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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. Mullerian 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, in spite of the XY chromosome constitution, 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 testes 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 examination should include a slit-lamp examination for Lisch 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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