Syndromes of Head and Neck – A Quick Review

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Abstract

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, biologically and physiologically. A group of deformation and malformation sequence etc that occurs together due to some identifiable underlying cause. The aim of this review is to describe a collective knowledge about various syndromes associated head and neck- A dentist point of view

Keywords: *Syndrome, Metabolism, Dental, Orofacial.*

Introduction

The real importance behind the learning of syndromes associated with conditions is of relevance to clinical examination of the head and neck. Knowledge of syndrome may quickly solve some difficult diagnostic problems and appropriate treatment instituted . The following list takes you through conditions met by the authors either in their clinical practice or in examinations and which could therefore be considered worth knowing and helpful in academic and clinical excellence.

In some instances, a syndrome is so closely associated with a pathogenesis and that the words yndrome, disease, and disorder end up being used interchangeably. This is especially true of inherited syndromes. For example, Down syndrome, Wolf–Hirschhorn syndrome, and Andersen syndrome are disorders with known pathogeneses, so each is more than just a set of signs and symptoms, despite the syndrome nomenclature[1,2]. In other instances, a syndrome is not specific to only one disease. For example, toxic shock syndrome can be caused by various toxins.

If an underlying genetic cause is suspected but not known, a condition may be referred to as a genetic association . By definition, an association indicates that the collection of signs and symptoms occurs in combination more frequently than would be likely by chance alone.

Syndromes are often named after the physician or group of physicians that discovered them or initially described the full clinical picture. Such eponymous syndrome names are examples of medical eponyms. Recently, there has been a shift towards naming conditions descriptively by symptoms or underlying cause rather than eponymously, but the eponymous syndrome names often persist in common usage

DIAGNOSIS BETWEEN A SYNDROME AND A DISEASE:

Representing medical knowledge is a highly complex endeavor. The improper use of the terms “syndrome”, “disease” and their relations to “diagnosis” is one of the difficulties with which medical informaticians must deal, especially when developing expert systems to support diagnoses. Although ubiquitous in medical and lay discourse, the term “disease” has no unambiguous,

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generally accepted definition. However, most of those using this term allow themselves the comfortable delusion that everyone knows what it means[3].

Diagnostic categories (diseases and most syndromes) are simply concepts. They are justified only if they provide a useful framework for organizing and explaining the complexity of clinical experience in order to derive inferences about outcome and if they guide decisions about treatment.

A **syndrome** is a recognizable complex of symptoms and physical findings which indicate a specific condition for which a direct cause is not necessarily understood. Thus in practice doctors refer to the infamous "viral syndrome" as such because of the uncertainty regarding the legion of viral agents that is causing the illness[4]. Once medical science identifies a causative agent or process with a fairly high degree of certainty, physicians may then refer to the process as a **disease**, not a syndrome. Mucocutaneous lymph node syndrome became Kawasaki syndrome which in turn metamorphosed into Kawasaki disease; the latter is properly a disease, no longer a syndrome, by virtue of its clearly identifiable diagnostic features and disease progression, and response to specific treatment.

Albert *et al.* catalogued six general views or concepts about what types of conditions may be said to constitute a disease. These views range from nominalism and cultural-relativistic theories (i.e. some conditions become a disease when a profession or a society labels it as such) to a "disease realism" view (objectively demonstrable departure from adaptive biological functioning). The latter model is the one best suited to the present state of medicine; it emphasizes that the clinical signs and symptoms do not constitute the disease and that it is not until causal mechanisms are clearly identified that we can say we have "really" discovered the disease[5,6].

Medical literature, even that from governmental organizations and institutions authorized to implement standards, is plagued with misleading assertions such as "a syndrome is a disease …", "a syndrome indicates a particular disease…" and "Lyme disease syndrome" (It is inappropriate to apply "syndrome" to Lyme disease because its causative agent is known).

Some syndromes such as "heart failure" are useful medical concepts but are not diagnoses, whereas more specific syndromes such as "congestive heart failure" or "right heart failure" are diagnoses[6]. Due to the imprecision of natural language, some syndromes could also imply a simple pathological finding (vasculitis) or just a physical finding. Frequently, for example, arthritis syndromes are simply referred to as "arthritis".

