# OCULAR SYNDROMES

Walter J. Geeraets, M.D.

THIRD EDITION

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# Preface

The third edition of *Ocular Syndromes* has been expanded to a total of 436 entries, included among which are syndromes of rare occurrence. Despite the attempted completeness, some syndromes may have been omitted unintentionally. Additions have also been made to tables and the bibliography. Three cross indices that provide identification of a syndrome from almost every point of view have been widely expanded and a new index of symptoms and clinical findings has also been added.

The purpose as outlined in the preface to the second edition has been maintained by compiling those clinical entities often difficult to remember. The tabulated presentation in alphabetical order offers a quick reference for daily clinical use. Terse and succinct wording has been used by necessity to ensure this goal.

Since many ocular manifestations and their association with other systemic findings provide guidance in recognizing an existing medical entity, *Ocular Syndromes* has proven to be of value not only for ophthalmologists, but also for internists and pediatricians as well. This is also reflected in the many references pertaining to those clinical fields. The book's usefulness in student and resident training has been well established.

Because of the many synonyms and eponyms of syndromes their listing has been printed as the first chapter of the book. The reader is well advised to consult this chapter first in order to find the syndrome in the alphabetically tabulated form under the name by which it is discussed in more detail.

I wish to extend my gratitude to Miss Florence Caywood Garrett and to Mrs. Fiona Porter Ellen for their assistance in the many facets of the preparation of the book. In reviewing the bibliography and the manuscript, their efforts have greatly contributed to the completeness and accuracy of this edition. My thanks and appreciation go as well to the staff of the Tompkins McCaw Medical Library for their help in reference research.

*Richmond, Virginia*

WALTER J. GEERAETS, M.D.

# Preface to the Second Edition

Encouraged by many readers of 'Ocular Syndromes' and by their stimulating remarks, several additions have been made in this second edition.

The number of syndromes covered has been expanded from the original 138 to 197 and includes those more recently described in the literature. Also the information given on each syndrome has been extended. References for further reading have been added and the original publication of the author(s) whose name(s) the syndrome bears is given. The 'Cross Reference of Syndromes based on Ocular Manifestations' also includes now the most outstanding systemic findings. Syndromes presenting similar features have been grouped and tabulated for easier comparison. Finally, a glossary has been added for the readers not familiar with ophthalmological terminology.

Some diseases, not truly belonging under the heading and definition of the term "syndrome," have been included since they are found occasionally referred to as such in the literature. This, however, will probably add rather than detract from the versatility of the book.

It is hoped that this second edition will serve the purpose of making these syndromes more easily recognizable in the constant striving for a more accurate diagnosis.

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# Contents

## NOTICE

Names of syndromes carrying the presyllable "von" or "van" are listed under V in the tabulated part of this book. In the "cross indices" they are listed under the authors' names, disregarding the presyllable, thus insuring adequate coverage (i.e., von Hippel-Lindau is quoted in the tabulated part under "V", but in the cross indices under "H"). As an exception they are listed under *both* in the alphabetical index of Syndromes: Their Synonyms and Eponyms. A few syndromes which are listed separately have been considered by some authors to be part or degrees of expression of others. In those instances reference has been made to this effect.

