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Review Article

A Review on Orofacial Syndromes Associated With Head and Neck Region - Part I

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***ABSTRACT*:**
Objectives: A syndrome is a condition presenting with a collection of signs & symptoms reflecting the presence of disease. Many syndromes have overlapping clinical and oral manifestations, and diagnostic uncertainty is frequently observed during clinical practice. Thorough basic knowledge about various orofacial syndromes would help us in diagnosis and perform successful management for patients. This paper briefly reviews various syndromes associated with the orofacial region. **Methods:** This study reviews on clear detailed representation of syndromes from original articles, overviews, case reports and reviews. **Results:** Several relevant reports were identified and collected to enlist the list of syndromes associated with craniofacial manifestations. **Conclusion**: This article is constructed to help the health care providers to understand and aid in the diagnosis to provide an optimal personalized care for individuals.
Keywords: Syndromes, Clinical features, Oral manifestations.

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INTRODUCTION

Syndrome is a condition characterized by "a set of signs and symptoms that tend to occur and reflect the presence of a particular disease or an increased chance of developing to a particular disease". They are caused by chromosomal anomalies, single gene mutations, teratogens, or other causes. There is a wide spectrum of syndromes associated with orofacial region and thorough knowledge of their manifestation & implication is pertinent for diagnosis to provide an optimal personalized care with an integral approach. [1] This article aims to provide an update on genetics, general features and oral & craniofacial manifestation of orofacial syndromes for early diagnosis.

OBJECTIVES

To enlist the various syndromes associated with head and neck region.
To provide a brief outline on etiopathogenesis, clinical features and oral manifestations.

MATERIALS & METHODS

Search Strategy

A systematic review from pertinent English literature was performed using Medline (through http:// www.ncbi.nlm.nih.gov/pubmed/). For this, a thorough literature search was carried out both manually and electronically. The databases PubMed and Google Scholar were searched using MeSH entry terms mixed with Boolean phrases 'AND' or 'OR'. The reference

sections of identified manuscripts were also explored for relevant reports and additional information.
Clear representation of the available insights regarding the topic of interest were done

Data collection and Extraction
The basic search included mainly original articles, overviews, case reports and reviews. Articles not relevant to the topic of interest were excluded. After completing the search, the selected documents were summarized and categorized based on the topic and its implications.

RESULTS
Several relevant reports were identified and collected to enlist the list of syndromes associated with craniofacial manifestations. The following is a broad overview of the etiopathogenesis, clinical and oral manifestations of orofacial syndromes. A detailed summary of syndromes are provided to assist the health care professionals to diagnose and construct an effective treatment plan.

SYNDROMES

***Acute chest syndrome*[2]**
Etiopathogenesis (E/P): due to sickling in small blood vessels in the lungs causing a pulmonary infarction/emboli or viral or bacterial pneumonia
Clinical features (C/F): chest pain, cough, fever, hypoxia (low oxygen level) and lung infiltrates.
Oral manifestation (O/M): aseptic pulpal necrosis, mucosal damage, dental eruption delays, bone pain and osteomyelitis of the maxilla, and oral neuropathies

***Acquired immunodeficiency* syndrome[2]**
E/P: the most common life threatening condition caused Human Immunodeficiency Virus that damages the CD4 T lymphocytes interfering the body's ability to fight the organism.
C/F: Affected patients are severely immunocompromised where they are vulnerable to pneumonia, tuberculosis, oral thrush, cryptococcal meningitis and toxoplasmosis.
O/M: xerostomia, candidiasis, oral hairy leukoplakia, periodontal diseases such as linear gingival erythema and necrotizing ulcerative periodontitis, Kaposi's sarcoma, human papilloma virus associated warts, and ulcerative conditions including herpes simplex viral infection.

***APECED syndrome*[2]**
E/P: inherited autoimmune disorder caused due to mutation of AIRE gene
C/F: hypoparathyroidism, adrenal gland insufficiency, vitamin B12 deficiency
O/M: enamel hypoplasia, sjogren syndrome, chronic mucocutaneous candidiasis.

***Apert syndrome*[2]**
E/P: developmental deformity caused due to mutation of FGFR2 gene
C/F: Craniosynostosis, midface hypoplasia, syndactyly, hyperhidrosis, exophthalmos, hypertelorism, downslanting palpebral fissures, strabismus, ocular proptosis
O/M: cleft palate, missing and crowded teeth.

***Ascher syndrome*[2]**
E/P: autosomal dominant disorder of unknown origin
C/F: Blepharochalasia, non-toxic thyroid enlargement
O/M: double lip.

