Vision Syndromes

A syndrome is a group of symptoms that occur together and may affect the whole body or any of its parts. The following selected syndromes affect the eye.

Syndrome	General Characteristics	Ocular Involvement
Bassen-Kornzweig Syndrome	It is an autosomal recessive condition that more often affects males. It is caused by a defect in a gene that tells the body to create lipoproteins (molecules of fat combined with protein). The defect makes it hard for the body to digest fat and essential vitamins properly.	• The progressive degeneration of the retina that can advance to near-blindness. • Night blindness is an early and prominent symptom • Atypical retinitis pigmentosa • The macula may or may not be affected, while peripheral fields are often severely constricted. • Loss of photoreceptors continues throughout life.
Batten-Vogt-Mayou Disease (also called Speilmeyer-Batten-Vogt)	Batten disease is the juvenile form of a group of progressive neurological diseases known as neuronal ceroid lipofuscinoses (NCL). It is characterized by the accumulation of a fatty substance in the brain and in tissue that does not contain nerve cells. This disorder is inherited and is marked by rapidly progressive vision failure (optic atrophy) and neurological disturbances, which may begin before eight years of age. The disorder affects the brain and may cause both	• Development of pigment disturbances resembling retinitis pigmentosa • Progressive primary optic atrophy • Granular pigmentary change of macula (macular degeneration)

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	deteriorations of intellect and neurological functions.	
Borneville Disease (also called Tuberous Sclerosis)	This condition is inherited as autosomal dominant. It is characterized by adenoma sebaceum (large "blackheads"), central nervous system tumors, renal tumors, multiple lung cysts, seizures, and intellectual disabilities. This disease appears at birth or within the first few years and results in death during the teenage years.	• Retinal tumors
C.H.A.R.G.E. Syndrome	CHARGE stands for **C**oloboma, **H**eart defect, **A**tresia choanae (a congenital disorder where the back of the nasal passage or choana is blocked), **R**etarded growth and development, **G**enital abnormality, and **E**ar abnormality.	• A coloboma may be present in one or both eyes and can affect vision, depending on its size and location • Some people also have microphthalmia (abnormally small eyes) • CHARGE syndrome is the leading cause of congenital deafblindness
Cri-du-chat	Cri-du-chat is a chromosomal condition that results when a piece of chromosome 5 is missing. Infants with this condition often have a high-pitched cry that sounds like that of a cat. The disorder is characterized by intellectual disability and delayed development, small head size (microcephaly), low birth weight, and weak muscle tone (hypotonia) in infancy. Affected individuals also have distinctive facial features,	• Strabismus • Eyes spaced wide apart • Folds of skin over the eyelids

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	including widely set eyes (hypertelorism), low-set ears, a small jaw, and a rounded face.	
Crouzon Syndrome	Crouzon is a rare genetic abnormality, affecting 15 in every million babies. Although brain development is normal, the plates of the skull fuse before birth. With no space for the brain to grow further, pressure builds up often causing a string of problems: the tubes behind the nose are constricted, teeth are bunched up, the throat is squeezed causing reflux, and ear infections are common.	• The optic nerves swell and become damaged
de Grouchy Syndrome	A person who has de Grouchy will have a short stature, hypotonia (lack of muscle tone), hearing impairment, and foot abnormalities. Poor coordination, seizures, a small head, underdeveloped mid-face, a carp-shaped mouth, and autistic behavior are all characteristics of this disorder.	• Nystagmus (involuntary eye movement) • Deep-set eyes • Slanted spaces between the eyelids • Widely spaced eyes • Small eyes • Corneal abnormalities
Down Syndrome (Trisomy 21)	Characteristics of Down syndrome include small stature, flattened/round faces with an extra fold on the eyelids, saddle nose (a nose in which the bridge has an externally visible concavity and loss of height), thick lower lip, large tongue, an inflammatory skin disorder, smooth hair, obesity, small genitalia, short fingers, congenital heart anomalies and intellectual disabilities.	• Hyperplasia of iris • Narrow palpebral fissures (the opening between upper and lower eyelids) • High myopia • Strabismus • Cataracts • Grey spots on the iris

