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Syndromes of oral and para-oral structures: A review

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Abstract
There is a wide spectrum of syndromes that affect the dental, oral, and craniofacial abnormalities. The primary aim of this review is to provide basic knowledge regarding syndromes, promote mutual understanding and interaction among all the medical and dental specialties involved in the early diagnosis and therapeutic guidance of patients presenting with these syndrome, in order to improve their quality of life.

Keywords: cleft lip, cleft palate, syndrome

Introduction
The word syndrome is a combination of two greek words (syn + dramein) which means "running together". It is recognized pattern of clinical abnormalities that have a single cause.[1] Cause of syndrome: When one or more chromosomes are missing or mutated, or if extra chromosomes are present, the proteins may not get made or made incorrectly or too many may be made. This can cause abnormal development and growth and result in a syndrome. Sometimes these abnormal genes or chromosomes are passed down from a parent, and sometimes they occur spontaneously without reason [1].

1. Ascher Syndrome
It is a combination of double lip, blepharochalasis (drooping of the tissue between the eyebrow and the edge of the upper eye lid), non toxic thyroid enlargement [2].

2. Apert Syndrome / Acrocephalosyndactyly / Syndactylic Oxycephaly
Mitten like syndactyly (fusion of fingers), Hallux varus (medial deviation of the great toe at the metatarsophalangeal joint), acrobrachycephaly (tower skull), kleeblattschadel deformity (cloverleaf skull), high palatal vault, v shaped maxillary arch, posterior palatal cleft, bifid uvula, Hypertelorism, depressed nasal bridge, exophthalmos, Crowding of teeth, Short stature, Mental retardation [3].

3. Behcet Syndrome
Recurrent oral ulceration, recurrent genital ulceration, skin lesion and ocular lesions like uveitis, retinal vasculitis, recurrent conjunctivitis and keratitis [4].

4. Beck With Wiedmann Hypoglycemic Syndrome
Macroglossia (enlarged tongue), neonatal hypoglycemia, mild microcephalu, umbilical hernia, fetal visceromegaly and postnatal somatic gigantism [1].

5. Burning Mouth Syndrome
It is characterized by pain and burning sensation of the mouth, altered taste sensation and xerostomia. No clinically detectable lesions in the oral cavity [5].

6. Chediak Higashi Syndrome
It includes oculocutaneous albinism, photophobia, nystagmus, recurrent infections of respiratory tract, skin and oral lesions [6].

7. Cleidocranial Dysplasia/Cleidocranial Dysostosis, Marie-Sainton Syndrome
It is characterized by frontal, parietal, occipital bossing, hypoplastic maxilla, high arched palate, hypoplasia or aplasia of paranasal sinus and clavicle, delayed exfoliation of primary tooth, delayed eruption of permanent tooth, malocclusion. Rarely dilacerations of roots and crown germinations can be seen [7].

8. Crouzon Syndrome/Craniofacial Dysostosis, Crouzon Disease, Apert Crouzon Syndrome
It includes craniosynostosis (premature fusion of cranial sutures), brachycephaly, scalocephaly, trigonocephaly, cloverleaf deformity of skull, Proptosis of eye leading to exposure keratitis, conjunctivitis, frontal bossing, midface hypoplasia. Parrot shaped prominence of nose, narrow maxillary arch, crowding of maxillary teeth, partial anodontia, peg shaped teeth. Rarely ocular malformations, cleft palate, hearing loss may be seen [8, 5].
Radiographically skull shows "Beaten silver" appearance/ beaten copper skull.

9. Carpenter Syndrome
A rare syndromic craniosynostosis with variable phenotypic expression characterized by craniosynostosis, high arched narrow palate, intellectual disability, distinctive facies, abnormalities of the fingers and toes (brachydactyly, polydactyly and syndactyly), short stature, congenital heart disease, skeletal defects, obesity, genital abnormalities and umbilical hernia [5].

10. Costen's Syndrome
It includes impairment of hearing, stuffy sensation in the ears especially while eating, burning sensation in the throat and mouth [5].

11. Cracked Tooth Syndrome
It is development of a crack in a restored tooth due to excessive occlusal force leading to sharp pain on biting [5].

12. Crest Syndrome
It includes calcinosis cutis, raynaud phenomenon, esophageal dysfunction, sclerodactyly (palmoplantar hyperkeratosis) and telangiectasia [9].

13. Cushing Syndrome/Hypercortisolism
It is characterized by adiposity about the upper portion of the body, mooning of the face and tendency to become round shoulders. There is dusky plethoric appearance with formation of purple striae. There is also muscular weakness, vascular hypertension, glycosuria, albuminuria, buffalo hump of neck. Osteoporosis and premature cessation of epiphyseal growth may be seen in children [5, 9].