***Ataxia telangiectasia syndrome or Louis Bar syndrome*[3]**
E/P: due to mutation of ATM gene which is responsible to recognize DNA damage
C/F: developmental difficulty in walking, problems with balance and hand coordination, involuntary jerking movements (chorea), muscle twitches (myoclonus), and disturbances in nerve function (neuropathy) poor growth pattern and dysphagia
O/M: recurrent aspiration of oral secretion

***Behcets syndrome*[4]**
E/P: a multisystem disease of autoimmune origin associated with HLA B51/B5 gene
C/F: uveitis, genital ulcers, erythema nodosum, arthritis, epididymitis
O/M: aphthous stomatitis.

***Beckwith Wiedemann syndrome*[5]**
E/P: chromosomal abnormality caused by imprinting errors in 11p15 chromosome
C/F: omphalocele, umbilical hernia, visceromegaly, hypoglycemia. It has high risk to develop Wilm's tumor and hepatoblastoma.
O/M:macroglossia, macrosomia and facial hemihyperplasia.

***Bloom syndrome*[6]**
E/P: chromosomal disorder caused due to mutation of BLM gene in chromosome 15 which maintains structural integrity of DNA
C/F: short stature, high pitch voice, skin rashes, polydactyly, infertility
O/M: micrognathia.

***Cartilage hair syndrome*[7]**
E/P: due to mutation of RMRP gene
C/F: dwarfism, hypotrichosis, immune deficiency.
O/M: candidiasis, micrognathia.

***Chediakhigashi syndrome*[8]**
E/P: due to mutation of LYST gene
C/F: Oculocutaneous albinism, peripheral neuropathy
O/M: periodontal disease

Cri du chat syndrome[9]
E/P: chromosomal abnormality caused due to deletion of short arm of chromosome 5
C/F: round face with broad nasal bridge, hypertelorism, microcephaly, malformed ears, laryngeal hypoplasia and mental retardation.
O/M: micrognathia, malocclusions, high but rarely cleft palate, anterior open bite, poor oral hygiene, enamel hypoplasia, chronic periodontitis, and retardation of tooth eruption

Cockayne syndrome (Neill-Dingwall syndrome) [10]
E/P: due to mutation in ERCC6 gene.
C/F: microcephaly, progeria, increased photosensitivity, developmental delay
O/M:delayed tooth eruption and malocclusion

Cohen syndrome[11]
E/P: due to mutation of COH1 gene.
C/F: retinal dystrophy, developmental delay, hypotonia, microcephaly.
O/M: high arch palate, micrognathia and short philtrum

Cowden syndrome[12]
E/P: due to mutation of PTEN gene.
C/F: Hamartomas, cutaneous papillomatosis, trichilemmomas,
O/M: oral fibromas and nodular gingival hyperplasia

CREST syndrome[13]
E/P: an autoimmune multisystem disease resulting in excess collagen production.
C/F: combination of Calcinosis, Raynaud's phenomenon, esophageal dysmotility, sclerodactyly, and telangiectasia.
O/M: microstomia, xerostomia, pseudoankylosis, widening of the periodontal ligament space, loss of mobility of tongue, mask like facial appearance.

Crouzon syndrome[14]
E/P: Craniosynostosis caused due to mutation of FGFR2 gene
C/F: Dolicofacial growth pattern with Frontal bossing, midfacial hypoplasia, shallow orbit, mental retardation, hypertelorism, strabismus, proptosis.
O/M: Narrow, high, or cleft palate, bifid uvula, oligodontia, macrodontia, peg-shaped teeth with a characteristic beaten copper skull radiographic appearance.

Coffin Lowry syndrome[15]
E/P: Mutation in RSK2 gene
C/F: Intellectual disability, osteopenia, growth retardation, delay in psychomotor development, Frontal bossing, hypertelorism, downward sloping of palpebral fissure,
O/M: high narrow palate, open bite, peg shaped incisors

Dunnigan syndrome[16]
E/P: Mutation of LMNA gene
C/F: Dyslipidemia, Muscle hypertrophy, cushingoid appearance, acanthosis nigricans, PCOS, diabetes
O/M: periodontal disease

Down syndrome[17]
E/P: common malformation syndrome caused due to trisomy 21
C/F: characteristic facial appearance of brachycephaly, midfacial hypoplasia, flattened nasal bridge with mongoloid palpebral fissure slant. There is intellectual disability, hypotonia, delayed development & behavioural changes
O/M: mouth breathing, open bite, enlarged & fissured tongue, drooling, malocclusion, low caries index and poor oral hygiene.

