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Congenital Adrenal Hyperplasia

Congenital adrenal hyperplasia was once considered a rare inherited disorder with severe manifestations. Mild congenital adrenal hyperplasia, however, is common, affecting up to 1% of all women in the United States.

Both classic and nonclassic forms of the disease are caused by deficiencies in the adrenal enzymes that are used to synthesize glucocorticoids. The net result is increased production from the adrenal gland of cortisol precursors and androgens. Even mild congenital adrenal hyperplasia can result in life-threatening sinus or pulmonary infections, orthostatic syncope, shortened stature and severe acne.

Women with mild congenital adrenal hyperplasia often present with excess hair growth (hirsutism), irregular & skipped periods (oligomenorrhea) or infertility. Congenital adrenal hyperplasia is diagnosed by demonstration of excess cortisol precursors in the blood, especially after a test injection of the drug ACTH. Diagnosis of congenital adrenal hyperplasia in an unborn child can be made with special testing after amniocentesis.

Treatment includes carefully monitored hormone replacement therapy. Recognition of the problem and timely replacement therapy can reduce problems and enhance quality of life in patients that are affected by congenital adrenal hyperplasia.