14. Down's Syndrome/Trisomy 21/ Mongolism
Oral manifestations include Gingivitis, periodontitis, greater resistance to tooth decay, macroglossia, bruxism, hypoplastic maxilla, narrow arch, malocclusion, delayed exfoliation and eruption of teeth. Cardiovascular defects include ventricular septal defects, ALV communication, patent ductus arteriousus, mitral valve prolapsed. V shaped high palate, chapped lower lip, fissured tongue, macroglossia, microdontia, hypodontia, supernumerary teeth, taurodontism, hypocalcification, delayed eruption of teeth and malocclusion [10].

15. Digeorge Syndrome
It includes congenital absence of the thymus, resulting in immunodeficiencies, hypoparathyroidism, which results in hypocalcemia, conotruncal heart defects (i.e., tetralogy of Fallot, interrupted aortic arch, ventricular septal defects, vascular rings), Cleft lip and/or palate [5, 9].

16. Eagle's Syndrome
It is characterized by elongation of the styloid process or ossification of stylohyoid ligament. Features include dysphagia, sore throat, otalgia, glossodynia, headache and vague orofacial pain [11].

17. Ehler Danlos Syndrome/Rubber Man/ Cutis Hyperelastica/ Dermatorrehxis
There is hyperelasticity of the skin, hyperextensibility of the joints, fragility of the skin and blood vessels, hypermobility of TMJ, enamel hypoplasia. It also includes swan neck deformity of fingers, kyphosis, scoliosis, subcutaneous nodules, hypertelorism, wide nasal bridge, epicanthic folds, frontal bossing [5, 9, 12].
Gorlin sign: patient can touch his nose with tip of tongue.

18. Frey's Syndrome/ Auriculotemporal Syndrome
It includes flushing and sweating of the involved side of face, chiefly in temporal area during eating, gustatory sweating when eating spicy food. Crocodile tears- profuse lacrimation when food is eaten. Presence of cutaneous hyperesthesia infront and above the ear is seen [5, 13].

19. Gardener Syndrome
It includes multiple polyposis of large intestine, multiple osteomas of jaws and skull, multiple epidermoid or sebaceous cysts of skin, scalp and back, hypercementosis, multiple unerupted supernumerary teeth and odontomas [14].

20. Goldenhar Syndrome
It includes unilateral microstomia, mental retardation, hypoplastic zygomatic arch. Other features include downward slanting of palpebral fissures, malformed pinna and iris colobama, high arched palate, palatal and uvular cleft with malocclusion [15].

21. Gorham Syndrome / Massive Osteolysis / Phantom Bone
It is charecterised by osteolysis of single or multiple bones followed by replacement with fibrous tissue, arthralgia and facial asymmetry [5].

22. Gorlin and Goltz Syndrome / Jaw Cyst Basal Cell Nevus Bifid Rib Syndrome
It includes basal cell carcinoma, dermal cyst, palmar plantar keratosis, dermal calcinosis, odontogenic keratocyst, bifid rib, mandibular prognathism, hypertelorism, hypogonadism, mental retardation [5, 9].

23. Grinspan Syndrome
It includes diabetes, hypertension and lichen planus [5].

24. Heerfordt's Syndrome
It includes firm painless bilateral enlargement of parotid glands, inflammation of uvael tract of eye and facial plasy [5].

25. Horton Syndrome / Sphenopalatine Neuralgia
It includes unilateral paroxysms of intense pain in eye,

maxilla, ear, mastoid region, base of nose, zygoma. It is characterized by classic alarm clock headache in which there is absence of trigger zones and occurrence of pain everyday exactly at same time [5, 9].

26. Hypohidrotic Ectodermal Dysplasia / Anhidrotic Ectodermal Dysplasia / Christ-Siemens-Touraine Syndrome
It includes hypohydrosis, hypodontia, hypotrichosis, saddle nose, frontal bossing, peri orbital hyperpigmentation and wrinkling, hypoplasia/ aplasia of eccrine glands, eczema and pruritis [16].

27. Jaw Winking Syndrome / Marcus Gunn Phenomenon
It is characterized by rapid elevation of ptotic eyelid occurring on rapid movement of mandible on contralateral side and congenital unilateral ptosis [5].

28. Marfan Syndrome
Skeletal features include excessive length of tubular bone resulting in disproportionate long thin extremities. Skull and face are long and narrow. Ears are large, eyes appear sunken and frontal bossing is seen. The others include TMJ joint dysarthrosis, multiple odontogenic cysts of maxilla and mandible, high arched palate and bifid uvula [5, 9, 16, 17].

The steinberg sign: Fold your thumb into closed fist. This test is positive if thumb tip extends from palm of your hand.

