Adrenal hyperplasia congenital pdf

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Congenital adrenal hyperplasia is group of inherited conditions that are present at birth (congenital) where the adrenal gland is larger than usual (hyperplasia). This page from Great Ormond Street Hospital (GOSH) explains about the medical condition congenital adrenal hyperplasia (CAH) and what to expect when your child comes to GOSH for assessment and treatment.In CAH, the body is missing an enzyme (chemical substance) that stimulates the adrenal glands to release the cortisol hormone. Lacking this hormone means that the body is less able to cope with physiological stress, which can be life threatening. It also makes the level of androgen (male hormone) increase, which causes male characteristics to appear early in boys or inappropriately in girls.The adrenal glands rest on the tops of the kidneys. They are part of the endocrine system, which organises the release of hormones within the body. Hormones are chemical messengers that switch on and off processes within the body. The adrenal glands consist of two parts:the medulla (inner section) which makes the hormone ‘adrenaline’ which is part of the ‘fight or flight’ response a person has when stressed. This is not usually affected in CAH.the cortex (outer section) which releases several hormones. The most important ones are: Aldosterone – this helps regulate the blood pressure by controlling how much salt is retained in the body.Cortisol – this is the body’s natural steroid and has three main functions: helping to control the blood sugar level helping the body deal with stress helping to control blood pressure and blood circulation. Androgen – male hormone produced by both males and females. CAH is an inherited disorder, that is, it is passed on from parent to child. Most types of CAH are autosomal recessive disorders; this means that both parents are carriers of the disease. Human beings have about 20,000 to 30,000 different genes, each of which has a function in making an individual person. The genes are arranged in pairs (one of the pair from each parent) on 23 chromosomes. Inevitably, some of these genes are faulty; a normal gene can overcome a faulty one, but if both genes in the pair are faulty, the genetic instructions cannot work.Most people carry different faulty genes but in CAH (and other recessive conditions) parents, though healthy themselves, carry the same faulty genes, and risk passing them on to their children. Each pregnancy carries:a 25 per cent chance of the child being affecteda 75 per cent chance of the child not being affected.Although the chance of the child being affected is 1 in 4 with each pregnancy, it is not unusual for consecutive children to be affected.In a small proportion of people, CAH is caused by a gene mutation (change) that happens by chance and cannot be predicted. A number of genes have been identified as causing different types of CAH – for instance, the most common form of CAH is called 21-hydroxylase deficiency and results from the gene labelled CYP21 being absent or changed. This means that aldosterone and cortisol are not produced but production of androgen is unaffected. The gene affected results in different types of CAH, affecting people to varying degrees from mild to severe.CAH affects boys and girls in equal numbers, affecting between one in 10,000 and one in 18,000 children born each year. The symptoms that present and the age at which they become apparent depends on the degree of cortisol and/or aldosterone deficiency. Boys with severe CAH show symptoms soon after birth, when a baby develops heart rhythm abnormalities, dehydration and vomiting. The levels of minerals (electrolytes) in the body are also affected, particularly showing low salt levels (hyponatraemia) and low blood sugar levels (hypoglycaemia).Females with severe CAH are born with virilised (male looking) genitalia, so confusion as to the sex of the baby may occur. This is referred to as atypical or ambiguous genitalia. They too have low levels of minerals, especially salt, so show similar symptoms to boys.Children with mild CAH may not have the same mineral imbalances as more severely affected children. Later, other symptoms caused by the increase in androgen develop. Girls will have abnormal or absent periods, a deep voice, early puberty and facial hair. Boys will have an enlarged penis, small testicles, early puberty, acne and a deep voice.Both girls and boys may appear tall for their age but usually end up being short as adults if the diagnosis is not made quickly and treatment commenced. Most cases of CAH are diagnosed soon after birth or in early childhood, but less severe CAH may not become apparent until a child does not start puberty at the expected age. CAH can be diagnosed before birth now that the affected gene has been identified.If a baby is born with ambiguous genitalia, blood tests are usually used to determine genetic gender. Blood and urine tests are also used to work out the levels of minerals and hormones in the body, so that they can be replaced. X-rays to look at a child’s bone age may also be suggested. Genetic testing may be used to identify the specific type of CAH and the associated gene deficiency or mutation. Children with CAH are usually cared for by a multidisciplinary team including endocrinologists (hormone specialists) and urologists (genitourinary system specialists). Initially, children with CAH will need to be stabilised with intravenous fluids to restore their electrolyte levels. Once stable, cortisol and/or aldosterone replacement therapy can start with repeated blood tests to monitor hormone levels so the most effective dose can be prescribed.Children with CAH will need to take replacement cortisol and aldosterone every day for the rest of their lives. However, if a person with cortisol deficiency becomes unwell, they are unable to increase the production of cortisol in their system to help the body cope and this could be life threatening – further details are available in our Cortisol deficiency and steroid replacement therapy information sheet.If precocious puberty (puberty starting before the expected age) occurs, further hormone treatment may be required. Long term follow up monitoring is essential for children with CAH. They will need regular blood tests to monitor hormone levels in the body and adjustments of steroid replacement doses. Men usually have normal fertility provided they have been optimally treated, but some women with CAH may have problems conceiving so may require additional help to have children. Once pregnant, women with CAH should carry the baby to term but may require a caesarean section to deliver. As CAH is an inherited disorder, genetic counselling may be helpful. The Endocrinology department in collaboration with the Child and Family Information