GTPIM105

Congenital adrenal hyperplasia (CAH)

Patient Information Material

What is Congenital adrenal hyperplasia?

Congenital adrenal hyperplasia (CAH) is a genetic disorder of the adrenal glands that affects the body's general health, growth, and development.

What causes CAH?

CAH is caused by impaired production of cortisol and aldosterome hormones due to deficiency of 21-hydroxylase enzyme in adrenal gland.

What is the incidence of CAH?

The worldwide incidence is estimated to be 1 in 15,000.

What are the symptoms of CAH?

Salt imbalance, tiredness, nausea, weight loss, abnormal growth, dysfunction in blood pressure and brain, learning disabilities, virilisation in girls, precocious puberty in boys, short stature are common.

What are the classic and non-classic forms of CAH?

A classic form with severe enzyme deficiency and prenatal onset of virilization is distinguished from a non-classic form with mild enzyme deficiency and postnatal onset.

How is CAH screened or diagnosed in lab?

Increased 17-OHP, a precursor steroid is indicative of CAH. Genetic test is highly confirmatory.

What are the symptoms in a new born?

Newborns with CAH may exhibit symptoms peaking at 3 weeks of age as poor feeding, vomiting, diarrhea, weak cry and failure to thrive. Sometimes symptoms may not be evident until adrenal crisis. If untreated, circulatory system can collapse leading to death in severe forms.

Is it required to screen every new born for congenital adrenal hyperplasia?

Yes, because clinical symptoms may not always be identified at birth.

How does Neonatal screening help?

1. Screening helps pre-symptomatic early diagnosis
2. Prevents life threatening adrenal crisis
3. Prevents virilisation (maleness) in females
4. Prevents short stature & psychosexual disturbances

What is the sampling procedure for screening?

3-4 blood drops of new born are collected on filter paper by heel prick method. Although screening can be done within the first 1 month of age, it is most sensitive when done between 2-5 days of age.

How is CAH confirmed?

A DNA test "Gene Sequencing for CAH" confirms or rules out congenital adrenal hyperplasia. The test looks for mutations on CyP21A2 gene.

How is CAH inherited?

CAH is an autosomal recessive disorder caused by mutations of CYP21 gene. That is two copies of mutated gene is required for the expression of disease. Most of the parents of affected children are unaffected carriers of mutated gene each.

What is the recurrence rate?

If parents are carriers of the genetic mutation recurrence risk is 25% for subsequent pregnancies. DNA tests are available to confirm carrier status.

Is prenatal diagnosis available?

Yes. Gene sequencing of CYP21A2 on fetal sample retrieved through prenatal sampling procedures like amniocentesis and chorionic villus sampling can diagnose or rule out CAH. However, it is important to have diagnosis of genetic mutation of the affected family member.

How is CAH treated?

Treatment for CAH involves cortisol replacement therapy to restore normal energy, glucose and electrolyte concentrations and fluid balance. In verilised female infants surgical intervention is required before 1yr of age. . In case of female fetuses diagnosed prenatally with CAH, prenatal therapy by dexamethasone may be considered.

How can a Genetic Counseling help?

A genetic counselor is a supportive professional who works with individuals and families to review genetic testing and diagnosis, review inheritance patterns, help identify possible risk in the family, discuss the symptoms and features of genetic disorders and suggest appropriate reproductive options. Due to the complexity of congenital adrenal hyperplasia diagnosis and inheritance, counseling is essential.

GeneTech is a pioneer in medical genetics testing in the country and offers the most comprehensive testing and screening services for genetic disorders in India

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