# Syndromes: Their Synonyms and Eponyms

Abducens-facial hemiplegia alternans (see: Millard-Gubler syndrome)
Aberfeld (blepharophimosis, exotropia, myopia, congenital cataract—myopathy, dwarfism, hypertrichosis, hypoplastic facial bones)
A-β-Lipoproteinemia (see: Bassen-Kornzweig syndrome)
Achard (myopia, lens dislocation—mandibulofacial dysostosis, skeletal anomalies, arachnodactyly)
Acosta (blurred vision, impaired color vision and light adaptation—restlessness, headaches, cyanosis, unconsciousness)
Acoustic neuroma (see: Cushing [2] syndrome)
Acrocephalosyndactylism syndrome (see: Apert syndrome)
Acrocranio-dysphalangy (see: Apert syndrome)
Acrodermatitis enteropathica (see: Danbolt-Closs syndrome)
Acrodynia (see: Feer syndrome)
Acrodysplasia (see: Apert syndrome)
Acute encephalopathy (see: Reye syndrome)
Acute histiocytosis (see: Letterer-Siwe syndrome)
Acute infectious neuritis (see: Guillain-Barré syndrome)
Acute polyradiculitis (see: Guillain-Barré syndrome)
Addison (keratoconjunctivitis, cataracts, blepharitis—moniliasis, tetany, skin pigmentation, weakness)
Addison's pernicious anemia (see: Biermer syndrome)
Adherence syndrome (see: Johnson syndrome)
Adherent lateral rectus syndrome (see: Johnson syndrome)
Adie (myotonia—absence of ankle or knee jerk)
Adrenal medulla tumor (see: Suprarenal-sympathetic syndrome)
Adrenal-sympathetic syndrome (see: Suprarenal-sympathetic syndrome)
Adrenocortical neuroblastoma with orbital metastasis (see: Hutchinson syndrome)
Adrenocortical syndrome (see: Cushing [1] syndrome)
Agranulocytic angina syndrome (see: Agranulocytosis syndrome)
Agranulocytosis syndrome (scleral and conjunctival icterus, conjunctival and retinal hemorrhages—malaise, swollen joints, sore throat, sepsis; hemorrhages, progressive severe pneumonia)
Aicardi (microphthalmia, retinopathy—infantile spasms, head deformities, skeletal anomalies, facial asymmetry)
Åland disease (see: Forsius-Eriksson)
Albers-Schönberg (optic atrophy—osteosclerosis, multiple fractures, jaundice, hepatosplenomegaly)
Albright (unilateral proptosis, optic nerve involvement—osteitis fibrosa, skin pigmentations, endocrine dysfunction)
Albright's hereditary osteodystrophy (see: Pseudohypoparathyroidism syndrome)
Alfano-Alfano (see: Superior vena cava syndrome)
α-Lipoprotein deficiency (see: Tangier syndrome)
Alport (lens changes, fundus albi punctatus, hemorrhagic nephritis, deafness)
Alternating oculomotor paralysis (see: Weber syndrome)
Amalric-Dialinas (see: Dialinas-Amalric syndrome)
Amaurosis congenita (see: Leber's tapetoretinal dystrophy)
Amaurosis fugax syndrome (blindness, retinal arteriolar spasm—hypertension)
Amèndola (entropium or ectropium, trichiasis, skin blisters around eyebrows, anterior lens pole haziness—Brazilian pemphigus resembling pemphigus foliaceous)
Amino diabetes (see: Fanconi syndrome)
Andersen-Warburg (bilateral microphthalmos—mental retardation)
Andogsky (subcapsular to dense cataracts, keratoconus—atopic dermatitis)
Aneurysm of internal carotid artery syndrome (see: Foramen lacerum syndrome)
Angelucci (conjunctivitis, tachycardia, excitability, labile vasomotor system)
Angiokeratoma corporis diffusum syndrome (see: Fabry-Anderson syndrome)
Angiomatosis retinae (see: von Hippel-Lindau syndrome)
Angioosteohypertrophy syndrome (see: Klippel-Trenaunay-Weber syndrome)
Angiospastic ophthalmo-auricular syndrome (see: Bazzana syndrome)
Angle tumor syndrome (see: Cushing [2] syndrome)

Anhidrotic ectodermal dysplasia (see: Hereditary ectodermal dysplasia)
Ankylosing spondylitis (see: von Bechterew-Strümpell syndrome)
Anomalous leukocytic inclusions with constitutional stigmata (see: Chediak-Higashi syndrome)
Anterior chamber cleavage syndrome (corneal opacities, iris-corneal adhesions, glaucoma, a. o.)
Anterior cleavage syndrome (see: Anterior chamber cleavage syndrome)
Anterior diencephalic autonomic epilepsy syndrome (see: Diencephalic epilepsy syndrome)
Anterior interosseous nerve syndrome (see: Kiloh-Nevin syndrome)
Anterior-segment traumatic syndrome (see: Frenkel syndrome)
Anterior spinal artery syndrome (nystagmus—quadriplegia, disturbed intestinal and bladder function with incontinence, loss of discriminative sensations)
Antimongolism (antimongolism slant of lid fissure, blepharochalasis—hypertony, large ear lobes, mental retardation, cardiac disease, pyloric stenosis, micrognathia)
Anton (blindness, denial of blindness—confabulation, allochiria)
Aortic arch syndrome (see: Takayasu syndrome)
Apert (cranial deformities, loss of vision, ophthalmoplegia—syndactyly)
Aphasia-agnosia-apraxia syndrome (see: Arnold Pick syndrome)
Aplasia axialis extracorticalis congenita (see: Pelizaeus-Merzbacher syndrome)
Arachnodactyly (see: Marfan syndrome)
Argyll Robertson (loss of pupillary reflex—involvement of the CNS)
Arnold-Chiari (visual field defect, nystagmus—pyramidal tract signs, vertebral malformations)
Arnold Pick (apperceptive blindness—progressive dementia, cerebral atrophy)
Ascher (lack of tone of orbital fascia, blepharochalasis, protrusion of lacrimal gland—goiter, reduplication upper lip)
Ataxia-telangiectasia syndrome (see: Louis-Bar syndrome)
Ataxic diplegia (see: Fanconi-Türler syndrome)
Atopic cataract syndrome (see: Andogsky syndrome)
Atresia of the foramen Magendie (see: Dandy-Walker syndrome)
Atrophia oculi congenita (see: Anderson-Warburg syndrome)
Atypical ectodermal dysplasia (see: Marshall syndrome)
Autonomic epilepsy syndrome (see: Diencephalic epilepsy syndrome)
Autoscopic syndrome (see: Mirror image syndrome)
AV syndrome (deviation on upward or downward gaze)
Avelli (see: Cestan-Chenais syndrome)
Axenfeld (glaucoma, posterior embryotoxon, ring-shaped corneal opacity)
Axenfeld-Schürenberg (congenital cyclic oculomotor paralysis)

