDC/NCHS 2000
- intrauterine
- standard deviation
- Tanner longitudinal
- growth velocity
- syndrome-specific

GROWTH AXIOMS

- Five percent of all *normal* children will be growing on or below the fifth centile.
- It's far less important *where you are* on the growth chart, and far more important *how you got there*.

HISTORY

- gestational age, birth weight and length: ? very premature, ?SGA/IUGR
- evidence of systemic disease, malabsorption
- failure to thrive *vs* growth failure
- pattern of growth: early *vs* late fall-off

FAMILY HISTORY

- size, weight of parents, siblings
- growth patterns of parents, siblings (age of menarche, when stopped growing)
- other “late bloomers”?

MIDPARENTAL HEIGHT

- is a rough estimate (± 10 cm) of genetic height potential:
    - BOYS: [father’s height + mother’s height + 13 cm]/2
    - GIRLS: [father’s height + mother’s height − 13 cm]/2
- (13 cm ≅ 5")

WEIGHT

- short and obese:
    - hormone deficiency
    - syndrome
- short and thin:
    - malnutrition
    - malabsorption
    - systemic disease
    - constitutional delay
- tall and obese:
    - exogenous obesity
- plotting weight-for-height or BMI often diagnostic!

GROWTH VELOCITY

- single most useful and reliable indicator to rule out an endocrinopathy affecting growth:
    - 0–6 mo 32 cm/yr (12"/yr)
    - 6–12 mo 16 cm/yr (6"/yr)
    - 1–2 yr 10 cm/yr (4"/yr)
    - 2–4 yr 7–8 cm/yr (2.5–3"/yr)
    - 4–10 yr 4.5–6 cm/yr (2–2.5"/yr)
- growth along a given centile is good evidence that the growth velocity is normal

PHYSICAL EXAMINATION

- accurate measurement of length/height, weight, head circumference
- arm span and sitting height or upper-to-lower segment ratio or sitting height
- obvious and subtle dysmorphologies
- evidence of systemic disease
- evidence of neglect, abuse
- dental eruption
- goiter, genitalia

LABORATORY EVALUATION

- CBC, differential, indices
- ESR
- electrolytes, BUN, creatinine, glucose
- LFT, Ca/Mg/P_i, protein/albumin
- urinalysis (check pH)
- bone age (left hand/wrist)
- free T4 and/or TSH
- IGF-1 and/or IGFBP-3
- [karyotype]

GENERALLY UNHELPFUL TESTS

- morning and/or afternoon cortisols
- lateral skull X-ray
- random growth hormone levels
- testosterone and/or estradiol in prepubertal children
- cranial MRI or CT without other evidence of endocrine or CNS disease

SYSTEMIC DISEASES PRESENTING AS SHORT STATURE

- poorly controlled diabetes
- cystic fibrosis
- celiac disease
- inflammatory bowel disease
- renal disease
- malnutrition
- psychosocial dwarfism/abuse/neglect
- chronic infection
- metabolic diseases

SYNDROMES ASSOCIATED WITH SHORT STATURE

- Turner syndrome (45,X and others)
- mixed gonadal dysgenesis (45,X / 46,XY)
- Noonan syndrome (PTPN11)
- Down syndrome (trisomy 21)
- Prader–Willi syndrome (paternal 15q deletion)
- Albright hereditary osteodystrophy (pseudo- and pseudopseudohypoparathyroidism)
- IUGR-related syndromes (*eg* Russell-Silver syndrome, UPD maternal 7)
- numerous forms of skeletal dysplasias (*eg* hypo- and achondroplasia)

ENDOCRINE CAUSES OF SHORT STATURE

- growth hormone deficiency
- hypothyroidism
- Cushing syndrome
- precocious puberty

CONSTITUTIONAL DELAY OF GROWTH AND ADOLESCENCE

- “late bloomer”
- exaggerated variation of normal physiology
- short stature ± delayed puberty
- normal growth velocity
- delayed bone age
- often thin
- more common in boys
- may have considerable psychosocial burden
- normal hormone studies
- may benefit from exogenous gonadal steroids

GROWTH HORMONE DEFICIENCY

- short stature ± delayed puberty
- obesity
- low growth velocity
- delayed bone age
- hypoglycemia in infancy
- microphallus in boys
- midfacial hypoplasia
- associated midline problems
- ± other pituitary deficiencies
- low IGF-I, low IGFBP-3, low maximal stimulated GH

ACQUIRED HYPOTHYROIDISM

- usually Hashimoto (autoimmune) thyroiditis
- short stature ± delayed puberty
- obesity
- low growth velocity
- delayed bone age ± epiphyseal stippling
- ± goiter
- decreased energy, increased sleep
- dry skin, constipation, cold intolerance
- low free T4, elevated TSH
- positive thyroid microsomal/peroxidase antibodies

CUSHING SYNDROME

- usually pituitary ACTH-secreting microadenoma
- short stature
- truncal obesity
- low growth velocity
- delayed bone age
- ± adrenarchal signs (acne, pubic hair)
- buffalo hump, purple striae, moon facies
- hypertension
- elevated 24-h urine free cortisol
- failed overnight dexamethasone suppression test