Eagle syndrome[18]
E/P: elongation of styloid process or calcification of stylohyoid ligament
C/F: difficulty in swallowing glossopharyngeal neuralgia, cluster headache, tinnitus, otalgia,
O/M: Shooting pain in jaw, base of tongue, myofascial pain dysfunction

Ehlers danlos syndrome[19]
E/P: an inherited collagen disorder caused due to mutation of COL5A1, COL5A2, and tenasin-X genes resulting in defective production & processing of collagen.
C/F: joint hypermobility, hypotonia, skin hyperextensibility, tissue fragility with extensive scarring.
O/M: increased mucosal fragility, hypermobility of TMJ, periodontitis, hypodontia, abnormal dentin & pulp structure, gorlin's sign, absence of frenum.

Ellis van crevald syndrome[20]
E/P: EVC gene mutation responsible for patterning of many parts of the body
C/F: dwarfism, polydactyly, ectodermal dysplasia
O/M: microdontia, enamel hypoplasia, delayed tooth eruption, malocclusion, taurodontism

Frey syndrome[21]
E/P: Due to aberrant reinnervation of postganglionic parasympathetic fibres of auriculotemporal nerve to salivary gland and overlying skin.
C/F: gustatory flushing, sweating, burning, neuralgia, and itching.

Gardner syndrome[22]
E/P: due to mutation of APC gene which leads to uncontrolled cell growth in the form of polyps, tumors and cancers
C/P: colonic polyposis, osteomas, epidermoid cysts, desmoid tumours
O/M: odontomas, supernumerary teeth, hypodontia, abnormal tooth morphology, impacted or unerupted teeth

***Goldenhar syndrome*[23]**
E/P: autosomal dominant disorder characterized by incomplete development of nose, ear, soft palate & jaw
C/F: micropthalmia, microtia, epibulbardermoids, scoliosis, hydrocephalus
O/M: microdontia, hemifacial macrosomia, mandibular hypoplasia, cleft lip & palate, malocclusion.

***Gorlingoltz syndrome*[24]**
E/P: mutation of PTCH gene.
C/F: ovarian fibroma & cyst, spina bifida, macrocephaly, frontal bossing, hypertelorism, basal cell carcinoma, hydrocephalus, meningioma, polydactyly.
O/M: multiple OKC, high arch palate, impacted teeth, coronoid hyperplasia

***Haim munk syndrome*[25]**
E/P: disorder caused due to mutation in the lysosomal protease cathepsin C gene
C/F: almoplantar hyperkeratosis, onychogryphosis, pes planus, arachnodactyly and acro-osteolysis
O/M: aggressive periodontitis with severe alveolar bone destruction.

***Hanhart syndrome*[26]**
E/P: tyrosine aminotransferase enzyme deficiency syndrome
C/F: telecanthus, ocular changes, palmo-plantar hyperkeratosis.
O/M: microstomia, micrognathia, hypoglossia, cleft palate, clefting of tongue and hypodontia.

***Heerfordt Waldenstrom syndrome*[27]**
E/P: is sarcoidosis associated disease
C/F: facial palsy, epineural granulomas and perineural inflammatory infiltrates of cranial nerve VII
O/M: parotid gland enlargement and anterior uvelitis.

***Herman syndrome*[28]**
E/P: mutation in HPS1 gene preventing the formation of lysosome related organelles
C/F: oculocutaneous albinism, decreased pigmentation, pulmonary fibrosis, granulomatous colitis
O/M: odontoma

***Hurler syndrome*[29]**
E/P: due to deficiency of Alpha-L-iduronidase enzyme
C/F: macrocephaly, cardiomyopathy, hearing loss, developmental delay, short stature.
O/M: Macroglossia, hypoplastic teeth, impacted teeth associated with developmental cysts.

***Hyperparathyroidism jaw tumour syndrome*[30]**
E/P: due to mutation of CDC73 gene.
C/F: Hypercalcemia, parathyroid adenoma, hamartomas, Wilms tumour, parathyroid carcinoma, renal & uterine tumours
O/M: ossifying fibroma

***Jaffe lichenstein syndrome*[31]**
E/P: Mutation in GNAS1 gene results in overactive G protein triggering abnormal proliferation of melanocytes, osteoblasts & endocrine glands
C/F: Expansion of bone, proptosis, vertical dystopia, café au lait spots
O/M: Polyostotic fibrous dysplasia

***Kabuki syndrome*[32]**
E/P: Mutation in KMT2D and KDM6A
C/F: Everted eyelids, strabismus, blue sclera, ptosis, lip pits
O/M: High arch palate, micrognathia, malocclusion, microdontia,