$B_1$ deletion syndrome (see: Cri-du-chat syndrome)
Baader's dermatostomatitis (see: Stevens-Johnson syndrome)
Babinski-Nageotte (Horner syndrome, nystagmus—cerebellar hemiataxia, hemiparesis)
Bakwin-Krida (see: Cranio-metaphyseal dysplasia)
Balint (optic ataxia, disturbance of visual fixation—loss of body coordination)
Bardet-Biedl (see: Laurence-Moon-Bardet-Biedl syndrome)
Barnard-Scholz (see: Ophthalmoplegic-retinal degeneration)
Barré-Liéou (corneal hypesthesia, reduced vision—headache, dizziness, ear noises, neck pain)
Basal-frontal syndrome (see: Foster Kennedy syndrome)
Basedow (exophthalmos, 'Graefe sign', disturbances in motility—tachycardia, restlessness, anxiety, atrial fibrillation)
Bassen-Kornzweig (retinitis pigmentosa—steatorrhea, acanthocytosis, neuropathy)
Batten-Mayou (retinal pigment disturbance, optic atrophy—amaurotic idiocy, convulsions)
Battered baby syndrome (see: Silverman syndrome)
Battered child syndrome (see: Silverman syndrome)
Bazzana (visual field contraction, retinal vessel tortuosity—progressive deafness)
Bechterew, von-Strümpell (nongranulomatous uveitis, optic atrophy—spondylitis of vertebrae, ankylosis)

Bechterew's disease (see: von Bechterew-Strümpell syndrome)
Beck syndrome (see: Anterior spinal artery syndrome)
Behçet (iritis, keratitis, uveitis—ulcerations of mucous membranes, skin lesions)
Behr (heredofamilial optic atrophy—abortive hereditary ataxia, mental deficiency)
Bends (see: Caisson syndrome)
Benedikt (oculomotor paralysis—contralateral hemichorea)
Benign retinohypophysary syndrome (see: Retinohypophysary syndrome)
Berardinelli-Seip (punctate corneal infiltrates—gigantism, liver cirrhosis, hepatosplenomegaly, skin hyperpigmentation, hypertrichosis)
Bernard-Horner (see: Schaumann syndrome)
Besnier-Boeck-Schaumann (see: Schaumann syndrome)
Bielschowsky-Jansky (see: Tay-Sachs syndrome)
Bielschowsky-Lutz-Cogan (paralysis internal rectus muscle, contralateral nystagmus on lateral gaze)
Biemond (retinal pigment degeneration, iris coloboma—genital dystrophy, mental retardation)
Biermer (retinal hemorrhages, optic neuritis and atrophy,—megaloblastic anemia, hypochlorhydria, stomatitis, diarrhea, paresthesias, incoordination)
Bing (see: Bing-Neel syndrome)
Bing-Neel (ptosis, reduced vision, paralysis extraocular muscles, glaucoma, retinopathy—macroglobulinemia associated with CNS symptoms, e.g. chronic encephalopathy, peripheral neuropathy, strokes, subarachnoidal hemorrhages)
Bird-headed dwarf syndrome (see: Seckel syndrome)
Blatt (microphthalmos, hypertelorism—cranial deformities)
Blepharochalasis with struma and double lip (see: Ascher syndrome)
Blind spot syndrome (see: Swan syndrome)
Bloch-Sulzberger (retinal pseudoglioma, optic nerve involvement, nystagmus—bullous skin eruptions and pigmentations)
Blue diaper syndrome (see: Drummond syndrome)
Boeck's sarcoid (see: Schaumann syndrome)
Bogaert, van-Hozay (hypertelorism, squint, astigmatism, myopia—facial dysplasia, short extremities, skeletal anomalies, acrocyanosis)
Bogaert, van-Nijssen syndrome (see: Greenfield syndrome)
Bogaert, van-Scherer-Epstein (xanthelasma, arcus juvenilis, lipid keratopathy, retinopathy—multiple xanthomatoses of skin, coronary insufficiency)
Bogorad (unilateral lacrimation—excessive salivation)
Bonnet-Dechaume-Blanc (arteriovenous retinal angiomas—angiomas of mesencephalon and thalamus)
Bonnevie-Ullrich (congenital cataracts—pterygial folds of neck, hyperelastic skin, edema of neck and extremities, hypertrichosis)
Bourneville (retinal tumor—adenoma sebaceum, epilepsy, mental deficiency)
Bourneville-Pringle (see: Bourneville syndrome)
Brachman-deLange (see: deLange syndrome)
Brachymorphy with spherophakia (see: Marchesani syndrome)
Brailsford-Morquio (see: Morquio-Brailsford syndrome)
Brain dysfunction syndrome (visual field defects, speech difficulties—physical brutality, manic behavior, seizures, sexual assault, impaired memory)
Brandt (see: Danbolt-Closs syndrome)
Bremer's status dysraphicus (see: Passow syndrome)
Brown (bilateral ptosis with backward head tilt, superior oblique palsy simulated by shortened superior oblique tendon sheath)
Brown-Marie ataxic syndrome (nystagmus, strabismus, retinitis pigmentosa, optic atrophy—hereditary ataxia)
Brown-Sequard (nystagmus, possible sluggish pupil reaction—sensory disturbances)
Bruns (photopsia, amaurosis, gaze paresis—headache, nausea, vertigo, ataxia, tachycardia, a.o.)
Burnett (band-shaped keratopathy, conjunctivitis—headache, nausea, dizziness, depression, mental confusion)
Burning feet syndrome (see: Gopalan syndrome)

