Geneticheck - Genetic Report

Gene

SPATA7

Associated Diseases

Severe Early-childhood-onset Retinal Dystrophy
Retinitis Pigmentosa
Leber Congenital Amaurosis-3 (lca3)/retinitis Pigmentosa, Juvenile, Autosomal Recessive
Leber Congenital Amaurosis

Phenotype

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

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.

Reduced visual acuity

Optic disc drusen
Optic disc drusen are acellular, calcified deposits within the optic nerve head. Optic disc drusen are congenital and developmental anomalies of the optic nerve head, representing hyaline-containing bodies that, over time, appear as elevated, lumpy irregularities on the anterior portion of the optic nerve.

Blurred vision
Lack of sharpness of vision resulting in the inability to see fine detail.

Hemiplegia/hemiparesis
Loss of strength in the arm, leg, and sometimes face on one side of the body. Hemiplegia refers to a severe or complete loss of strength, whereas hemiparesis refers to a relatively mild loss of strength.

Peripheral visual field loss
Loss of peripheral vision with retention of central vision, resulting in a constricted circular tunnel-like field of vision.

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.

Retinal detachment
Separation of the inner layers of the retina (neural retina) from the pigment epithelium.

Posterior synechiae of the anterior chamber
Adhesions between the iris and the lens.

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

Encephalocele
A neural tube defect characterized by sac-like protrusions of the brain and the membranes that cover it through openings in the skull.

Macular coloboma
A congenital defect of the macula distinct from coloboma associated with optic fissure closure defects. Macular coloboma is characterized by a sharply defined, rather large defect in the central area of the fundus that is oval or round, and coarsely pigmented.

Optic disc pallor
A pale yellow discoloration of the optic disk (the area of the optic nerve head in the retina). The optic disc normally has a pinkish hue with a central yellowish depression.

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).

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 .

Color vision defect
An anomaly in the ability to discriminate between or recognize colors.

Exotropia
A form of strabismus with one or both eyes deviated outward.

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).

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.

Postural instability
A tendency to fall or the inability to keep oneself from falling; imbalance. The retropulsion test is widely regarded as the gold standard to evaluate postural instability, Use of the retropulsion test includes a rapid balance perturbation in the backward direction, and the number of balance correcting steps (or total absence thereof) is used to rate the degree of postural instability. Healthy subjects correct such perturbations with either one or two large steps, or without taking any steps, hinging rapidly at the hips while swinging the arms forward as a counterweight. In patients with balance impairment, balance correcting steps are often too small, forcing patients to take more than two steps. Taking three or more steps is generally considered to be abnormal, and taking more than five steps is regarded as being clearly abnormal. Markedly affected patients continue to step backward without ever regaining their balance and must be caught by the examiner (this would be called true retropulsion). Even more severely affected patients fail to correct entirely, and fall backward like a pushed toy soldier, without taking any corrective steps.

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

Chorioretinal atrophy
Atrophy of the choroid and retinal layers of the fundus.

Abnormality of the optic disc
A morphological abnormality of the optic disc, i.e., of the portion of the optic nerve clinically visible on fundoscopic examination.

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

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.

Hypoplasia of penis

Obesity
Accumulation of substantial excess body fat.

Hypogonadism
A decreased functionality of the gonad.

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.

Posterior subcapsular cataract
A type of cataract affecting the posterior pole of lens immediately adjacent to ('beneath') the Lens capsule.

Retinal pigment epithelial atrophy
Atrophy (loss or wasting) of the retinal pigment epithelium observed on fundoscopy or fundus imaging.

Abnormality of neuronal migration
An abnormality resulting from an anomaly of neuronal migration, i.e., of the process by which neurons travel from their origin to their final position in the brain.

Abnormal pupillary light reflex
An abnormality of the reflex that controls the diameter of the pupil, in response to the intensity of light that falls on the retina of the eye.

Abnormal macular morphology
A structural abnormality of the macula lutea, which is an oval-shaped highly pigmented yellow spot near the center of the retina.

Retinal pigment epithelial mottling
Mottling (spots or blotches with different shades) of the retinal pigment epithelium, i.e., localized or generalized fundal pigment granularity associated with processes at the level of the retinal pigment epithelium.

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.

Aplasia/Hypoplasia of the cerebellar vermis
Absence or underdevelopment of the vermis of cerebellum.

Rhegmatogenous retinal detachment
A type of retinal detachment associated with a retinal tear, that is, with a break in the retina that allows fluid to pass from the vitreous space into the subretinal space between the sensory retina and the retinal pigment epithelium.

Attenuation of retinal blood vessels

Undetectable electroretinogram
Lack of any response to stimulation upon electroretinography.

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.

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

Severely reduced visual acuity
Severe reduction of the ability to see defined as visual acuity less than 6/60 (20/200 in US notation; 0.1 in decimal notation) but at least 3/60 (20/400 in US notation; 0.05 in decimal notation).

Progressive night blindness

Constriction of peripheral visual field
An absolute or relative decrease in retinal sensitivity extending from edge (periphery) of the visual field in a concentric pattern. The visual field is the area that is perceived simultaneously by a fixating eye.

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.

Mild global developmental delay
A mild delay in the achievement of motor or mental milestones in the domains of development of a child.

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.

Delayed social development
A failure to meet one or more age-related milestones of social behavior.

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).

Granular macular appearance
Mottled (spotted or blotched with different shades) pigmentary abnormality of the macula lutea.

Retinal degeneration
A nonspecific term denoting degeneration of the retinal pigment epithelium and/or retinal photoreceptor cells.

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.

Unsteady gait

Abnormal corneal endothelium morphology
Abnormality of the corneal endothelium, that is, the single layer of cells on the inner surface of the cornea.

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.