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IAP Textbook on

PEDIATRIC ENDOCRINOLOGY



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CHAPTER

Clinical Pointers in Endocrine Disorders that may be Easily Overlooked by General Pediatricians

Yeshwant Krishna Amdekar

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General pediatricians face a challenge as they are expected to know "bit" of everything while specialists may know everything of a "bit," albeit it is impossible to know everything even about a "bit." Generalist has to find out which system and which part of the system are affected in a given patient while specialist starts with an advantage of knowing the system involved. Hence, generalist must achieve competence in picking up clinical pointers related to common disorders in each system, which in turn helps timely referral to a specialist. It is possible only with detailed history and thorough physical examination. In fact, detailed history mostly offers clue to a probable diagnosis, and focused physical examination helps to arrive at most likely diagnosis. Experience makes it possible even in a short time. This article focuses on clinical pointers in endocrine disorders.

CLINICAL POINTERS IN NEONATES

Maternal prenatal history of hypo- or hyperthyroidism, diabetes mellitus, and vitamin D or iodine deficiency should alert need for relevant action at birth with appropriate blood tests. In fact, routine neonatal screening helps to diagnose hypothyroidism and congenital adrenal hyperplasia (CAH) at birth before clinical symptoms appear. Neonate may present with *lethargy and refusal of feeds* simulating sepsis; and if subtle clinical pointers, such as *delayed passage of meconium, prolonged jaundice, hypotonia, and bradycardia*, are missed, one may overlook severe—athyrotic congenital hypothyroidism. Examination of genitals is often overlooked in neonate. *Hypospadias with undescended testes* may represent disorder of sexual development. *Ambiguous genitalia* is obvious at birth and needs caution to counsel parents before announcing sex of the baby. *Seizure* may be a manifestation of transient short-lasting hypoglycemia or hypocalcemia in neonates, which often present within first 2–3 days after birth. However, when seizure occurs after 5–7 days of life and is difficult to control, it may represent endocrine disorder. Hypopituitarism may be suspected in the presence of *micropenis and hypoglycemia, especially if there is a midline defect*. Hyperinsulinism may be another disorder presenting with hypoglycemia. Similarly, late onset of persistent hypocalcemia may be due to hypoparathyroidism. Neonate born with intrauterine growth restriction may be due to many causes. If such a neonate presents with *failure to thrive* and *tachypnea*, neonatal diabetes may be considered. *Polyuria* is easily overlooked at this age. During subsequent 2–4 weeks, neonate presenting with *poor feeding, vomiting, and failure to thrive* is typical of CAH, often missed as gastrointestinal infection. Female neonates with CAH are easier to detect due to ambiguous genitalia.

CLINICAL POINTERS BEYOND NEONATAL AGE

Growth Chart—Most Important Clinical Tool

Tracking growth parameters on standard growth chart is the best clinical tool in diagnosis of endocrine disorders. Faltering growth curve is an early clinical pointer to many endocrine disorders that present with *short or tall stature* and/or obesity. In fact, it is an excellent screening tool to suspect cause of the disorder that can be further confirmed by specific laboratory tests. In addition to tracking weight and length/height, *growth velocity* is an important

parameter. Early deviation in height is better picked up by change in growth velocity, even before height curve falters. *Weight for height* is another clinical parameter that differentiates short stature due to endocrine disorder from that due to other chronic diseases, including malnutrition. *Proportionate or disproportionate short stature* would separate hypothyroidism from growth hormone deficiency along with the involvement of mental faculty, abnormal facies, and other clues in the case of hypothyroidism. One must also look for *short-limb or short-trunk disproportion* and be cautious to differentiate between hypothyroidism from other bone disorders, including rickets. *Tall stature* may also be a manifestation of endocrine disorder, though not so common. Excess of growth hormone presents with tall stature along with syndromic disorders and genetic or constitutional factors. Sudden upward deviation of height curve may point out to adolescent phase, and if it occurs early, it would suggest precocious puberty. *Obesity* may be due to endocrine disorder. Short and obese must always be considered to be due to endocrine problem while tall and obese is mostly an exogenous obesity. Cushing's syndrome, hypothyroidism, and hypothalamic disorders are examples of endocrine obesity. Presence of *hypertension* in Cushing's and typical features of hypothyroidism are clinical pointers to specific diagnosis in obese child. Hypothalamic disorder may also present with other symptoms, such as *abnormal sleep rhythm, high fever not responding to antipyretics*, and *mood changes*. In the case of hypothalamic tumor, symptoms and signs of raised intracranial pressure as well as vision abnormalities help to diagnose the condition. Growth tracking helps to pick up early upward deviation in weight centiles much before child becomes overweight and thereafter obese. Many babies born small tend to show extreme upward trend in weight centiles and run risk of *syndrome X* in young adulthood. Thus early deviation in the trend of growth centiles should be addressed promptly.