Caffey (circum-ocular edema and tenderness, transient proptosis, conjunctivitis—cortical hyperostosis with tissue swelling, anemia, fever, pleurisy, a.o.)
Caisson (diplopia, nystagmus, transient blindness—severe joint pain, dyspnea, vertigo, coma)
Calcinosis universalis (see: Conradi syndrome)
Camurati-Engelmann (see: Engelmann syndrome)
Canalis opticus syndrome (reversible or irreversible blindness after head injury, absent pupil reactions)
Capgras (illusion of double perception—hallucinations)
Capsular exfoliation syndrome (see: Exfoliation syndrome)
Cardiorespiratory-obesity syndrome (venous congestion with retinal exudates and hemorrhages, papilledema—excessive obesity, cyanosis, dyspnea, drowsiness, heart failure, reduced consciousness)
Carotid artery syndrome (homolateral transient visual loss, photopsia, cholesterol plaques in retinal arteries, hemianopsia—contralateral weakness of arm and leg, hemisensory disturbances, confusion)
Carotid artery-cavernous sinus fistula syndrome (exophthalmos, secondary glaucoma, papilledema, retinopathy—unilateral headache, subjective buzzing intracranial noise)
Carotid vascular insufficiency syndrome (see: Carotid artery syndrome)
Carpal tunnel syndrome (see: Lefi syndrome)
Carpenter (see: Apert syndrome)
Carrefour hypothalamique (see: Hypothalamique-carrefour syndrome)
Cartilaginous-arthritic-ophthalmic-deafness syndrome (uveitis—deafness, rheumatic arthritis, joint dislocations)
Cat-cry (5p−) syndrome (see: Cri-du-chat syndrome)
Cat-eye syndrome (iris coloboma, hypertelorism—preauricular fistulae, umbilical hernia)
Cat-scratch-oculoglandular syndrome (see: Parinaud oculoglandular syndrome)
Cavernous sinus-nasopharyngeal tumor syndrome (see: Foix syndrome)
Cavernous sinus neuralgia syndrome (see: Foix syndrome)
Cavernous sinus syndrome (see: Foix syndrome)
Central nervous system deficiency syndrome (visual loss, bitemporal optic atrophy—deafness, tinnitus, unsteady gait, tingling in legs)
Cephalo-oculo-cutaneous telangiectasis (see: Louis-Bar syndrome)
Cerebellar peduncle syndrome (see: Weber syndrome)
Cerebellomedullary malformation (see: Arnold-Chiari syndrome)
Cerebellopontine angle syndrome (see: Cushing [2] syndrome)
Cerebral gigantism (see: Sotos syndrome)
Cerebrofacial-reno-arthro-syndactylia (trichiasis; slanted, asymmetric lid fissures, blepharitis, visual field contraction—mild oligophrenia, microcephaly, arthropathy, renal anomaly)
Cerebro-hepato-renal syndrome (see: Zellweger syndrome)
Cerebro-hepato-renal syndrome (see: Smith-Lemli-Opitz syndrome)
Cerebro-oculo-facio-skeletal syndrome (microphthalmia, cataracts, blepharophimosis—microcephaly, hypotonia, skeletal deformities, respiratory infections, facial anomalies)
Cerebroretinal arteriovenous aneurysm syndrome (see: Bonnet-Dechaume-Blanc syndrome)
Cerebroretinal degeneration (see: Batten-Mayou syndrome)
Cervical sympathetic paralysis syndrome (see: Horner syndrome)
Cervico-oculo-acusticus syndrome (see: Wildervanck syndrome)
Cestan (1) (see: Cestan-Chenais syndrome)
Cestan (2) (see: Raymond syndrome)
Cestan-Chenais (Horner syndrome, nystagmus—flaccid paralysis of soft palate and vocal cord, contralateral hemiplegia, ataxia)
Chandler (glaucoma, iris atrophy, corneal dystrophy, corneal edema)
Charcot-Marie-Tooth (nystagmus, optic atrophy—progressive muscular atrophy)
Charcot-Wilbrand (visual agnosis, loss of ability to revisualize images)
Charlin (inflammation of anterior segment of the eye, orbital pain—rhinorrhea)
Chauffard-Still (see: Felty syndrome)

