Orofacial Syndromes: A Review

1Shyam Sunder, 2D Rama Raju, 3Srinivas, 4Appaji Athota

1Professor, Department of Oral Pathology, Gandhi Dental College, Bhubaneswar, Odisha, India
2Professor, Department of Oral Medicine and Radiology, Purvanchal Institute of Dental Sciences, Gorakhpur, Uttar Pradesh, India
3Reader, Pedodontics, Panineeya Institute of Dental Sciences, Hyderabad, Andhra Pradesh, India
4Assistant Professor, Department of Oral Medicine and Radiology, Rungta College of Dental Science and Research, Bhilai, Chhattisgarh, India

Correspondence: Appaji Athota, Assistant Professor, Department of Oral Medicine and Radiology, Rungta College of Dental Science and Research, Bhilai, Chhattisgarh, India, e-mail: drappaji_a@rediffmail.com

ABSTRACT

A syndrome is a set of signs and symptoms that tend to occur together and reflect the presence of a particular disease or an increased chance of developing to a particular disease. There are numerous orofacial syndromes and a thorough knowledge of their manifestations and implications is pertinent in good oral health care delivery. The aim of this review is to describe collective esoteric knowledge, about various malformations and syndromes associated with orofacial region.

Keywords: Facial syndromes, Oral syndromes.

INTRODUCTION

The real importance behind the learning of syndromes associated with conditions is of relevance to clinical examination of the head and neck. Knowledge of syndromes may quickly solve some difficult diagnostic problems and appropriate treatment instituted. The following list takes you through conditions met by the authors either in their clinical practice or in examinations and which could, therefore, be considered worth knowing and helpful in academic and clinical excellence.

DEFINITIONS

• Malformation: A primary structural defect resulting from a localized error of morphogenesis.
• Anomalad: A pattern of development initiated by a single structural defect which subsequently leads to associated secondary defects.
• Syndrome: A recognized pattern of malformation, presumed to have the same etiology, constitutes group of symptoms that collectively indicate or characterize a disease, psychological disorder or other abnormal condition.

SYNDROMES

Aschers syndrome is a combination of blepharochalasis, doublelip and nontoxic goiter. Onset at young age with transient, recurrent edemas of angioneurotic in both eyelids. Then with the progress of the disease, prolapse of the orbital fat and lacrimal glands and blepharoptosis. Unknown etiology, genetic cause has been suggested.1

Apert syndrome is a rare developmental deformity consisting of a craniosynostosis, premature fusion of cranial sutures and syndactyly, fusion of fingers or toes. Severe midface retrusion leads to exophthalmoses of varying severity. Early surgical intervention may be indicated for raised intracranial pressure or to prevent blindness from subluxation of the globe of the eye.2

Behcet syndrome is, classically, oral ulceration, genital ulceration and uveitis. It is, in fact, a multisystem disease of immunological origin, it tends to affect young adults, especially males and there is an association with HLA-B5. It undergoes spontaneous remission, a variety of drugs including thalidomide are in use to treat it.3

Chediak-Higashi syndrome is a combination of defective neutrophil function, abnormal skin pigmentation and increase susceptibility to infection leading to severe gingivitis, periodontitis and aphthae in young children. It is a genetic disease.4

Cleidocranial dysostosis—cleidocranial dysplasia is an autosomal dominant inherited condition consisting of hypoplasia or aplasia of the clavicles, delayed ossification of the cranial fontanelles, and a large, short skull. Associated features are shortness of stature, frontal and parietal bossing, failure to pneumatize the air sinuses, a high arched palate and clefting, midface hypoplasia, and failure of tooth eruption with multiple supernumerary teeth. Many of the teeth present have inherent abnormalities such as dilaceration of roots or crown gemination. Hypoplasia of secondary cementum may occur. 5

‘Cri du chat’ syndrome is a chromosomal abnormality caused by deletion of part of the short arm of chromosome no. 5, resulting in microcephaly, hypertelorism, and a round face with a broad nasal bridge and malformed ears. Associated laryngeal hypoplasia causes a characteristic shrill cry. There is associated severe mental retardation.6

Crouzon syndrome is the commonest of the craniosynostoses. It is an autosomal dominant condition consisting of premature fusion of cranial sutures, midface hypoplasia, and,
due to this, shallow orbits with ptosis of the globe of the eye. Radiographically the appearance of a ‘beaten copper skull’ is characteristic. The enlarging brain is entrapped by the prematurely fused sutures, and increased intracranial pressure can lead to cerebral damage and resulting intellectual deficiency.

