Geneticheck - Genetic Report

Gene

STK11

Associated Diseases

Peutz-jeghers Syndrome
Peutz-jeghers Syndrome
Testicular Tumor, Somatic
Melanoma, Cutaneous Malignant
Pancreatic Cancer

Phenotype

Uveal melanoma
A malignant melanoma originating within the eye. The tumor originates from the melanocytes in the uvea (which comprises the iris, ciliary body, and choroid).

Abnormality of the respiratory system
An abnormality of the respiratory system, which include the airways, lungs, and the respiratory muscles.

Abnormality of the gastrointestinal tract
An abnormality of the gastrointestinal tract.

Biliary tract neoplasm
A tumor (abnormal growth of tissue) of the biliary system.

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

Nasal polyposis
Polypoidal masses arising mainly from the mucous membranes of the nose and paranasal sinuses. They are freely movable and nontender overgrowths of the mucosa that frequently accompany allergic rhinitis.

Neoplasm of the pancreas
A tumor (abnormal growth of tissue) of the pancreas.

Enlarged polycystic ovaries

Atypical nevi in non-sun exposed areas

Breast carcinoma
The presence of a carcinoma of the breast.

Gastrointestinal infarctions

Gastrointestinal carcinoma

Oral melanotic macule
Flat, distinct, discolored area of oral mucosal membrane less than 1 cm wide not associated with a change in the thickness or texture of the affected mucosal membrane. The lesions are small, solitary, well-circumscribed and often uniformly pigmented.

Cutaneous melanoma
The presence of a melanoma of the skin.

Clubbing
Broadening of the soft tissues (non-edematous swelling of soft tissues) of the digital tips in all dimensions associated with an increased longitudinal and lateral curvature of the nails.

Ovarian cyst
The presence of one or more cysts of the ovary.

Numerous nevi

Azoospermia
Absence of any measurable level of sperm in his semen.

Abnormality of the ureter
An abnormality of the ureter. The ureter is the duct by which urine passes from the kidney to the bladder.

Intestinal bleeding
Bleeding from the intestines.

Macule
A flat, distinct, discolored area of skin less than 1 cm wide that does not involve any change in the thickness or texture of the skin.

Intussusception
An abnormality of the intestine in which part of the intestine invaginates (telescopes) into another part of the intestine.

Biliary tract abnormality
An abnormality of the biliary tree.

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.

Abdominal pain

An unpleasant sensation characterized by physical discomfort (such as pricking, throbbing, or aching) and perceived to originate in the abdomen.

Bladder polyp
An abnormal growth that projects from the mucous membrane of the urinary bladder.

Choriocarcinoma
A malignant, trophoblastic and aggressive cancer, usually of the placenta. It is characterized by early hematogenous spread to the lungs and belongs to the far end of the spectrum of gestational trophoblastic disease (GTD), a subset of germ cell tumors.

Abnormality of the nose
An abnormality of the nose.

Hypermelanotic macule
A hyperpigmented circumscribed area of change in normal skin color without elevation or depression of any size.

Gastrointestinal hemorrhage
Hemorrhage affecting the gastrointestinal tract.

Pancreatic adenocarcinoma
The presence of an adenocarcinoma of the pancreas.

Teratoma
The presence of a teratoma.

Rectal prolapse
Protrusion of the rectal mucous membrane through the anus.

Neoplasm of the colon

Atypical nevus
A large pigmented lesion measuring 5-15 mm in diameter with irregular, notched, and ill defined border and with color that may range from tan to dark brown to pink.

Hamartomatous polyposis
Polyp-like protrusions which are histologically hamartomas. These can occur throughout the gastrointestinal tract. Hamartomatous polyps are composed of the normal cellular elements of the gastrointestinal tract, but have a markedly distorted architecture.

Iron deficiency anemia

Esophageal neoplasm
A tumor (abnormal growth of tissue) of the esophagus.

Neoplasm of the rectum

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

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).

Neoplasm of the nose
Tumor (An abnormal mass of tissue resulting from abnormally dividing cells) of the nasal cavity.

Elevated alpha-fetoprotein
An increased concentration of alpha-fetoprotein.

Melanonychia
Brown or black discoloration of the nails.

Abnormal pigmentation of the oral mucosa
An abnormality of the pigmentation of the mucosa of the mouth.

Anemia
A reduction in erythrocytes volume or hemoglobin concentration.

Clubbing of fingers
Terminal broadening of the fingers (distal phalanges of the fingers).

Precocious puberty with Sertoli cell tumor

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

Neoplasm of the small intestine
The presence of a neoplasm of the small intestine.

Embryonal neoplasm

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.

Increased level of L-fucose in urine
An increase in the level of L-fucose in the urine.

Multiple renal cysts
The presence of many cysts in the kidney.

Sporadic
Cases of the disease in question occur without a previous family history, i.e., as isolated cases without being transmitted from a parent and without other siblings being affected.

Labial melanotic macule
Flat, distinct, discolored area on the lip less than 1 cm wide not associated with a change in the thickness or texture.

Renal cell carcinoma
A type of carcinoma of the kidney with origin in the epithelium of the proximal convoluted renal tubule.

Abnormality of the gallbladder
An abnormality of the gallbladder.

Cervix cancer
A tumor of the uterine cervix.

Multiple lentigines
Presence of an unusually high number of lentigines (singular: lentigo), which are flat, tan to brown oval spots.

Neoplasm of the lung
Tumor of the lung.

Intestinal obstruction
Blockage or impairment of the normal flow of the contents of the intestine towards the anal canal.

Uterine neoplasm
A tumor (abnormal growth of tissue) of the uterus.