Chediak-Higashi (decreased iris pigmentation, corneal edema, elevated disc-hepatosplenomegaly, recurrent infections)
Chiasmal syndrome (see: Cushing [2] syndrome)
Child abuse syndrome (see: Silverman syndrome)
Chondrodystrophia foetalis hypoplastica (see: Conradi syndrome)
Chondroectodermal dysplasia (see: Ellis-van Creveld syndrome)
Chondro-osteo-dystrophy (see: Hurler syndrome)
Chorioretinal infarction syndrome (see: Hollenhorst syndrome)
Christ-Siemens-Touraine (see: Hereditary ectodermal dysplasia)
Chromosome 4 partial deletion syndrome (see: Wolf syndrome)
Chromosome 13q− partial deletion (long arm) syndrome (antimongoloid slant of lid fissures, epicanthus, choroidal coloboma—mental and physical retardation, meningocele, brain anomalies, simian crease, hypoplasia of phalanges, short neck, dental anomalies)
Chromosome 18 partial deletion (long arm) syndrome (nystagmus, posterior staphyloma, oblique optic disc—dwarfism, microcephaly, mental retardation, midface dysplasia, abnormal fingerprints, prominent antihelix)
Chromosome 18 partial deletion (short arm) syndrome (hypertelorism, epicanthal folds, ptosis, strabismus—short stature, mental retardation, low set ears, dysphagia, moon face, microcephaly, congenital alopecia)
Chromosome 21 partial deletion syndrome (see: Antimongolism syndrome)
Chronic idiopathic jaundice (see: Dubin-Johnson syndrome)
Chronic renal tubular insufficiency syndrome (see: pseudohypoparathyroidism syndrome)
Claude (III and IV nerve paralysis—contralateral hemianesthesia and hemiataxia)
Claude Bernard-Horner syndrome (see: Horner syndrome)
Clefting syndrome (see: Wagner syndrome)
Cleidocranial dysostosis (see: Cranio-cleido-dysostosis syndrome)
Cloverleaf skull (see: Extreme hydrocephalus syndrome)
Coats' disease (see: General information in van Bogaert-Scherer-Epstein syndrome)
Cockayne (retinal pigmentary degeneration, optic atrophy—dwarfism, deafness, mental retardation)
COFS syndrome (see: Cerebro-oculo-facio-skeletal syndrome)
Cogan (1) (interstitial keratitis—vestibuloauditory symptoms)
Cogan (2) (conjugate muscle palsy, head tilt, frequent blinking—cerebellar signs)
Cogan-Guerry (wavy and curly corneal epithelial lines with epithelial microcysts and geographic superficial corneal pattern, abnormal thickened corneal epithelial basement membrane)
Cole-Rauschkolb-Toomey (see: Zinsser-Engman-Cole syndrome)
Compressed air illness (see: Caisson syndrome)
Cone dysfunction (macular degeneration, field defects, decreased visual acuity)
Congenital brevicollis (see: Klippel-Feil syndrome)
Congenital calcifying chondrodystrophy (see: Conradi syndrome)
Congenital dyslexia syndrome (convergence insufficiency, refractive errors, abnormal optokinetic nystagmus [all rare]—signs of mild parietal dysfunction, lack of integration of visual and auditory stimuli)
Congenital encephalo-ophthalmic dysplasia (see: Krause syndrome)
Congenital epiblepharon-inferior oblique insufficiency syndrome (narrow interpupillary distance, epicanthus, inferior oblique insufficiency, keratitis—chubby cheeks)
Congenital facial diplegia (see: Möbius syndrome)
Congenital familial dysautonomia (see: Riley-Day syndrome)
Congenital generalized lipodystrophy (see: Berardinelli-Seip syndrome)
Congenital hyperphosphatasemia (see: Paget syndrome)
Congenital keratoconus posticus circumscriptus syndrome (see: Haney-Falls syndrome)
Congenital muscular hypertrophy-cerebral (see: de Lange syndrome)
Congenital myotonia syndrome (see: Thomsen syndrome)
Congenital oculofacial paralysis (see: Möbius syndrome)
Congenital paralysis of 6th and 7th nerves (see: Möbius syndrome)
Congenital poikiloderma with juvenile cataract (see: Rothmund syndrome)
Congenital retinoschisis (see: Wagner syndrome)