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Duane Syndrome	A congenital rare type of strabismus. Duane's is most commonly characterized by the inability of the eye to move outwards. While usually isolated to the eye abnormalities, Duane's syndrome can be associated with other problems including cervical spine abnormalities.	• Limitation of outward movement of the affected eye • Less marked limitation of inward movement of the same eye • Poor convergence • Often uses a head turn to the side of the affected eye to compensate for the movement limitations of the eye(s) and to maintain binocular vision
Edwards Syndrome (Trisomy18)	Edwards is characterized by intellectual disabilities and developmental delays, congenital heart defects, and renal abnormalities.	• Corneal and lenticular opacities • Unilateral ptosis (droopy eyelids) • Optic atrophy
Galactosemia Syndrome	This syndrome is characterized by allergy to milk; autosomal recessive inheritance pattern. If not identified and treated (withdrawal of milk products), can cause enlarged liver and intellectual disabilities.	• If untreated, cataracts develop
Gillespie Syndrome	Gillespie Syndrome is a rare genetic disorder in which there is absence of all or part of the iris (aniridia). Gillespie Syndrome is described to look like "a fixed dilated pupil." Systemic features frequently include hand tremors and hypertonia. There is usually some degree of developmental delay ranging from fine motor difficulties to intellectual disabilities.	• Visual acuity is usually about 20/60 which can be classified as reduced but functional vision • Nystagmus (involuntary eye movements) • Ptosis (droopy eyelids)

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Goldman-Favre Syndrome	This is an inherited eye disease. There are no general systemic manifestations associated with Goldman-Favre syndrome.	• Increased sensitivity to blue light • Varying degrees of red and green vision • Night blindness occurring from early life • Vision loss • Retinal degeneration
Graves' Disease (Hyperthyroidism)	Graves' is an endocrine and immune response problem that results in overproduction of thyroid hormones. It is sometimes referred to as hyperthyroidism. All body systems are affected. Some common symptoms include rapid heartbeat, shortness of breath, changes in eyes and vision, tremors and muscle weakness, anxiety, difficulty concentrating, and insomnia. This is most common in women.	• Lid retraction • Proptosis (abnormal protrusion or bulging of the eyeball) • Extraocular (outside of eyeball) muscle involvement • Corneal involvement • Optic nerve involvement
Hallerman-Streiff-Francois Syndrome	This is a rare genetic disorder in which the primary symptom is the malformation of the bones of the head and face. This leads to an abnormal facial structure involving the mouth, teeth, nose, and eyes. This can result in developmental problems affecting speech, normal chewing and swallowing patterns, and vision.	• Abnormally small eyes • Cataracts • Detached retinas developed by teen years
Hermansky-Pudlac Syndrome	This syndrome is characterized by a condition called oculocutaneous albinism, which causes abnormally light coloring (pigmentation) of the skin, hair, and eyes; problems with blood	• Oculocutaneous albinism, which causes vision problems such as nystagmus and photophobia

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	clotting; breathing problems due to a lung disease called pulmonary fibrosis; inflammation of the large intestine (colitis) and kidney failure.	• Vision problems usually remain stable after early childhood
Hurler Syndrome (Type I)	Hurler's is an autosomal recessive condition that is characterized by gargoylism, a thickened tongue, puffy cheeks, umbilical hernia, flat nose-bridge and intellectual disability.	• Corneal clouding • Buphthalmos (abnormally large eyeball) • Esotropia (eye deviates inward) • Slight ptosis (droopy eyelids) • Pigmentary retinopathy • Optic atrophy
Laurence-Moon-Bardet-Biedl Syndrome	This is an autosomal recessive condition that is characterized by obesity, intellectual disabilities, polydactyly, and hypogenitalism.	• Retinitis Pigmentosa
Lowe Syndrome	Lowe syndrome is an X-linked syndrome resulting in cerebral defects, intellectual disabilities, dwarfism, renal dysfunction and high early mortality rate. This occurs only in males.	• Congenital cataracts • Infantile glaucoma • Nystagmus
Marchesani Syndrome	This is an autosomal recessive syndrome that results in multiple skeletal abnormalities. Individuals are short and stocky with well-developed muscles, with hands and feet that are spade-shaped. Childhood x-rays show delayed carpal and tarsal ossification.	• Spherophakia (congenital bilateral anomaly in which the lenses of the eye are small, spherical and prone to subluxation • Ectopia lentis (partial displacement of the lens) which leads to lenticular myopia • Iridonesis (an iris that is not held in place by the lens) • Glaucoma (which resists treatment)

