X. Metabolic disorders

A. Diagnosis

1. General

Know when genetic counseling is appropriate for the family of a child with a metabolic disease 2009;30:131, e22
2007;28:363

Recognize the signs and symptoms of inborn errors of carbohydrate metabolism 2009;30:131, e22

Recognize the signs and symptoms of hyperinsulinism 2009;30:131, e22

Recognize lipoprotein disorders 2009;30:131, e22

Recognize Gaucher disease and lipid storage disease 2009;30:131, e22

Recognize urea cycle defects 2009;30:131, e22
2008;29:355

Recognize glycogen storage disease 2009;30:131, e22
2006;27:107

Recognize the signs and symptoms of Tay-Sachs disease 2009;30:131, e22

Know how to evaluate patients with suspected metabolic disease who are comatose 2009;30:131, e22
2008;29:355

Know how to evaluate patients with suspected metabolic disease who are hypoglycemic 2009;30:131, e22
2009:167

Know how to evaluate patients with suspected metabolic disease who are acidic 2009;30:357
2009;30:131, e22
2009:183

Recognize the clinical features of disorders of fatty acid and carnitine metabolism 2009;30:131, e22

Recognize the clinical findings of a disorder of amino acid metabolism 2009;30:131, e22
2010:199

Recognize the signs and symptoms of the mucopolysaccharidoses, including Hurler syndrome 2008:72

B. Treatment

1. General

2. Diet

3. Medication

Know how to manage carbohydrate metabolic disorders (eg, glycogen storage diseases type I) 2009:39

Know how to treat hypoglycemia 2009;30:217

4. Prognosis and long-term care

Be familiar with the long-term prognosis and chronic management of Tay-Sachs disease 2011:231
XI. Endocrine disorders

A. Sex differentiation

1. Normal development

2. Ambiguous genitalia

Recognize the signs and symptoms of congenital adrenal hyperplasia

Know the laboratory evaluation of congenital adrenal hyperplasia

Know that congenital adrenal hyperplasia can be diagnosed prenatally

Plan the treatment for an adrenal crisis in a patient with congenital adrenal hyperplasia

Understand the value of neonatal screening for salt-losing congenital adrenal hyperplasia in male infants with normal genitalia

Understand that maternal exposure to androgens or progestins can cause virilization in female infants

B. Growth

1. Short stature

Know the most common causes of short stature

Distinguish among constitutional short stature, genetic (familial) short stature, and growth hormone or thyroid deficiencies by growth chart evaluation

Know how to distinguish between familial short stature and other conditions

Know the natural history of familial short stature

Know how to use laboratory tests effectively to distinguish between constitutional growth delay and other conditions

Know the natural history of constitutional growth delay

Recognize the signs and symptoms of acquired and congenital growth hormone deficiency (eg, micropenis)

2. Tall stature

Differentiate among the causes of tall stature

C. Puberty

1. Normal

Distinguish between the variations of normal (eg, thelarche, pubarche) and precocious puberty

Know the pathophysiology and differentiating features of normal
vs abnormal gynecomastia in males

Understand the significance of a breast mass in an adolescent girl 2011:196

2. Precocious puberty

Recognize the signs and symptoms of precocious puberty 2007;28:419 2008:122
2006;27:373

Know the differential diagnosis of precocious puberty 2007;28:419 2008:122
2006;27:373

Know that premature thelarche occurs without other signs of puberty, is most common among those 1 to 4 years of age, and often regresses spontaneously 2006;27:373 2011:138

Recognize the importance of obtaining the history of medication use, including phytoestrogens and estrogen-based creams, when evaluating a child with premature breast development 2006;27:373 2011:138

Recognize the tumors that may produce precocious puberty (eg, in liver, CNS, ovary, testes, adrenal glands) 2006;27:373 2009:218

Know how to use laboratory tests effectively to distinguish an adrenal etiology of precocious puberty 2008:10

Recognize that testosterone creams used by parents can cause virilization in male or female children 2010:122

3. Delayed puberty

Recognize the signs and symptoms of delayed puberty 2007:42

Recognize the signs and symptoms of gonadal dysgenesis (Turner syndrome) 2006;27:351

Know the laboratory evaluation of gonadal dysgenesis (Turner syndrome), including karyotype, and serum concentrations of luteinizing hormone, follicle-stimulating hormone, and estradiol) 2009:10

Understand the importance of evaluating for cardiac and renal disorders in gonadal dysgenesis (Turner syndrome) 2008;29:219

Understand the familial influences on the onset of puberty 2010:106

Know how to interpret the growth chart and bone age x-ray when evaluating constitutional delayed puberty 2007:10

Know the natural history of constitutional delayed puberty 2007:10

Know when treatment for constitutional delayed puberty is indicated and understand the therapeutic options 2007:26

D. Thyroid disorders

1. Hashimoto thyroiditis

Know the laboratory studies that distinguish among Hashimoto thyroiditis, other causes of thyroid enlargement, and hypothyroidism 2008:26

Know the natural history of Hashimoto thyroiditis 2008:26
Know that Hashimoto thyroiditis is the most common cause of goiter in adolescents

Know that Hashimoto thyroiditis may be associated with other autoimmune disorders