Congenital rubella syndrome (see: Rubella syndrome)
Congenital spinocerebellar ataxia-cataract-oligophrenia syndrome (see: Marinesco-Sjögren syndrome)
Congenital word blindness of Hermann (see: Congenital dyslexia syndrome)
Conjunctivo-urethro-synovial (see: Reiter syndrome)
Conradi (hypertelorism, bilateral congenital cataracts—short limbs, hip contraction, abnormal facies, mental retardation, heart defect, skin anomalies)
Cortico-striato-spinal degeneration (see: Creutzfeldt-Jakob syndrome)
Costen (supraorbital pain—dysfunction temporomandibular joint, neuralgia, facial pain, dizziness, impaired hearing)
Cranial arteritis (see: Temporal-arteritis syndrome)
Cranio-cervical syndrome (convergence insufficiency, nystagmus, vestibular impairment, disturbed accommodation, asthenopia, fogging vision, double vision—headache, vertigo, dizziness)
Cranio-cleido-dysostosis (proptosis, antimongoloid slant of lid fissure—saddle nose, prominent forehead, hyperlaxia of joints, hemiplegia, mental deficiency)
Cranio-facial dysostosis (see: Crouzon syndrome)
Cranio-metaphyseal dysplasia (hypertelorism, nystagmus, lagophthalmos, optic atrophy—thick and dense base of bony skull, deafness, late dentition)
Cranio-orbito-ocular dysraphia (see: Blatt syndrome)
Craniotelencephalic dysplasia (see: Extreme hydrocephalus syndrome)
Creutzfeldt-Jakob (cortical blindness—pyramidal signs, loss of reflexes, tremor, rigidity, ataxia, mental retardation)
Cri-du-chat syndrome (hypertelorism, strabismus, epicanthal folds, antimongoloid slanting—mental retardation; abnormal dermatoglyphics, broad nasal root, low-set ears, micrognathia, growth retardation)
Critical allergic conjunctivitis (see: Angelucci syndrome)
Crocodile tear (see: Bogorad syndrome)
Crouzon (exophthalmos, strabismus, papilledema—prognathism, maxilla atrophy, deformity anterior fontanel)
CRST syndrome (see: Diffuse keratoses)
Crying cat syndrome (see: Cri-du-chat syndrome)
Cryptophthalmia syndrome (eyebrows partially missing, skin of forehead covers one or both eyes, microphthalmia, lens anomalies—syndactyly, coloboma alae nasi, urogenital abnormalities, abnormal hairline)
Cryptophthalmos-syndactyly syndrome (see: Cryptophthalmia syndrome)
Curtius (hypertelorism, amblyopia, hypotrichosis—hypodontia, reduced thermal regulation, a.o.)
Cushing (1) (ocular muscle palsies, visual field changes, optic atrophy—obesity, hirsutism, skin pigmentation, osteoporosis)
Cushing (2) (paralysis V-VIII, decreased corneal sensitivity—tinnitus, deafness, defect in labyrinthine function)
Cushing (3) (bilateral hemianopsia, optic atrophy)
Cutis hyperelastica (see: Ehlers-Danlos syndrome)
Cyclic oculomotor paralysis (see: Axenfeld-Schürenberg syndrome)
Cystic fibrosis syndrome (ischemic retinopathy—recurrent pulmonary infections, pancreatic achylia, salty skin)
Cystine storage-aminoaciduria-dwarfism syndrome (see: Lignac-Fanconi syndrome)
Cystinosis syndrome (see: Lignac-Fanconi syndrome)