The walker Murdoch sign: Grip your wrist with your opposite hand. If thumb and the fifth finger of your hand overlap each other, this represents positive walker Murdoch sign.

29. MC Cune Albright Syndrome / Osteitis Fibrosa Disseminate
This includes severe fibrous dysplasia involving nearly all bones of skeleton, pigmented lesions of the skin (café au lait spots) and endocrine disturbances like precocious puberty, hyperthyroidism, macrocephaly, acromegaly and cushing syndrome [9, 16].

30. Melkersson Rosenthal Syndrome
It is a traid of chelitis granulomatosa, facial paralysis and scrotal tongue [9, 16].

31. Noonan Syndrome / Female Pseudo-Turner Syndrome / Male Turner Syndrome
It is characterized by congenital heart disease, chest deformity, mental retardation, short statute, facial bone anomalies and cryptochidism [5].

32. Papillon Leferve Syndrome
It is characterized by juvenile periodontitis, gingival ulcers, inflammatory gingival enlargement and formation of pockets. Palmar plantar keratosis, generalized hyperhydrosis and dirty coloured skin are also seen [18].

33. Parry Romberg Syndrome / Progressive Facial Hemiatrophy / Progressive Hemifacial Atrophy
Oral manifestations include Hemiatrophy lip, tongue, muscles of mastication, mandible and hemi masticatory spasm. Unusually there may be small root of teeth, malocclusion and trigeminal neuralgia [5].

34. Peutz Jeghers Syndrome / Hereditary Intestinal Polyposis Syndrome
There is familial generalized intestinal polyposis, pigmentation of the face, oral cavity, sometimes hand and feet, there may be gastric, duodenal and colonic adenocarcinomas [5].

35. Pierre Robin Syndrome
There is u shaped cleft palate, micrognathia, glossoptosis. Occasionally hydrocephaly or microcephaly may be seen [5, 16].

36. Plummer Vinson Syndrome / Sideropenic Dysphagia / Paterson's Syndrome / Paterson- Brown-Kelly Syndrome
It is characterized by iron deficiency anemia (microcytic, hypochromic), dysphagia (due to esophageal webs), palor, weakness and fatigue. Others include angular chelietis, atrophic oral mucosa, atrophic glossitis and burning tongue [5, 9].

37. Ramsay Hunt Syndrome
It is zoster infection of the genicualte ganglion with involvement of external ear and oral mucosa. It includes facial paralysis, pain of external auditory meatus, vesicles on pinna of ear, vesicular eruptions in oral cavity, oropharynx, hoaarmess, tinnitus and vertigo [9].

38. Rubinstein Taybi Syndrome / Broad Thumb-Hallux Syndrome
It is associated with talon's cusp, developmental retardation, broad thumb and great toes. It also includes high arched palate, under developed maxilla and mandible, retrognathia, crowding and bifid uvula [5, 16].

39. Sjogren's Syndrome
Primary: Keratocinjunctivitis sicca and xerostomia**.**

Scondary: Along with primary lesions there may be lupus erythematous, polyarteritis nodosa, scleroderma and rheumatoid arthritis [9].

40. Steven Johnson's Syndrome
It is severe form of erythema multiforme. Eye lesions photophobia, conjunctivitis, corneal ulveration. Genital ulcers include non specific urethritis, balanitis and vaginal ulcers [9].

41. Sturge Weber Syndrome
It is characterized by angiomatosis of face (nevus flammeus), leptomeningeal angiomas, intracranial calcifications and contralateral hemiplagia [16].

42. Treacher Collin Syndrome / Mandibulofacial Dysostosis
Facial features include downward sloping of palpebral fissures (antimongoloid obliquity). recceding chin and malformation of external ear, hypoplasia of malar bone and mandible, blind fistula between angle of the mouth and angle of ear, fish like mouth, macrostomia, high arched palate and malocclusion [19].

43. Trotter's Syndrome
It is carcinoma of nasopharynx often producing trigeminal neuralgia like pain in the mandible, tongue and side of the head [5].

44. Vander Woude Syndrome / LIP PITS Syndrome
It is occurrence of pits of lower lip. Rarely may it include cleft lip and palate [16].

45. Weber Cockayne Syndrome
It is localized form of epidermolysis bullosa charecterised by bulla on hand and feet and are related to frictional trauma [5].

Conclusion
The diagnosis, genetic counseling and long-term treatment planning can be improved by the early detection of syndromes affecting craniofacial and dental structures. As the development of dental and facial structures is lagging in these syndromic patients, a deep phenotyping of craniofacial syndromes is possible in only young adulthood. This early detection also helps in early intervention of orthodontic treatment and maxillofacial surgery, especially in patients with disrupted development in the craniofacial and dental structures.

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