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Review

Ocular Syndrome and their Systemic Association

***[1]Samia Iqbal, [1]Anwaar ul haq Hashim, [1]Muhammad Mujahid, [1]Farah Tariq, [1]Aisha Azam and [2]Saleem Ullah**

[1]The University of Lahore teaching hospital.

[2]Mayo hospital Lahore

***Corresponding Author E-mail: samiaiqbal988@gmail.com**

Abstract

Coexistence of several ocular syndromes is more frequent than suspected. In spite of the ocular syndrome prevalence, one or more of the following can be detected simultaneously: Down syndrome, Goldenhar syndrome, Duane Retraction syndrome, Brown syndrome, and Mobius syndrome. In addition, as people age, ocular comorbidities are much more usually seen. Specific diseases are openly acknowledged to affect the eyes and vision, such as systemic diseases and congenital neurodegenerative disorders, hematologic malignancies, and/or systemic infections. Recent advances in early diagnosis and therapy of the ophthalmic syndrome and their systemic association have reinforced patient options to prevent visual impairment and blindness. Because of this, it is essential not to overlook sight-threatening conditions such as the ocular comorbidities and/or the eye involvement in the context of systemic disorders. Moreover, the important role of the multidisciplinary cooperation to improve and sustain management of patients affected with eclectic ocular comorbidities and/or systemic disorders with eye repercussion is specifically addressed. This study intends to shed light on these topics to help in making opportune diagnosis and appropriately managing the affected patients.

Keywords: Ocular syndrome, systemic association, infection, mobius syndrome, blindness.

INTRODUCTION

Achoo Syndrome

Ocular features include photic sneeze reflex by sudden exposure of dark-adapted subject to bright light. Systemic features include continous sneezing.

Achard Syndrome

Ocular features include aphakia myopia, lens dislocation. Various systemic features include arachnodactyly, high-arched palate, mandibulofacial dysostosis, skeletal anomalies, and heart condition.

Dental-Ocular-Cutaneous Syndrome

Ocular features are glaucoma (juvenile type) entropion lower eyelids.

Systemic features include unusual upper lip with lack of “cupid’s bow” and thickening and widening of the philtrum, syndactyly, cutaneous hyperpigmentation overlying the interphalangeal joints, clinodactyly, single conical roots altogether primary teeth and permanent first molars, scant hair, horizontal ridgingof fingernails (Hassan et al., 2018).

Dialinas-Amalric Syndrome

Ocular features aren’t any nyctalopia but heterochromia iridis, atypical retinitis pigmentosa with small, scattered, fine-pigmented deposits within the macular region with some accumulations and accompanied by small white and yellow spots. Systemic features include deaf mutism.

Diffuse Unilateral Subacute Neuroretinitis Syndrome

Ocular features are vitritis, gray-white lesions of the retina,optic atrophy, papillitis, retinal vessel narrowing, diffuse pigment epithelial degeneration, endophthalmitis, nematode within the fundus, the pathognomonic finding in DUSN is that the presence of a motile intraocular nematode. Systemic features include weight loss, lack of appetite, cough, fever, pulmonary infiltration, hepatomegaly, leukocytosis, persistent eosinophilia.

Dollinger-Bielschowsky Syndrome

Ocular features are optic nerve atrophy, macular pigmentation. Systemic features are cerebrospinal degeneration, cerebellar ataxia, defective hearing, convulsions, spasticity, contractures, and progressive mental deterioration.

ACL Syndrome

Ocular features include bilateral corneal leukoma, keratitis. Systemic features are unusually tall, large hands, feet and chin, skin of hands very soft, skin of scalp lies in folds, frontal bosses, ear calcification, pituitary tumors, abnormal dermal ridge patterns, enlargement of supra orbital arch of os frontale.

Aarskog's syndrome

Ocular symptoms include hypertelorism, antimongoloid palpebral fissures and megalocornea. Systemic features are short stature, syndactyly. Doyne Honeycomb Choroiditis (Dominant Orbruch Membrane Drusen, Holthouse-Batten Superficial Choroiditis, Hutchinson-Tays Central Guttate Choroiditis, Malattia-Leventinese Syndrome) Ocular features include drusen with multiple yellow lesions becoming calcified and presenting crystalline appearance.

Duane Syndrome

Ocular features include narrowing of palpebral fissure on adduction, widening on abduction, primary global retraction, deficiency of medial and lateral recti motility, limitation of abduction within the affected eye usually is complete and retraction of the planet with attempted adduction varies from 1 to 10 mm, convergence insufficiency, heterochromia irides, left eye ismore frequently involved. It is further divided into four types according to the movement. Systemic features are associated Klippel-Feil syndrome; malformation of face, ears, and teeth.

Aberfeld Syndrome

Ocular features include exotropia, myopia, congenital cataracts blepharophimosis, and microcornea. Systemic features are myopathy, arachnodactyly, dwarfism, hypoplastic facial bones, hypertrichosis bone deformities, kyphoscoliosis.

Duck-Bill Lips and Ptosis

Ocular features are ptosis, strabismus, and hypertelorism. Systemic features are short philtrum, duck-bill lips, low-set ears, a broad forehead, slightly anteverted nose, and flat nasal bridge, slightly wide-spaced teeth and high arched palate, slightly receding chin, slightly wide-set nipples, two phalanges in both fifth fingers, impaired speech.

Acosta Syndrome

Ocular features include acute blurred vision, and decreased acuity difficulties in color discrimination, impaired adaptation, retinal hemorrhage, chronic lid edema, bluish scleral injection. Systemic features are restlessness and irritability, headaches, impaired judgment at approximately 15,000 feet, confusion, cyanosis, muscular in coordination and possible loss of consciousness at approximately 18,000–20,000 feet, exertional dyspnea, epistaxis, gum bleeding, hemoptysis, anorexia, nausea, vomiting, tinnitus, cough, loss of libido, paresthesia extremities, coma, clubbing of fingers and hepatosplenomegaly.

Acroreno-ocular Syndrome

Ocular features include coloboma of optic nerve, complete coloboma, ptosis and Duane anomaly. Systemic features are renal anomalies, hypoplasia of distal a neighborhood of thumb with lack of motion at phalangeal joint, renal ectopia without fusion, bladder diverticula, malrotation of both kidneys, absence of kidney, clubhand or absence of thumb.

Aicardi's Syndrome

Ocular features include chorioretinal lacunar defects and colobomata. Systemic features are corpus callosal agenesis and other CNS abnormalities, infantile spasms, retardation, vertebral and rib malformations.

Alagille's Syndrome

Ocular features are pale fundi, posterior embryotoxon, and blind spot drusen hypertelorism. Systemic features are intrahepatic common bile duct hypoplasia, butterfly vertebrae, congenital heart disease.

Allgrove Syndrome

Ocular features are distichiasis, conjuncitivitis, keratitis, congenital alacrima. Systemic features are adreno corticotropic hormone (ACTH) insensitivity, achalasia.

Alport syndrome

Ocular features are anterior lenticonus, anterior polar and cortical cataracts, fleck retina. Systemic features are sensorineural deafness, nephritis.

Alstrom-Olsen syndrome

Ocular features are cone-rod dystrophy with features of retinitis pigmentosa, posterior subcapsular cataracts. Systemic features include DM, sensorineural deafness, nephropathy, obesity, acanthosis nigricans.

Amendola Syndrome

Blisters around eyebrows, entropion, ectropion, trichiasis, iritis. Systemic features are fevers and chills.

Ankyloblepharon Filiforme Adnatum and congenital anomaly Syndrome (AEC Syndrome, Hay-Wells Syndrome).

Ocular features are a filiform fusion of eyelids, pterygium, keratoconus. Systemic features include harelip and palate, paramedian mucous pits of lower lip, ectodermal dysplasia, infrequent association with trisomy 18, partial-thickness fusion of a central portion of lid margins.

Annette von Droste-Hulshoff Syndrome

Ocular features are myopia, detachment of the retina, negative and positive angle kappa, esotropia, temporal macular ectopia, chorioretinal colobomata, falciform folds, persistent hyaloids artery, abnormal position of the blind spot, epicanthus, telecanthus, blepharophimosis, hypertelorism, asymmetrical orbits, exophthalmos, enophthalmos. Systemic features include face turn, angioma, neoplasia.

Anoxic Overwear Syndrome

Ocular features include refractive error changes, endothelial cell changes, physical trauma to the anterior surface of the cornea, corneal neovascularization, giant papillary conjunctivitis, contact deposits, acute red eye syndrome (George et al., 2018).

Apert Syndrome

Various ocular features found are hypertelorism, proptosis, strabismus, keratoconus, ectopia lentis, congenital glaucoma, optic atrophy. Systemic features include craniosynostosis, syndactyly, broad distal phalanx of great thumb/toe, mental handicap.

Anterior Chamber Cleavage Syndrome (Peters-Plus Syndrome, Reese-Ellsworth Syndrome)

Ocular features include increased pressure , adhesions between the iris and cornea, persistence of mesenchymal tissue within the chamber angle, usually shallow anterior chamber, iris coloboma and hypoplasia, prominent Schwalbe ring, contiguous membrane, corneal opacities of various density with or without edema, usually at the situation of iris adhesion, anterior pole cataract, remains of hyaloid artery. Systemic features include dental anomalies, retardation, congenital anomaly, syndactyly, craniofacial dysostosis and myotonic dystrophy.

Balint Syndrome (Psychic Paralysis of Visual Fixation Syndrome)

Ocular features include psychic paralysis of visual fixation, lack of full voluntary control of eye movements, unstable visual fixation. Systemic features include tonic and motor phenomena of upper limbs, loss of body coordination (bilateral), optic ataxia, it's been reported to occur in association.

Baraitser-Winter Syndrome

Ocular features include ptosis, hypertelorism, down-slanting palpebral fissures. May be confused with Noonan syndrome, phenotypic features appear to be variable.

Laron Syndrome

Ocular features include microphthalmia, reduced retinal

vascularization, optic nerve hypoplasia, pseudo papilledema. Systemic features are short stature, abnormally small extremities, subnormal head circumference, increased bodyfat and delayed sexual development (Przeździecka-Dołyk et al., 2020).

Larsen Syndrome

Ocular features are hypertelorism, bilateral chronic keratitis, corneal neovascularization, lower lid entropion. Systemic features include frontal bossing, depressed nasal bridge, flat face, flat and broad thumbs, skeletal dysplasia with multiple joint dislocations, unusual faces, long, cylindrical fingers, spatulate thumbs, dental abnormalities, cardiac defects, hydrocephalus, laryngo tracheomalacia, dislocation of the cervicalspine, trachea malacias, heart condition, severe respiratory infection, clubfeet, multiple joint deformities, hydro cephalus, tracheal stenosis.

Barrier Deprivation Syndrome

Various ocular features found are cystoid macular edema, corneal endothelialdystrophy, detachment of the retina, leakage inperipheral retina and macula, iris pigment loss, uveitis, vitreous in anterior chamber, retinal holes, band keratopathy, glaucoma, iritis (Przeździecka-Dołyk et al., 2020).

Basedow Syndrome

Ocular features include exophthalmos, swelling of eyelids and discoloration of upper eyelids, lid lag (von Graefe), globe lag (Koeber), lid trembling on gentle closure (Rosenbach sign), reduced blinking (Stellwag), retraction of upper lid, difficulty in everting upper lid (Gifford sign), convergence weakness (Möbius), impaired fixation on extreme lateral gaze (Suker), possible external ophthalmoplegia (Ballet), Dalrymple sign (staring appearance), tearing, photophobia, epiphora, prolapse of lacrimal gland, neuroretinal edema, tortuous vessels, papilledema and papillitis, anisocoria, keratitis, increased pressure, increased intraocular pressure on upgaze, decreased visualacuity, enlargement of the extraocular muscles, increased volume of the extraorbital fat, superiorrectus muscle enlargement, decreased venous out flow. Systemic features include tachycardia, anxiety, insomnia, loss of weight, hyperhidrosis, restlessness, myocarditis (toxic), atrial fibrillation.

Laurence–Moon Syndrome

Grouped with Bardet–Biedl syndrome but no obesity or polydactyly.