***Kallmann syndrome*[33]**
E/P: Deficiency of gonadotrophin releasing hormone
C/F: cryptorchidism, micropenis, or associated non-reproductive signs or at the time of puberty,
O/M: Retrognathia of both jaws, cleft palate, tooth agenesis, microdontia,

***Kleinfelter syndrome*[34]**
E/P: Extra copy of X chromosome
C/F: Tall stature, cognitive retardation, infertility, small testicles, gynecomastia,
O/M: Macrodontia, dental agenesis, taurodontism

***Kostmann syndrome*[35]**
E/P: Autosomal recessive disorder
C/F: Congenital neutropenia, mild splenomegaly, moderate thrombocytopenia, osteoporosis, and malignant transformation into myelodysplasia (MDS)/leukemia.
O/M: Oral ulcers, periodontal disease, early loss of permanent teeth

***Larsen syndrome*[36]**
E/P: Mutation in FLNB gene
C/F: Club feet, dislocation of hip elbow, frontal bossing, mid face hypoplasia, kyphosis or scoliosis, hydrocephalus
O/M: Cleft palate

***Leopard syndrome*[37]**
E/P: Mutation in PTPN11 gene
C/F: Lentigines, electrocardiographic conduction defect, ocular hypertelorism, pulmonary stenosis, short stature, deafness
O/M: Agenesis of permanent teeth, osseous hypodevelopment

Leschnyan syndrome[38]
E/P: Mutation in HPRT1 gene
C/F: Involuntary muscle movement, neurological impairment, gouty arthritis,
O/M: Oral self mutilation

Lofgrens syndrome[39]
E/P: Acute form of sarcoidosis
C/F: erythema nodosum, bilateral hilar lymphadenopathy (BHL), and polyarthralgia or polyarthritis
O/M: Oral sarcoid granulomas, ulcers, nodular growth

Maffuci syndrome[40]
E/P: Mutation in IDH1 gene
C/F: Multiple enchondromas, hemangiomas, phlebolith, lymphangioma, short stature, underdeveloped muscles
O/M: Hemangioma

MAGIC syndrome[41]
E/P: Autoimmune disorder (HLA-DR4)
C/F: Genital ulcers, relapsing polychondritis, myelodysplasia, arthritis, uveitis
O/M: Oral ulcers

Marfan syndrome[42]
E/P: Inherited disorder, Mutation in FBN1 gene
C/F: Flexible joints, scoliosis, ectopialentis, aneursyms
O/M: dolichocephaly, frontal bossae, prominent supraorbital ridges, malar hypoplasia, long and narrow face, retrognathic jaw, skeletal malocclusion, hypermobility of the temporomandibular joint

Mazabraud syndrome[43]
E/P: Mutation of GNAS1 gene results in overactive G protein triggering abnormal proliferation of melanocytes, osteoblasts & endocrine glands
C/F: Intramuscular myxomas, café au lait spots
O/M: Polyostotic Fibrous dysplasia

Mccune Albright syndrome[44]
E/P: Mutation of GNAS1 gene results in overactive G protein triggering abnormal proliferation of melanocytes, osteoblasts & endocrine glands
C/F: cafe au lait spots, endocrinal dysfunction
O/M: Polyostotic Fibrous dysplasia

Melkersonrosental syndrome[45]
E/P: Unknown origin
C/F: Facial palsy
O/M: Granulomatous chelitis, fissured tongue

Mikulicz syndrome[46]
E/P: rare chronic condition characterized by the abnormal enlargement of glandular tissue
C/F: Enlargement of lacrimal gland
O/M: Enlargement of salivary gland

Mohr syndrome[47]
E/P: Autosomal recessive disorder
C/F: polydactyly of hands and feet
O/M: median cleft lip, poly lobed tongue, absence of medial incisors

Mobius syndrome[48]
E/P: Rare congenital disorder caused due to vascular interruption in the subclavian artery territory, infections, hyperthermia, trauma, and teratogens
C/F: Sucking impairment, excess drooling, orofacial dysmorphology
O/M: High arch & cleft palate, hypoplastic upper lip, tooth agenesis, short & fissured tongue

Muir Torre Syndrome[49]
E/P: Autosomal dominant - Germ-line mutations in hMSH2 and hMLH1 genes - alteration or inactivation of tumor suppressor genes.
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).
O/M: Salivary gland tumors, Keratoacanthoma.

Murray–Puretic–Drescher syndrome / Juvenile hyaline fibromatosis / Fibromatosis hyalinica multiplex juvenilis / Systemic hyalinosis[50]
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

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