Geneticheck - Genetic Report

Gene

DHCR7

Associated Diseases

Smith-lemli-opitz Syndrome
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Phenotype

Gingival overgrowth
Hyperplasia of the gingiva (that is, a thickening of the soft tissue overlying the alveolar ridge. The degree of thickening ranges from involvement of the interdental papillae alone to gingival overgrowth covering the entire tooth crown.

Autism
Autism is a neurodevelopmental disorder characterized by impaired social interaction and communication, and by restricted and repetitive behavior. Autism begins in childhood. It is marked by the presence of markedly abnormal or impaired development in social interaction and communication and a markedly restricted repertoire of activity and interest. Manifestations of the disorder vary greatly depending on the developmental level and chronological age of the individual (DSM-IV).

Advanced eruption of teeth
Premature tooth eruption, which can be defined as tooth eruption more than 2 SD earlier than the mean eruption age.

Short toe
A toe that appears disproportionately short compared to the foot.

Low-set, posteriorly rotated ears
Ears that are low-set (HP:0000369) and posteriorly rotated (HP:0000358).

Self-mutilation

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.

Renal hypoplasia
Hypoplasia of the kidney.

Aplasia/Hypoplasia of the radius
A small/hypoplastic or absent/aplastic radius.

Aggressive behavior
Aggressive behavior can denote verbal aggression, physical aggression against objects, physical aggression against people, and may also include aggression towards oneself.

Severe photosensitivity
A severe degree of photosensitivity of the skin.

Atrioventricular canal defect
A defect of the atrioventricular septum of the heart.

Ptosis
The upper eyelid margin is positioned 3 mm or more lower than usual and covers the superior portion of the iris (objective); or, the upper lid margin obscures at least part of the pupil (subjective).

Bifid tongue
Tongue with a median apical indentation or fork.

Microcephaly
Head circumference below 2 standard deviations below the mean for age and gender.

Epicanthus
A fold of skin starting above the medial aspect of the upper eyelid and arching downward to cover, pass in front of and lateral to the medial canthus.

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

Overlapping toe
Describes a foot digit resting on the dorsal surface of an adjacent digit when the foot is at rest.

Abnormality of the larynx
An abnormality of the larynx.

Ureteropelvic junction obstruction
Blockage of urine flow from the renal pelvis to the proximal ureter.

Downslanted palpebral fissures
The palpebral fissure inclination is more than two standard deviations below the mean.

Dandy-Walker malformation
A congenital brain malformation typically characterized by incomplete formation of the cerebellar vermis, dilation of the fourth ventricle, and enlargement of the posterior fossa. In layman's terms, Dandy Walker malformation is a cyst in the cerebellum (typically symmetrical) that is involved with the fourth ventricle. This may interfere with the ability to drain cerebrospinal fluid from the brain, resulting in hydrocephalus. Dandy Walker cysts are formed during early embryonic development, while the brain forms. The cyst in the cerebellum typically has several blood vessels running through it connecting to the brain, thereby prohibiting surgical removal.

Aganglionic megacolon
An abnormality resulting from a lack of intestinal ganglion cells (i.e., an aganglionic section of bowel) that results in bowel obstruction with enlargement of the colon.

Sleep-wake cycle disturbance

Any abnormal alteration of an individual's circadian rhythm that affects the timing of sleeping and being awake.

Abnormal lung lobation
A developmental defect in the formation of pulmonary lobes.

Tracheal stenosis

Elevated 7-dehydrocholesterol
Elevated 7-dehydrocholesterol levels.

Unilateral renal agenesis
A unilateral form of agenesis of the kidney.

Bicornuate uterus
The presence of a bicornuate uterus.

Aplasia/Hypoplasia affecting the eye

Abnormality of cardiovascular system morphology
Any structural anomaly of the heart and great vessels.

Periventricular heterotopia
A form of gray matter heterotopia were the mislocalized gray matter is typically located periventricularly, also sometimes called subependymal heterotopia. Periventricular means beside the ventricles. This is by far the most common location for heterotopia. Subependymal heterotopia present in a wide array of variations. There can be a small single node or a large number of nodes, can exist on either or both sides of the brain at any point along the higher ventricle margins, can be small or large, single or multiple, and can form a small node or a large wavy or curved mass.