Leber's Congenital Amaurosis

Systemic features include autosomal recessive, Ocular features are blindness from birth, eyepoking (oculodigital sign), hypermetropia, sluggish or paradoxical pupillary reflexes, macular dysplasia but fairly normal fundus appearance (Couser et al., 2017).

Anton Syndrome (Denial-Visual Hallucination Syndrome)

Ocular features include denial of blindness, patients may persistently deny having any loss of beholding, and the objects the patient describes and claims to ascertain are considered visual hallucinations, field of vision hemianopsia. Systemic features include confabulation, allocheiria (reference of a sensation is formed to the other side to which the stimulus is applied).

Arylsulfatase A Deficiency (Van Bogaert-Nyssen-Peiffer Disease)

Ocular features are Visual loss in association with optic atrophy, strabismus, macular cherry-red spot, corneal opacification, oculomotor disorders (nystagmus, strabismus), nervus opticus and retinal demyelination. Systemic features include motor and mental deterioration with spasticity, paralysis, seizures, dementia, death in infancy, although attenuated and adult sorts of the disease occur, schizophrenia, temporo occipital demyelination, unreactive to visual and auditory stimuli, adult form: moodiness, withdrawal, megalomania, hallucinations, violent reactions and dementia.

Autoimmunologically Mediated Syndrome

Ocular features include dacryoadenitis.

Arnold–Chiari malformation

Systemic features include congenital herniation of the cerebellum/ brainstem through the foramen may cause hydrocephalus, cerebellar signs (e.g., ataxia) and should be related to syringomyelia. Ocular feature is nystagmus.

Axenfeld-Schürenberg Syndrome (Cyclic Oculomotor Paralysis)

Ocular features include cyclic oculomotor paralysis

(paralysis alternating with spasm), during times of paralysis, lid exhibits ptosis and affected eye is abducted, during spasm, lid is raised, deviation of affected eye is either inward or outward, and pupil is fixed and contracted.

Bardet–Biedl and Laurence–Moon Syndromes

Ocular features include retinitis pigmentosa with early macular involvement, polydactyly, hypogonadism, obesity, microcephaly and nephropathy.

Bassen–Kornzweig Syndrome (Abetalipo proteinaemia)

Ocular features are retinitis pigmentosa, cataract, spinocerebellar degeneration, steatorrhoea, and acanthosis (of erythrocytes).

Batten-Mayou Syndrome (Batten Disease, Vogt-Spielmeyer Syndrome)

Ocular features include vision initially reduced, getting to totalblindness, fat deposition within the retina with gradual development of pigment disturbances resembling retinitis pigmentosa, progressive primaryoptic atrophy, granular pigmentary change of macula, there's clinical evidence supporting the concept the first lesion of the retina is in the inner layers. Systemic features include mental disturbances, convulsions (later), apathy, irritability, ataxia, upper and lower motor neuronpalsies, rigidity, complete paralysis adementiain terminal stage, hypertonus, death from intercurrent infection (Borrego-Sanz et al., 2019).

Arndt-Gottron Syndrome (Scleromyxedema)

Ocular features include corneal opacities of amyloid deposits, thickening of eyelids, lagophthalmos, ectropion, thickened eyebrow or eyelid skin, corneal opacities. Systemic features include exaggerated facial folds impair opening of the mouth, flexion contractures from poor joint mobility, erythema, scaling of skin, phimosis, urethral stenosis.

Axenfeld-Rieger Syndrome (Axenfeld Syndrome)

Ocular features include posterior embryotoxon: ring-like opacity of cornea, long trabecula, prominent Schwalbe line, iris adhesions to Schwalbe line and cornea with large abnormal iris processes or broad sheets of tissues of varying size and site, anterior layer of iris may appear hypoplastic, ectopia of the pupil not uncommon, polycoria occurs, ring like opacity of the deep corneal layers extending several millimeters from the limbus in continuity with the sclera, keratoconus.

BBB Syndrome (Hypertelorism-Hypospadias Syndrome, Opitz Syndrome)

Ocular features include epicanthal folds, strabismus, blepharophimosis, telecanthus, widely spaced eyebrows. Systemic features are high nasal bridge, hypospadias, cryptorchidism, cleft palate and lip, urinary mal formations, mental retardation, osteochondritis dissecans, congenital heart defects, upper urinary tractanomalies.

Benson Disease (Asteroid Bodies of the Vitreous, Asteroid Hyalitis, Scintillatio Albescens, Snowball Opacities of the Vitreous)

Ocular features include small, solid, stellate, spherical bodies in another wise normal vitreous, creamy, flat white,or shiny when viewed with an ophthalmoscope,may interfere with accurate measurement of axial length. Systemic features are the increased prevalence of DM, hypertension, atherosclerosis, and hyperopia (Brydak-Godowska et al., 2017).

Blepharophimosis Syndrome (Simosa Syndrome)

Ocular features are scarred or contracted in secondary blepharophimosis because of ocular pemphigus or trachoma, ectropion, epicanthus inversus, lacrimal puncta displacement, ptosis, telecanthus, optic nervecoloboma, angle dysgenesis, nervus opticus hypoplasia, amblyopia, strabismus. Systemic features include low-set ears, low nasal bridge.

Blocked Nystagmus Syndrome

Ocular features are bilateral or monocular convergence where theadducted eye(s) can't be abducted to the midline, if monocular, it's going to alternate, esotropia increases with prolonged fixation, head turn, nystagmus.

Bogorad Syndrome (Crocodile Tear Syndrome, Paroxysmal Lacrimation Syndrome)

Ocular features include unilateral lacrimation while eating or drinking due to misdirected nerve fibre regeneration. Systemic features include excessive salivation

(occasionally), diffuse facialmuscle response, or facial contracture with lacrimation.

Bloch–Sulzberger Syndrome (Incontinentia Pigmenti)

Ocular features are abnormal peripheral retinal vasculature, gliosis, tractional detachment of the retina. Systemic features are abnormal teeth, cutaneous pigment whorls, and CNS anomalies.

Albers-Schonberg Disease

Ocular features are, ptosis, exophthalmos, papilledema, nystagmus, anisocoria, congenital cataracts, oculomotor paralysis, nerve VII (facial) palsy, optic atrophy hypertelorism, visual loss in infancy, duct obstruction, keratoconus. Systemic features are cartilage and bone thickening, multiple fractures, hyperchromic anemia, osteomyelitis, severe forms: jaundice, hepato splenomegaly, skeleton sclerosis, lymphadenopathy and hydrocephalus in infants, mild forms: neurological disorder, fractures and milder kind of anemia, pancytopenia from marrow obliteration, low serum calcium and elevated phosphorus.

Albright syndrome

Ocular features are orbital involvement may cause proptosis, sinus mucoceles, and compressive optic neuropathy. Systemic features are polyostotic fibrous dysplasia (of bone), endocrine abnormalities (including precocious puberty), and cafe-au-lait spots.

Bourneville Disease (Tuberous Sclerosis)

Ocular features are retinal astrocytomas,

Brown Syndrome

Ocular features include mechanical restriction syndrome attributed to the superior oblique tendon sheath,.

Canalis Opticus Syndrome

Ocular features are spontaneous unilateral or bilateral, reversible orirreversible amaurosis, absent pupil reaction incases of complete blindness, spontaneous visualrecovery has been reported anecdotally. Usually related to blunt head injury.

Canine Tooth Syndrome (Class VII Superior Oblique Palsy)

Ocular features include underaction of the superior oblique and underactionof the inferior oblique on an equivalent side.

Capsular Bag Distension Syndrome

Ocular features include shallow anterior chamber, pupil peaking, and accumulation of turbid fluid within the capsular bag.

Capsular Block Syndrome

Complete sealing of the anterior capsule openingby the optic and displacement of the posteriorcapsule far behind the posterior optic surface.

Cestan-Chenais Syndrome [Cestan (1) Syndrome]

Ocular features include enophthalmos, ptosis, nystagmus, miosis.Systemic features include pharyngolaryngeal or glossopharyngeal paralysis, cerebellar hemiataxia, disturbance of sensibility, contralateral side of lesion.

Charge Association (Atresia, coloboma, ear malformation association, genital hypoplasia, heart condition, multiple congenital anomalies syndrome, retarded growth)

Ocular features include blepharoptosis, iris coloboma, nervus opticus coloboma, macular hypoplasia, lacrimal canalicularatresia, duct obstruction. Systemic features are microcephaly, brachycephaly, malformed ear, bilateral finger contractures, heart condition, genitalhypoplasia, heart condition, choanal atresia, retarded growth, deafness , facial palsies, mental retardation.

Cockayne Syndrome (Dwarfism with Retinal Atrophy and Deafness, Mickey Mouse Syndrome)

Ocular features are enophthalmos, cataracts, pigmentary degenerationof the retina, optic atrophy, band keratopathy, exotropia, nystagmus, absence of foveal reflex, corneal dystrophy, corneal perforation, anhidrosis, exposure keratitis, decreased blinking. Systemic features include dwarfism (nanism) with disproportionately

longlimbs, large hands and enormous feet, kyphosis, deformed limbs, thickened skull, intracranial calcifications, retardation, prognathism, deafness (often partial), precociously senileappearance, sensitivity to sunlight, with skinpigmentation and scarring, cavity .

Cogan's Syndrome

Ocular features include interstitial keratitis reduced vision mainly with the involvement of center of the cornea, very fine wavy lines resembling fingerprints within or very on the brink of corneal epithelium and best seen on biomicroscopy with retroillumination, fine grayish spheres (0.1–0.5 mm diameter) in superficial corneal epithelium, maplike irregular border-lined slightly grayish area. Systemic features are sensorineural deafness, tinnitus, vertigo, systemic vasculitis (including life-threatening aortitis).

Computer User Syndrome

Various ocular features found are ocular pain, asthenopia, excyclotorsion, depressionof gaze, ocular synkinesis.Systemic features include hand-wrist pronation, ulnar abduction, headache, fatigue, various sorts of head and shoulderdistress, carpal tunnel syndrome.

Congenital Dyslexia Syndrome (Attention deficit disorder, congenital visual aphasia congenital visual aphasia of hermann, developmental dyslexia of critchley, dyslexia syndrome, minimalbrain dysfunction syndrome, primary dyslexia)

Ocular features include abnormal optokinetic nystagmus, metamorphopsia, defective chromatic vision, convergence insufficiency, muscle imbalance, refractive errors, low accommodative converge/accommodation related to decreased visual acuity and contrast sensitivity. Systemic features are general clumsiness, disorientation (time-space, right-left), behavioral changes, lack of integrationof visual and auditory stimuli.

De Morsier's Syndrome

Ocular features include optic nerve hypoplasia, blepharitis, keratoconus and cataracts. Systemic features are midline brain abnormalities including absent septum pellucidum and corpus callosal hypo/ aplasia. Mongolism mongolism, 1 in 650 live births, musculoskeletal abnormalities, congenital heart disease.

Dejean Syndrome (Orbital Floor Syndrome)

Ocular features are enophthalmos, exophthalmos, lid hematoma,diplopia because of displacement of the planet orrestricted function of the inferior rectus muscle muscle and/orinferior oblique muscles, orbital emphysema.Systemic features include severe pain in superior maxillary region,numbness in area of first and second branchesof trigeminal, nausea, and vomiting.

CAR Syndrome (Cancer-Associated Retinopathy Syndrome)

Ocular features include vision loss usually progressive, retinal degeneration, retinal whole, abnormal visual fields, loss ofcolor vision, detachment of the retina, optic atrophy, ring-like scotoma, nyctalopia, retinal phlebitis. Systemic features are carcinoma with or without metastasis to any partof the body.

Cebocephalia

Ocular features are hypotelorism, mongoloid obliquity.Systemic features are flat, incomplete nose, full cheeks, medial nostril, no palate or cleft lips.

Central systema nervosum Deficiency Syndrome (GarlandSyndrome, Spillan-Scott Syndrome)

Various ocular features found are greatly reduced vision, particularly sight, increasing over weeks or months but rarelyprogressing to finish blindness, relative orabsolute central or paracentral scotomata, bitemporalpallor of the disks, optic neuropathy. Systemic features include incomplete bilateral deafness, never proceeding to complete deafness, tinnitus, numbness and tingling within the legs, rarely within the hands, unsteadinessof gait, abnormal tendon reflexes (bothhyper active or absent), peripheral neuropathy.