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		• Poor visual prognosis
Marfan Syndrome	Marfan's is an autosomal dominant, hereditary disorder. Marfan's is characterized by arachnodactyly (increased length of long bones especially in fingers and toes); scanty subcutaneous fat (relaxed ligaments); congenital heart disease; spine/joint deformities; high infant mortality rate.	• Dislocated lens (usually superiorly and nasally) • Severe refractive errors • Megalocornea (front third of the eye is larger than normal) • Cataracts • Uveal colobomas • Secondary glaucoma
Moebius Syndrome	This is an extremely rare congenital neurological disorder, which is characterized by facial paralysis due to underdevelopment of cranial nerves. Respiratory problems, speech and swallowing disorders, visual impairments, sensory integration dysfunction, sleep disorders, and weak upper body strength may also be present.	• Eye sensitivity due to inability to squint • Absence of lateral eye movement • Absence of blinking • Strabismus
Norrie Disease	This is a rare X-linked genetic disorder that causes abnormal development of the retina. About one third of individuals with Norrie's disease develop progressive hearing loss, and more than half experience developmental delays in motor skills such as sitting up and walking. Other problems may include mild to moderate intellectual disability, often with psychosis, and abnormalities that can affect	• Cataracts • Leukocoria (a condition in which the pupils appear white when light is shone on them) • Developmental issues in the eye, such as shrinking of the eyeball (phthisis bulbi)

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	circulation, breathing, digestion, excretion, or reproduction.	
Peters Plus Syndrome	This congenital condition is characterized by developmental delays, short stature, cleft lip, distinctive facial features, and abnormal development of the front part of the eye (Peter's anomaly). Most cases occur by chance, but it sometimes runs in families (autosomal dominant). A child may then "inherit" the condition from one parent.	• Central corneal opacity • Corneal thinning • Adhesions on the iris and cornea, which usually result in glaucoma • Frequently leads to amblyopia (suppression of visual information from the affected eye) • May be associated with cataracts and other lens abnormalities
Refsum Disease	This congenital, genetic disease affects neurological and metabolic processes that break down and eliminate certain fatty acids and toxic substances from the body. It results in Spinocerebellar ataxia, deafness and polyneuritis. It is also characterized by retinitis pigmentosa, anosmia (loss of sense of smell), and neuropathy in the hands and feet. The condition is progressive, and symptoms may not appear until middle age.	• Retinal degeneration
Reiter's Syndrome	Reiter syndrome is a type of reactive arthritis that happens as a reaction to a bacterial infection in the body. The infection usually happens in the intestines, genitals, or urinary tract. Reiter syndrome includes joint swelling and pain, often in knees, ankles, and feet, along with inflammation of the eyes and urinary tract. It is	• Conjunctivitis (inflammation of the eye) • Iritis (inflammation of the iris of the eye that needs immediate treatment to avoid eye damage)

