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Review Article

Orofacial Syndromes Part II

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INTRODUCTION

Syndromes is a group of deformations and malformation sequences, etc. that occur together due to some identifiable underlying cause. It is defined as "The aggregate of signs and symptoms associated with any morbid process and constituting together the picture of the disease and related to each other anatomically, biochemically or physiologically". They are caused by chromosomal anomalies, single gene mutations, teratogens, or other causes. The head and neck region constitutes a wide variety of syndromes and this article aims to give a brief update on the etiopathogenesis, clinical features and oral manifestations of various orofacial syndromes which would be helpful for early diagnosis.

MATERIALS & METHODS

A systematic review from pertinent English literature was performed using Medline (through http:// www.ncbi.nlm.nih.gov/pubmed/). The search strategy included the combinations of Orofacial OR syndrome AND each of the relevant clinical features and oral manifestations were used. The reference sections of identified manuscripts were also explored for relevant reports and additional information.

Muir Torre Syndrome[1]

Etiopathogenesis (E/P)– Autosomal dominant - Germ-line mutations in hMSH2 and hMLH1 genes - alteration or inactivation of tumor suppressor genes.

Clinical features (C/F)- At least a single sebaceous gland tumor (sebaceous adenoma, sebaceous carcinoma, sebaceoma (sebaceous epitheliomas) and keratoacanthoma (KA) with sebaceous differentiation.) And a minimum of one internal malignancy (colorectal, genitourinary, breast carcinoma, hematological disorders, endometrial carcinoma, and rarely gastric carcinoma).

Oral manifestation (O/M)- Salivary gland tumors, Keratoacanthoma.

Murray–Puretic–Drescher syndrome / Juvenile hyaline fibromatosis / Fibromatosis hyalinica multiplex juvenilis / Systemic hyalinosis[2]

E/P - Autosomal recessive disease – mutant gene - 4q21- Abnormal biosynthesis of glycosaminoglycans and collagen III- VI

C/F - Papules distributed around the nose, the ears, in the genital area and on the thighs, Excessive skin stretching, rogressive joint involvement, joint contractures and cutaneous thickening

O/M - Gingival hypertrophy

Nevoid Basal Cell Carcinoma Syndrome (NBCCS) / basal cell nevus syndrome (BCNS) / Gorlin-Goltz syndrome / Gorlin syndrome / Hermans-Herzberg phakomatosis[3]

E/P - Rare, complex genetic disorder - autosomal dominant - mutations in the PTCH1 gene

C/F - Basal cell carcinomas, multiple distinctive palmar pits, calcification of the structures in the brain including the dura mater, the outermost layer of the three membranes that cover the brain and spinal cord.

O/M - Recurrent keratocystic odontogenic tumors of the jaws

Numb Chin Syndrome / mental nerve neuropathy[4]

E/P - Numb chin syndrome (NCS) is a rare sensory neuropathic condition that can be associated with many local or systemic causes.

C/F - NCS is also referred to as mental nerve neuropathy because the facial numbness occurs along the distribution of the mental branch of the inferior alveolar portion of the trigeminal nerve.

O/M- unilateral hypoesthesia, dysesthesia, or paresthesia that is localized to the chin, jaw, or lower lip, and may be accompanied by an abnormal sensation of "thickening" of the lower lip, which is similar to the experience of dental anesthesia

Oculocerebrocutaneous syndrome (OCC) / Delleman Oorthuys syndrome / orbital cyst with cerebral and focal dermal malformations[5]

E/P - Rare genetic disorder, geneticists suggest that OCC syndrome is caused by a genetic change (a mutation) that appears to be present in part of the cells of the body only (somatic mosaicism). The exact cause of OCC is still unknown and difficult to determine.

C/F - orbital cyst with periorbital skin appendages which may be associated with anopthalmia or microphthalmia, major cerebral malformation, and focal dermal hypoplasia or aplasia. Characteristic of this syndrome are pink-colored or flesh-colored outgrowths of skin (cutaneous tag) within certain facial areas, most commonly the periocular area.

