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REVIEW ARTICLE

SYNDROMES AND THEIR ORAL MANIFESTATIONS: A REVIEW

[1*]Dr. Himanshu Singh, [2]Dr. Avijit Avasthi, [3]Dr. Sanjay Talnia and [4]Dr. Kundendu Arya Bishen

[1]Senior Lecturer, Department of oral Pathology and Microbiology, Index Institute of dental Sciences, Indore, Madhya Pradesh, India
[2]Senior Lecturer, Department of Public Health Dentistry, Bhojia Dental College And Hospital,Budh (BADDI), Solan , Himachal Pradesh, India
[3]Senior Lecturer, Department of Oral and Maxillofacial Surgery, Swami Devi Dyal Hospital and Dental College, Barwala, Haryana, India
[4]Head and Professor, Department of oral Pathology and Microbiology, Index institute of dental Sciences, Indore, Madhya Pradesh, India

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Corresponding author: Dr. Himanshu Singh

ABSTRACT

Variety of syndromes shows number of manifestations in oral cavity. Knowledge of these syndromes and their manifestations is important as this will help in diagnosis and management of syndromes as well as dental diseases. In this review article, we are aimed at discussing various syndromes which predominantly shows oral manifestations.

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INTRODUCTION

In the past years, there has been tremendous growth in genetic researches, which increases the awareness regarding the various genetic disorders and syndromes. There are variety of syndromes which affects directly the various organs or structures of body. One of the most common site is oral cavity, where number of syndromes shows their manifestations in various form of dental diseases. Some syndromes occurs in children whereas some are predominantly occurs in adults.

Syndromes and their oral manifestation aarskog syndrome: Aarskog syndrome is an x-linked disorder, which induces by variation in fgd-1 gene .Other names of aarskog syndrome are facio-digito-genital syndrome, aarskog-scott syndrome, or faciogenital dysplasia. This syndrome principally affects male, which is distinguised by skeletal, facial and genital anomalies. The females shows insignificant manifestations of this syndrome (Moraes *et al.,* 2006). General prevalent features of this syndromes comprises of short distal extremities, hyper flexible joints, bifid scrotum, delayed puberty. It also shows internally twisted little finger and webbed representation of feets and hands (Orrico *et al.,* 2010; Bozorgmehr, 2006). Scoliosis and spina bifida occulta with cervical spine abnormalities have also been observed (Jogiya, 2005). The most common facial features shows by aarskog syndrome are hypertelorism ,long philtrum, broad nasal bridge and widow's peak (Halse, 1979).

Oral manifestations: This syndrome shows various dental manifestations which includes hypodontia, dental malocclusion, overcrowded teeth and delay in permanent teeth eruption. Also, some features like decrease in vertical facial height and maxillary hypoplasia are also seen (Melnick, 1976; Dayal, 1990; Jones, 1965).

Angelman syndrome: Angelman syndrome,first reported by Harry angelman in 1965,is neurological or neuro-developmental disorder of genetic origin. In his report, angelman described three children that he mentioned them as puppet children as these childrens shows abnormal arm posture and jerky gesture (Angelman, 1997). The prevalence of angelman syndrome is approximate to be 1 out of 15000-

30000.the reason for appearance of angelman syndrome is thought due to insufficient expression of UBE3A (ubiquitin-protein ligase e3a) gene in brain (Kishino, 1997). Angelman syndrome shows some behavioral hallmark .These are easily provoked laughter, short attention span, happy demeanor, affinity for water and disturbance in sleep (Williams, 2010; Pelc, 2008). Seizures happen in 80%-95 % cases having angelma syndrome and it typically appear in childhood (Thibert, 2013).

Oral manifestations: Angelman syndrome shows dental manifestation. These includes presence of thin upper lip, prominent mandible, uncontrolled chewing behaviour and widely spaced teeth with midline diastema (Clayton-Smith, 2003; Clayton-Smith, 1992). Some other features which also contribute to dental manifestations are long as well as narrow face, anteriorized tongues, anterior open bite. Also, tongue thrusting and uncontrolled drooling can be seen (Faria *et al.,* 2002; Jefferson, 2010).

Hallermann-streiff syndrome: In 1948, Aubry was the first person who reported Hallermann-streiff syndrome. Later, a comprehensive clinical explanation of the disease was described by Hallermann in 1948 and by Streiff in 1950 (Kirzioğlu, 2009). It apparently occurs because of developmental disorder which happens in 5^{th}-6^{th} gestational week which outcomes as defect in 2^{nd} branchial arch (Shiomi, 1999).