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	not contagious, but the bacteria that trigger it can be passed from one person to another.	• Uvieitis (inflammation of the inner eye that needs immediate treatment to avoid eye damage)
Rubella Syndrome	Rubella syndrome is caused by maternal infection of the fetus resulting in heart abnormalities, vision and hearing defects and sometimes-intellectual disabilities.	• Bilateral cataracts • Uveal colobomas • Searching nystagmus • Microphthalmus • Strabismus • Retinopathy • Glaucoma
Scheie Syndrome	Scheie syndrome is a condition characterized by corneal clouding, facial abnormalities and normal lifespan. Also known as "MPS I-S"; it is a less severe version of Hurler syndrome.	• Corneal clouding
Septo-Optic Dysplasia (SOD, DeMorsier's Syndrome)	SOD is a rare disorder, present at birth, in which the optic nerve is underdeveloped, the pituitary gland does not function properly, and often a portion of the brain tissue is not formed (septum pellucidum). Some children with SOD have normal intelligence, while others may be developmentally delayed, learning disabled, or intellectually disabled. Some symptoms of this disorder can be treated, but the visual impairment usually cannot be corrected.	• Optic nerve hypoplasia (underdevelopment of the optic nerve) • Hypoplasia is generally manifested by nystagmus (involuntary eye movements, often side-to-side) and a smaller-than-usual optic disc • The degree of visual impairment is variable, and ranges from near normal vision to complete blindness
Stevens-Johnson Syndrome	SJS is a rare, life-threatening disorder caused by a reaction to medication, infection, or food. A rash with blisters and lesions on the skin and mucus	• Purulent conjunctivitis • Occlusion of lacrimal gland ducts, which produces a "dry eye"

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	membranes develops, which damages the eyes and causes the top layer of the skin to separate and die.	• Corneal ulcers • Panophthalmitis (infection in all structures of the eyeball)
Still's Disease (Juvenile Rheumatoid Arthritis & Systemic-Onset Juvenile Idiomatic Arthritis)	This is an inflammatory disorder characterized by arthritis (especially in the knee joints), high spiking fever, and intermittent rash. Also, inflammation of the lungs, lymph glands, ocular tissues, and around the heart may occur. Ocular complications occur three times as often in girls.	• Chronic uveitis • Cataracts • Secondary glaucoma • Band-shaped keratopathy (corneal abnormality)
Sturge-Weber Syndrome	This is an autosomal dominant condition characterized by port-wine-stain tumor on one side of the face, central nervous system disorders and seizures. There is no treatment and it often results in death by age thirty.	• Unilateral infantile glaucoma on the effected side which appears at birth • Choroidal tumor
Tay-Sachs Disease	Tay-Sach's is an autosomal recessive syndrome that results in mental and physical deterioration in the first two to three years of life. Death occurs shortly after.	• Cerebromacular degeneration • Degeneration of the inner layer of the retina • Retinal pigmentary changes • Optic atrophy • "Cherry red" spot in macula
Turner Syndrome	This syndrome affects females. It is characterized by retarded growth, rudimentary ovaries in female genitalia, and amenorrhea (absence of menstruation).	• Pterygium (wedge-shaped growth that may affect conjunctiva cornea) • Epicanthus (vertical skin fold on either side of the nose • Ptosis (droopy eyelids) • Color blindness.

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Usher Syndrome	Usher's Syndrome is a genetic disorder that causes abnormalities of the inner ear and retinal degeneration. It is characterized by some degree of congenital hearing loss and progressive visual impairment. Vision usually deteriorates in the teen years.	• Retinitis Pigmentosa
WAGR Syndrome	This is a rare genetic syndrome in which affected children are predisposed to develop Wilm's tumor (a tumor of the kidneys), Aniridia (absence of the colored part of the eye, the iris), Genitourinary anomalies, and mental Retardation (intellectual disability)	• Absence of the iris which can cause reduction in the sharpness of vision (visual acuity) and increased sensitivity to light (photophobia) • Cataracts • Glaucoma • Nystagmus
Wilms Tumor	Wilms tumor results in malignancy of the kidney, small stature, and developmental delays. This often accompanies aniridia.	• Total or partial absence of the iris • Cataracts • Ptosis • Decreased visual acuity • Photophobia • Nystagmus
Wilson's Disease	This is a recessive inheritance pattern, which includes neurological symptoms, cirrhosis of the liver, and faulty renal function. It is linked to defects in copper metabolism.	• Pigment ring in periphery of cornea (progressive, but rarely occludes the pupil) • Sometimes cataracts
Zellweger Syndrome	This is one of a group of diseases which are caused by defects in Peroxisomes which are required for normal brain development and function and the formation of myelin, the whitish	• Glaucoma • Retinal degeneration

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	substance that coats nerve fibers. Peroxisomes are also required for normal eye, liver, kidney and bone functions.	