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REVIEW

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The Influence of Some Rare Syndromes on Oral and Perioral Health: A Literature Review

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Abstract

Rare medical syndromes may directly relate to the oral health status and, so far, affect the oral and dental management provided to the patients by dentists. Among many clinical features of these rare syndromes, the oral findings may include oral exophytic lesions, recurrent oral ulcerations, hemangiomas, vascular lesions, bone lesions, altered immune response with poor wound healing, and oral and craniofacial features which may require management by a team of different dental specialties. Some characteristic oral findings are considered prognostic or diagnostic for more serious or acute conditions that require urgent interventions. The aim of this review is to offer the general dental practitioner, dental specialists, and the oral medicine specialists with brief reviews about some rare syndromes that are characterized by multiple oral, dental, and craniofacial features and findings. Google Scholar, the National Library of Medicine (PubMed/MEDLINE), PubMed Central (PMC), and ScienceDirect are the main electronic databases that have been searched. Research articles, case reports, and review literature were involved in the present paper. In conclusion, early oral diagnosis and intervention in some rare conditions associated with characteristic oral, cranial, and facial features may reduce the need for more complicated treatment plans.

Keywords: Syndrome, Rare disorders, Oral ulceration, Oral findings, Oral mucosa, Oral manifestations

1. Introduction

The meaning of the word "rare" as a medical term is that a health condition affects a small group of the population. The number of affected patients may vary and depend on the prevalences reported documents [1, 2]. The obvious development of genetic and molecular investigations, the advances in technological and biomedical fields, the increasing prevalence of rare conditions that affected different populations, and the medication requirement for certain rare conditions; all these together increased the attention to enhance the knowledge about the rare conditions [2, 3]. This paper provides brief reviews about groups of rare syndromes with different etiologies and pathogeneses. These syndromes show variable oral manifestations and craniofacial characteristics that dentists should be aware of and have the complete knowledge about them.

2. Methodology

Google scholar, National library of Medicine (PubMed/MEDLINE), PubMed Central (PMC), and ScienceDirect are the main electronic databases that have been searched. Research articles, case reports, and review literatures were involved in the present paper. The rare syndromes included in this paper were randomly selected and reviewed from the medical and dental studies in the last five years. Oral findings and oral manifestations were the keywords that have been used during the search for syndromes articles.

- **Ascher syndrome**

It is a skin disorder, benign and is rarely occur. *Clinically*, this syndrome presented as a triad of signs including nontoxic thyroid gland enlargement, a double upper lip, and blepharochalasis. The

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inflammation and the hypertrophic enlargement of the minor salivary glands in the labial mucosal surfaces result in progressive upper lip enlargement. There is no specific cause for this condition but hormonal dysfunction and trauma are reported [4–6].

- **Bare lymphocyte syndrome (Qualitative T-Cell Defects)**

It is rare, autosomal dominant, its early manifest and includes the major histocompatibility complex MHC II defects. these defects result in insufficient activation cytotoxic T-cell and reduced in serum immunoglobulin levels, as T cells fail to present antigen to B cells. *Clinically*, Cytomegalovirus, Salmonella, and Cryptosporidium infections can occur, as well as opportunistic infections in the oral cavity as Candidiasis, stomatitis, herpes infection, and oral warts (Human Papilloma virus) [7–10].

- **Bazex syndrome**

Its acrokeratosis paraneoplastica; a rare syndrome. Its showed higher male predilection. It is accompanying malignancies (Squamous cell carcinoma) particularly of the upper digestive and respiratory systems, as well as other types of malignancies have been also reported. *Clinically*, it is associated with skin lesions appear symmetrical scaly plaques resemble the psoriatic lesions. The characteristic feature of these scaly lesion is its appearance on the ear lobes and nose in addition to other skin areas. Oral cancer and lip cancer have been reported to be associated with Bazex syndrome. The pathogenesis may associate with immune reaction to certain antigens, or due to growth factor productions [11–14].