Cerebro-Oculo-Facio-Skeletal Syndrome (COFS Syndrome)

Ocular features are microphthalmia, blepharophimosis and cataracts. Systemic features include microcephaly, hypotonia, prominent nasal root, large ear pinnae, flexion contractures at elbows and knees, camptodactylia, osteoporosis, kyphosis, scoliosis, congenital dystrophy.

East-West Syndrome

Ocular features include glare, halos, monocular diplopia, irregular pupil microphthalmos, glaucoma, cataracts.Systemic features include Edwards' syndrome Trisomy 18, 1 in 8000 live births, failure to thrive, congenital heart disease, anticipation.

Gapo Syndrome

Ocular features are progressive optic atrophy, glaucoma and keratoconus. Systemic features include growth retardation, alopecia, pseudoanodontia, frontal bossing, high forehead, midfacial hypoplasia, wide-open fontanelle, retardedbone age, premature aged appearance, hypogonadism, hepatomegaly, muscular body build (Kordyś et al., 2018).

Lenoble-Aubineau Syndrome (Nystagmus-Myoclonia Syndrome)

Ocular features include congenital nystagmus related to fasciculationsof muscles spontaneously elicited by mechanical stimulation or cold. Systemic features include tremors of head and limbs, myoclonic movements of extremities and trunk, hypospadias, abnormalities of teeth, facial asymmetry, localize dedema. Löfgren syndrome: presentation of sarcoidosis with fever, erythema, bihilar lymphadenopathy.

Gardner's Syndrome

Ocular features are exophthalmos, congenital hypertrophy of retinalpigment epithelium (RPE), multiple lesions ofthe eye, bilateral occurrence, orbital osteoma, highly pleomorphic pigmentation, unilateralor bilateral retinal lesions, pilomatrixoma-like epidermal cysts, presence of pigmented funduslesions appears to cluster within families. Systemic features include intestinal polyps, dermoid tumors, neurofibrous osteomatosis, carcinoma, supernumerary teeth.

General Fibrosis Syndrome

Ocular features are ptosis, enophthalmos, disk hypoplasia, astigmatism, esotropia, exotropia, hypotropia, nystagmus, visual loss, positive forced duction test, could even be associated with Marcus Gunn jaw-winkingand synergistic divergence in attempted rightgaze.

Majewski Syndrome

Ocular features include cataract, blind spot edema, optic atrophy, hypertelorism, absent lashes and brows, persistentpupillary membrane. Systemic features include short rib polydactyly, cleft lip, birth defect, narrowthorax, short tibia, hypoplastic epiglottis, lung and visceral abnormalities.

Gerstmann's Syndrome

Ocular features include dominant lobe lesion resulting in finger agnosia, right/left confusion Macular cherry-red spot, corneal clouding, cerebromacular degeneration., Systemic features include dysgraphia, acalculia, could even be associated with failure of ipsilateral pursuit movements. Dwarfism, gargoyle facies, retardation, seizures, hearing disorder.

Gillespie Syndrome

Systemic features are retardation and cerebellar ataxia. Ocular features include congenital cataracts, incomplete formation of iris, bilateral congenital mydriasis.

Goldenhar Syndrome (Goldenhar-Gorlin Syndrome, Oculoauriculovertebral Dysplasia)

Ocular features include anophthalmia, colobomata of the choroid, iris andeyelid, antimongolian slant of lid fissure, epibulbardermoid, or lipodermoids of the conjunctiva, cornea and orbit, tilted blind spot, nerve hypoplasia, microphthalmia, macular heterotopia, tortuous retinal vessels. Systemic features include frontal bulging of the skull, receding chin, malarhypoplasia, micrognathia and macrostomia, auricular appendices (single or multiple), multiple vertebral anomalies, preauricular fistulas, mental retardation.

Louis–Bar Syndrome

Ocular features are progressive oculomotor apraxia. Systemic features include (ataxia telangiectasia), conjunctival telangiectasia, cerebellar ataxia, Decreased IQ, immunodeficiency.

Lower syndrome

Ocular features are (oculocerebrorenal syndrome), congenital cataract, microspherophakia, bluesclera, Systemic features include anterior segment dysgenesis, glaucoma, dIQ, hypotonia, vitamin D–resistant rickets. X-linked disorder of aminoalkanoic acid metabolism (Kordyś et al., 2018).

Goldmann–Favre disease

Systemic features areautosomal recessive, Ocular features are optically empty vitreous, macular retinoschisis, macular changes, peripheral pigmentary retinopathy.

Good Acuity Plus Photosensitivity (Gapp), Track Related Iridiocyclitis and Scleritis (Trisc), Transient Light Sensitivity (Tls)

Ocular features are photophobia, glare, uveitis, iridocyclitis, scleritis associated with refractive surgery and theuse of Intralase technology starts 6–8 weekspostoperatively and resolves by 4–5 months.

Gorlin's syndrome

Systemic features include multiple basal cell carcinomas, jaw cysts, skeletal abnormalities, ectopic calcification (e.g., falx cerebri). Ocular features include hypertelorism, prominent supraorbital ridges.

Gradenigo's syndrome

Ocular features include VI nerve palsy and pain in V nerve distribution because of lesion at the apex of the petrous temporal bone, Systemic features include chronic middle ear infection.

Grayson-Wilbrandt Syndrome (Corneal Dystrophy of Reis-Buecklers, Reis-Buecklers Syndrome)

Ocular features include corneal changes variable from a mottled scarring to gray macular opacities of the anterior limiting membrane of the cornea, strabismus.

Greig Syndrome (Hypertelorism, Hypertelorism Ocularis, Ocular Hypertelorism Syndrome, Primary Embryonic Hypertelorism)

Ocular features are hypertelorism (wide spacing of orbits), enophthalmos, epicanthus, deformities of eyelids andbrows, defects of the palpebral fissure, bilateral sixth nerve paralysis, esotropia, astigmatism, optic atrophy by tension on the opticnerve, strabismus. Systemic features arethat skull may show mild malformations, including bitemporal eminences and decreased anteroposterior diameter, harelip, high-arched palate, cleft palate, broad and flat nasal root and mental impairment.

Behçet Syndrome

Ocular features include muscle palsies (occasional), nystagmus (occasional), conjunctivitis, hypopyon, iritis, recurrentuveitis, keratoconjunctivitis sicca, keratitis, vitreoushemorrhages, thrombophlebitis retinal veins (occasional), retinal hemorrhages, optic neuritis (occasional), macular edema, optic nerveatrophy, retinitis, secondary glaucoma, retinalvasculitis, disk edema, panophthalmitis, optic neuropathy, skin lesions, posterior uveitis, andsystemic complications are associated with loss of vision with this disorder, cornealimmune ring opacity. Systemic features are aphthous lesions of mucous membranes of themouth and genitalia, cerebellar signs, convulsions, paraplegia, skin erythema (multiforme, bullosum), arthritis, urethritis, glossitis, recurrent fever.

Behr Syndrome (Optic Atrophy Ataxia Syndrome)

Ocular features are Nystagmus, scotoma, severe progressivetemporal atrophy of the nervus opticus, bilateral retrobulbar neuritis, and horizontal nystagmus. Systemic features include pyramidal motor system signs (increased tendon reflexesand positive Babinski sign), ataxia anddisturbance of coordination, moronity, vesical sphincter weakness, muscularhypertonia, clubfoot, progressive spastic paraplegia, dysarthria, head nodding.

Gronblad–Strandberg syndrome

Ocular features are angioid streaks, Systemic features include pseudoxanthoma elasticum.

Kiloh-Nevin Syndrome (Muscular Dystrophy of External Ocular Muscles, Ocular Myopathy)

Ocular features include ptosis, orbicularis muscle weakness, ocular myopathy, diplopia getting to bilateral myopathic ophthalmoplegia, could also be associated with pigmentary retinopathy and heart block (see Kearns-Sayre syndrome). Systemic features include progressive dystrophy during which facial muscles could also be involved, occasionally, hereditary ataxia, pain, myokymia.

Kinsbourne Syndrome (Dancing Eyes Syndrome, Opsoclonus-Myoclonus Syndrome)

Ocular features include twitching of lids and eyebrows once in a while, more pronounced with activity than at rest, irregular vertical movements and jerky in

appearance and sometimes with some lateral nystagmic components. Systemic features are sporadic, jerky movements of head, trunk and limbs, usually more pronounced when the childis active, lack of coordination, ataxia, irritability, mental retardation, chronic neurologic deficits.

Oculocerebral Syndrome with Hypopigmentation

Ocular features include spastic ectropion, microphthalmos, enophthalmos, microcornea, corneal opacification, corneal vascularization, palpebral conjunctival injection, narrow lid fissures, aniridia, nystagmus, bilateraloptic atrophy. Systemic features include spastic diplegia, cutaneous hypopigmentation, mental retardation, hypogonadism, growth retardation, developmental defects of the CNS, such as cystic malformation of the posterior fossa of the Dandy-Walker type.

Oculorenocerebellar Syndrome (Orc Syndrome)

Ocular features include progressive tapetoretinal degeneration with loss of retinal vessels. Systemic features include retardation, continuous jerky movements, spastic diplegia, glomerulopathy with most renal glomeruli completely sclerosed.

Oguchi Disease

Ocular features include non-progressive nyctalopia (CSNB), Systemic features are autosomal recessive pseudotapetal reflex which normalizes with adaptation (Mizuo phenomenon).

Kirk Syndrome

Ocular features are photophobia, excessive lacrimation, amyloid corneal deposits.

Kloepfer Syndrome

Ocular features are progressive loss of vision to finish blindness associated with progressive dementia. Systemic features include severe blistering in sunlight, no increase in weight and height after erythema subsides at age 5–6 years, age doesn't progress beyond the level of imbeciles, progressive degenerative dementia occurs during or immediately after adolescence.

Knobloch Syndrome

Ocular features are high myopia, detachment of the retina, vitreo retinal degeneration, persistent papillary membrane, posterior vitreous detachment, retino choroidal staphylomas. Systemic features include occipital encephalocele, normal intelligence, congenital midline scalp defect and unusual plantarcreases.

Koby Syndrome (Floriform Cataract)

Ocular features are multiple opacities of various shapes (annular, floriform and polychromatic), found especially around embryonic nucleus.

Komoto Syndrome

Ocular features are Ptosis, epicanthus inversus, blepharophimosis and telecanthus.

Krause Syndrome

Ocular features include microphthalmos, enophthalmos, ptosis, strabismus, secondary glaucoma, iris atrophy, anterior and posterior synechiae, scleral atrophy, persistent remnants of hyaloids artery, intraocular hemorrhages and exudates, cyclitic membranes, cataracts, retinal hypoplasia and hyperplasia, choroidal and retinal malformation, retinal glial membranes, detachment of the retina, choroidal atrophy, optic nerve malformation, and optic atrophy. Systemic features are congenital cerebral dysplasia, hydrocephalus ormicrocephaly, retardation and heterotopia.

Leber's Syndrome

Systemic features are hereditary optic neuropathy mitochondrial inheritance. Ocular features include rapid sequential visual loss in 20s to 30s thanks to optic neuropathy.

Marcus Gunn Syndrome

Ocular features are unilateral congenital ptosis in additional than 90% of cases, 10% have spontaneous onset, usually inolder persons, lid elevates rapidly when the mouth is opened or mandible is moved to at least one or the otherside, the left eye seems to be more frequently affected than the proper eye, high incidences of strabismus (36%), amblyopia (34%), bilateral jaw-winking, decreased abduction. Systemic features are stimulation of ipsilateral pterygoid with chewing, opening the mouth, sucking, or contralateral jaw thrusts.

Meckel–Gruber Syndrome

Ocular features are coloboma, microcephaly, occipital

encephalocele. Systemic features include cleft lip/palate, polydactyly, polycystic renal disorder, autosomal recessive.

MELAS Syndrome

Ocular features include ophthalmoplegia, blindness, optic atrophy, pigmentary retinopathy. Systemic features include migraines, sensorineural deafness, grand malseizures, stroke-like episodes, lactic acidosis, ragged-red muscle fibers.

