### Why a Parent’s Handbook?

![Image of a mother and child]

Why a Parent’s Handbook?

<table>
<thead>
<tr>
<th>What Matters Most to Patients?</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>DEATH</strong></td>
</tr>
<tr>
<td>If untimely</td>
</tr>
<tr>
<td><strong>DISEASE</strong></td>
</tr>
<tr>
<td>Symptoms, Signs, Derangements</td>
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<tr>
<td><strong>DISCOMFORT</strong></td>
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<tr>
<td>Pain, Nausea, Fatigue, Itching, etc.</td>
</tr>
<tr>
<td><strong>DISABILITY</strong></td>
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<tr>
<td>Impaired Daily Function</td>
</tr>
<tr>
<td><strong>DISSATISFACTION</strong></td>
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<tr>
<td>Sadness, Anger, Depression</td>
</tr>
<tr>
<td><strong>DESTITUTION</strong></td>
</tr>
<tr>
<td>Financial Burden of Illness</td>
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</tbody>
</table>
WHAT IS CONGENITAL ADRENAL HYPERPLASIA (CAH)?

**Congenital** – present at birth

**Adrenal** – involving the adrenal glands

**Hyperplasia** – results in overgrown glands
<table>
<thead>
<tr>
<th>CHROMOSOME</th>
<th>GENE NAME</th>
<th>ENZYME AFFECTED</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>HSD3B3</td>
<td>3-Beta Hydroxysteroid Dehydrogenase</td>
</tr>
<tr>
<td>6</td>
<td>CYP21</td>
<td>21-Hydroxylase</td>
</tr>
<tr>
<td>8</td>
<td>CYP11B1</td>
<td>11-Beta Hydroxylase</td>
</tr>
</tbody>
</table>

LOCATION OF CAH GENES
SYMPTOMS OF CAH
Treatment is based on the individual and the type of CAH they have. Treatment includes…

- Glucocorticoid Treatment
- Mineralocorticoid Treatment
- Stress Dosing
The Beginning....

- The Clinic for Special Children
- PBH course
- Family Day
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**Ambiguous genitalia:**
External genitals that do not appear to be clearly male or female, have not fully formed, or appear to be of the opposite sex of the internal sexual organs.
FOR THE SUMMER....
• 6 very different drafts to edit
• Review
• Amish Home Visits
• Design Elements
• Medical Reviews
• Editorial Review
Normal Adrenal Feedback Loop

- ACTH
- Cortisol
- Aldosterone
- Androstenedione
- Testosterone
- ACTH
- Cortisol
- Androgens

Cortisol:
- Sufficient to "turn off" pituitary ACTH production

Androgens:
- * Testes only start producing androgens when puberty begins.*
Frequently Asked Questions

Is newborn screening available for CAH?

Yes. newborn screening is available to help detect CAH, although it may miss some of the more rare variants. Keep in mind, this test only indicates that there may be a problem. A second diagnostic test is needed to confirm if the child has CAH. For more information on newborn screening, refer to Chapter 9.

Is genetic testing available for CAH?

Yes. genetic testing is available to determine the specific gene error in those with CAH. Genetic testing can also determine an individual’s carrier status, which can be useful for family planning. Prenatal testing is also available. Please refer to Chapter 8 for more details.

How common is CAH?

CAH affects about 1 in 15,000 babies born in the United States. However, the condition is more common in certain areas or populations. For instance, in the Ashkenazi community in the United States, more children are born with CAH.

Are boys or girls more likely to have CAH?

Both males and females have an equal chance of developing CAH. The odds of having a child with CAH are based on whether or not the parents are carriers. It has nothing to do with the sex of the child.

What is the Endocrine System?

The endocrine system is a group of glands and tissues that regulate diverse processes such as growth, development, and response to environmental stress. In individuals with CAH, the endocrine system does not work properly, resulting in the hormonal imbalance that causes the symptoms of CAH.

Glands are groups of cells that produce and secrete chemical messengers known as hormones. Hormones travel to target cells, which then respond to the hormone signal with specific changes in activity. Although different hormones circulate throughout the bloodstream, each one affects only the cells that are able to receive and respond to its message. The same hormone may impact various cells in different ways.

The major glands that make up the human endocrine system are the pituitary, thyroid, parathyroid, pancreas, adrenal, and reproductive glands, which include the ovary in females and the testes in males.

What is the Adrenal Cortex?

Located atop the kidneys, the body has two cone-shaped adrenal glands (Figure 1.3). Each gland has two parts. The outer part, called the adrenal cortex, produces steroid hormones, hormones made from the conversion of cholesterol. Corticosteroids such as mineralocorticoids, glucocorticoids, and androgens are all termed steroid hormones. The principle mineralocorticoid is aldosterone, which regulates the mineral or salt levels in the body. Glucocorticoids, such as cortisol, control the availability of glucose as fuel for the body and regulate the body’s overall response to physical stress such as starvation, dehydration, infection, and trauma. Androgens such as testosterone play an important part in the sexual development of boys and girls.

Hormones are the chemical messengers of the body produced and secreted by glands and received by target organs.
THE END-PRODUCT?

A handbook that serve as a simplified guide for parents with children who have CAH

How Will I Recognize the Symptoms of the Condition?

Symptoms of CAH in Infants

Symptoms of CAH in Older Children
Thank You!!!