Dabre-Lamy-Lyell (see: Fuchs-Lyell syndrome)
Dacryosialoadenopathy (see: Mikulicz-Radecki syndrome)
Danbolt-Closs (blepharitis, conjunctivitis, ectropion, photophobia—symmetrical skin eruptions, stomatitis, alopecia)
Dancing eyes syndrome (see: Kinsbourine syndrome)
Dandy-Walker (ptosis, paralysis VI nerve, papilledema—hydrocephalus)
Darier-White (bilateral corneal subepithelial infiltrations—reddish firm papules, seborrheic dermatitis, alopecia, genital lesions)
Deaf-mutism-retinal degeneration syndrome (see: Dialinas-Amalric syndrome)

Degos (atrophic skin of eyelids, diplopia, choroidal changes—skin lesions, gastrointestinal involvement)
de Grouchy (see: Chromosome 18 partial deletion [long arm] syndrome)
Déjean (exophthalmia, diplopia—superior maxillary pain, numbness V 1 and 2)
Déjérine-Klumpke (Horner syndrome—paralysis and atrophy of small muscles of upper extremities)
Déjérine-Roussy (hemianopsia—transient hemiplegia, hemiataxia, choreo-athetotic movements)
deLange (1) (antimongoloid slant, palpebral fissures, myopia, anisocoria, pale discs—mental retardation, multiple skeletal abnormalities)
Denial-visual hallucination syndrome (see: Anton syndrome)
Dental-ocular-cutaneous syndrome (glaucoma, entropion—skin lesions, syndactyly, abnormal dentition)
Dermatogenous cataract syndrome (see: Andogsky syndrome)
Dermatostomatitis (see: Stevens-Johnson syndrome)
Dermatostomatoophthalmic syndrome (see: Behçet syndrome)
Destructive iridocyclitis and multiple joint dislocations (see: Hilding syndrome)
Developmental dyslexia of Critchley (see: Congenital dyslexia syndrome)
Devic (optic neuritis—ascending myelitis)
Diabetic glomerulosclerosis (see: Kimmelstiel-Wilson syndrome)
Dialinas-Amalric (retinal pigmentary anomalies—deaf-mutism)
Diaphyseal dysplasia (see: Engelmann syndrome)
Diencephalic epilepsy (proptosis, lacrimation, pupillary anomalies—abdominal pain, headache, restlessness, vasodilatation of the skin, salivation, elevated blood pressure)
Diffuse angiokeratosis (see: Fabry-Anderson syndrome)
Diffuse keratoses (skin lesion of lids, visual impairment, hydrophthalmos, corneal changes, retinopathy—ichthyosis, allergic manifestations, keratosis palmoplantaris, telangiectasia, a.o.)
Dissociation of lateral gaze syndrome (see: Raymond syndrome)
Diver's palsy (see: Caisson syndrome)
Divergence paralysis (see: Parinaud syndrome)
Dolichosternomelia (see: Marfan syndrome)
Dolichosternomelia-arachnodactyly-hydrochondroplasia-dystrophia mesodermalis congenita (see: Marfan syndrome)
Dominant hemisphere syndrome (see: Gerstmann syndrome)
Dorsolateral medullary syndrome (see: Wallenberg syndrome)
Double whammy syndrome (voluntary propulsion of eyes)
Down (hypertelorism, slanted eyelid fissures, myopia, iris spots, lens opacities—mental retardation, skeletal and heart anomalies)
Drepanocytic anemia (see: Herrick syndrome)
Dresbach's syndrome (see: Herrick syndrome)
Drummond (epicanthal folds, decreased vision, nystagmus, optic and peripheral retinal atrophy, papilledema—depressed bridge of nose, vomiting, constipation)
D-trisomy syndrome (see: Trisomy-D syndrome)
Duane (primary global retraction, narrowing palpebral fissure)
Dubin-Johnson (scleral and conjunctival jaundice—abnormal pain, right hypochondrium, a.o.)
Dwarf-cardiopathy syndrome (see: Schönenberg syndrome)
Dwarfism-hepatomegaly-obesity-juvenile diabetes syndrome (see: Juvenile diabetes-dwarfism-obesity syndrome)
Dwarfism with retinal atrophy and deafness (see: Cockayne syndrome)
Dyscephalic-mandibulo-oculo-facial syndrome (see: Hallermann-Streiff syndrome)
Dyschondroplasia syndrome (narrow optic foramen and supraorbital fissure, ophthalmoplegia, optic atrophy—joint deformities, scoliosis, unilateral shortening of leg, facial asymmetry)
Dyscontrol syndrome (see: Brain dysfunction syndrome)
Dyscraniopylophalangy (see: Ullrich syndrome)
Dysencephalia splanchnocystica (see: Gruber syndrome)
Dysgenesis mesodermalis corneae et irides (see: Riegers syndrome)