Menke's Disease

Ocular features are optic atrophy, retinal dystrophy. Systemic features include x-linked recessive deficiency of copper transport protein, wiry hair, ataxia, neurodegeneration.

Meretoja's Syndrome

Ocular features are corneal dystrophy, thin lines within the corneal stroma.

Meyer-Schwickerath-Weyers Syndrome (Micro phthalmos Syndrome, Oculodentodigital Dysplasia)

Ocular features are microphthalmos, hypotrichosis, glaucoma, irisanomalies (eccentric pupil, changes in normal iris texture, and remnants of the pupillary membrane along iris margins), microcornea, hypertelorism, myopia, hyperopia, keratoconus. Systemic features include thin, small nose with anteverted nostrils andhypoplastic alae, syndactyly, camptodactyly (fourth and fifth fingers), anomalies of middle phalanx of fifth finger and toe, hypoplastic teeth, wide mandible, gum ridge , sparse hair growth, visceral malformations.

Micropsia Syndrome (Lilliputian Syndrome)

Ocular features are Illusions, with misjudging of distance, position, and size of known objects (regarded as apsychovisual phenomenon). Systemic features include fixed hallucinations or dreams are expressions ofillusions and are misinterpreted by the patient.

Midas Syndrome (Dermal Aplasia and Sclerocornea, Microphthalmia)

Ocular features are bilateral microphthalmia, sclerocornea, blepharophimosis. Systemic features include dermal aplasia, microcephaly, cardiomyopathy, ventricular fibrillation, congenital heart defect.

Mietens Syndrome (Mietens-Weber Syndrome)

Ocular features are bilateral corneal opacities, horizontal and rotational nystagmus, strabismus, bushy eyebrows, ptosis. Systemic features include growth failure, flexion contracture of the elbows, dislocation of the top of the radii, mental retardation, small pointed nose with a depressedroot, low hairline, outer ear defects, digital defects, and hypertrichosis.

Mikulicz's Syndrome

Ocular features are lacrimal glands. Bilateral painless enlargement of lacrimal glands with bulging of upper lid, decreased or absent lacrimation, conjunctivitis, uveitis, optic atrophy, optic neuritis, phlyctenules, kerato conjunctivitis, dacryoadenitis, retinal candle wax spots, periphlebitis. Systemic features include infiltrative swelling of salivary symmetrical, perhaps marked, enlargement of salivary glands, dryness of mouth and pharynx, hoarseness, neurologic complications.

Millard–Gubler Syndrome

Ocular features are lesion of the facial colliculus (dorsal pons) leading to ipsilateral CN VI and VII palsies, contralateral hemiparesis.

Miller–Fisher Syndrome

The variant of Guillan–Barre syndrome characterized byocular features like acute external ophthalmoplegia, ataxia, and are flexia.

Miller Syndrome

Ocular features are glaucoma, bilateral aniridia (aniridia often notcomplete, with remnants of iris root present asrudimentary forms), cataract. Systemic features include Wilms tumor, retardation with microcephaly, genital malformations with cryptorchidismand hypospadias, hemihypertrophy, kidney anomalies (horse shoe kidney). Möbius I Syndrome (Hemicrania, Hemiplegic, Hemiplegic-Ophthalmoplegic Migraine, Hemiplegic Familial Migraine) Ocular features include extraocular palsy, permanent damage ofoculomotor nerve III.Systemic features are hemicrania, hemiparesis, aneurysm of the internalcarotid, neoplasia, headache.

Möbius Ii Syndrome

Ocular features are proptosis, ptosis, weakness of abductor muscles, normal convergence, limitation to internal rotation in lateral movements, esotropia. Systemic features include facial diplegia, deafness, loss of vestibular responses, webbed fingers or toes, club foot.

Monbrun-Benisty Syndrome (Ocular Stump Causalgia)

Ocular features are severe refractory pain of eye socket. Systemic features include pain of face and therefore the corresponding hemicranium, congestion and hyperhidrosis of region involved.

Monofixation Syndrome

Ocular features are deviation of 8 prism diopters or less by simultaneous prism and canopy test, scotoma, stereopsis, good fusional vergences found in patients with congenital esotropia, unilateral syphilitic optic perineuritis (rare), congenital esotropia (inherited during a multifactorial fashion). Systemic features include syphilis (rare).

Gruner-Bertolotti Syndrome

Ocular features include hemianopia, lid retraction, ptosis, extraocularmuscle paralysis, papilledema. Systemic features include vertigo, hemiplegia, sensory disturbances, and brain tumors.

Hallgren Syndrome

Ocular features are horizontal nystagmus (10%), cataract, retinitis pigmentosa, retinal atrophy, narrow retinal vessels, optic atrophy, keratoconus Systemic features include congenital deafness (complete or a minimum of severe auditory impairment), moronity (25%), vestibulo-cerebellar ataxia (90%), schizophrenia-likesymptoms (25%).

Hallermann–Streiff–Francois syndrome

Ocular features include micropthalmos, cataract, and blue sclera. Systemic features are hypotrichosis, dyscephaly, and short stature.

Harboyan Syndrome (Congenital corneal dystrophy and sensorineural deafness, congenital hereditary endothelial, corneal dystrophy, maumenee syndrome)

Ocular features include bluish-white opacities of cornea with normalsensitivity and no vascularization, nystagmus, keratoconus. Systemic features are sensorineural deafness with childhood onset.

Heerfordt's syndrome

Ocular features include (uveoparotid fever) uveitis. Systemic features are presentation of sarcoidosis with fever, parotid enlargement.

Hennebert Syndrome (Luetic-Otitic-Nystagmus Syndrome)

Ocular features include spontaneous nystagmus when the column of airin the auditory meatus is compressed, interstitialkeratitis, disseminated syphilitic chorioretinitismay be present. Systemic features include vertigo, fistula within the labyrinth, deafness, other clinical manifestations of congenital syphilismay be present, like "saddle" nose and Hutchinson teeth.

Hermansky–Pudlak Syndrome

Ocular features include type II oculocutaneous albinism. Systemic features are platelet dysfunction, pulmonary fibrosis, granulomatous colitis.

Andersen-Warburg Syndrome

Ocular features are bilateral microphthalmos with extensive destruction of all ocular structures often resembling a pseudotumor, blindness at birth, iris atrophy, iritis, corneal opacification and lenticular destruction with a mass visible behind the lens as long because the lens remains clear, malformed retina and choroid with retinal pseudotumors, detachment of the retina , retrolental vascular mass. Systemic features are retardation ranging from imbecility to idiocy (may begin at any age) in about two-thirds of cases, deafness of differing severity.

Andogsky Syndrome

Ocular features are dense subcapsular cataract

developing to a whole dense opacification and atopic kerato conjunctivitis, keratoconus, uveitis. Systemic features are erythematous thickening of the skin with papular hyperpigmented and scaly changes, most frequently found in regions of the wrist, popliteal fossa, neck and sometimes forehead.

Angelucci Syndrome

Ocular features include chemosis, conjunctivitis (papillary type), severe itching and burning, photophobia. Systemic features include tachycardia, vasomotor lability, excitability, allergies (asthma, urticaria, edema), dystrophic conditions and endocrine disorders are frequently associated findings.

Herrick Syndrome (Dresbach Syndrome, sickle-cell anemia, red blood cell Disease)

Ocular features are secondary glaucoma, telangiectasis of conjunctivalvessels, scleral icterus, vitreous hemorrhages,cataract, retinal hemorrhages, exudatesand neovascularization, retinitis proliferans,microaneurysms, thrombosis of retinal venules, retinal vascular sheathing, central vein occlusion, angioid streaks, retinopathy with “blacksunburst sign” in patients with SS hemoglobin, “sea fan sign” in patients with SC hemoglobin, comma signs of conjunctiva, fan-shaped neovascularization of iris, sector ischemic atrophyof iris, optic atrophy, the white cotton massof vitreous, retinal holes, chromatic vision defects, central retinal artery obstruction, branch retinal artery obstruction, white without pressure, venous tortuosity, sickling maculopathy. Systemic features include severe anemia with hemolytic crises, bone andjoint aches, hemarthrosis, jaundice, hepatosplenomegaly.

Hollenhorst Syndrome (Chorioretinal Infarction Syndrome)

Ocular features include slight proptosis, ecchymosis of lid, marked lidedema, dilated and glued pupil, hazy cornea, retinal edema, serous detachment of the retina ,cherry-red spot of the macula, attenuations of retinal arteries, pigmentary retinopathy, opticnerve atrophy, ophthalmoplegia.Horner Syndrome (Bernard-Horner Syndrome, Cervical Sympathetic Paralysis Syndrome, Claude-Bernard-Horner Syndrome, Horner Oculopupillary Syndrome) Ocular features include enophthalmos, ptosis or narrowing of palpebral fissure, ocular hypotony, miosis (degree of miosis depends on site of lesion, most pronounced when roots of cranial nerves VII and VIII and first spinal nerve are involved), hypo chromicheterochromia (children quite adults), pupildoes not dilate with cocaine. Systemic features are anhidrosis on ipsilateral side of face and neck, transitory rise in facial temperature, hemifacialatrophy, may result from a spread of conditions,including histamine headache, parasellar neoplasmsor aneurysms, internal carotid dissection orocclusion, and Tolosa-Hunt syndrome.

Hunt Syndrome

Ocular features are diminished lacrimation, absence of motor corneal reflex on the affected side, whereas consensual reflexof the non-involved eye remains normal.Systemic features are herpes zoster lesions of the outer ear and oralmucosa, severe pain within the area of external auditorymeatus and pinna, diminished hearing, tinnitus, vertigo, facial palsy, diminution or total loss ofsuperficial and deep facial reflexes, zoster lesions may involve the scalp, face, and neck, hoarseness, absence of auricular lesions has been reported, progressive dementia, extensive frontal white matter change, myoclonus, ataxia, facial paralysis, tinnitus, deafness, hyperacusis, vertigo, dysgeusia, seizures, cerebellar ataxia, schizophrenia-like symptoms.

Hypomelanosis of Ito Syndrome

Ocular features are iridal heterochromia, myopia, esotropia, microphthalmia, hypertelorism, nystagmus, strabismus,corneal opacity, choroidal atrophy, exotropia,small nervus opticus , hypopigmentation of thefundus, corneal asymmetry, pannus, atrophicirides with irregular pupillary margins, cataract,retinal detachment.Systemic features are cutaneous manifestations consisting of macularhypopigmented whorls, streaks and patchesin a bilateral or unilateral distribution affectingalmost any portion of the body surface, 50% haveassociated noncutaneous abnormalities, includingcentral systema nervosum dysfunction (seizure,delayed development) and musculoskeletal anomalies.

Iridal Adhesion Syndrome

Ocular features are posterior synechiae, irregular pupil.

Iris Nevus Syndrome

Ocular features are Unilateral glaucoma in eyes with peripheral anterior synechiae, multiple iris nodules, ectopic descemet’s membrane, corneal edema, stromaliris atrophy, iris pigment epithelial atrophy, ectropion uveae, ectopic pupil, keratoconus, herpes simplex virus DNA has been detected in patients with iridocorneal endothelial syndrome from corneal

specimens. Systemic features include Glass like membrane covering the anterior iris surface, corneal endothelial degeneration and accompanying ectopic endothelial membranes are liable for occlusion of the filtration mesh work and subsequent pressure increase.

Marshall-Smith Syndrome

Ocular features are hypertelorism, protuberant eyes with shalloworbits.Systemic features include feeding and respiratory difficulties, developmentaldelay, advanced age, characteristic facies.

Martsolf Syndrome

Ocular features are cataracts.Systemic features include retardation , short stature, hypogonadism.

Ivic Syndrome (Hearing Impairment, Internal Ophthalmoplegia, Radial Ray Defects, Thrombocytopenia)

Ocular features include strabismus, internal ophthalmoplegia. Systemic features are malformed upper limb, short distal phalanx, hearing loss, thrombocytopenia, leukocytosis, imperforate anus, radial ray defect.