- **Beckwith-Wiedemann syndrome**

it is congenital disorder with chromosomal abnormality. *Clinically*, this condition characterized with many features include the craniofacial areas, heart defect, kidney and liver tumors, and other body characteristics. The most common oral feature is the large size of the tongue (macroglossia), that presented from the first moment of child birth. This may further associate with problems in the growth of jaws and teeth. Dry and fissured tongue, skeletal class III, prognathism, apparent long facial Hight, teeth spacing with anterior open bite, speech and chewing problems, drooling, breathing problems, aesthetic issues, and other associated oral features have been reported in patients with BWS [15–18].

- **Bernard–Soulier syndrome**

The syndrome is a rare hereditary qualitative bleeding disorder arises from recognized absence in platelet membrane glycoprotein Ib-IX-V complex and in turn the platelets will be unable to interact with von-Willebrand Factor. *Clinically*, the patients experience spontaneous haemorrhage. The management of this syndrome is mainly depended on multiple transfusions of platelets which are usually reserved for more acute bleeding episodes or for surgical procedures. Prolonged bleeding is the main dental complication after dental procedures like tooth extraction and gingival bleeding [19–22].

- **CREST syndrome**

CREST is the acronym of clinical features which diagnosed the CREST syndrome and mean "calcinosis, Raynaud's phenomenon, esophageal dysmotility, sclerodactyly, and telangiectasia". This syndrome required three out of these five features and considered as a rare form of systemic scleroderma. It is an autoimmune slowly progressed connective tissue disorder and may show multiorgan involvement. *Clinically*, the syndrome associated with multiple orofacial manifestations among them; pale mucosa, progress limitation in mouth opening, oral pigmentations, hardening of the oral mucosa and tongue, low salivary flow, and oral telangiectasia [23, 24].

- **Familial atypical multiple mole melanoma syndrome**

It is an inherited disorder; an autosomal dominant associated with numerous melanocytic atypical nevi > 50 in number. Carcinomas and different malignancies are also reported to be developed in this syndrome, particularly the pancreatic cancer. *Clinically*, the nevi which presented within the oral mucosa; related to malignant melanoma. Oral squamous cell carcinoma currently reported associated with this syndrome, mainly tongue SCC, palatal SCC, and head and neck SCC [25–28].

- **Felty's syndrome**

It is rare an autoimmune condition (HLA-DR4 genotype) associated with thrombocytopenia, neutropenia, and splenomegaly along with rheumatoid arthritis RA. The patients have vulnerabilities to infection particularly bacterial in case of severe neutropenia present. Clinically, the associated genotype act as an indicator for more destructive seropositive rheumatoid arthritis along with more manifestations in extra-articular body areas. Temporomandibular joints have been also reported affected by the rheumatoid arthritis [29–31].

- **Gorlin-Goltz syndrome**

It is rarely inherited disorder; an autosomal dominant trait. *Clinically*, among many other multisystemic and clinical manifestations of the syndrome,

its characterized with multiple odontogenic tumors (keratocystic tumors) and basal cell carcinomas in the skin particularly may present on the non-sun exposed skin areas (Nevoid basal cell carcinoma). The proper management is symptomatic; the odontogenic tumors treatment is simple curettage or marsupialization [32–34].

- **Heerfordt-Waldenström syndrome**

This rare subacute type of sarcoid which may occur whether systemic sarcoidosis present or not. Its etiology showed from the granulomas, T lymphocytes, and mononuclear phagocytes which destructed the affected tissue. *Clinically*, parotid glands are the most frequently involved from the major salivary glands. It is distinct by the triad of eye inflammation, particularly the uveal tract, facial nerve palsy, and parotid swelling along with xerostomia [35, 36].

