Geneticheck - Genetic Report

Gene

CDH1

Associated Diseases

Endometrial Carcinoma, Somatic
Gastric Cancer, Hereditary Diffuse
Prostate Cancer
Breast Cancer
Cleft Lip/palate
Blepharo-cheilo-odontic Syndrome
Blepharocheilodontic Syndrome 1
Ovarian Cancer

Phenotype

Small nail
A nail that is diminished in length and width, i.e., underdeveloped nail.

Cutaneous syndactyly
A soft tissue continuity in the A/P axis between two digits that extends distally to at least the level of the proximal interphalangeal joints, or a soft tissue continuity in the A/P axis between two digits that lies significantly distal to the flexion crease that overlies the metacarpophalangeal or metatarsophalangeal joint of the adjacent digits.

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

Endometrial carcinoma
A carcinoma of the endometrium, the mucous lining of the uterus.

Anal atresia
Congenital absence of the anus, i.e., the opening at the bottom end of the intestinal tract.

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).

Abnormality of the eye
Any abnormality of the eye, including location, spacing, and intraocular abnormalities.

Abnormality of vision
Abnormality of eyesight (visual perception).

Hypodontia
The absence of five or less teeth from the normal series by a failure to develop.

High anterior hairline

Distance between the hairline (trichion) and the glabella (the most prominent point on the frontal bone above the root of the nose), in the midline, more than two SD above the mean. Alternatively, an apparently increased distance between the hairline and the glabella.

Euryblepharon
Euryblepharon is a congenital eyelid anomaly characterized by horizontal enlargement of the palpebral fissure. The eyelid is shortened vertically compared with the horizontal dimension, with associated lateral canthal malpositioning and lateral ectropion abnormally wide lid opening.

Oral-pharyngeal dysphagia

Breast carcinoma
The presence of a carcinoma of the breast.

High forehead
An abnormally increased height of the forehead.

Abnormal hair quantity
An abnormal amount of hair.

Peg-shaped maxillary lateral incisors
A tooth crown with its mesial and distal sides converging or tapering toward the incisal edge causing severe reduction of mesiodistal diameter

Carious teeth
Caries is a multifactorial bacterial infection affecting the structure of the tooth. This term has been used to describe the presence of more than expected dental caries.

Autosomal dominant 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 heterozygotes. In the context of medical genetics, an autosomal dominant disorder is caused when a single copy of the mutant allele is present. Males and females are affected equally, and can both transmit the disorder with a risk of 50% for each child of inheriting the mutant allele.

Nasal speech
A type of speech characterized by the presence of an abnormally increased nasal airflow during speech.

Hypoplasia of the maxilla
Abnormally small dimension of the Maxilla. Usually creating a malocclusion or malalignment between the upper and lower teeth or resulting in a deficient amount of projection of the base of the nose and lower midface region.

Conductive hearing impairment
An abnormality of vibrational conductance of sound to the inner ear leading to impairment of sensory perception of sound.

Conical tooth

An abnormal conical form of the teeth, that is, a tooth whose sides converge or taper together incisally.

Oral cleft
The presence of a cleft in the oral cavity, the two main types of which are cleft lip and cleft palate. In cleft lip, there is the congenital failure of the maxillary and median nasal processes to fuse, forming a groove or fissure in the lip. In cleft palate, there is a congenital failure of the palate to fuse properly, forming a grooved depression or fissure in the roof of the mouth. Clefts of the lip and palate can occur individually or together. It is preferable to code each defect separately.

Ovarian papillary adenocarcinoma
The presence of a papillary adenocarcinoma of the ovary.

Speech articulation difficulties
Impairment in the physical production of speech sounds.

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

Neural tube defect
A neural tube defect arises when the neural tube, the embryonic precursor of the brain and spinal cord, fails to close during neurulation. The cranial region (anencephaly) or the low spine (open spina bifida; myelomeningocele) are most commonly affected although, in the severe NTD craniorachischisis, almost the entire neural tube remains open, from midbrain to low spine.

Chronic atrophic gastritis
A form of chronic gastritis associated with atrophic gastric mucous membrane.

Distichiasis
Double rows of eyelashes.

Cleft upper lip
A gap in the upper lip. This is a congenital defect resulting from nonfusion of tissues of the lip during embryonal development.

Abnormal eyelid morphology
An abnormality of the eyelids.

Unilateral cleft palate

Sparse hair
Reduced density of hairs.

Neoplasm
An organ or organ-system abnormality that consists of uncontrolled autonomous cell-proliferation which can occur in any part of the body as a benign or malignant neoplasm (tumour).

Abnormality of dental eruption
An abnormality of tooth eruption.

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.

Dental malocclusion
Dental malocclusion refers to an abnormality of the occlusion, or alignment, of the teeth and the way the upper and lower teeth fit together, resulting in overcrowding of teeth or in abnormal bite patterns.

Clinodactyly
An angulation of a digit at an interphalangeal joint in the plane of the palm (finger) or sole (toe).

Agenesis of lateral incisor

Delayed speech and language development
A degree of language development that is significantly below the norm for a child of a specified age.

Ectropion of lower eyelids

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".

Bilateral cleft lip and palate
Cleft lip and cleft palate affecting both sides of the face.

Abnormal number of permanent teeth
The presence of an altered number of of permanent teeth.

Dysgerminoma
The presence of a dysgerminoma, i.e., an undifferentiated germ cell tumor of the ovary.

Flat face
Absence of concavity or convexity of the face when viewed in profile.

Somatic mutation
A mode of inheritance in which a trait or disorder results from a de novo mutation occurring after conception, rather than being inherited from a preceding generation.

Stomach cancer
A cancer arising in any part of the stomach.

Hypertelorism

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

Heterogeneous

Malnutrition
A deficiency in the intake of energy and nutrients.

Velopharyngeal insufficiency
Inability of velopharyngeal sphincter to sufficiently separate the nasal cavity from the oral cavity during speech.

Prostate cancer
A cancer of the prostate.

Bilateral cleft palate
Nonmidline cleft palate on the left and right sides.

Palate fistula
A fistula which connects the oral cavity and the pharyngeal area via the aspects of the soft palate.

Epidermoid cyst
Nontender, round and firm, but slightly compressible, intradermal or subcutaneous cyst measuring 0.5-5 cm in diameter. Epidermal cysts are intradermal or subcutaneous tumors, grow slowly and occur on the face, neck, back and scrotum. They usually appear at or around puberty, and as a rule an affected individual has one solitary or a few cysts.