Group Congenital adrenal hyperplasia (CAH) describes a group of genetic disorders affecting your adrenal glands. You have one adrenal gland on top of each kidney. The adrenal glands produce several important hormones your body needs. In CAH, genetic mutations (changes) cause shortages of certain enzymes, such as 21-hydroxylase, that help your body produce needed hormones. For many people with CAH, the enzyme shortage causes the adrenal glands to overproduce other hormones, like testosterone. There are two main types of CAH: classic CAH and nonclassic CAH. The symptoms depend on the type of CAH you have. There are much more rare forms of CAH with different symptoms. Who is most at risk for getting congenital adrenal hyperplasia (CAH)? Congenital means a condition you are born with. All forms of CAH are genetic, or inherited. To develop CAH, you must inherit certain genetic mutations from each of your parents. How common is congenital adrenal hyperplasia (CAH)? Classic CAH, the most common form of the disease, affects as many as 1 in every 10,000 to 15,000 people living in the U.S. and Europe. Both classic and nonclassic CAH affect people worldwide. Specific genetic mutations cause CAH. These genetic changes happen during a baby’s development in the womb. People living with CAH have all inherited mutated genes from their parents. What are the symptoms of congenital adrenal hyperplasia (CAH)? The symptoms of CAH may appear differently depending on factors including your age at diagnosis, and whether you are male or female. Each type of CAH has different symptoms. Classic CAH For people living with classic CAH, symptoms occur due to a lack of the enzyme 21-hydroxylase. Doctors group classic CAH into 2 subcategories: salt-wasting CAH and simple virilizing (non-salt-wasting) CAH. Salt-wasting CAH is the most severe form of this disease. People living with salt-wasting CAH have adrenal glands that don’t produce enough aldosterone, a hormone that helps regulate sodium (salt) levels in the body. Low levels of aldosterone result in too much sodium being lost from the body in urine. The hormonal imbalances caused by salt-wasting CAH result in severe symptoms, including: Dehydration Hyponatremia (low blood sodium levels) Cardiovascular problems, including low blood pressure and arrhythmias (heart rhythm problems) Vomiting Low blood glucose (blood sugar) Metabolic acidosis (too much acid content in the blood) Shock Additionally, people living with salt-wasting CAH have high levels of androgens, or male sex hormones, in their bodies. Elevated androgen levels cause other symptoms, including: Enlarged or ambiguous (abnormal) external genitalia—for example, clitoris enlargement in a female infant Rapid growth Premature signs of puberty, including voice changes, early pubic, armpit, and facial hair, and severe acne Abnormal menstrual periods Noncancerous testicular tumors Infertility Simple virilizing (non-salt-wasting) CAH causes a less-severe aldosterone deficiency. Androgen levels are also elevated in this type of CAH. Symptoms of simple virilizing CAH may include: Ambiguous (unusual) external genitalia, particularly in females Development of male characteristics, such as bulky muscles and a deep voice, in females or prematurely in boys Nonclassic CAH People with nonclassic CAH also have too little 21-hydroxylase enzyme activity, but this deficiency isn’t as severe as in other forms of the disease. You may be unaware you have this form of CAH because your symptoms are more mild. Children, teenagers and adults are all diagnosed with nonclassic CAH. The symptoms of nonclassic CAH include: Early signs of puberty, including acne and excess facial or body hair in females Rapid growth in childhood and as a teenager Irregular menstrual periods Male-pattern baldness Early puberty changes in a boy, for example early pubic hair and an enlarged penis but with small testicles Infertility In the U.S., doctors screen all newborn babies for 21-hydroxylase deficiency before they leave the hospital. This screening determines whether a child has classic CAH. In almost all cases, doctors diagnose classic CAH in infants and young children. Diagnosis of nonclassic CAH may not occur until a person begins to show symptoms of the disease. In some cases, this may not happen until early adulthood. Some couples at higher risk, including those with family members diagnosed with CAH, may have genetic testing to determine the risk of passing on genetic mutations to their children. Treatment for congenital adrenal hyperplasia depends on the type of CAH you have and how severe your symptoms are. There is no cure for CAH, but many people find symptom relief with medications. Doctors prescribe several types of medications, including salt supplements and steroids, to people living with classic CAH. Your doctor monitors your condition with regular blood tests to check hormone levels. If classic CAH causes genital abnormalities, surgery two to six months after birth can correct the appearance of external genitalia. In some cases, it may be appropriate to delay surgery for several years. People living with nonclassic CAH who have very mild or no symptoms may not need treatment. For some, medications like steroids or fertility drugs improve symptoms. What complications are associated with congenital adrenal hyperplasia (CAH)? Children living with classic CAH lose too much water and salt in their urine. They are at risk for serious complications, including imbalances of electrolytes like potassium. Left untreated, these imbalances lead to other problems, like heart arrhythmias (irregular heartbeat), cardiac arrest, and death. Untreated nonclassic CAH can lead to early puberty and short stature in a male, and permanent male type body changes in a female child, or irregular periods and infertility in a young woman. Surgery to correct ambiguous genitalia carries its own risks for complications, including infection, bleeding and scarring. There is no way to prevent congenital adrenal hyperplasia. With treatment, most people living with congenital adrenal hyperplasia lead healthy, productive lives. If you become pregnant, ask your doctor about genetic testing for CAH. Your infant is tested for specific enzyme deficiencies before leaving the hospital. If you or an older child begins to experience symptoms of CAH, contact your doctor. Last reviewed by a Cleveland Clinic medical professional on 10/16/2018. References Genetic and Rare Disease Information Center. Congenital adrenal hyperplasia. (Accessed 10/17/2018. National Organization for Rare Disorders. Congenital Adrenal Hyperplasia. (Accessed 10/17/2018. National Institute of Child Health and Human Development. Congenital Adrenal Hyperplasia (CAH). (Accessed 10/17/2018. National Institute of Child Health and Human Development. 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