2. Cyst, tumor, nodule

Understand the importance of referral in a child with a thyroid mass/nodule

Know the significance of a previous history of irradiation to the head and neck in a patient with a thyroid mass/nodule

Know that a solitary thyroid nodule may be a sign of thyroid cancer

Recognize a thyroglossal duct cyst

3. Hypothyroidism

Know the consequences of untreated hypothyroidism in the neonate

Recognize the signs and symptoms of congenital and acquired hypothyroidism

Know the varying causes of congenital and acquired hypothyroidism

Know how to manage and treat congenital and acquired hypothyroidism and the use of thyroid-stimulating hormone to guide treatment

Know the prognosis for a patient with congenital or acquired hypothyroidism

Know how to recognize thyroid-binding globulin (TBG) deficiency

4. Hyperthyroidism

Recognize the signs and symptoms of hyperthyroidism

Know how to use history, physical examination, and laboratory tests effectively to diagnose hyperthyroidism

Be aware of various modalities for treatment of hyperthyroidism

Recognize the signs and symptoms of neonatal hyperthyroidism

E. Parathyroid disorders

Recognize the typical laboratory findings associated with hypoparathyroidism

Know that DiGeorge syndrome (22q-) can be a cause of hypoparathyroidism
F. Adrenal gland disorders

1. General

2. Addison disease
   - Recognize the signs and symptoms of Addison disease 2009;30:447 2008:169
   - Know how to use laboratory tests effectively for the diagnosis of Addison disease 2009;30:447 2010:138
   - Plan the treatment of an adrenal crisis in a patient with Addison disease 2009;30:447
   - Recognize that Addison disease is usually an autoimmune disorder 2009;30:491 2010:234
   - Recognize the signs and symptoms of adrenal insufficiency after discontinuation of exogenous corticosteroid therapy 2009;30:447 2008:214

3. Cushing syndrome
   - Recognize the signs and symptoms of Cushing syndrome 2009:58

G. Pituitary gland disorders

- Recognize the clinical manifestations of hypopituitarism 2011:42
- Recognize the clinical manifestations of craniopharyngioma 2011:42

H. Diabetes

1. General
   - Recognize the signs and symptoms of type 1 diabetes 2008;29:374 2010:154
   - Know how to treat type 1 diabetes effectively to achieve good control: insulin, diet, exercise, and psychologic acceptance of the disease 2008;29:374
   - Know the natural history of type 1 diabetes (eg, "honeymoon" period) 2008;29:374
   - Counsel patients on the self-management of type 1 diabetes (hyperglycemia, sick days) 2008;29:374
   - Know how to manage sick days in diabetic patients 2008;29:374 2011:186
   - Know the long-term complications of type 1 diabetes 2008;29:374 2008:184
   - Know the importance of blood glucose control in the prevention of long-term complications of type 1 diabetes 2008;29:374 2009:106
   - Recognize the association between type 1 diabetes and other autoimmune disorders, including celiac disease 2008;29:374 2008:199
   - Recognize that ketotic hypoglycemia is the most common type of hypoglycemia presenting in early childhood 2011:58
   - Recognize the typical presentation of ketotic hypoglycemia in a young child 2011:58

Unit 4: Diabetes, Metabolic Syndrome, Disorders of PTH, Calcium and Phosphate Metabolism
2. **Diabetic ketoacidosis**

Know the complications of type 1 diabetes, particularly diabetic ketoacidosis and its pathophysiology, treatment, and complications (hypokalemia, hypoglycemia, cerebral edema, shock)  

Recognize cerebral edema as a complication of the treatment of diabetic ketoacidosis  

Understand the risks of using bicarbonate in diabetic ketoacidosis  

Know that noncompliance is a major cause of recurrent diabetic ketoacidosis  

Plan the management of a child who has mild to moderate diabetic ketoacidosis

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3. **Type 2 diabetes**

Understand the difference between type 1 diabetes and type 2 diabetes  

Know that acanthosis nigricans is a marker for insulin resistance  

Plan appropriate screening tests for type 2 diabetes  

Recognize the long-term complications of type 2 diabetes  

Recognize that complications of type 2 diabetes may be present at diagnosis

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I. **Metabolic syndrome**

Plan appropriate screening tests for metabolic syndrome  

Identify the risk factors that necessitate screening tests for metabolic syndrome  

Plan appropriate initial management of a patient with metabolic syndrome (eg, lifestyle modification with diet and physical activity)  

Understand the significance of metabolic syndrome (cardiovascular risk factors)

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J. **Disorders of PTH, calcium, and phosphate metabolism**

1. **Hypocalcemia**

Recognize the signs and symptoms of hypocalcemia  

Know the causes of hypocalcemia in a neonate  

Know that hypocalcemia with hypophosphatemia suggests vitamin D deficiency

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2. **Hypercalcemia**

Recognize the signs and symptoms of hypercalcemia  

Recognize the possibility of hypercalcemia and its complications following prolonged immobilization
3. Hypophosphatemia

Recognize the typical clinical and laboratory findings associated with familial hypophosphatemic rickets 2008:58

Plan the treatment of a child with familial hypophosphatemic rickets 2011:106