

Gene

EGFR

Associated Diseases

Inflammatory Skin And Bowel Disease, Neonatal, 2
Lung Cancer, Susceptibility To
Neonatal Inflammatory Skin And Bowel Disease

Phenotype

Vomiting

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

Recurrent bacterial skin infections

Increased susceptibility to bacterial infections of the skin, as manifested by recurrent episodes of infectious dermatitis.

Recurrent gastroenteritis

Increased susceptibility to gastroenteritis, an infectious inflammation of the stomach and small intestines manifested by signs and symptoms such as diarheas and abdominal pain, as manifested by recurrent episodes of gastroenteritis.

Scaling skin

Refers to the loss of the outer layer of the epidermis in large, scale-like flakes.

Long eyelashes

Mid upper eyelash length >10 mm or increased length of the eyelashes (subjective).

Horizontal eyebrow

An eyebrow that extends straight across the brow, without curve.

Generalized abnormality of skin

An abnormality of the skin that is not localized to any one particular region.

Blepharitis

Inflammation of the eyelids.

Slow-growing scalp hair

Scalp hair whose growth is slower than normal.

Onychogryposis

Nail that appears thick when viewed on end.

Non-small cell lung carcinoma

Edema

An abnormal accumulation of fluid beneath the skin, or in one or more cavities of the body.

Recurrent bronchiolitis

An increased susceptibility to bronchiolitis as manifested by a history of recurrent bronchiolitis.

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).

Anal fissure

A small tear in the thin, moist tissue (mucosa) that lines the anus. It appears as a crack or slit in the mucous membrane of the anus.

Failure to thrive

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

Erythema

Redness of the skin, caused by hyperemia of the capillaries in the lower layers of the skin.

Lung adenocarcinoma**Psoriasiform dermatitis**

A skin abnormality characterized by redness and irritation, with thick, red skin that displays flaky, silver-white patches (scales).

Chapped lip

Cracking, fissuring, and peeling of the skin of the lips.

Diarrhea

Abnormally increased frequency of loose or watery bowel movements.

Bloody diarrhea

Passage of many stools containing blood.

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.

Epidermal acanthosis

Diffuse hypertrophy or thickening of the stratum spinosum of the epidermis (prickle cell layer of the skin).

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.

Chronic monilial nail infection

Chronic infection of the nails by *Candida* species.

Papule

A circumscribed, solid elevation of skin with no visible fluid, varying in size from a pinhead to less than 10mm in diameter at the widest point.

Left ventricular hypertrophy

Enlargement or increased size of the heart left ventricle.

Hypertension

The presence of chronic increased pressure in the systemic arterial system.

Pustule

A small elevation of the skin containing cloudy or purulent material usually consisting of necrotic inflammatory cells.

Dehydration**Otitis externa**

Inflammation or infection of the external auditory canal (EAC), the auricle, or both.

Recurrent pneumonia

An increased susceptibility to pneumonia as manifested by a history of recurrent episodes of pneumonia.

Alveolar cell carcinoma

Adenocarcinoma of the Bronchus.

Perianal rash

The presence of a rash (change of color and texture) of the perianal skin.