Jabs Syndrome (Granulomatous Uveitis, And Cranial Neuropathies, Synovitis)

Ocular features are granulomatous uveitis, iritis, VI nerve palsy. Systemic features are granulomatous synovitis, corticosteroid-responsive hearing loss, boggy polysynovitis, boutonne use deformities, granulomatous arthritis, skin involvement

Norrie Disease

Ocular features include retinal dysplasia, detachment of the retina, leukocoria, cataract. Systemic features include phthisis, deafness, X-linked, vitreous hemorrhage.

Nothnagel Syndrome (Ophthalmoplegia-Cerebellar Ataxia Syndrome)

Ocular features include oculomotor paresis, gaze paralysis most frequently upward, combined with a point of internal or external ophthalmoplegia. Systemic features include cerebellar ataxia, poor upper extremity movements, neoplasia, infarction, midbrain lesion.

Joubert Syndrome (Familial Cerebellar Vermis Agenesis)

Ocular features include choroidal coloboma, nystagmus, ocular fibrosis, and telecanthus. Systemic features include episodic hyperpnea, apnea, ataxia, psychomotorretardation, rhythmic protrusion of tongue, mental retardation, micrognathia, complex cardiacmal formation, cutaneous dimples over wrists and elbows.

Congenital Vertical Retraction Syndrome

Ocular features include aberrant regeneration of the oculomotor, concurrent protective eyelid closure, congenital alterations within the extraocular muscle, its insertion and its peripheral innervation, nystagmus retractorius, surgical or traumatic rearrangement of orbital structures may account for retraction.

Maffuci's syndrome

Ocular features are multiple hemangiomas. Systemic features are enchondromas (which may cause limb deformities), with risk of malignant transformation.

Marfan Syndrome

Ocular features are ectopia lentis, detachment of the retina, glaucoma, axial myopia. Systemic features are arachnodactyly, long-limbed, aortic dissection.

Cranio-Oro-Digital Syndrome

Ocular features include downward-slanting palpebral fissures (antimongoloid obliquity). Systemic features are microcephaly, small mouth, midface hypoplasia, cleft palate, flexed, overlapping fingers with syndactyly of digits 3 and 4, syndactyly of toes 2 and 5, bifid uvula, slight deviation of the terminal phalanges of the third fingers, radial deviation of the terminal phalanx of the proper fourth finger,short first toe and long second toe, short first metacarpal, extra bone within the capitate-hamate complex, small thorax, bowed limbs with absentfibula, mild frontal bossing, conductive hearing impairment, flat facies, broad nasal base, wavy irregular clavicles and ribs, widely spaced eyes, prominent forehead.

Criswick-Schepens Syndrome

Ocular features include posterior vitreous detachment of organized membranes of vitreous, snowflake-like

opacities of vitreous, heterotropia of macula, subretinal exudates, detachment of the retina, degenerative retinal changes, retinal hemorrhage, retinal folds, enophthalmos, phthisis, intraretinal exudate, vitreous hemorrhage, amblyopia, falciform retinal fold. Systemic features are normal general development, normal birth weight.

Crouzon's Syndrome

Ocular features include proptosis, strabismus, micro/megalocornea, iris coloboma, cataract, ectopia lentis, and glaucoma. Systemic features are craniosynostosis, maxillary hypoplasia, prognathism, hooked nose.

Crowded Disk Syndrome (Bilateral Choroidal Folds and Optic Neuropathy)

Ocular features are bilateral choroidal folds, blind spot congestion, optic atrophy, hyperopia, shortened axial length.

Cryptophthalmia Syndrome (Cryptophthalmos Syndactyly Syndrome, Fraser Syndrome)

Ocular features are microphthalmia, epibulbar dermoid, cryptophthalmos, enophthalmia, eyebrows partially or completely missing, skin from forehead completely covers one or both eyes, but the globes can be palpated beneath the skin, in unilateral cases, the guy eye may present lid coloboma, buphthalmos, conjunctival sac partially or totally obliterated, absence of trabeculae, Schlemm canaland ciliary muscles, cornea is differentiated from the sclera, lens anomalies from complete absence to hypoplasia, dislocation and calcification. Systemic features are syndactyly (finger, toes) (about 40%), coloboma of alae nasi and nostrils, urogenital abnormalities, including pseudo hermaphroditism and renal hypoplasia, abnormal, bizarre hairline, narrow external acoustic meatus and malformation of ossicles, harelip and palate may occur, atresia orhypoplasia of larynx in some cases, hoarse voice, dysplastic pinna, meatal stenosis, glottic web and subglottic stenosis.

Curly Hair-Ankyloblepharon-Nail Dysplasia Syndrome (CHANDS)

Ocular features include congenital ankyloblepharon (fused eyelids). Systemic features are curly hair, hypoplastic nails.

Jugular Foramen Syndrome (Vernet Syndrome)

Ocular features include enophthalmos, ptosis, and miosis. Systemic features are paralysis of the ix, x, and xi cranial nerves withresulting impairment of related function, i.e.dysphagia, loss of taste on the posterior third of the tongue and nasal regurgitation, anhidrosis, paralysis of the sternocleido mastoid muscle and a part of Trapezium (upper portion), hoarseness, tachycardia, dysarthria, weight loss.

Karsch-Neugebauer Syndrome (Nystagus-Split Hand Syndrome)

Ocular features are horizontal nystagmus, strabismus, cataract, fundus changes. Systemic features include split hand and split foot deformities, monodactylous hands.

Sluder Syndrome (Lower Facial Neuralgia Syndrome, Sphenopalatine Ganglion Neuralgia Syndrome)

Ocular features are severe orbital pain; increased lacrimation during episodes of pain. Systemic features include unilateral facial pain, mainly root of nose, orbit and mastoid area, episodes of headaches, nasal congestion.

Snuff-Out Syndrome (Snuff Syndrome)

Ocular features are loss of central fixation, reduction in acuity, reduction in field of vision, cataract, glaucoma.

NARP Syndrome

Ocular features include retinitis pigmentosa, bulls-eye maculopathy, salt-and-pepper retinopathy. N Systemic features include ARP syndrome patients develop ataxia, weakness, and have retinitis pigmentosa, causing gradual field of vision constriction.

Nematode Ophthalmia Syndrome (Toxocariasis, Visceral Larva Migrans Syndrome)

Ocular features are leukocoria, uveitis, cataract, marked vitreousreaction with large floaters, choroiditis, large, cystlike white masses extending into vitreous, optic neuritis, papillitis, strabismus, hemorrhagic, exudative or granulomatous retinitis, retinal detachment, endophthalmitis, larvae present inthe cornea. Systemic features are hepatosplenomegaly, pulmonary infiltration,

fever, cough, lack of appetite.

Kasabach–Merritt syndrome

Ocular features are giant hemangioma. Systemic features include localized intravascular coagulation causing low platelets and fibrinogen.

Kearns–Sayre syndrome

Ocular features are CPEO, pigmentary retinopathy Pigmentary degeneration of the retina, progressive external ophthalmoplegia, corneal decompensation, optic neuritis. Systemic features include mitochondrial inheritance, (granular pigmentation, peripapillary atrophy), and Adams-Stokes syndrome, usually present before 20 years. Abnormal mitochondria with paracrystalline inclusion within the muscle fiber, Adams-Stokes syndrome, limb weakness, hyperglycemic acidotic coma, death, cerebellar dysfunction. Abnormal mitochondria with paracrystalline inclusion within the muscle fiber, Adams-Stokes syndrome, limb weakness, hyperglycemic acidotic coma, death, cerebellar dysfunction.

Mulibrey Nanism Syndrome (Perheentupa Syndrome)

Ocular features are alternating esotropia and exotropia, yellowish retinal dots and scattered pigment dispersionin clusters (especially within the midperiphery), drusen of Bruch membrane, hypoplasia of choriocapillaries (diagnostic sign).

Keratoconus Posticus Circumscriptus (KPC, KPC related to Malformations)

Ocular features are corneal opacities, retinal coloboma, ptosis, hyperopia, iridocorneal adhesions, and hypertelorism. Systemic features are harelip, birth defect, neck webbing, short stature, retardation, hernia, undescended testes, tight heel cords, vertebral anomalies, delayed age, double ureters, cone-shaped epiphyses, stubby limbs and digits, limitation of extension and supination of the elbows, brachydactyly, fifth finger clinodactyly, frequent tract infections, prominent nose, mild maxillary hypoplasia, low posterior hairline, short, broad feet with bilateral pescavus, bilateral ureteric reflux.

Posthypoxic Encephalopathy Syndrome (Parieto-Occipital Syndrome, Posthypoxic Syndrome)

Ocular features include nystagmus, nuclear ophthalmoplegia, visual hallucinations, partial cerebral blindness (predominant defects within the sphere of psychic elaboration rather than in primary visual perception), complete cortical blindness, central scotomata, pupillary paresis, retinal atrophy, optic atrophy. Systemic features are confusion, irritability and agitation, alexia, disorientation (mainly spatial), spasm.

Potter Syndrome (Renal Agenesis Syndrome, Renofacial Syndrome)

Ocular features include hypertelorism, pronounced epicanthal folds extending down the cheeks, antimongoloid slant of palpebral fissure. Systemic features include flat bridge of the nose, low-set ears, facial deformities, micrognathia, pulmonary hypoplasia, cystic dysplasia of kidney to agenesis, oligohydramnios, clubbing of hands and feet, spinabifida, prominent infracanthal folds, flattened beaked nose, creased skin, and positional deformities of the limbs.

Progressive Intracranial Arterial Occlusion Syndrome (Taveras Syndrome)

Ocular features include unilateral ptosis, defective optokinetic nystagmus, visual agnosia, amaurosis fugax. Systemic features include progressive intracranial arterial occlusion with both internal carotid arteries involved, memoryloss, muteness, localized numbness, cryingspells, catatonic states and episodes, staring, seizures.

Morning Glory Syndrome

Ocular features are strabismus, abnormality of embryologic developmentof anterior chamber (anterior chambercleavage syndrome), remnants of hyaloid system,chorioretinal pigment surrounding opticdisk, narrow branches of retinal arteries at edgeof blind spot , retinal exudates and detachment,subretinal hemorrhages and retinal neovascularization,enlarged pink blind spot , funnel-shapedwith a central white fluffy dot, nerve head surroundedby elevated annulus of chorioretinalpigment, unilateralSystemic features include midline cranial facial defects like hypertelorism,cleft lip/palate, basal encephalocele,agenesis of nerve pathway , sphenoid encephaloceledefects within the floor of the sella turcica,cranial, facial and neurologic associations, pituitarydwarfism, association with the CHARGE syndrome.

Mort D'amour Syndrome

Ocular features are pupillary dilation. Systemic features

include hypertension, arrhythmia, heart ischemia, rupture of aneurysm.

Parinaud Syndrome

Systemic features are lesion of the dorsal midbrain Ocular features include light-near dissociation, supranuclear upgaze palsy, convergence retraction nystagmus, and failure of convergence and accommodation.

Patau syndrome

Systemic features are Trisomy 13, 1 in 14,000 live births, microcephaly, anticipation < 3 months. Ocular features are cyclopia, colobomata, retinal dysplasia.

Mulvihill-Smith Syndrome

Ocular features are keratoconus, conjunctivitis. Systemic features include patients have short stature, microcephaly, unusual facies, numerous pigmented nevi, hypodontia, sensorineural deafness, immunodeficiency (low IgG) and a high-pitched voice.

Neu Syndrome

Ocular features include hypertelorism; absent eyelids. Systemic features include flexion deformities, overlapping fingers, rocker bottom feet, protruding heels, toe syndactyly, microcephaly, short neck, tiny nose, brain atrophy.

Niemann–Pick Disease

Ocular features include cherry-red spot, vertical supranuclear gaze palsy rarely cherryred. Systemic features, including autosomal recessive, deficiency of sphingomyelinase. A is infantile onset with visceromegaly, neurodegeneration, and B juvenile-onset with visceromegaly, spot, type C features a variable onset, ataxia, and neurodegeneration. Nonne-Milroy-Meige Disease (Blepharospasm-Oromandibular Dystonia, Chronic Hereditary Edema, Chronic Hereditary Trophedema, Chronic Trophedema, Chronic Hereditary Lymphedema, Congenital Trophedema, Elephantiasis Arabum Congenita, Elephantiasis Congenita Hereditaria, Familial HereditaryEdema, Hereditary Edema, Idiopathic Hereditary Lymphedema, Meige Disease, Meige-Milroy Syndrome, Milroy Disease, Nonnemilroy Syndrome, Oromandibular Dystonia, Pseudoedematous Hypodermal Hypertrophy, Pseudoelephantiasis Neuroarthritica, Tropholymphedema, Trophoneurosis) Ocular features include lid and conjunctival edema, blepharoptosis, distichiasis, strabismus, buphthalmos, ectropion. Systemic features include lymphedema, mandibulo facial dysostosis, unilateral or bilateral edema of ankle ascending to the knee and eventually above, rough, pigmented skin over swollen parts.