Broad alveolar ridges

Coarctation of aorta
Coarctation of the aorta is a narrowing or constriction of a segment of the aorta.

Pyloric stenosis
An abnormal narrowing of the pylorus.

Sensorineural hearing impairment
A type of hearing impairment in one or both ears related to an abnormal functionality of the cochlear nerve.

Breech presentation
A position of the fetus at delivery in which the fetus enters the birth canal with the buttocks or feet first.

Short thumb
Hypoplasia (congenital reduction in size) of the thumb.

Bifid scrotum
Midline indentation or cleft of the scrotum.

Septate vagina
The presence of a vaginal septum, thereby creating a vaginal duplication. The septum is longitudinal in the majority of cases.

Micromelia
The presence of abnormally small extremities.

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

Renal hypoplasia/aplasia
Absence or underdevelopment of the kidney.

Low-set ears
Upper insertion of the ear to the scalp below an imaginary horizontal line drawn between the inner canthi of the eye and extending posteriorly to the ear.

Aplasia/Hypoplasia of the corpus callosum
Absence or underdevelopment of the corpus callosum.

Abnormal dental enamel morphology
An abnormality of the dental enamel.

Eczema
Eczema is a form of dermatitis. The term eczema is broadly applied to a range of persistent skin conditions and can be related to a number of underlying conditions. Manifestations of eczema can include dryness and recurring skin rashes with redness, skin edema, itching and dryness, crusting, flaking, blistering, cracking, oozing, or bleeding.

Atrial septal defect
Atrial septal defect (ASD) is a congenital abnormality of the interatrial septum that enables blood flow between the left and right atria via the interatrial septum.

Finger syndactyly
Webbing or fusion of the fingers, involving soft parts only or including bone structure. Bony fusions are referred to as "bony" Syndactyly if the fusion occurs in a radio-ulnar axis. Fusions of bones of the fingers in a proximo-distal axis are referred to as "Symphalangism".

Microglossia
Decreased length and width of the tongue.

Intrauterine growth retardation
An abnormal restriction of fetal growth with fetal weight below the tenth percentile for gestational age.

Sclerocornea

A congenital anomaly in which a part or the whole of the cornea acquires the characteristics of sclera, resulting in clouding of the cornea.

Depressed nasal bridge
Posterior positioning of the nasal root in relation to the overall facial profile for age.

Optic atrophy
Atrophy of the optic nerve. Optic atrophy results from the death of the retinal ganglion cell axons that comprise the optic nerve and manifesting as a pale optic nerve on fundoscopy.

Attention deficit hyperactivity disorder
Attention deficit hyperactivity disorder (ADHD) manifests at age 2-3 years or by first grade at the latest. The main symptoms are distractibility, impulsivity, hyperactivity, and often trouble organizing tasks and projects, difficulty going to sleep, and social problems from being aggressive, loud, or impatient.

Cholestatic liver disease

Scrotal hypoplasia

Upslanted palpebral fissure
The palpebral fissure inclination is more than two standard deviations above the mean for age (objective); or, the inclination of the palpebral fissure is greater than typical for age.

Abnormal dermatoglyphics
An abnormality of dermatoglyphs (fingerprints), which are present on fingers, palms, toes, and soles.

Abnormal form of the vertebral bodies
Abnormal morphology of vertebral body.

Ambiguous genitalia
A genital phenotype that is not clearly assignable to a single gender. Ambiguous genitalia can be evaluated using the Prader scale: Prader 0: Normal female external genitalia. Prader 1: Female external genitalia with clitoromegaly. Prader 2: Clitoromegaly with partial labial fusion forming a funnel-shaped urogenital sinus. Prader 3: Increased phallic enlargement. Complete labioscrotal fusion forming a urogenital sinus with a single opening. Prader 4: Complete scrotal fusion with urogenital opening at the base or on the shaft of the phallus. Prader 5: Normal male external genitalia. The diagnosis of ambiguous genitalia is made for Prader 1-4.

Pulmonary hypoplasia

Recurrent infections
Increased susceptibility to infections.

Proptosis
An eye that is protruding anterior to the plane of the face to a greater extent than is typical.

Hypertonia

A condition in which there is increased muscle tone so that arms or legs, for example, are stiff and difficult to move.