O/M - Delleman Oorthuys syndrome shows overlapping clinical features with Goldenhar syndrome and Goltz syndrome

Oculo glandular syndrome of Parinaud[6]

E/P - Rare eye disease caused by different etiologic agents, including bacteria, viruses (*herpes simplex virus)* and fungi (sporotrichosis (*S. schenckii*), blastomycosis (*Blastomyces dermatitidis*) and coccidioidomycosis (*Coccidioides immitis)*

C/F - Granulomatous conjunctivitis, accompanied by adjacent preauricular lymphadenopathy which is almost always caused by local trauma

O/M - The preauricular and submandibular lymph nodes are grossly enlarged and may suppurate

Osler–Weber–Rendu syndrome / Hereditary hemorrhagic telangiectasia (HHT) / Osler–Weber–Rendu disease[7]

E/P - Rare autosomal dominant genetic disorder that leads to abnormal blood vessel formation in the skin, mucous membranes, and often in organs such as the lungs, liver, and brain.

2 types – HHT1 & HHT2

HHT1 - mutation in endoglin (ENG). With this type, patients, especially women, are at a higher risk of getting pulmonary and cerebral AVMs

HHT2 - mutation in activin A receptor-like type 1 (ACVRL1), also known as ALK1. Patients with HHT2 have a higher risk of getting liver AVMs

C/F - epistaxis that begins during childhood or adolescence at a mean age of 12 years. Telangiectasias do not usually appear until after puberty but may not occur until adulthood

O/M - They typically occur on the face, lips, tongue, palms, and fingers including the periungual area and the nail bed. Telangiectasias are dilated blood vessels that appear as thin spiderweb-like red and dark purple lesions that blanch with pressure. AVMs are abnormal connections between arteries and veins that bypass the capillary system.

Papillon–Lefèvre syndrome[8]

E/P- Autosomal recessive inherited disorder of keratinization - mutations in *cathepsin C* (CTSC) gene, immunologic alterations, and the role of bacteria

C/F- Diffuse palmoplantar keratoderma, hyperhidrosis, arachnodactyly, intracranial calcification, increased susceptibility to infections, and mental retardation

O/M- Precocious aggressive periodontitis, leading to premature loss of deciduous and permanent dentition at a very young age.

Paraneoplastic autoimmune multiorgan syndrome[9]

E/P- The exact pathogenetic mechanisms of PAMS are still unknown

C/F- Skin manifestations i) pemphigus-like, ii) bullous pemphigoidlike, iii) erythema multiforme-like, iv) graft-vs-host disease-like and v) lichen planus-like

O/M- Persistent, painful erosions and ulcerations, affecting the gingivae, the lateral borders of the tongue, and subsequently extend to the entire oral cavity and the vermilion border of the lips, usually in the form of crusts, resembling erythema multiforme or even advanced cases of Stevens-Johnson syndrome

Parry-Romberg Syndrome / Progressive hemifacial atrophy[10]

E/P - The etiology of hemifacial atrophy has been the subject of numerous theories, which include heredity, viral infection, trauma, endocrine disturbances, autoimmunity, sympathetic malfunctions, trigeminal neuritis, and association with a connective tissue disorder, particularly scleroderma.

C/F- Uncommon degenerative condition characterized by a slow and progressive atrophy, generally unilateral, of facial tissues, including muscles, bones, and skin

Some patients present a demarcation line between normal and abnormal skin, reminding a big linear scar, known as "*coup de saber*"

O/M- Mouth and nose are deviated to the affected side, deviating also facial and dental midlines. Atrophy of superior lip led the anterior teeth to be exposed, and there may be also unilateral atrophy of tongue.

Peutz-Jeghers syndrome / intestinal polyposis-cutaneous pigmentation syndrome / periorificial lentiginosis syndrome / Peutz-Jeghers polyposis / polyps-and-spots syndrome[11]

E/P - Autosomal dominant - development of noncancerous growths called hamartomatous polyps in the gastrointestinal tract

Mutations in the *STK11* gene (also known as *LKB1*)

C/F - Abdominal pain, intestinal bleeding, menstrual irregularities in females, cutaneous pigmentation in perioral areas

O/M - The brown pigmented macules -at birth. Pigmented lesions - skin around the lips and the vermilion zone of the lips

Intra orally - Usually flat, brown pigmented, painless patches on the buccal mucosa, tongue or labial mucosa

PFAPA syndrome (Periodic Fever, Aphthous Stomatitis, Pharyngitis, Adenitis)[12]

E/P - Etiology – unknown

C/F - Mouth sores (aphthous stomatitis)**,** Sore throat with redness (pharyngitis)**,** Enlarged lymph nodes of the neck (adenitis)**,** White patches on the tonsils

O/M - Aphthous stomatitis

PHACE Syndrome (Posterior fossa brain malformations, Hemangioma, Arterial lesions, Cardiac abnormalities, and Eye abnormalities)[13]

E/P - The pathogenesis of PHACE syndrome is unknown.