Oral manifestation: Hallermann-streiff syndrome present with various dental manifestations. It includes high arched palate, small and retracted tongue, open bite and class II malocclusion, natal teeth and supernumerary teeth (Jain, 2011; Gungor, 2015). Other significant dental manifestation are enamel hypoplasia, maxillary hypoplasia, poorly matured paranasal sinuses and severe dental caries (Kirzioğlu, 2009; Shiomi *et al.,* 1999).

Fraser Syndrome: Fraser syndome was first reported in 1962 by George R Fraser, an canadian Genetician. This syndrome is considered as autosomal recessive disorder. The distinctive feature of this syndrome are cryptophthalmos, mental retardation, genitourinary tract and laryx deformity, syndactyly. Other names of Fraser syndrome are Meyer-Schwickerath's syndrome, Ullrich-Feichtiger syndrome, Fraser-Francois syndrome and Cryptophthalmos syndrome (Francannet *et al.,* 1990; Van haelst, 2007; Fraser, 1962). It is observed that Fraser syndrome results from mutation in Fras 1 and Frem 2 human genes (Mcgregor, 2003).

Oral manifestation: The Fraser syndrome shows various dental manifestations. These includes agenesis of second premolars, delayed development of teeth, retained deciduous teeth and microdontia of deciduous molars (Kantaputra *et al.,* 2001; Ide, 1969). Other important features are, malocclusion, cleftlip or cleft palate, hypoplastic teeth, tooth crowding and ankyloglossia (Boyd, 1998).

Moebius Syndrome: Moebius syndrome was initially reported in 1880 by Von Graefe and in detailed by Paul Julius Moebius in 1888. Predominantly, this syndrome is distinguished by 6^{th} and 7^{th} cranial nerve palsy (Ouanounou, 2005). Other names of Moebius syndromes are nuclear agenesis, congenital facial diplegia, Congenital oculofacial paralysis, congenital nuclear hypoplasia, and congenital abducens-facial paralysis (Jones, 1988).

Oral manifestation: Various dental manifestations of Moebius syndrome are cleft palate ,incomplete lip closure, hypodontia, abnormal tongue movements and mandibular hypoplasia (Strömland, 2002; Domingos, 2004). Other features contributing to dental manifestations are gingivits, open bite, nursing bottle caries, bifid uvula, micrognathia and atrophy of tongue (Rizos *et al.,* 1998; Sensat, 2003).

Hutchinson–Gilford Progeria Syndrome: This syndrome was first described by Jonathon Hutchinson in 1886 (Hutchinson, 1886). Hutchinson-Gilford progeria syndrome occurs because of mutations seen in LMNA (lamin a/c) gene. Patients suffering from this syndrome shows premature aging as a typical feature. The common characteristics of this syndromes includes thin skin, interupt post natal growth, mottled hyperpigmentation, high pitched voice etc (Gilford, 1904; Gordon, 2007; Hutchinson, 1886).

Oral manifestation: In dentistry, this syndromes shows variety of oral feautres. These features includes delayed tooth eruption, micrognathia, hypodontia, severe crowding, hypoplastic mandible. In some patients, delayed anterior and vertical growth is also seen (Domingo, 2009). Microscopic investigation of the dentition discloses the irregularity in size as well as shape of odontoblast, with reticular pulp atrophy and obstruction in coronal calcification (Yu, 1991).

Rett Syndrome: Rett syndrome is known as neurological disorder which was initially described in 1966 by Rett. This syndrome is characterized by retarded growth of head, delay in psychomotor development and seizures (Chahrour, 2007; Rett, 1966). Other prevalent features are peripheral vasomotor disturbances, periodic apnea, hyperventilation, bloating of abdomen, loss of weight and growth retardation (Hagberg, 1989).

Oral manifestation: Various oral findings seen in Rett syndrome are bruxism, hand/digit sucking, gingivitis, anterior open bite, micrognathia, masseter muscle hypertrophy (Buccino, 1989; Fuertes-González, 2011).

Conclusion

In the present era, we are rewarded with latest and advanced technology in medical sciences. It is important for all people in healthcare professions to have excellent knowledge of syndromes. This knowledge helps us in diagnosing and prevention of the syndromes along with their management.

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