Constipation
Infrequent or difficult evacuation of feces.

Intellectual disability
Subnormal intellectual functioning which originates during the developmental period. Intellectual disability, previously referred to as mental retardation, has been defined as an IQ score below 70.

Hammertoe
Hyperextension of the metatarsal-phalangeal joint with hyperflexion of the proximal interphalangeal (PIP) joint.

Gastroesophageal reflux
A condition in which the stomach contents leak backwards from the stomach into the esophagus through the lower esophageal sphincter.

Glaucoma
Glaucoma refers loss of retinal ganglion cells in a characteristic pattern of optic neuropathy usually associated with increased intraocular pressure.

Abnormality of dental morphology
An abnormality of the morphology of the tooth.

Seizure
A seizure is an intermittent abnormality of nervous system physiology characterised by a transient occurrence of signs and/or symptoms due to abnormal excessive or synchronous neuronal activity in the brain.

Choanal atresia
Absence or abnormal closure of the choana (the posterior nasal aperture).

Cleft palate
Cleft palate is a developmental defect of the palate resulting from a failure of fusion of the palatine processes and manifesting as a separation of the roof of the mouth (soft and hard palate).

Cutis marmorata
A reticular discoloration of the skin with cyanotic (reddish-blue appearing) areas surrounding pale central areas due to dilation of capillary blood vessels and stagnation of blood within the vessels. Cutis marmorata generally occurs on the legs, arms and trunk and is often more severe in cold weather.

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

Epiphyseal stippling
The presence of abnormal punctate (speckled, dot-like) calcifications in one or more epiphyses.

Ventriculomegaly
An increase in size of the ventricular system of the brain.

Micrognathia
Developmental hypoplasia of the mandible.

Narrow forehead
Width of the forehead or distance between the frontotemporales is more than two standard deviations below the mean (objective); or apparently narrow intertemporal region (subjective).

Hearing impairment
A decreased magnitude of the sensory perception of sound.

Metatarsus adductus
The metatarsals are deviated medially (tibially), that is, the bones in the front half of the foot bend or turn in toward the body.

Autosomal recessive inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in individuals with two pathogenic alleles, either homozygotes (two copies of the same mutant allele) or compound heterozygotes (whereby each copy of a gene has a distinct mutant allele).

Postaxial foot polydactyly
Polydactyly of the foot most commonly refers to the presence of six toes on one foot. Postaxial polydactyly affects the lateral ray and the duplication may range from a well-formed articulated digit to a rudimentary digit.

Brachydactyly
Digits that appear disproportionately short compared to the hand/foot. The word brachydactyly is used here to describe a series distinct patterns of shortened digits (brachydactyly types A-E). This is the sense used here.

Patent ductus arteriosus
In utero, the ductus arteriosus (DA) serves to divert ventricular output away from the lungs and toward the placenta by connecting the main pulmonary artery to the descending aorta. A patent ductus arteriosus (PDA) in the first 3 days of life is a physiologic shunt in healthy term and preterm newborn infants, and normally is substantially closed within about 24 hours after bith and completely closed after about three weeks. Failure of physiologcal closure is referred to a persistent or patent ductus arteriosus (PDA). Depending on the degree of left-to-right shunting, PDA can have clinical consequences.

Wide intermamillary distance
A larger than usual distance between the left and right nipple.

Kyphosis
Exaggerated anterior convexity of the thoracic vertebral column.

Abnormal eyelash morphology
An abnormality of the eyelashes.

Polyhydramnios
The presence of excess amniotic fluid in the uterus during pregnancy.

Wide mouth
Distance between the oral commissures more than 2 SD above the mean. Alternatively, an apparently increased width of the oral aperture (subjective).

Ulnar deviation of finger
Bending or curvature of a finger toward the ulnar side (i.e., away from the thumb). The deviation is at the metacarpal-phalangeal joint, and this finding is distinct from clinodactyly.

Hypoplasia of the frontal lobes
Underdevelopment of the frontal lobe of the cerebrum.

Hypocholesterolemia
An decreased concentration of cholesterol in the blood.

Multicystic kidney dysplasia
Multicystic dysplasia of the kidney is characterized by multiple cysts of varying size in the kidney and the absence of a normal pelvicaliceal system. The condition is associated with ureteral or ureteropelvic atresia, and the affected kidney is nonfunctional.