In addition to evaluating probable endocrine disorder, general pediatricians must recognize the importance of growth chart that helps them a lot even in routine practice. Growth chart is useful to define onset, duration, and progress of every disease, to differentiate between worsening and improving disease, recurrent and persistent disease and to assess severity of disease. Thus every pediatrician must maintain growth chart of every patient, irrespective of type of disease.

Precocious puberty is considered when sexual characteristics are seen in girls before the age of 8 years and in boys before the age of 9 years. However, any physical change due to sex hormone effect, such as *premature thelarche* or *pubarche*, needs careful assessment. *Delayed puberty* is considered in the absence of any sexual characteristics beyond 13 years of age in girls and beyond 14 years in boys, especially in the urban setting. *Change in sequence of development of sexual characteristics* and presence of heterosexual precocity is definitely abnormal and demands proper evaluation. Young girls may present with menstruation with other abnormalities such as *café-au-let spots* and *bone fracture* that suggest McCune–Albright syndrome. Precocious puberty may also be a manifestation of hypothyroidism. *Hematuria* in a girl may be mistaken for renal disease when it actually represents menstruation. Short-lasting and self-limiting hematuria reported in young girls presenting every month is then diagnosed as precocious puberty. Hence, *sexual maturity* must be assessed normally in every child at appropriate age and earlier if need be. *Breast development* in girls and *testicular size* in boys must be monitored along with the development of *pubic hair*. As mentioned above, sudden increase in height and body structure may suggest puberty.

Constipation is extremely common symptom in general population. However, stubborn constipation not responding to usual measures should arouse suspicion of hypothyroidism, even in the absence of other features. Similarly, *mild developmental delay* and *subnormal school performance, lethargy,* and constipation may be the only clues to a missed mild congenital hypothyroidism, while children with juvenile hypothyroidism, which generally occurs beyond 3 years of age, are academically very good because they prefer to sit and study rather than play. It being a treatable condition, even isolated symptom demands evaluation of thyroid function. *Thyromegaly*, if any, is obvious and certainly demands evaluation of thyroid function. *Asthenia*—generalized weakness—may be due to many diseases, but one may easily overlook diabetes mellitus, adrenal insufficiency, or parathyroid disorders. *Hyperpigmentation* offers clue to Addison's disease. *Polyuria* is often missed in the history as focus is often on oliguria and so also polyphagia and polydipsia are not evident unless specially enquired. Diabetic child may present with *tachypnea* due to ketoacidosis and *change in sensorium* and *dehydration*. Diabetes insipidus also presents with polyuria and polydipsia. *Irritability* of undefined origin may be due to hyperthyroidism or hypernatremia in diabetes insipidus. Vague *muscle aches* and *tingling-numbness and renal stones* may be a feature of parathyroid disorders that can be easily missed. *Abdominal pain* is such a common symptom in children and so also *vomiting*. While many common diseases present with such symptoms, diabetes, Addison's disease, and hyperparathyroidism may be overlooked. *Diarrhea* may be rare manifestation of endocrine disorder, such as in vipoma or hyperthyroidism or autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy. *Fever* may rarely be due to endocrine disorders, such as hyperthyroidism or hypothalamic disease. *Hypertension*

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may be due to pheochromocytoma, rare forms of CAH and Cushing's syndrome.

Endocrine disorder may also present as life-threatening emergency and should be kept in mind. *Coma* in diabetes mellitus may the first presenting feature of diabetes when earlier subtle symptoms are often missed. Sudden *shock* may be due to acute adrenal failure due to severe infection or hemorrhage. Thyroid crisis may present with *cardiac failure*.

In summary, specific clues to endocrine disorders are easily picked up by growth charts, and hence, it should be routine for every pediatrician to monitor growth. However, many common symptoms presenting in children may also be due to endocrine disorders. While pediatricians must think of common causes of common symptoms first, when confronted with difficulty in diagnosis, uncommon causes of common symptoms should also include endocrine disorders.

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