Geneticheck - Genetic Report

Gene

CYP

Associated Diseases

Adrenal Insufficiency, Congenital, With 46xy Sex Reversal, Partial Or Complete
Inherited Isolated Adrenal Insufficiency Due To Partial Cyp11a1 Deficiency
46,xy Disorder Of Sex Development-adrenal Insufficiency Due To Cyp11a1 Deficiency

Phenotype

Feeding difficulties
Impaired ability to eat related to problems gathering food and getting ready to suck, chew, or swallow it.

Decreased circulating androgen concentration
A reduction in the blood concentration of an androgen, that is, of a steroid hormone that controls development and maintenance of masculine characteristics. The androgens include testosterone and Dehydroepiandrosterone.

Hypovolemia
An decrease in the amount of intravascular fluid, particularly in the volume of the circulating blood.

Adrenal calcification
Calcification within the adrenal glands.

Premature birth
The birth of a baby of less than 37 weeks of gestational age.

Cryptorchidism
Testis in inguinal canal. That is, absence of one or both testes from the scrotum owing to failure of the testis or testes to descend through the inguinal canal to the scrotum.

Hyperaldosteronism
Overproduction of the mineralocorticoid aldosterone by the adrenal cortex.

Adrenal insufficiency
Insufficient production of steroid hormones (primarily cortisol) by the adrenal glands.

Urogenital sinus anomaly
A rare birth defect in women where the urethra and vagina both open into a common channel.

Adrenal hypoplasia
Developmental hypoplasia of the adrenal glands.

Failure to thrive
Failure to thrive (FTT) refers to a child whose physical growth is substantially below the norm.

Abnormal vagina morphology

Any structural abnormality of the vagina.

Decreased circulating cortisol level
Abnormally reduced concentration of cortisol in the blood.

Low maternal serum estriol
An abnormally high concentration of serum conjugated estriol as compared to normal values for gestational-age.

Delayed puberty
Passing the age when puberty normally occurs with no physical or hormonal signs of the onset of puberty.

Abnormal sex determination
Anomaly of primary or secondary sexual development or characteristics.

Renal salt wasting
A high concentration of one or more electrolytes in the urine in the presence of low serum concentrations of the electrolyte(s).

Clitoral hypertrophy
Hypertrophy of the clitoris.

Increased circulating ACTH level
An abnormal increased in the concentration of corticotropin, also known as adrenocorticotropic hormone (ACTH), in the blood.

Female external genitalia in individual with 46,XY karyotype
The presence of female external genitalia in a person with a male karyotype.

Hyperkalemia
An abnormally increased potassium concentration in the blood.

Elevated circulating luteinizing hormone level
An elevated concentration of luteinizing hormone in the blood.

Hypotension
Low Blood Pressure, vascular hypotension.

Increased circulating renin level
An increased level of renin in the blood.

Induced vaginal delivery
Vaginal delivery following induction of labor, a procedure used to stimulate uterine contractions during pregnancy before labor begins on its own.

Generalized hyperpigmentation

Vomiting

Forceful ejection of the contents of the stomach through the mouth by means of a series of involuntary spasmic contractions.

Adrenocorticotropic hormone excess
Overproduction of adrenocorticotropic hormone (ACTH), which generally leads secondarily to overproduction of cortisol by the adrenal cortex.

Abnormal urine potassium concentration
An abnormal concentration of potassium(1+) in the urine.

Aplasia of the uterus
Aplasia of the uterus.

Delayed skeletal maturation
A decreased rate of skeletal maturation. Delayed skeletal maturation can be diagnosed on the basis of an estimation of the bone age from radiographs of specific bones in the human body.

Decreased circulating aldosterone level
Abnormally reduced levels of aldosterone.

Reduced bone mineral density
A reduction of bone mineral density, that is, of the amount of matter per cubic centimeter of bones.

Acidosis
Abnormal acid accumulation or depletion of base.

Agenesis of corpus callosum
Absence of the corpus callosum as a result of the failure of the corpus callosum to develop, which can be the result of a failure in any one of the multiple steps of callosal development including cellular proliferation and migration, axonal growth or glial patterning at the midline.

Decreased testicular size
Reduced volume of the testicle (the male gonad).

Ambiguous genitalia, male
Ambiguous genitalia in an individual with XY genetic gender.

Osteoporosis
Osteoporosis is a systemic skeletal disease characterized by low bone density and microarchitectural deterioration of bone tissue with a consequent increase in bone fragility. According to the WHO criteria, osteoporosis is defined as a BMD that lies 2.5 standard deviations or more below the average value for young healthy adults (a T-score below -2.5 SD).

Hyponatremia
An abnormally decreased sodium concentration in the blood.

Abnormal circulating cholesterol concentration
Any deviation from the normal concentration of cholesterol in the blood circulation.

Generalized bronze hyperpigmentation

Hyperpigmentation of the skin
A darkening of the skin related to an increase in melanin production and deposition.

Sex reversal
Development of the reproductive system is inconsistent with the chromosomal sex.

Gynecomastia
Abnormal development of large mammary glands in males resulting in breast enlargement.

Neonatal hypoglycemia

Decreased fertility

Elevated circulating follicle stimulating hormone level
An elevated concentration of follicle-stimulating hormone in the blood.

Abnormality of prenatal development or birth
An abnormality of the fetus or the birth of the fetus, excluding structural abnormalities.

Absence of secondary sex characteristics
No secondary sexual characteristics are present at puberty.

Abnormality of the Leydig cells

Male pseudohermaphroditism
Hermaphroditism refers to a discrepancy between the morphology of the gonads and that of the external genitalia. In male pseudohermaphroditism, the genotype is male (XY) and the external genitalia are imcompletely virilized, ambiguous, or complete female. If gonads are present, they are testes.

Hypernatriuria
An increased concentration of sodium(1+) in the urine.

Primary adrenal insufficiency
Insufficient production of steroid hormones (primarily cortisol) by the adrenal glands as a result of a primary defect in the glands themselves.

Dehydration

Midshaft hypospadias
Hypospadias with location of the urethral meatus in the middle of the inferior shaft of the penis.