Dysgenesis mesostromalis (see: Riegers syndrome)
Dysgenesis neuroepithelialis retinae (see: Leber's tapetoretinal dystrophy)
Dyskeratosis congenita with pigmentation (see: Zinsser-Engman-Cole syndrome)
Dyskeratosis follicularis syndrome (see: Darier-White syndrome)
Dyslexia syndrome (see: Congenital dyslexia syndrome)
Dysostosis cranio-facialis (see: Crouzon syndrome)
Dysostosis multiplex (see: Hurler syndrome)
Dysplasia epiphysealis congenita (see: Conradi syndrome)
Dystrophia adiposogenitalis (see: Frölich syndrome)
Dystrophia mesodermalis congenita (see: Marfan syndrome)
Dystrophia mesodermalis congenita hyperplastica (see: Marchesani syndrome)
Dystrophia myotonica syndrome (see: Myotonic dystrophy syndrome)

E-syndrome (see: Trisomy-18 syndrome)
Eaton-Lambert (ocular myoclonus, decreased vision, corneal haze—weakness, fatigue, peripheral paresthesia)
Economo, von (nystagmus, strabismus, diplopia, dimness of vision—fever, headache, delirium, athetoid movements, psychic disturbances)
Ectodermal dysplasia with ocular malformations (see: Curtius syndrome)
Ectodermal syndrome (see: Rothmund syndrome)
Edward (see: Trisomy-18 syndrome)
Ehlers-Danlos (hyperelasticity of palpebral skin, ptosis, epicanthus, hypotonic extraocular muscles, increased intraocular pressure, thin sclera and cornea, keratoconus, subluxated lens, retinopathy—cutaneous hyperelasticity, atrophic skin, excessive articular laxity)
Ekman (see: van der Hoeve syndrome)
Elastorrhexis (see: Groenblad-Strandberg syndrome)
Ellis-van Creveld (congenital cataract, strabismus—talipes, polydactyly, skeletal anomalies, heart defects, mental deficiency)
Embryonic fixation syndrome (see: Waardenburg syndrome)
Empty-sella (decreased vision, optic atrophy, visual field defects—acromegalic features)
Encephalitis hemorrhagica superioris (see: Wernicke syndrome)
Encephalitis lethargica (see: von Economo syndrome)
Encephalitis periaxialis diffusa (see: Schilder syndrome)
Encephalofacial angiomatosis (see: Sturge-Weber syndrome)
Encephalo-ophthalmic syndrome (see: Krause syndrome)
Enchondromatosis (see: Dyschondroplasia syndrome)
Engelmann (exophthalmos, lagophthalmos, ptosis, ophthalmoplegia, epiphora, hypertelorism, cataract, papilledema, optic atrophy, tortuosity retinal vessels, convergence insufficiency—skeletal anomalies, waddling gait, hepatosplenomegaly, hypogonadism, scaly skin, deafness, carious teeth)
Eosinophilic pneumonitis (see: Loeffler syndrome)
Epidermolysis acuta toxica (see: Lyell syndrome)
Epidermolysis bullosa (see: Goldscheider syndrome)
Epiloia syndrome (see: Bourneville syndrome)
Epithelial erosion syndrome (recurrent erosion of corneal epithelium, fever, herpetic skin blisters occasionally)
Epstein (see: Nephrotic syndrome)
Erb-Goldflam (ptosis, strabismus—general muscle weakness)
Erythema multiforme exudativum (see: Stevens-Johnson syndrome)
Espildora-Luque (ophthalmic artery emboli—temporary contralateral hemiplegia)
Essential lipoid histiocytosis (see: Niemann-Pick syndrome)
Exfoliation syndrome (iridodonesis, rubeosis iridis, cataract, phacodonesis, dislocated lens, corneal dystrophy, choroidal sclerosis, optic atrophy)
Exhaustive psychosis syndrome (see: Nielsen syndrome)
Exophthalmic goiter (see: Basedow syndrome)
Extreme hydrocephalus (exophthalmos, downward placement of globes, upper lid retraction, nystagmds, strabismus, visual loss, optic atrophy—extreme hydrocephalus, convulsions, thin and spastic limbs, low-set ears)
Eyelid-malar mandible syndrome (see: Franceschetti syndrome)