Congenital Adrenal Hyperplasia (CAH) is a family of inherited disorders affecting the adrenal glands. 21-hydroxylase deficiency is the most common form of CAH. The disorder is divided into classical (severe) form of CAH (further divided into salt-wasting and simple-virilizing types) and the mild form known as non-classical CAH.

Today babies born with classical 21-hydroxylase deficiency are diagnosed soon after birth on the newborn screen. All 50 states screen for the disorder. Early diagnosis is important since children with the salt-wasting form of the disease can become ill soon after birth because their bodies cannot hold on to sodium or salt, which can lead to vomiting, dehydration, lethargy and even death. In addition, the level of potassium can become very high, which can lead to disturbances in the heart rhythm. Girls born with classical CAH exhibit abnormalities of the genitalia and are therefore quickly brought to medical attention. Boys, however, do not exhibit any overt symptoms at birth, which is why the newborn screen is so crucial in identifying baby boys affected with classical CAH.

Immediately after a baby is diagnosed with classical CAH, a medication called hydrocortisone is given. It is a small pill that can be crushed easily and can be mixed with a small amount of water, breast milk or formula. Giving this life-long treatment to the baby is not difficult. Those children who are affected with salt-wasting CAH are also given salt-supplementation in the form of Sodium Chloride, as well as fludrocortisone, a medication that helps the body hold on to salt. Patients with salt-wasting CAH are deficient in a chemical called aldosterone, which is responsible for the sodium and potassium balance in our body; fludrocortisone and salt supplementation restore that balance. Sodium Chloride and fludrocortisone are easy to give to the baby as well.

Your child should be monitored very closely, especially in infancy. Medication doses are adjusted based on laboratory results as well as your child’s growth pattern.

We know it may feel overwhelming that your child was diagnosed CAH. Our team is here to help your child and your family every step of the way.

- Our team of physicians will see your child frequently in the beginning, and eventually the visits will take place every 3 months. We are available 24 hours a day, 7 days a week should an urgent or emergent situation arise. Our number is 212-746-3975.
- Our surgical team is one of the leading teams in the world specializing in genital reconstructive surgery. To make an appointment with a surgeon, please call 212-746-5337.
- Our psychologist, with years of experience with patients and families living with chronic illness, will provide ongoing psychological support to families taking care of children with CAH.