C/F - The most remarkable feature of PHACE syndrome is Infantile hemangioma (IH), but abnormalities in the brain, aortic, thoracic, and cervical arteries, which are not obvious on clinical examination, have considerable potential for morbidity

Pierre Robinson syndrome / Pierre Robin Sequence[14]

E/P - Cleft palate is associated with deletions on 2q and 4p, and duplications on 3p, 3q, 7q, 78q, 10 p, 14q, 16p, and 22q. Micrognathia is associated with deletions in 4p, 4q, 6q, and 11q, and duplications on 10q and 18q

C/F - Triad of micrognathia, glossoptosis, and airway obstruction

O/M - Micrognathia (which he termed "mandibular hypotrophy") and glossoptosis (an abnormal posterior placement of the tongue), which result in airway obstruction and feeding difficulties

Plummer Vinson Syndrome[15]

C/F - Triad of dysphagia, iron-deficiency anemia and esophageal webs

E/P - The pathogenesis of Plummer-Vinson syndrome is unknown.

O/M - Glossitis, angular cheilitis and koilonychia (spoon-shaped finger nails). Enlargement of the spleen and thyroid. Postcricoid dysphagia, upper esophageal webs and iron deficiency anemia,

Popliteal pterygium syndrome[16]

E/P - Mutations in the IRF6 gene

C/F - Webs of skin on the backs of the legs across the knee joint,

O/M – Cleft lip, Cleft palate

Proteus syndrome[17]

E/P - Unknown

C/F - Hemihypertrophy, symmetric megalodactyly, sub-cutaneous masses (vascular, lymphatic and lipomatous), epidermal nevi and skull hyperostosis, Partial gigantism of a limb or digital overgrowth is pathognomonic with an unusual body habitus and often, cerebriform thickening of soles of feet

O/M - Gingival overgrowth and malposition of teeth, as well as unilateral enamel hypoplasia

Ramsay Hunt Syndrome[18]

E/P - Infectious cranial polyneuropathy caused by *Varicella zoster* virus infection. Reactivation of herpes zoster virus that has previously caused chickenpox in the patient

C/F - Facial nerve paralysis associated with herpetic eruptions on the pinna, and is frequently complicated by vestibulocochlear dysfunction

O/M - Paralysis of the facial muscles on the same side of the face as the infection. So, the virus infects the facial nerve that normally controls the muscles on one side of the face. Peripheral facial nerve palsy accompanied by an erythematous vesicular rash on the ear (zoster oticus) or in the mouth.

Rasmussen syndrome / Rasmussen Encephalitis / chronic encephalitis and epilepsy / chronic localized (focal) encephalitis / epilepsy, hemiplegia and intellectual disabilities[19]

E/P - The exact cause of this disorder is not known.

C/F - Rare disorder of the central nervous system characterized by chronic progressive inflammation (encephalitis) of one cerebral hemisphere

O/M - Progressive weakness of one side of the body (hemiparesis), language problems (if on the left side of the brain) and intellectual disabilities.

Reiter's Syndrome / reactive arthritis[20]

E/P - The infection is most commonly chlamydia

C/F - Triad of conjunctivitis, urethritis, and arthritis occurring after an infection, particularly those in the urogenital or gastrointestinal tract

O/M - Papules and ulcerations on the buccal mucosa, gingiva and lips. Lesions on the tongue resemble "geographic tongue"

Reye's syndrome[21]

E/P - The exact cause of Reye's syndrome is unknown.