Outer Retinal Ischemic Infarction Syndrome

Ocular features are acute loss of central and paracentral vision, whitening of the outer retinal layers in posterior fundus, mottled changes within the pigment epithelium

Nystagmus Blockage Syndrome (Nbs)

Ocular features include esotropia, nystagmus, amblyopia, most patients with this syndrome like better to fixate with one eye, but others show alternating fixation. Systemic features include abnormal head position.

Senter Syndrome

Ocular features are corneal involvement. Systemic features include ichthyosiform erythroderma, deafness, hepatomegaly, hepatic cirrhosis, glycogen storage, short stature, retardation, hepatitis.

Shy-Drager Syndrome

Ocular features include external ophthalmoplegia, iris atrophy, ocular sympathetic and parasympathetic insufficiency (alternating Horner syndrome, cholinergic supersensitivity, decreased lacrimation, and corneal hypesthesia). Systemic features are postural hypotension, rigidity, tremor, adiadochokinesia, wasting of muscles, retardation, impotence, dysphagia, bilateral vocalcord paralysis, bladder disorder, anhydrosis, extremity weakness and paresthesia, dizziness, abnormal postural balance.

Oculo-Orogenital Syndrome

Ocular features include conjunctivitis, varying from mild to severe, keratitis, optic atrophy, corneal vascularization. Systemic features include stomatitis, glossitis, scrotal dermatitis with pruritus, erythema, erythema of pharynx and taste bud ,small sensitive ulcers of buccal membranes, diarrhea, fatigue, muscular weakness, painful feet with erythema, exfoliation and ulceration, burning, itching, mental depression, dizziness, oral mucosa becomes pale and macerated with

fissuring of skin.

Optic Atrophy, Juvenile Syndrome

Ocular features include scotoma, color defects, choroidalsclerosis, optic neuritis, temporal optic atrophy, aggregation of retinal pigment epithelium, tortuosity of retinal arteries and veins, reduced central vision, retinal lesions, may present with mild-to-moderate reduction of acuity with 50% of patients having vision between 20/60 and 20/200, field of vision defect associated could also be acentral, paracentral or cecocentral scotoma. Systemic features arekeratosis pilaris on the extremities, approximately 10% of patients present with mental abnormalities and approximately 80% of patients with neuralhearing loss.

Optic Disc Traction Syndrome

Systemic features include posterior vitreous detachment or vitreopapillary fibrous membrane could also be related to retinal surgery. Ocular features are central retinal vein occlusion, blind spot traction (vitreopapillary), localized detachment of the retina.

Orf Syndrome (Ecthyma Infectiosum)

Ocular features include pigmentation of lids. Systemic features include single or multiple lesions of hands and other parts of body, itching, fever, concurrent aseptic meningitis caused by enterovirus.

Pallidal Degeneration, Progressive, with Retinitis Pigmentosa syndrome

Ocular features are retinitis pigmentosa. Systemic features include progressive extrapyramidal rigidity, dysarthria.

Pigmentary Ocular Dispersion Syndrome (Pigmentary Glaucoma)

Ocular features include myopia, glaucomatous field changes, ocular hypertension, iris translucency, abnormal number of iris processes, insertion of iris anterior to scleral spur, pigmentation of posterior trabecular meshwork, grades 3–4. Systemic features include krukenberg spindles, presence of pigmentation on equatorial border of membrane, glaucomatous cupping and myopic nervus opticus changes.

Pillay Syndrome (Ophthalmom and ibulomelic Dysplasia)

Ocular features include temporomandibular fusion, obtuse mandibularangle, and short forearms. Systemic features are diagnosis is formed by clinical findings.

Plummer-Vinson Syndrome

Ocular features include reduced tear formation, pale conjunctiva, dry eyes, retinal hemorrhages, and papilledema. Systemic features include dysphagia for solid food with main difficulties originating within the upper portion of the esophagus, glossitis and gastritis, anemia, atrophy ofmucous membranes, dystrophy of the fingernails (koilonychia), fatigue.

Poems Syndrome

Ocular features are bilateral blind spot edema and bilateral cystoidmacular edema. Systemic features are hyperpigmentaion and lower extremity edema, hypertrichosis, angiomas, motor deficiency, deeptendon reflexes are diminished, endocrinopathy, hypogonadism, pulmonary hypertension, hepatomegaly, splenomegaly and lymphadenopathy.

Posner-Schlossman Syndrome (Glaucomatocyclitic Crisis)

Ocular features include slight blurring of vision and colored halos duringepisodes of high intraocular tension, high intraocular pressure (unilateral), glaucomato cycliticcrisis (benign and typically unilateral), enlarged pupil, anisocoria, absence of ciliary or conjunctival injection, only trace of aqueous flare, no posterior synechiae, chamber angleopen, heterochro Systemic features include allergy, related to gastrointestinal disease (peptic ulcers).

Posterior Iris Chafing Syndrome

Ocular features include iris transillumination defects, recurrent microhyphemas, pigment dispersion glaucoma, pigment deposition in trabecular meshwork, iris pigmentatrophy.

Pseudo-Ophthalmoplegia Syndrome (Roth-Bielschowsky Syndrome)

Ocular features include paralysis of lateral gaze in one

direction, vestibularnystagmus during which the fast phase is absent on the ipsilateral side but the slow phase is present. Systemic features include basal ganglia or tectum lesion.

Purtscher Syndrome (Duane Retinopathy, embolism Syndrome, Traumatic Retinal Angiopathy, Traumatic Liporrhagia, Valsalva Retinopathy of Duane)

Ocular features are retinal and preretinal hemorrhages over entire fundus, cotton-wool exudates, mainly posterior aspect, retinal edema, posterior and macularserous detachment, venous congestion and engorgement, papilledema, usually bilateral, although unilateral causes are reported. Systemic features include multiple fractures (mainly extensive crushing), lung congestion, dyspnea, lymphorrhagia, pancreatitis, scleroderma, dermatomyositis, lupus erythematosus, child birth.

Raymond Syndrome [Cestan (2) Syndrome, Disassociation of Lateral Gaze Syndrome, Pontine Syndrome, Raymond-Cestan Syndrome]

Ocular features include ipsilateral abducens palsy, paralysis of lateral conjugate gaze. Systemic features are contralateral hemiplegia, anesthesia of the face, limbs, and trunk.

Relapsing Polychondritis (Jaksch-Wartenhost Syndrome, Meyenburg-Altherz-Vehlinger Syndrome, Von Meyenberg Ii Syndrome)

Ocular features include Conjunctivitis, corneal ulcer, exophthalmos, panophthalmitis,phthisis bulbi, proptosis, opticneuritis, papilledema, detachment of the retina , bluesclera, episcleritis, scleromalacia, vitreous opacity,cataracts, nystagmus, retinal artery thrombosis, keratoconjunctivitis sicca, secondary glaucoma, scotoma, uveitis, paresis of third or sixth nerve, conjunctival mass (salmon patch), chorioretinitis. Systemic features include destruction of cartilage and eventual replacement with animal tissue, polyarthritis, chondritis, tracheal collapse, bronchial collapse, anemia, liver dysfunction, death, malaise, fever, dyspnea,changes in pitch of voice, hearing disorder ,vertigo, deformed ears, semilunar valve insufficiency.

Refsum's Disease

Systemic features are autosomal recessive, deficiency of phytanic acid A-hydrolase leads to accumulation of phytanic acid, ichthyosis, deafness, cardiomyopathy, ataxia. Riley–Day syndrome (familial dysautonomia) autosomal recessive, sensory neuropathy, autonomic dysfunction/crises, morecommon in Ashkenazi Jews, tear deficiency, commonly with ulceration ocular features are keratoconjunctivitis sicca, reduced corneal sensation, pigmentary retinopathy, optic atrophy.

Retinohypophysary Syndrome (Benign Retinohypophysary Syndrome, Lijo Pavia-Lis Syndrome)

Ocular features include superior nasal field contraction, narrowing ofretinal vessels, macular edema, optic neuritis, optic atrophy, field of vision defects. Systemic features include glycosuria, headache, vertigo, psychic disturbances.

Retinopathy, Pigmentary, And retardation (Mirhosseini-Holmes-Walton Syndrome)

Ocular features include pigmentary retinal degeneration, cataract, and keratoconus. Systemic features include microcephaly, severe retardation, hyperextensiblejoints, scoliosis, arachnodactyly, hypogonadism.

Retroparotid Space Syndrome (Posterior Retroparotid Space Syndrome, Villaret Syndrome)

Ocular features are enophthalmos, ptosis, lagophthalmos, epiphora, miosis, may produce sympathetic overactivity resulting in increased sympathetic outflow (i.e. pupillary dilation, widened palpebral fissureand facial sweating). Systemic features include homolateral paralysis cranial nerves IX to XII,with dysphagia and loss of taste in posteriorthird of the tongue, dysphonia, paralysis of sternocleidomastoid and trapezium muscles,paralysis nerve VII occasionally.

Reye Syndrome (Acute Encephalopathy Syndrome)

Ocular features include cortical blindness, dilated pupils with absent orsluggish reaction to light, papilledema. Systemic features include respiratory infections with recovery between3 and 21 days, vomiting after recovery from infection, dyspnea, hypotonia, coma, convulsions, fever, flexion of elbows and hands.

Rollet Syndrome (Orbital Apex-Sphenoidal Syndrome)

Ocular features include exophthalmos, ptosis, hyperesthesia or anesthesia of the upper lid, ophthalmo-

plegia (partial or complete), wide pupil with loss of reactionon accommodation, neuralgic pain within the regionof the ophthalmic branch of nerve V, anesthesia of the cornea, papilledema, opticneuritis, optic atrophy, diplopia, herpes zosterophthalmicus. Systemic features include hyperesthesia or anesthesia of the forehead, inflammation of cavernous sinuses, meningoencephalitis.

Rosenberg-Chutorian Syndrome

Ocular features include optic atrophy. Systemic features include polyneuropathy, neural deafness.

Pancoast Syndrome

Ocular features include mild enophthalmos, ptosis, narrowing of the palpebral fissure, miosis. Systemic features include pulmonary apical tumor, severe shoulder pain, paresthesia, pain and paresis of the homolater alarm with atrophy of arm and hand muscles.

Petzetakis-Takos Syndrome

Ocular features include superficial keratitis, palpebral edema, corneal hyperesthesia, photophobia, blepharospasm, decreased pupillary response, and xerophthalmia. Systemic features include lymph gland hypertrophy.

Rothmund Syndrome

Ocular features include eyebrows could also be sparse or absent, hypertelorism, cilia sometimes are diminished or absent, trichiasis, epiphora, cataracts (anterior subcapsular, posterior stellate or perinuclear type), corneal lesions, retinal hyper pigmentation, keratoconus, strabismus, epibulbar dermoids. Systemic features are poikiloderma, hypogonadism, hypomenorrhea, head deformity (enlarged with depressed nasal bridge also as microcephaly), small stature, with short or malformed distal phalanges, aplasiacutis congenita (congenital absence of skin inone or more areas), alopecia.

Rubella Syndrome

Ocular features include nystagmus, glaucoma, corneal haziness, cataracts, retinal pigmentary changes, appearance and central distribution of lesions are quite distinguishable from retinitis pigmentosa, retinopathy isn't progressiveand has little, if any, effect on vision, waxyatrophy of blind spot, conjunctivitis, megalocorneaor microcornea, buphthalmos, microphthalmos, uveitis, iris atrophy, spherophakia, strabismus. Systemic features include low-birth-weight, diarrhea, pneumonia, urinary infection, deafness, heart condition, hepatosplenomegaly, mental retardation, inguinal hernias, ataxia, cardiac abnormalities.