Short stature
A height below that which is expected according to age and gender norms. Although there is no universally accepted definition of short stature, many refer to "short stature" as height more than 2 standard deviations below the mean for age and gender (or below the 3rd percentile for age and gender dependent norms).

Aplasia/Hypoplasia of the cerebellum

Tooth agenesis
The absence of one or more teeth from the normal series by a failure
to develop

Micropenis
Abnormally small penis. At birth, the normal penis is about 3 cm (stretched length from pubic tubercle to tip of penis) with micropenis less than 2.0-2.5 cm.

Iris coloboma
A coloboma of the iris.

Anteverted nares
Anteriorly-facing nostrils viewed with the head in the Frankfurt horizontal and the eyes of the observer level with the eyes of the subject. This gives the appearance of an upturned nose (upturned nasal tip).

Precocious puberty
The onset of secondary sexual characteristics before a normal age. Although it is difficult to define normal age ranges because of the marked variation with which puberty begins in normal children, precocious puberty can be defined as the onset of puberty before the age of 8 years in girls or 9 years in boys.

Nystagmus
Rhythmic, involuntary oscillations of one or both eyes related to abnormality in fixation, conjugate gaze, or

vestibular mechanisms.

Sleep disturbance
An abnormality of sleep including such phenomena as 1) insomnia/hypersomnia, 2) non-restorative sleep, 3) sleep schedule disorder, 4) excessive daytime somnolence, 5) sleep apnea, and 6) restlessness.

Hip dislocation
Displacement of the femur from its normal location in the hip joint.

Cataract
A cataract is an opacity or clouding that develops in the crystalline lens of the eye or in its capsule.

Scoliosis
The presence of an abnormal lateral curvature of the spine.

Clitoral hypertrophy
Hypertrophy of the clitoris.

Congenital diaphragmatic hernia
The presence of a hernia of the diaphragm present at birth.

Hydrocephalus
Hydrocephalus is an active distension of the ventricular system of the brain resulting from inadequate passage of CSF from its point of production within the cerebral ventricles to its point of absorption into the systemic circulation.

Abnormality of the metacarpal bones
An abnormality of the metacarpal bones.

Hypotonia
Hypotonia is an abnormally low muscle tone (the amount of tension or resistance to movement in a muscle). Even when relaxed, muscles have a continuous and passive partial contraction which provides some resistance to passive stretching. Hypotonia thus manifests as diminished resistance to passive stretching. Hypotonia is not the same as muscle weakness, although the two conditions can co-exist.

Decreased fetal movement
An abnormal reduction in quantity or strength of fetal movements.

Poor suck
An inadequate sucking reflex, resulting in the difficult of newborns to be breast-fed.

Holoprosencephaly
Holoprosencephaly is a structural anomaly of the brain in which the developing forebrain fails to divide into two separate hemispheres and ventricles.

Gastroschisis
A type of congenital ventral incomplete closure of the abdominal wall in which the intestines and sometimes other organs extend freely into the amniotic fluid space through a small opening in the abdomen, usually to the right of the umbilicus.

Talipes calcaneovalgus
Talipes calcaneovalgus is a flexible foot deformity (as opposed to a rigid congenital vertical talus foot deformity) that can either present as a positional or structural foot deformity depending on severity and/or causality. The axis of calcaneovalgus deformity is in the tibiotalar joint, where the foot is positioned in extreme hyperextension. On inspection, the foot has an "up and out" appearance, with the dorsal forefoot practically touching the anterior aspect of the ankle and lower leg.

Hypoplasia of penis

Hypopigmentation of hair

Hyperactivity
Hyperactivity is a state of constantly being unusually or abnormally active, including in situations in which it is not appropriate.

Excessive daytime somnolence
A state of abnormally strong desire for sleep during the daytime.

Generalized hypotonia
Generalized muscular hypotonia (abnormally low muscle tone).

Abnormality of the ribs
An anomaly of the rib.

Intestinal malrotation
An abnormality of the intestinal rotation and fixation that normally occurs during the development of the gut. This can lead to volvulus, or twisting of the intestine that causes obstruction and necrosis.