C/F - Confusion, seizures and loss of consciousness require emergency treatment

Romberg syndrome / Progressive hemifacial atrophy / Parry Romberg Syndrome[22]
E/P - Unknown cause
C/F - Slow and progressive atrophy affecting one side of the face generally unilateral, of facial tissues, including muscles, bones, and skin
O/M - Deviation of mouth and nose to the affected side, and unilateral exposition of teeth, when lips are involved. atrophy of half of the lip and tongue, shortening of the body of the mandible and/or ramus of the mandible, retarded tooth eruption, and malformed tooth roots

Rombo Syndrome[23]
E/P - Autosomal dominant
C/F - Characterized by vermiculate atrophoderma, milia, hypotrichosis, trichoepitheliomas, peripheral vasodilation with cyanosis and basal cell carcinomas,
O/M - Follicular skin atrophy of cheeks, cyanotic redness of the lips

Rothmund-Thomson syndrome/ congenital poikiloderma / poikiloderma atrophicans and cataract / poikiloderma congenitale / poikiloderma congenitale of Rothmund-Thomson[24]
E/P - Autosomal, recessive inheritance - Mutations in the *RECQL4* gene
C/F - Sparse hair, eyebrows, and eyelashes; slow growth and small stature; abnormalities of the teeth and nails; and gastrointestinal problems in infancy, such as chronic diarrhea and vomiting. skeletal abnormalities including absent or malformed bones, fused bones, and low bone mineral density (osteopenia or osteoporosis), radial ray defects, premature aging
O/M – Microdontia, rudimentary or hypoplastic teeth, multiple crown malformations, increase in prevalence of caries, malocclusion, hypodontia/oligodontia or hypodontia, ectopic eruption, delay in eruption, bifid uvula

Rubinstein-Taybi syndrome / Broad Thumb-Hallux Syndrome[25]
E/P - Mutations in the *CREBBP* gene. Mutations in the *EP300* gene cause a small percentage of cases of Rubinstein-Taybi syndrome
C/F - Short stature, moderate to severe intellectual disability, distinctive facial features, and broad thumbs and first toes. Additional features of the **disorder** can include eye abnormalities, heart and kidney defects, dental problems, and obesity.
O/M - Thin upper lip, small oral opening, pouting lower lip, retro/micrognathia, and apparently higher arched, narrow palate. Cleft uvula, cleft palate, or, rarely, cleft upper lip

SAPHO Syndrome[26]
E/P - Caused by autoimmune reactions in genetically predisposed organisms, triggered by some infectious agent
C/F - Chronic **disorder** that involves the skin, bone, and joints. **SAPHO** is an acronym for the combination of synovitis, acne, pustulosis, hyperostosis, and osteitis, osteoarticular lesions
O/M – Arthritis of TMJ may be involved

Shwachman–Diamond syndrome (SDS) / Shwachman–Bodian–Diamond syndrome / Congenital Lipomatosis of Pancreas / Metaphyseal chondrodysplasia, Shwachman type / Shwachman-Bodian syndrome / Shwachman-Diamond-Oski Syndrome / Shwachman syndrome[27]
E/P - Autosomal recessive - Mutations in the *SBDS* gene
C/F - Exocrine pancreatic insufficiency, bone marrow dysfunction, skeletal abnormalities and short stature.
O/M - Abnormally decreased saliva production**,** delays in tooth eruption, and their teeth may develop improperly (dental dysplasia)**,** dental caries, mouth ulcers, periodontal disease

Sezary syndrome[28]
E/P - Unknown
C/F- Triad of erythroderma, generalized lymphadenopathy and the presence of clonally related neoplastic T cells with cerebriform nuclei (Sézary cells) in skin, lymph nodes and peripheral blood. Pruritus, alopecia, ectropion, palmar / plantar hyperkeratosis, onychodystrophy, leonine facies
Sezary syndrome is an aggressive form of a type of blood cancer called cutaneous T-cell lymphoma.

Sicca syndrome / Sjogren syndrome[29]
E/P – Autoimmune-induced inflammation of the lacrimal and salivary glands
C/F - Dryness of the eyes, mouth, and vagina.
O/M- Xerostomia

Steven Johnson Syndrome/ Toxic Epidermal Necrolysis (SJS/TEN)/ Lyell's syndrome[30]
E/P – Variation in the HLA-Bgene
C/F -Severe exfoliative reactions affecting mainly the skin and mucous membranes, erythematous macules, blisters and denuded skin
O/M - Erosive areas with epithelial detachment in the oral cavity, lip ulceration

Stickler's syndrome/ hereditary arthro-ophthalmo-dystrophy[31]
E/P – Mutations in the *COL2A1* gene
C/F - Distinctive facial appearance, eye abnormalities, hearing loss, and joint problems
O/M - Flat cheeks and nasal bridge, Small jaw, Split uvula

Sturge-Weber syndrome/ angiomatosis aculoorbital-thalamic syndrome[32]
E/P – Mutation in the *GNAQ* gene
C/F - Red or pink birthmark called a port-wine birthmark, a brain abnormality called a

leptomeningeal angioma, and increased pressure in the eye
O/M - Gingival haemangioma on the right side of the maxilla.

