structures is ultrasonography, a noninvasive, painless imaging technique that does not require anesthesia or sedation. It is especially useful in CAH because it readily identifies the absence or presence of female reproductive organs or male testes in a newborn or child with ambiguous genitalia. Because ultrasonography yields immediate results, it has the advantage of determining the child's gender long before the more complex laboratory results for chromosome analysis or steroid levels are available.

Therapeutic Management

After diagnosis is confirmed, medical management includes administration of glucocorticoids to suppress the abnormally high secretions of ACTH and adrenal androgens. If cortisone is begun early enough, it is very effective. Cortisone depresses the secretion of ACTH by the anterior pituitary, which in turn inhibits the secretion of adrenocorticosteroids, which stems the progressive virilization. The signs and symptoms of masculinization in girls gradually disappear, and excessive early linear growth is slowed. Puberty occurs normally at the appropriate age.

The recommended oral dosage is divided to simulate the normal diurnal pattern of ACTH secretion. Because these children are unable to produce cortisol in response to stress, it is necessary to increase the dosage during episodes of infection, fever, surgery, or other stresses. Acute emergencies require immediate IV or intramuscular administration. Children with the salt-losing type of CAH require aldosterone replacement, as outlined under chronic adrenal insufficiency, and supplementary dietary salt. Frequent laboratory tests are conducted to assess the effects on electrolytes, hormonal profiles, and renin levels. The frequency of testing is individualized to the child.

Gender assignment and surgical intervention in the newborn with ambiguous genitalia is complex and controversial. It is a significant stress for families, who need support from a multidisciplinary team of experienced specialists. Factors that influence gender assignment include genetic diagnosis, genital appearance, surgical options, fertility, and family and cultural preferences. Generally, genetically female (46XX) infants should be raised as girls. Early reconstructive surgery should be considered only in the case of severe virilization (Lee, Houk, Ahmed, et al,