Self-injurious behavior
Aggression towards oneself.

Dental crowding
Changes in alignment of teeth in the dental arch

Feeding difficulties in infancy
Impaired feeding performance of an infant as manifested by difficulties such as weak and ineffective sucking, brief bursts of sucking, and falling asleep during sucking. There may be difficulties with chewing or maintaining attention.

Recurrent otitis media
Increased susceptibility to otitis media, as manifested by recurrent episodes of otitis media.

Hypoplasia of the corpus callosum
Underdevelopment of the corpus callosum.

Posteriorly rotated ears
A type of abnormal location of the ears in which the position of the ears is characterized by posterior rotation

(the superior part of the ears is rotated towards the back of the head, and the inferior part of the ears towards the front).

Renal cyst
A fluid filled sac in the kidney.

Biparietal narrowing
A narrowing of the biparietal diameter (i.e., of the transverse distance between the protuberances of the two parietal bones of the skull).

Global developmental delay
A delay in the achievement of motor or mental milestones in the domains of development of a child, including motor skills, speech and language, cognitive skills, and social and emotional skills. This term should only be used to describe children younger than five years of age.

Hip subluxation
A partial dislocation of the hip joint, whereby the head of the femur is partially displaced from the socket.

Wide nasal bridge
Increased breadth of the nasal bridge (and with it, the nasal root).

Renal agenesis
Agenesis, that is, failure of the kidney to develop during embryogenesis and development.

Growth delay
A deficiency or slowing down of growth pre- and postnatally.

Rhizomelia
Disproportionate shortening of the proximal segment of limbs (i.e. the femur and humerus).

Proximal placement of thumb
Proximal mislocalization of the thumb.

Short neck
Diminished length of the neck.

Ventricular septal defect
A hole between the two bottom chambers (ventricles) of the heart. The defect is centered around the most superior aspect of the ventricular septum.

Hypospadias
Abnormal position of urethral meatus on the ventral penile shaft (underside) characterized by displacement of the urethral meatus from the tip of the glans penis to the ventral surface of the penis, scrotum, or perineum.

Postaxial hand polydactyly
Supernumerary digits located at the ulnar side of the hand (that is, on the side with the fifth finger).

2-3 toe syndactyly
Syndactyly with fusion of toes two and three.

Mesomelia
Shortening of the middle parts of the limbs (forearm and lower leg) in relation to the upper and terminal segments.

Facial capillary hemangioma
Hemangioma, a benign tumor of the vascular endothelial cells with small endothelial spaces, occurring in the face.

Hydronephrosis
Severe distention of the kidney with dilation of the renal pelvis and calices.

Hypertelorism
Interpupillary distance more than 2 SD above the mean (alternatively, the appearance of an increased interpupillary distance or widely spaced eyes).

Strabismus
A misalignment of the eyes so that the visual axes deviate from bifoveal fixation. The classification of strabismus may be based on a number of features including the relative position of the eyes, whether the deviation is latent or manifest, intermittent or constant, concomitant or otherwise and according to the age of onset and the relevance of any associated refractive error.

Long philtrum
Distance between nasal base and midline upper lip vermilion border more than 2 SD above the mean. Alternatively, an apparently increased distance between nasal base and midline upper lip vermilion border.

Cutaneous photosensitivity
An increased sensitivity of the skin to light. Photosensitivity may result in a rash upon exposure to the sun (which is known as photodermatosis). Photosensitivity can be diagnosed by phototests in which light is shone on small areas of skin.

Abnormality of the gallbladder
An abnormality of the gallbladder.

Supernumerary tooth
The presence of one or more teeth additional to the normal number.

Gastrointestinal dysmotility
Abnormal intestinal contractions, such as spasms and intestinal paralysis, related to the loss of the ability of the gut to coordinate muscular activity because of endogenous or exogenous causes.

Increased nuchal translucency
The presence of an abnormally large hypoechoic space in the posterior fetal neck (usually detected on prenatal ultrasound examination).

Split hand
A condition in which middle parts of the hand (fingers and metacarpals) are missing giving a cleft appearance. The severity is very variable ranging from slightly hypoplastic middle fingers over absent middel fingers as far as oligo- or monodactyl hands.

Abnormal localization of kidney
An abnormal site of the kidney.