Sabin-Feldman Syndrome (Chorioretinitis)

Ocular features include Microphthalmia, strabismus, fixed pupils, posterior lenticonus, microcornea, chorioretinitis or atrophic degenerative chorioretinal changes, optic atrophy. Systemic features arecerebral calcifications (infrequent), convulsions (frequent), microcephaly, hydrocephalus.

Saldino-Mainzer Syndrome (Retinitis Pigmentosa)

Ocular features include tape to retinal degeneration, retinal atrophy, Leber congenital amaurosis, retinitis pigmentosa. Systemic features are nephronophthisis, cone shaped epiphyses of hands and feet, flared ribs, hypoplastic pelvis, brachydactyly, hyperparathyroidism, osteomalacia, osteopetrosis, kidney failure.

Sandifer Syndrome (Hiatal Hernia-Torticollis Syndrome)

Ocular features include strabismus (not associated with existing torticollis).Systemic features include rotation of the top to at least one shoulder withstretching of the neck (more pronounced duringeating and reading), epigastric pain associatedwith vomiting, primarily in infancy, malnutrition, hiatal hernia, asthenia. Systemic features are deficiency of hexosominadase A and B, cherry-red spot, optic atrophy, splenomegaly, neurodegeneration.

Sands of the Sahara Syndrome (Diffuse Lamellar Keratitis)

Ocular features include Interface inflammation after LASIK may be a rare,but potential sightthreateningcomplication,syndrome presents 1–5 days after LASIK, affected patients often complain of decreased or cloudy vision, foreign body sensation, and photophobia, symptoms could also be mild or severe, explanation for the interface debris is unknown, but microkeratome material is implicated.

Rubinstein–Taybi Syndrome

Ocular features include hypertelorism, colobomas, Antimongoloid slant of lid fissure, epicanthus, long

eyelashes and highly arched brows, strabismus, myopia, hyperopia, iris coloboma, cataract, optic atrophy, ptosis, detachment of the retina. Systemic features include developmental abnormality broad thumbs/big toes, maxillary/ mandibular hypoplasia, hypertrichosis, motor and retardation, broad thumbs and toes, highly arched palate, allergies, heart murmurs, anomalies of size, shape, and position of ears, dwarfism, cryptorchidism.

Russell Syndrome

Ocular features include lid retraction, nystagmus (horizontal, vertical, or rotatory), homonymous hemianopsia, opticnerve atrophy. Systemic features include extreme emaciation, euphoria, pale skin.

Sandwich Infectious Keratitis Syndrome (SIK Syndrome)

Ocular features include infectious infiltrates within the interface of the corneal which are white small irregular or circular.

Schnyder's Crystalline Corneal Dystrophy

Ocular features are hypercholesterolemia and genu valgam. Systemic features are central corneal haze, subepithelial cholesterolcrystal deposition, midperipheral, and panstromal haze and arcus lipoides.

Schomberg Disease (Subconjunctival Hemorrhage)

Ocular features are subconjunctival hemorrhage. Systemic features include petechiae may occur in any tissue, bleeding fromany orifice.

Schwartz Syndrome (Retinal Detachment)

Ocular features are secondary open angle glaucoma, detachment of the retina, uveitis, myopia, blepharophimosis, long eyelashes, and microcornea. Systemic features are small stature, myotonia, expressionless facies, joint limitation in hips, dystrophy of epiphyseal cartilage, vertical shortness of vertebrae, shortneck, and low hairline.

Second Eye Syndrome

Ocular features include increased pain during the second surgery. Systemic features include diagnosis is formed by clinical findings.

Zinsser-Engman-Cole Syndrome (Cole-Rauschkolb-Toomey Syndrome, Dyskeratosis Congenita with Pigmentation)

Ocular features are ectropion, chronic blepharitis, obstruction oflacrimal puncta, conjunctival keratinization, bullous conjunctivitis, epiphora, nasolacrimal duct obstruction, loss of eyelashes, cataract, glaucoma, strabismus, abnormal fundi. Systemic features include congenital dyskeratosis with the pigmentation of "marble" configuration or "gunmetal" appearance,atrophic areas and telangiectasis, dystrophyof nails, vesicular and bullous lesions of mouth followed by ulceration, mucosalatrophy, leukoplakia, a plastic anemia, defect of teeth, physical and mental development may beretarded, tufts of hairs on the limbs, keratinized basal cell, papillomas on the trunk (Przeździecka-Dołyk et al., 2020).

Spasmus Nutans Syndrome

Ocular features are bilateral nystagmus; attempt at gaze fixation intensifies manifestations. Systemic features are rhythmic movements of head in upright position.

Stargardt's Disease (Fundus Flavimaculatus)

Ocular features are macular dystrophies, with two clinical presentations: Stargardt's ("beaten-bronze" atrophy, yellowish flecks of the posterior pole, significant dVA) and fundus flavimaculatus (widespread pisciform flecks with relative preservation of vision), p. 459. Systemic features are autosomal recessive (usually Ch1p, ABCA4),

Steele–Richardson–Olszewski (Progressive Supra nuclear Palsy)

Systemic features include neuro degenerative disease of the elderly Parkinsonism, pseudobulbar palsy, and dementia, Ocular features include supranuclear vertical gaze, postural instability.

Sturge-Weber Syndrome (Encephalofacial Angiomatosis, Encephalotrigeminal Syndrome, Meningocutaneous Syndrome, Neuro-Oculocutaneous Angiomatosis, Vascular Encephalotrigeminal Syndrome)

Ocular features are unilateral hydrophthalmos, secondary glaucoma (late) conjunctival angiomata (telangiectases), iris decoloration, nevoid marks or vascular dilation of the episclera, glioma, serous retinal detachment, choroidal angiomata, deep anterior chamber angle, nevus

flammeus of eyelid, buphthalmos, nervus opticus cupping, anisometropia, hemianopsia, increased corneal diameter, enophthalmos, exophthalmos, optic atrophy,choroidal hemangioma, anterior chamber angle vascularization. Systemic features include vascular port-wine nevus (face, scalp, limbs, trunk, leptomeninges), acromegaly, facial hemihypertrophy, intracranial angiomas, convulsion, mental retardation, obesity, limb atrophy.

Stickler's Syndrome (Hereditary Arthro-Ophthalmopathy)

Ocular features are high myopia, optically empty vitreous, detachment of the retinas, cataract, ectopia lentis, glaucoma. Systemic features include autosomal dominant (Ch12q, COL2A1), abnormality of type II collagen, arthropathy, Pierre Robin sequence (micrognathia, high arched/cleft palate), sensorineural deafness, bicuspid valve prolapse, p. 389.

X-Linked Cone Dysfunction Syndrome

Ocular features are myopia, visual loss, color vision abnormality. Systemic features include diagnosis is made by clinical findings.

Scaphocephaly Syndrome

Ocular features include shallow orbits, proptosis, nystagmus, exotropia, aniridia, cataract, papilledema, optic atrophy, aniridia, dislocated lens. Systemic features are long anteroposterior head diameter, short transverse diameter of the top, increase dintracranial pressure, flat forehead with absent superciliary arches, prominent nose, mental retardation.

Schaumann Syndrome

Ocular features are orbital granulomatous mass, bony defects, cutaneous and subcutaneous nodules, myogenicpalsy, lachrymal gland adenopathy, decreased tear Ocular Syndromes and Systemic Diseases 707 formation, secondary glaucoma, granuloma tousuveitis with iris nodules, cells, and flare, muttonfat keratitic precipitates, keratitis sicca, vitreousfloaters, band shaped keratitis, complicated cataract, inflammatory retinal exudates, "candlewax drippings", nervus opticus atrophy, neuritis, eyelid nodules, ocular nerve enlargement (granuloma). Systemic features are lymphadenopathy, hilar nodes, fatigue, cystic, punched outor reticulated changes in small bones (mainly hands and feet), muscle wasting, contractures, weakness in legs and arms (Kordyś et al., 2018).

Xxxxx Syndrome (Penta X Syndrome, Tetra X Syndrome)

Ocular features include epicanthal folds, hypertelorism, antimongoloid (upward slant) of palpebral fissures. Systemic features are growth retardation, bilateral.

Young-Simpson Syndrome

Ocular features include blepharophimosis. Systemic features are congenital hypothyroidism, congenital heart defects, facial dysmorphism (microcephaly, bulbousnose, low-set ears, micrognathia), cryptorchidismin males, hypotonia, retardation, postnatal growth retardation.

Walker-Clodius Syndrome

Ocular features are hypertelorism, nasolacrimal obstruction with constant epiphora, mucopurulent conjunctival discharge, keratitis, nanocanalization of thelacrimal duct. Systemic features are deformities of hands and feet ("lobster claw"), absence of both index and middle fingers and second metacarpals with rudimentary third metacarpals, syndactylism, congenital anomaly and lips, deafness, ear malformation, renal anomalies.

West Syndrome (Massive Myoclonia, Jack knife Convulsion)

Ocular features include nystagmus. Systemic features are convulsion, nodding of the highest, opisthotonos, mental retardation.

Wildervanck Syndrome (Cervico-Oculo-Acousticus Syndrome, Cervico-Oculo-Facial Dysmorphia, Cervicooculofacial Syndrome, Franceschetti-Klein-Wildervanck Syndrome, Wildervanck-Waardenburg Syndrome)

Ocular features are abducens paresis, nystagmus, heterochromiairidis. Systemic features are deafness or deaf-mutism, torticollis with short, webbed neck, epilepsy, retardation, cleft palate, scoliosis, ventricular congenital heart defect, ectopic kidney, hydrocephalus, hypoplastic thumb, and growth retardation.

Windshield Wiper Syndrome

Ocular features include ruptured zonules, lateral lens tilt, lateral decentration of the lens, decreased acuity, and glare.

Wolf Syndrome (Chromosome 4 Partial Deletion Syndrome, Hirschhorn-Cooper Syndrome, Monosomy 4 Partial Syndrome)

Ocular features are hypertelorism, antimongoloid slanting of palpebral fissures, ptosis, nystagmus, strabismus, iris coloboma, retinal coloboma. Systemic features are microcephaly, retardation, seizures, earmalformations, hypospadias, beaked nose, broad nasal root, harelip and palate, hypotonia.

Walker-Warburg Syndrome (Cerebro-OcularDysplasia-Muscular Dystrophy, Cod-Md Syndrome, Fukuyama Congenital dystrophy, Hard + or - E Syndrome, Warburg Syndrome)

Ocular features are microphthalmia, cataract, immature anterior chamber angle, retinal dysplasia, detachment of the retina, persistent hyperplastic primary vitreous, optic nerve hypoplasia, iris coloboma, opaque cornea, myopia, orbicularis weakness, irregular gray subretinal mottling, optic atrophy. Systemic features include cerebral and cerebellar agyria-micropolygyria, cortical disorganization, glialmesodermal proli feration, neuronal heterotopias, hypoplasia of nerve tracts, hydrocephalus, encephalocele, muscular dystrophy, seizures, retardation, hypotonia, abnormal facies (Przeździecka-Dołyk et al., 2020).

Ward Syndrome (Epitheliomatous Phakomatosis, Nevus-Jaw Cyst Syndrome)

Ocular features include hypertelorism, dystopia canthorum, nevi of eyelids, congenital cataracts, congenital corneal opacities, and colobomata. Systemic features are basal cell nevi with multiple basalomatous nodules on face, neck and trunk, epithelioma adenoides cysticum.

Wermer Syndrome (Endocrine Adenoma-Peptic Ulcer Complex, Multiple Endocrine Neoplasia 1, Men 1, Multiple Endocrine Adenomatosis 1, Mea 1, Pluriglandular Adenomatosis N)

Ocular features are field of vision defects secondary to pituitaryadenoma. Systemic features include parathyroid adenomas or hyperplasia, pancreatic adenomas, pituitary adenomas, thyroid adenomas, adrenocortical adenomas, subcutaneous lipomas, hypoglycemic crisis, headaches, amenorrhea, diarrhea, weight loss, acromegaly, Cushing syndrome, hyperthyroidism, ulcer, aneurysm.

