

Prenatal Diagnosis of Congenital Adrenal Hyperplasia

The use of a combination of DNA probes for polymorphisms both within the 21-hydroxylase gene and in and around the closely linked HLA region permits reliable prenatal diagnosis of 21-hydroxylase deficiency in more than 95% of families at risk for having a child with congenital adrenal hyperplasia. Chorionic villus sampling during the first trimester permits early detection of the defect, and also provides the option of using intrauterine therapy in families where there is severe salt loss and significant masculinization of females.

Since masculinization is likely to have occurred prior to the prenatal diagnostic procedure, the current

recommendation is to treat the mother with dexamethasone as soon as the first menstrual cycle is missed. Treatment is continued until the results of chorionic villus sampling determine whether the fetus is male or female and whether it is affected or not. If the fetus is female and affected, treatment is continued throughout pregnancy.

Dreno B, Meignier M, Bignon JD, et al. *Lancet* 1987;ii:1272-1273.

Editor's comment—*Although many families will opt for prenatal treatment of 21-hydroxylase deficiency, others will opt for termination of affected pregnancies. The new linkage techniques allow accurate detection and, therefore, offer more options to family and physicians.*

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