- **Hyperparathyroidism jaw tumor syndrome (HPT-JT)**

It is a rare syndrome; an autosomal dominant disorder. *Clinically*, the syndrome is characterized by numerous ossifying fibromas, including both jaws, along with cystic adenomas or cystic carcinomas of the parathyroid glands and other renal disorders. These adenomas and jaw tumors are occurred in separation to each other and so far, the jaw tumors do not heal after the removal parathyroid gland. Laboratory investigation in cases of bone lesions showed an increase in level of alkaline phosphatase. The adenoma assessment required radiographic methods [37–39].

- **Job's Syndrome**

The syndrome is also called Hyper-IgE Syndrome HIES with mutations in STAT3 were shown to underlie HIES. It's an autosomal dominant disorder include neutrophil disorder, a qualitative disorder, and phagocyte activation. Job's syndrome is characterized by respiratory infections, recurrent skin infections, and eczematoid dermatitis, which present as a triad. *Clinically*, oral features showed craniofacial characteristics, dental features, and the susceptibility to fungal infection. These features become distinguished in late puberty. Delayed exfoliation of primary teeth is also a frequent feature. Delayed or absent root resorption in primary teeth, a fissured tongue, deep grooves on the buccal mucosa, and a high deep palate are also observed [40–42].

- **Kostmann's syndrome**

It is a condition that showed a severe congenital neutropenia (SCN) with a low absolute neutrophil count (ANC) for more than 6 months and there is no specific underlying pathology. In SCN, children typically have ANC of $<500/\mu$L. SCN is a heterogeneous disease that results from mutations in a variety of genes (ELANE, HAX1, and GFI1 mutation). Patients experience deep tissue infections and abscesses, secondary anemia, and thrombocytosis [43, 44]. *Clinically*, other findings often initiate early in childhood as gingivitis, severe periodontitis, and serious bacterial infections. Younger patients affected with the manifestation of severe periodontitis, which results in early loss of the deciduous teeth, may indicate the occurrence of chronic benign idiopathic neutropenia. The management of oral ulceration and the breakdowns that result from the severe periodontitis may initiate with G-CSF along with good oral hygiene and dental care for a couple of weeks, or may require the correction of the ANC levels in some patients with SCN [45].

- **Lacrimo-auriculo-dento-digital syndrome (Levy-Hollister syndrome)**

It is a rare congenital disorder, also known as Levy-Hollister syndrome. *Clinically*, the syndrome is associated with numerous anomalies, including the shape and position of ears; low-set and cup-shaped ears with hearing problems. Anomalies in digital features and extremities, keratoconjunctivitis and less commonly, alacrima and xerophthalmia. Salivary gland anomalies may result in xerostomia and increase susceptibility to tooth decay. Anomalies in dentition include microdontia, enamel hypoplasia, delayed eruption [46, 47].

- **Maffucci's syndrome.**

It's Considered one of the hemangioma syndromes, accompanied by numerous chondromas (chondrodysplasia); it is associated with vascular and bone lesions. *Clinically*, tumors containing cartilage are rarely found in jaws, and if detected within the oral cavity or jaws, they should be diagnosed cautiously to eliminate malignancies. The main concern with these tumors is that the diagnosis must be accurate. The tumors diagnosed as non-malignant can be treated conservatively by surgical excision [48–51].

- **Melkersson–Rosenthal syndrome**

It is a rare, neurological disorder. This syndrome shows respiratory symptoms (Granulomatous diseases), Facial palsy, Parotid gland hypoplasia, Fissured or furrowed tongue. *Clinically*, the facial and labial swelling is more common and recurrent and can develop into progressive fibrosis. A cobblestone pattern within the buccal mucosa has also been reported. The etiology of this condition remains unclear, although genetic, inflammatory, immunological, and infectious factors may be associated with its incidence [52–54].