Down syndrome—trisomy 21 is the commonest of all malformation syndromes, affecting up to 1:600 births. Down’s children account for 1/3 of severely mentally handicapped children. Facial appearance is characteristic with brachycephaly, midface retrusion, small nose with flattened nasal bridge and upward sloping palpebral fissures (mongoloid slant). There is relative macroGLOSSIA and delayed eruption of teeth. Heart defects, atlantoaxial subluxation, anemia and an increase risk of leukemia.

Eagle syndrome is dysphagia and pain on chewing and turning the head associated with an elongated styloid process. Ehlers-Danlos syndrome is a group of disorders characterized by hyperflexibility of joints, increase bleeding, bruising and hyperextensible skin. There appears to be an underlying molecular abnormality of collagen in this inherited disorder. Bleeding is common in type IV, early onset periodontal disease in type VIII. Pulp stones may be seen in all types.

Frey syndrome is a condition in which gustatory sweating and flushing of skin occur. It follows trauma to skin overlying a salivary gland and is thought to be due to posttraumatic crossover of sympathetic and parasympathetic innervation to the gland and skin, respectively.

Gardener syndrome comprises multiple osteomas particularly of the jaws and facial bones, multiple polyps of the large intestine, epidermoid cysts and fibromas of the skin. It shows autosomal dominant inheritance. The discovery on clinical or X-ray examination of facial osteomas mandates examination of the lower gastrointestinal tract, as these polyps have a tendency to rapid malignant change.

Goldenhar syndrome is a variant of hemifacial microsomia and consists of microtia—small ears, macrostomia, agenesis of the mandibular ramus and condyle, vertebral abnormalities and epibulbar dermoids. Cardiac, renal or skeletal abnormalities can occur.

Gorlin-Goltz syndrome—multiple basal cell Naevi syndrome. consists of multiple basal cell carcinomas, multiple jaw cysts-odontogenic keratocysts, vertebral and rib anomalies, usually bifid ribs and calcification of the falk cerebi. Frontal bossing, mandibular prognathism, hypertelorism, hydrocephalus, eye and endocrine abnormalities are seen.

Heerfordt syndrome—uveoparotid fever is sarcoidosis with associated lacrimal and salivary gland swelling, uveitis and fever. Sometimes there are associated neuropathies, e.g. facial palsy.

Horner syndrome consists of a constricted pupil miosis, drooping eyelid ptosis, unilateral loss of sweating on the face and occasionally enophthalmos. It is caused by interruption of sympathetic nerve fibers at the cervical ganglion secondary to, bronchogenic carcinoma, invading the ganglion or neck trauma.

Hurler syndrome is a mucopolysaccharidosis causing growth failure and mental retardation. A large head, frontal bossing, hypertelorism and coarse features give it its classical appearance. Multiple skeletal abnormalities, corneal clouding and serum and urinary acid mucopolysaccharide abnormalities also occur.

Hypohydrotic ectodermal dysplasia-hypodontia found in association with lack of hair, sweating and saddle nose.

Larsen syndrome is a mainly autosomal dominant condition, with a predilection for females, consisting of cleft palate, flattened face, multiple congenital dislocations, and deformities of the feet. Sufferers are usually of short stature. Larynx may be affected.

Marfan syndrome is an autosomal dominant condition characterized by tall, thin stature and arachnodactyly - long, thin spider-like hands, dislocation of the lens, dissecting aneurysms of the thoracic aorta, aortic regurgitation, floppy mitral valve and high arched palate. Joint laxity is also common.

McCune-Albright syndrome consists of polyostotic fibrous dysplasia, patchy skin pigmentation referred to as cafe-au-lait spots and an endocrine abnormality, usually precocious puberty in girls. Facial asymmetry is seen.

Merkel-Rosenthal syndrome consists of facial paralysis, facial edema and fissured tongue. It is probably a variant of the group of conditions now known as orofacial granulomatosis.

Multiple endocrine neoplasia is a group of conditions affecting the endocrine glands. MEN IIb is of particular relevance as it consists of multiple mucosal neuromas, pheochromocytoma, medullary thyroid carcinoma. Calcitonin levels are elevated if medullary thyroid carcinoma is present. Index of suspicion should be high in tall, thin, wasted-looking children and young adults presenting with lumps in the mouth.

Orofacial-Digital syndrome or Mohr syndrome is characterized by a lobulate tongue, midline cleft of lip, high arched or cleft palate, broad nasal root with wide bifid nasal tip, hypertelorism, micrognathia, brachydactyly, syndactyly and polydactyly, conductive hearing loss and normal intelligence.

