CONTENTS

ARTICLES

267 What the General Pediatrician Should Know About Developmental Anomalies
John M. Opitz

273 Male Pseudohermaphroditism
Donna J. Levy, Lenore S. Levine, and Maria I. New

285 Narcotic Withdrawal Syndrome in the Newborn
Avron Y. Sweet

293 The Multiple Forms of Neurofibromatosis
Vincent M. Riccardi

ABSTRACTS

271 Tuberculosis Screening

272 Congenital Adrenal Hyperplasia

272 Routine Urinalysis Questioned

283 Bladder Paralysis with Myelomeningocele

284 Dysmenorrhea

284 Zinc Deficiency

291 Groups in Well Child Care

292 Small Bowel Resection

292 Pseudoephilepsy

298 Rotavirus Diarrhea

299 Self-Assessment


Discussion of some of the questions follows.

(1) Incompletely differentiated external genitalia in an XY patient is due to inadequate male hormones at the cellular level. Müllerian inhibiting factor prevents the development of the uterus, fallopian tubes, and upper vagina.

(2) In the complete form of testicular feminization syndrome, the patient has female characteristics, despite the XY chromosome constellation, because of the resistance to testosterone at the cellular level. The gonad is a testis; although the external genitalia is female, no virilization occurs, and adrenal hormones are normal. There is no uterus, thus amenorrhea, and the testicles may present as an inguinal mass.

(5) The infant with fetal alcohol syndrome is also fretful and small. The symptoms of cerebral palsy are usually apparent only after 6 months of age.

(7) A child with seven cafe au lait spots (CLS) should be suspected of having neurofibromatosis. A base line evaluation should include a slit-lamp examination for Liisch nodules, auditory neurofibromas, cranial tumors (EEG), and pheochromocytomas (catecholamines). CLS are usually sufficiently characteristic that biopsy is not warranted.

(8) Periodic monitoring should include the evaluation of height, head size, presence of scoliosis, and school performance. Neurofibromatosis patients are not especially prone to dental problems or hand-eye incoordination.

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