Geneticheck - Genetic Report

Gene

USH2A

Associated Diseases

Usher Syndrome, Type Iia
Retinitis Pigmentosa 39
Usher Syndrome Type 2
Retinitis Pigmentosa

Phenotype

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

Hemianopia
Partial or complete loss of vision in one half of the visual field of one or both eyes.

Visual loss
Loss of visual acuity (implying that vision was better at a certain time point in life). Otherwise the term reduced visual acuity should be used (or a subclass of that).

Abnormal retinal vascular morphology
A structural abnormality of retinal vasculature.

Abnormality of dental color
A developmental defect of tooth color.

Anxiety
Intense feelings of nervousness, tenseness, or panic, often in reaction to interpersonal stresses; worry about the negative effects of past unpleasant experiences and future negative possibilities; feeling fearful, apprehensive, or threatened by uncertainty; fears of falling apart or losing control.

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).

Subcortical cerebral atrophy
Atrophy of the cerebral subcortical white and gray matter, termed subcortical atrophy, reflects loss of nerve cells in the basal ganglia or fibers in the deep white matter.

Photophobia
Excessive sensitivity to light with the sensation of discomfort or pain in the eyes due to exposure to bright light.

Atypical scarring of skin
Atypically scarred skin .

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.

Aplasia/Hypoplasia of the cerebellum

Hallucinations
Perceptions in a conscious and awake state in the absence of external stimuli which have qualities of real perception, in that they are vivid, substantial, and located in external objective space.

Keratoconus
A cone-shaped deformity of the cornea characterized by the presence of corneal distortion secondary to thinning of the apex.

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).

Ataxia
Cerebellar ataxia refers to ataxia due to dysfunction of the cerebellum. This causes a variety of elementary neurological deficits including asynergy (lack of coordination between muscles, limbs and joints), dysmetria (lack of ability to judge distances that can lead to under- or overshoot in grasping movements), and dysdiadochokinesia (inability to perform rapid movements requiring antagonizing muscle groups to be switched on and off repeatedly).

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

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

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

Visual field defect

Myopia
An abnormality of refraction characterized by the ability to see objects nearby clearly, while objects in the distance appear blurry.

Hypoplasia of penis

Obesity
Accumulation of substantial excess body fat.

Hypogonadism
A decreased functionality of the gonad.

Congenital sensorineural hearing impairment
A type of hearing impairment caused by an abnormal functionality of the cochlear nerve with congenital onset.

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

Abnormal electroretinogram
Any abnormality of the electrical responses of various cell types in the retina as measured by electroretinography.

Iris hypopigmentation
An abnormal reduction in the amount of pigmentation of the iris.

Rod-cone dystrophy
An inherited retinal disease subtype in which the rod photoreceptors appear to be more severely affected than the cone photoreceptors. Typical presentation is with nyctalopia (due to rod dysfunction) followed by loss of mid-peripheral field of vision, which gradually extends and leaves many patients with a small central island of vision due to the preservation of macular cones.

Type II diabetes mellitus
A type of diabetes mellitus initially characterized by insulin resistance and hyperinsulinemia and subsequently by glucose interolerance and hyperglycemia.

Hyperinsulinemia
An increased concentration of insulin in the blood.

Abnormal dental enamel morphology
An abnormality of the dental enamel.

Cerebral cortical atrophy
Atrophy of the cortex of the cerebrum.

Depression
Frequent feelings of being down, miserable, and/or hopeless; difficulty recovering from such moods; pessimism about the future; pervasive shame; feeling of inferior self-worth; thoughts of suicide and suicidal behavior.

Microdontia
Decreased size of the teeth, which can be defined as a mesiodistal tooth diameter (width) more than 2 SD below mean. Alternatively, an apparently decreased maximum width of tooth.

Attenuation of retinal blood vessels

Scotoma
A regional and pathological increase of the light detection threshold in any region of the visual field surrounded by a field of normal or relatively well-preserved vision.

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

Progressive night blindness

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.

Abnormality of retinal pigmentation

Visual impairment
Visual impairment (or vision impairment) is vision loss (of a person) to such a degree as to qualify as an additional support need through a significant limitation of visual capability resulting from either disease, trauma, or congenital or degenerative conditions that cannot be corrected by conventional means, such as refractive correction, medication, or surgery.

Abnormality of the inner ear
An abnormality of the inner ear.

Ophthalmoplegia
Paralysis of one or more extraocular muscles that are responsible for eye movements.

Nyctalopia
Inability to see well at night or in poor light.

Abnormal testis morphology
An anomaly of the testicle (the male gonad).

Bone spicule pigmentation of the retina
Pigment migration into the retina in a bone-spicule configuration (resembling the nucleated cells within the lacuna of bone).

Schizophrenia
A mental disorder characterized by a disintegration of thought processes and of emotional responsiveness. It most commonly manifests as auditory hallucinations, paranoid or bizarre delusions, or disorganized speech and thinking, and it is accompanied by significant social or occupational dysfunction. The onset of symptoms typically occurs in young adulthood, with a global lifetime prevalence of about 0.3-0.7%.

Blindness
Blindness is the condition of lacking visual perception defined as visual perception below 3/60 and/or a visual field of no greater than 10 degrees in radius around central fixation.

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.

Hyperreflexia
Hyperreflexia is the presence of hyperactive stretch reflexes of the muscles.