Sturge–Weber syndrome

Systemic features are phakomatosis with nevus flammeus of the face. Ocular features are ocular and CNS hemangiomas.

Sunrise Syndrome

Ocular features are fringe of IOL in pupil, decreased acuity, and glare.

Sunset Syndrome

Ocular features are superior fringe of IOL in pupil, IOL in vitreous humor, retinal detachment, decreased acuity, glare, zonular disinsertions.

Susac Syndrome

Ocular features include cotton-wool spot, central retinal vein occlusion. Systemic features include deafness, encephalopathy.

Tangier Syndrome (Alpha-Lipoprotein Deficiency, Familial HDL Deficiency)

Ocular features are corneal infiltrates, fine, dotted stromal opacities, most marked in posterior central third of corneal stroma, wasting of orbicularis oculi muscle. Systemic features include maculopapular rash, orange-yellow striped tonsils, hepatosplenomegaly, lymphadenopathy, intermittent diarrhea, bilateral motor weakness.

Tapetal-Like Reflex Syndrome

Ocular features are ring scotoma, discrete bright yellow spots inposterior polar region deep to the retinal vessels, tapetal-like reflex and retinitis pigmentosa maybe present in members of an equivalent family.

Tay–Sachs Disease

Systemic features include autosomal recessive (Ch15q, HEXA), GM2 gangliosidosis with deficiency of hexosominadase A, neurodegeneration.

Ocular features are cherry-red spot, optic atrophy.

Terson Syndrome (Subarachnoid Hemorrhage Syndrome)

Ocular features are Weakness of extraocular muscles, disarrangedand uncoordinated gaze, severe intra-ocular hemorrhage, preretinal hemorrhages, peripapillary

hemorrhages, papilledema secondary to nervus opticus sheath hemorrhages, pigmentary changes in macula and retina, preretinal membrane formation, vitreous detachment, amblyopia, anisocoria, bilateral detachment of the retina shave been associated with this disorder, epiretinal membranes (sequelae). Systemic features include sudden unconsciousness, elevated cerebrospinal fluid pressure.

Tolosa-Hunt Syndrome (Painful Ophthalmoplegia)

Ocular features are Steadily “growing” retro-orbital pain, ptosis, involvement of cranial nerves III, IV, VI, and first division of V, scintillating scotomata, sluggish pupil reaction to light, corneal sensitivity diminished, optic neuritis. Systemic features include inflammatory lesions of sinus cavernosus.

Transient Light Sensitivity Syndrome (Uveitis)

Ocular features are transient postoperative photosensitivity.

Traumatic Encephalopathy Syndrome (Postconcussion Syndrome, Post-Traumatic General Cerebral Syndrome, Punch-Drunk Syndrome)

Ocular features are nystagmus or Nystagmoid ocular movements, lower lid colobomas, dermoids. Systemic features are personality change, rigid face blankly, staggering gait, dysphonia.

Treacher–Collins Syndrome (Mandibulo facial Dysostosis)

Ocular features are antimongoloid palpebral fissures. Systemic features include mandibular hypoplasia, zygoma hypoplasia, choanal atresia autosomal dominant (Ch5q), clefting syndrome.

Treft Syndrome

Ocular features are optic atrophy, visual loss, ptosis; ophthalmoplegia. Systemic features include deafness by age of 14 years, myopathicchanges, balance difficulty.

Turcot syndrome

Systemic features are variant of familial adenomatous polyposis (autosomal dominant) with CNS neuroepithelial tumors, especially medulloblastoma and glioma, Ocular features are a typical CHRPE.

Turner Syndrome

Ocular features are antimongoloid palpebral fissures, cataracts; convergence insufficiency. Systemic features are XO, 1 in 2000 live female births, short stature, wide carrying angle, low hair line, webbed neck, primary gonadal failure, congenital heart defects.

UGH Syndrome (Uveitis-Glaucoma-Hyphema Syndrome)

Ocular features are uveitis, glaucoma, hyphema (UGH).

Ullrich Syndrome (Dyscraniopylophalangy, Ullrich-Feichtiger Syndrome)

Ocular features are microphthalmia to anophthalmia, hypertelorism, narrow lid fissures, strabismus, glaucoma, aniridia, cloudy cornea, corneal ulcers, and chorioretinal coloboma. Systemic features include hypoplastic mandible, broad nose, polydactyly, spina bifida, bicornuate uterus or septa vagina, congenital heart condition.

Unverricht Syndrome (Familial Myoclonia Syndrome, Lafora Disease)

Ocular features include amaurosis, laminated Lafora bodies in ganglion cell and inner nuclear layers of the retina, either intracellular or extracellular, in inner plexiform and nerve fibre layers, and in the optic nerve. Systemic features are major epilepsy, widespread myoclonus, dementia, tetraplegia, pseudobulbar palsy, generalized tonic-clonic seizure, behavioral changes, brisk tendon reflexes, cerebellar signs.

Urrets-Zavalia Syndrome

Ocular features are fixed dilated pupil, iris atrophy ssen after penetrating keratoplasty.

Usher Syndrome (Hereditary Retinitis Pigmentosa-Deafness Syndrome)

Ocular features are concentric contraction of visual fields, retinitis pigmentosa with dotted, fine pigmentation in midperiphery, bone-corpuscle configured pigment deposits mainly along the vessels toward the periphery, yellow-white dots in outer retina and choroid, poor night-sight. Systemic features include deaf-mutism, however, deafness isn't always complete, MS.

Uvea Touch Syndrome

Ocular features are corneal decompensation, endothelial dystrophy, retinal edema, pigment dispersion, painful eye, disorders of motility. Systemic features include uveal effusion syndrome: Idiopathic exudative detachments of choroid, membrane and retina, thought to arise from impaired posterior segment drainage usually related to scleral thickening.

Uyemura Syndrome (Fundus Albipunctatus with Hemeralopia and Xerosis)

Ocular features are nyctalopia, conjunctival xerosis, Bitot spots, white spots on the fundus.

Van Bogaert-Hozay Syndrome (Esotropia)

Ocular features are hypertelorism, hypoplastic cilia and eyebrows, ptosis, esotropia, astigmatism, myopia. Systemic features include facial dysplasia, broad nasal bridge and zygomatic arch, flat, wide nose, arched palate, skeletal anomalies with short, thick phalangeal joints, finger and toes appear infantile, flat nasal bridge, thickened cheeks, deformed ears, micrognathia.

Van Bogaert-Scherer-Epstein Syndrome (Familial Hypercholesterolemia Syndrome, Primary Hyper lipidemia)

Ocular features include xanthelasma, arcus juveniles of the cornea, lipid keratopathy, cataract, retinopathy with yellowish deposits and cholesterol crystals have been reported but are more rare manifestations. Systemic features are xanthelasmatosis of skin and tendons, progressive atherosclerosis, cardiac insufficiency, cardiac infarcts, dementia, progressive ataxia, cerebral infarction, polyneuropathy.

Van Der Hoeve Syndrome (Brittle Bone Disease, Eddowes Syndrome, Ekman Syndrome, Lobstein Syndrome, Osteogenesis Imperfecta, Osteopsathyrosis, Spurway Syndrome, Vrolik Syndrome)

Ocular features are glaucoma, blue sclera, keratoconus, cataract, optic nerve atrophy, retinopathy, retinal detachment. Systemic features include brittle bones, deafness, and hyperflexibility of ligaments, dental defects, and developmental delay.

Velocardiofacial Syndrome (Di-George Syndrome)

Ocular features are retinal vascular tortuosity, posterior embryotoxon, narrow palpebral fissures, suborbital discoloration, small optic nerves, iris nodules, cataracts, prominent corneal nerves, strabismus, hyperopia, myopia, astigmatism, anisometropica stigmatism. Systemic features are birth defect, learning disorder, ventricular septal defect with or without the tetralogy of Fallot, rightside daortic arch, prominent nose, retrognathia, helical thickening, small auricles, auricular protrusion, microcephaly, small stature, inguinalor omphalocele , scoliosis, slender hands and digits, small vermis, small posterior fossa, developmental delay, heart malformations, late onset psychosis (Hassan et al., 2018).

Visual Disorientation Syndrome (Riddoch Syndrome)

Ocular features include agnosia, stereoscopy and central vision unimpaired, homonymous quadrant anopsia. Systemic features are contralateral numbness and tingling, loss of static or postural sensation when post central convolution affected.

Vitreous Tug Syndrome (Vitreous Wick Syndrome)

Ocular features are sensation of sunshine flashes thanks to vitreous pullon the retina, irregular pupil, vitreous strands passing through pupil to connect to corneal woundor scar, loss of foveal reflex on ophthalmo scopic examination, circumscribed retinal edema, occasional posterior detachment of the retina.

Vogt-Koyanagi–Harada syndrome

Ocular features are bilateral granulomatous panuveitis. Systemic features include multisystem disease, vitiligo, alopecia, deafness, tinnitus, sterile meningo encephalitis and cranial neuropathies.

Zollinger-Ellison Syndrome (Multiple Endocrine Adenomatosis Partial Syndrome, Polyglandular Adenomatosis Syndrome)

Ocular features are scotomata according to size and position of pituitary tumors, optic nerve atrophy, papilledema, bilateral extraocular muscle metastases. Systemic features include enter itis and/or peptic ulcers, malignant or benign tumor of islet cell of the pancreas, hypersecretion, vomiting, diarrhea, poly glandular

adenomatosis, and endocrine involvement.

Von Bekhterev-Strumpell Syndrome (Ankylosing Spondylitis, Bekhterev Disease, Marie-Strumpell Spondylitis, Pierre-Marie Syndrome, Rheumatoid Spondylitis)

Ocular features are nongranulomatous anterior uveitis, optic nerveatrophy (occasionally), hypopyon, band keratopathy, spontaneous hyphema. Systemic features are spondylitis of vertebra and sacroiliac joints, ankylosis, general arthralgia, kyphosis, scoliosis, displaced head and total rigidity of spine.

Von Reuss Syndrome (Galactokinase Deficiency, Galactosemia, Galactosemic Syndrome)

Ocular features are searching-type nystagmus, bilateral nuclear orcortical cataracts appear clinically as oil droplets, bilateral zonular cataracts with fine punctateopacities within the lens periphery. Systemic features include vomiting, refusal of food, diarrhea, weight loss, hepatomegaly with ascites, jaundice, galactosuria, amino aciduria, dehydration, hypoglycemic crisis, failure to thrive, hypotonia, lethargy, severe mental and neurologic manifestations. Von-Hippel Lindau Autosomal dominant (Ch3p, VHL gene), phakomatosis with retinal capillary hemangiomas, CNS hemangioblastomas, renal cell carcinomas, and other tumors.

Waardenburg Syndrome

Ocular features are heterochromia, hypertelorism Hyperplasia of the medial portions of the eye brows, hypertelorism, blepharophimosis, strabismus, heterochromia iridis, aniridia, microcornea, cornea plana, microphakia, abnormal fundus pigmentation, hypoplasia of optic nerve, synophrys, poliosis, hypo pigmentation and hypoplasia of retina and choroid, epicanthus, lateral displacement of inferior puncta, lenticonus, under development of orbital bones, lateral displacement of inner canthi, hypopigmented iris. Systemic features are autosomal dominant (PAX3), white forelock, deafness. Congenital deafness, unilateral deafness or deaf mutism, broad and high nasal root with absent nasofrontal angle, albinotic hair strain (unilateral), faint patches of skin pigmentation, pituitary tumor, nasal atresia, white forelock.

Werner Syndrome

Ocular features are absence of eyelashes and scanty eyebrows, blue sclera, juvenile cataracts, bullous keratitis, trophic corneal defects, paramacular retinal degeneration, proptosis, telangiectasia of lid, astigmatism, nystagmus, presbyopia, uveitis. Systemic features include leanness, short stature (160 cm maximum), thin limbs, short, deformed fingers, small mouth, early baldness, stretched, atrophic skin (scleropoikiloderma), telangiectasia and trophic indolent ulcers on toes, heels and ankles, arteriosclerosis with secondary heart failure .

CONCLUSION

It is concluded that, the important role of the multidisciplinary cooperation to improve and sustain management of patients affected with eclectic ocular syndrome and their systemic disorders with eye repercussion is specifically addressed.

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