Sweet syndrome/ Acute Febrile Neutrophilic Dermatosis[33]
E/P – Unknown
C/F - Sudden onset of fever and painful rash on the arms, legs, trunk, face, or neck
O/M - Mouth lesions (sores or tumors)

Tourette syndrome[34]
E/P – Genetic and nongenetic (epidemiological) factors
C/F – Sudden repetitive motor and phonic tics
O/M – Oral self-injurious behaviour

Treacher collin syndrome/ Franceschetti-Zwahlen-Klein syndrome / mandibulofacial dysostosis[35]
E/P – Mutations in the *TCOF1*, *POLR1C*, or *POLR1D* gene
C/F –Down slanting eyes with notched lower lids, sunken cheekbones, pointed nasal prominence, conducting hearing loss
O/M - Cleft lip and/or palate, high arched palate

Tricho-dento-osseous syndrome[36]
E/P – Autosomal dominant disorder- mutation in the DLX3 gene
C/F – Kinky or tightly curled hair at birth, Sclerosis of bone, Splitting of the superficial layers of the nails, Frontal bossing
O/M - Impacted teeth, Mandibular prognathism, Maxillary retrusion, Yellow-brown discolored teeth, Dental abscesses, Taurodontism

Trotters's syndrome/ Syndrome of the Sinus of Morgagni[37]
E/P – Unknown
C/F - Unilateral deafness, neuralgia affecting branches of the trigeminal nerve, and defective mobility of the soft palate
O/M - Defective mobility of the soft palate due to ipsilateral infiltration

Turner syndrome[38]
E/P - Complete monosomy X
C/F – Short stature, ovarian failure, webbed neck, cardiac abnormalities, impaired glucose tolerance, thyroid disease and hearing loss
O/M - Retrognathic lower face (including a recessed and small mandible) Increased cranial base angle and abnormal palate , Distal molar occlusion. Large overjet and lateral cross-bite.

Vander woude syndrome[39]
E/P – Mutations in the *IRF6* gene
C/F – Clefting syndrome includes bilateral midline lower lip pits, cleft lip, and cleft palate along with hypodontia
O/M – Cleft lip, cleft palate, hypodontia

Von Recklinghausen's disease / Neurofibromatosis type I[40]
E/P - Alteration of the *NF-1* gene
C/F - Café-au-lait spots, axillary and inguinal freckling, optic gliomas, Lisch nodules, spinal and peripheral nerve neurofibromas, neurological or cognitive impairment, scoliosis, abnormalities in the oral and maxillofacial region, malignant tumors of the nerve sheath, pheochromocytoma, vasculopathy
O/M - Asymptomatic nodules covered by normally colored mucosa

Wiskott–Aldrich syndrome[41]
E/P - Rare X-linked recessive disease
C/F - Eczema, thrombocytopenia (low platelet count), immune deficiency, and bloody diarrhea (secondary to the thrombocytopenia).
O/M - Gingival bleeding and palatal petechiae.

Witkop von sallmann syndrome/ Hereditary benign intra-epithelial dyskeratosis[42]
E/P – Rare hereditary disease – oral and ocular lesions
C/F – White granular to gelatinous triangular, perilimbal and bulbar conjunctival plaques, red eye
O/M – Oral candidiasis

Zimmermann laband syndrome[43]
E/P – Autosomal dominant inheritance – mutation of KCNH1 gene
C/F - Abnormalities of the nose and ears, absence or hyperplasia of the nails or terminal phalanges of the hands and feet
O/M - Gingival fibromatosis

Zinsser-Engman-Cole syndrome/ Dyskeratosis congenita[44]
E/P – Mutations in DKC1 gene
C/F – Reticulated skin hyperpigmentation, nail dystrophy, and oral leukoplakia
O/M – Oral leukoplakia

CONCLUSION

This article concentrates on the importance of syndromes to be noted especially in the orofacial region which enables the clinician to detect the defect as soon as possible in order to identify and treat the patient, thus preventing them from further complications. A more detailed study should be done on each syndrome to know the exact feature and pathology behind it.

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