

PRENATAL TREATMENT AND DIAGNOSIS OF CONGENITAL ADRENAL HYPERPLASIA OWING TO 21-HYDROXYLASE DEFICIENCY

S. Nimkarn, M.I. New

Department of Pediatrics, Mount Sinai School of Medicine, New York, NY, USA

Congenital adrenal hyperplasia is a group of inherited disorders caused by an enzyme deficiency in steroid biosynthesis. The most common form of congenital adrenal hyperplasia is 21-hydroxylase deficiency, which in its severe form can cause genital ambiguity in females. Steroid 21-hydroxylase deficiency can be diagnosed in utero through molecular genetic analysis of fetal DNA. Prenatal treatment successfully reduces genital ambiguity, and the subsequent problems of sex misassignment and gender confusion. Data from current studies show that prenatal diagnosis and treatment are safe for the mother and the fetus. The evidence also suggests that it is safe over the long-term, but all subjects exposed to dexamethasone treatment during embryonic and fetal life should have their physical, cognitive and emotional developments recorded.