Papillon-Lefèvre syndrome is palmoplantar hyperkeratosis and juvenile periodontitis, which affects both primary and secondary dentition. Normal dental development occurs until the appearance of the hyperkeratosis of the palms and soles, and then simultaneously an aggressive gingivitis and periodontitis begin.

Patterson-Brown-Kelly syndrome and Plummer-Vinson syndrome is the occurrence of dysphagia, microcytic hypochromic anemia, koilonychia—spoon-shaped nails and angular cheilitis. The dysphagia is due to a posteriocr web, usually a membrane on the anterior esophageal wall, which is premalignant. The koilonychia and angular cheilitis are secondary to the anemia.
Peutz-Jeghers syndrome is an autosomal dominant condition of melanotic pigmentation of skin especially perioral skin and mucosa and intestinal polyposis. These polyps, unlike those of the Gardener syndrome, have no particular propensity to malignant change, being hamartomatous, and are found in the small intestine. They may, however, cause intussusception or other forms of gut obstruction. Ovarian tumors are sometimes associated with the condition.27

Progeria is probably a collagen abnormality. It causes dwarfism and premature ageing. Characteristic facial appearance occurs due to a disproportionately small face with mandibular retrognathia and a beak-like nose; creating an unforgettable appearance, death occurs in the second decade.28

Ramsay Hunt syndrome is a lower motor neuron facial palsy, with vesicles on the same side in the pharynx, external auditory canal, and on the face. Tinnitus, deafness and vertigo are seen. It is thought to be due to herpes zoster of the geniculate ganglion.29

Pierre Robin syndrome—consists of micrognathia, cleft palate and glossophtosis.30

Romberg syndrome—hemifacial atrophy consists of progressive atrophy of the soft tissues of half the face, associated with contralateral Jacksonian epilepsy and trigeminal neuralgia. Rarely, half the body may be affected. It starts in the first decade and lasts for 3 years before it becomes quiescent.31

Sicca syndrome and Primary Sjogren syndrome is xerostomia and keratoconjunctivitis sicca, i.e. dry mouth and dry eyes. There is an increase risk of developing parotid lymphoma with this condition.32

Sjogren syndrome and secondary Sjogren syndrome: In addition to dry eyes and dry mouth this has both the serology and symptomatology of an autoimmune condition, usually rheumatoid arthritis, but sometimes SLE, systemic sclerosis or primary biliary cirrhosis are present.33

Stevens-Johnson syndrome a severe version of erythema multiforme, a mucocutaneous condition that is probably autoimmune in nature and precipitated particularly by drugs. Classical signs are the target lesions, concentric red rings which especially affect the hands and feet. Stevens-Johnson syndrome is said to be present when the condition is particularly severe and is associated with fever and multiple mucosal involvement. Viral infections, e.g. herpes simplex, are the second commonest cause.34

Stickler syndrome is the commonest syndrome associated with cleft palate. Consists of flat mid-face, cleft palate, myopia, retinal detachment, hearing loss and arthropathy.35

Sturge-Weber anomalad is due to a hamartomatous angioma affecting the upper part of the face, which may extend intracranially. There may be associated convulsions, hemiplegia on the contralateral side of the body, or intellectual impairment.36

Treacher-Collins syndrome—mandibulofacial dysostosis basically involves defects in structures derived from the first branchial arch. It is inherited as an autosomal dominant trait with variable expressivity and consists of downward-sloping (antimongoloid slant) palpebral fissures, hypoplastic malar complexes, mandibular retrognathia with a high gonial angle, deformed pinna, hypoplastic air sinuses, colobomas in the outer third of the eye, and middle and inner ear hypoplasia and deafness. Cleft palate and an unusual tongue-like projection of hair pointing toward the cheek are present.37

Trotter syndrome is unilateral deafness, pain in the mandibular division of the trigeminal nerve, ipsilateral immobility of the palate and trismus, due to invasion of the lateral wall of the nasopharynx by malignant tumor. Pterygopalatine fossa syndrome is a similar condition where the first and second divisions of the trigeminal are affected.38

Von Recklinghausen syndrome multiple neurofibromas with skin pigmentation, skeletal abnormalities, CNS involvement and a predisposition to malignancy are the basics of this syndrome. It undergoes autosomal dominant transmission and has a large and varied number of manifestations. Lesions of the face can be particularly disfiguring.39

REFERENCES