- **Multiple Endocrine Neoplasia Syndrome2 MEN2**

The syndrome is very rare, occurring at any age and affecting both males and females equally. It is an inherited disorder associated with a mutation in the proto-oncogene (RET) and presented as multiple hyperplasia and tumors within the neuroendocrine tissues. The syndrome has a variant mentioned as MEN 2B,characterized by a specific phenotype and unique presentation and signs, as marfanoid body habitus, thyroid carcinoma, and pheochromocytoma. *Clinically,* this variant also involves lip enlargement and oral mucosal neuromas. The oral neuromas may appear before the other signs of the syndrome [55–57].

- **Paraneoplastic autoimmune multiorgan syndrome**

It is a severe, multiorgan, and blistering disease. This condition is connected with an underlying neoplasm. The most frequent neoplasms included chronic lymphocytic leukemia, non-Hodgkin lymphoma, thymoma and other neoplasms [58]. Systemic involvement, particularly in lungs,with different presentation on the skin. The lesions of PNPP in histopathological investigations showed inflammation at the junction between dermis and epidermis, keratinocyte necrosis, and acantholysis. Direct immunofluorescence assays showed IgG deposition at the basement membrane as in pemphigoid, besides the keratinocyte surface creating a lattice pattern as in Pemphigus Vulgaris. Indirect immunofluorescence showed antibodies binding to epithelium and to bladder, heart, and liver tissues. *Clinically*, the oral mucosal lesions presented painful erosive lesions, erythema multiforme-like, lichenoid reaction-like, as most commonly reported [58–61].

- **Lesch–Nyhan syndrome**

This condition is rare, X- linked inherited disorder. *Clinically*, patients often develop ulcerative lesions in lips, tongue, and oral mucosa because they are uncontrolunable the self-injury. During their childhood, they have self-mutilation behaviors and are always biting their lips. Later in their life, they develop more aggressive actions. A mouth guard may be required in order to protect the lips, oral cavity, and oral mucosa. Extraction of all the teeth may be recommended in some situations. The patients may also suffer from growth and mental retardation in addition to neurological dysfunction [62–64].

- **Sturge-Weber syndrome**

This syndrome is rare and associated with hemangiomas (encephalotrigeminal angiomatosis). Epilepsy and mental problems have also been reported with this syndrome. *Clinically*, this syndrome is also associated with oral hemangiomas presented unilaterally in the mandible, maxilla, tongue, gingiva, and other oral and perioral areas [65, 66].

- **Sweet syndrome**

It is an acute febrile neutrophilic dermatosis associated with recurrent oral ulceration. *Clinically*, the disease is characterized by fever, multiple painful pustular ulcers and elevated lesions on the extremities, neck, and face. The syndrome may be presented with severe painful oral pustular lesions on the buccal mucosa, tongue, and lips. This syndrome is associated with vasculitis and clinically shows similarity in presentation with Behçet disease, erythema multiforme, pemphigus vulgaris, and pemphigoid, but has a different disease course and different etiopathogeneses [67–70].

- **Wiskott–Aldrich syndrome**

Rare X-linked recessive condition associated with thrombocytopenic purpura, eczema, and an opportunistic infection as a result of a defect in immunity. There is a defect in the quantity and the quality of platelets. *Clinically,* oral manifestations include petechiae seen in the oral cavity, particularly on palate and bleeding from the gingiva. Clinical manifestations that can be seen directly after birth include bruises, bloody diarrhea, infections like skin infections and middle ear infections, and bacterial pneumonia, but manifestations may be delayed to a later age. Other complications such as autoimmune hemolytic anemia, autoimmune neutropenia, and vasculitis have also been found, along with an increased incidence of malignancy, particularly lymphomas [71, 72].

3. Conclusion

The early oral diagnosis and intervention in some rare conditions which are associated with characteristic oral, cranial, and facial features may reduce the need for more complicated treatment plan. Oral health providers may not only save the patient's teeth and provide aesthetic value, but also can improve the patient's general health by expanding their knowledge about the influences of rare general health conditions on oral, dental, and perioral health status.

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Ethical issue

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Conflicts of interest

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