CAUSES FOR A SYNDROME:

When one or more chromosome is missing or mutated or if extra chromosome are present, then there is incorrect number of proteins made , this may cause abnormal development and growth and result in syndrome. Sometimes these abnormal genes or chromosomes are passed down from the parent and sometimes they occur spontaneously without reason[7,8].

CLASSIFICATION OF SYNDROME[9-11]:

1) Etiologic classification
2) Embryologic or histologic classification
3) Syndrome prototype
4) Polythetic classification
5) Monothetic classification
6) Mixed classification
7) Morphogenetic classification

1.ETIOLOGIC CLASSIFICATION

Syndromes can be classified according to broad etiologies suc as

i) Monogenic
ii) Chromosomal
iii) Environmental induced

Such classification usually requires supplementary category such as : Multifactorial , Unknown, and Disruptive

2.EMBRYOLOGIC / HISTOLOGIC CLASSIFICATION

Developmental disturbances of the tissue structure are basis of some classification. Thus hematoneoplastic

syndrome are sometimes classified on the basis of the germ layer involved.

EXAMPLE : 1) Multiple osteochondroma involves only one germ layer- the mesoderm

2) Gardner syndrome involves all three germ layer

3.Polythetic classification: syndromes sharing a large portion of their principal anomalies are grouped together . They are even better aid in differential diagnosis.

4.Syndrome prototype by Herrmann and Opitz 1974 and Cohen 1982:

i) Dysmetabolic syndrome (metabolism)

Normal birth, uniform clinical feature, no congenital malformation, biochemically defined, recessive mode of inheritance Example : Hurler syndrome , Tay- Sachs Disease.

ii) Deformation syndrome (region)

Changes in shape of previously normal structure, lack of movement – mechanical andfunctional, commonly affects musculoskeletal system

iii) Malformation syndrome (organ or field)

Non-contiguous malformation , Embryonic pleiotropy, Lack of biochemical definition example- Trisomy 13 syndrome, Rubenstein Tyabi syndrome

iv) Dyshistogenic syndrome (Tissue)

A) SIMPLE : Involvement of only 1 germ layer , Inheritance- dominant or recessive Example: Achondroplasia

B) HEMARTONEOPLASTIC SYNDROME : Hamartomas or neoplasias, involves more than 1 germ layer, commonly dominant. Example : Peutz Jeghers syndrome, Gardners syndrome

5.MONOTHETIC CLASSIFICATION

In this type of classification , syndromes are grouped together because they share a single feature such as for example cleft palate. Such groupings are often used as an aid in differential diagnosis, for example :

i) Syndrome with arthrogryposis

ii) Syndrome with craniosynostosis

iii) Syndrome with propensity

LIST OF SYNDROMES ACCORDING TO ORAL CAVITY SITE

SYNDROMES ASSOCIATED WITH DEFORMATION IN LIP[12]

FEATURES	SYNDROME
Double lip	Ascher's syndrome
Pebbly lesions of lip	Cowden's syndrome
Everted lip	Down's syndrome
Fusion of upper lip to maxillary gingival	Fusion of upper lip to maxillary gingival
Thick lips	Hurler syndrome
Pigmented lip	Laugier – Hunziker syndrome
Neuromas of lip	Multiple endocrine neoplasia syndrome
Cleft lip	Goltz Gorlin syndrome
Protruding lip	Reiger's syndrome
Occurrence of pits of lip	Van Der Woude's Syndrome
Whisting Lips	Whistling face syndrome
Dryness and fissuring of lips	Mucocutaneous lymph node syndrome

SYNDROMES ASSOCIATED WITH DEFORMATION IN BUCCAL MUCOSA[12]

FEATURES	SYNDROME
Lichen planus lesion on buccal mucosa	Graham little syndrome and Grinspan syndrome
Bilateral oral white lesion	Jadassohn Lewandowsky syndrome
Neuromas of Buccal Mucosa	multiple endocrine neoplasia syndrome
Oral Leukoplakia	Zinsser-Engaman – Cole syndrome

SYNDROMES ASSOCIATED WITH DEFORMATION IN TONGUE[12]

FEATURES	SYNDROME
Macroglossia	Down's syndrome / Beckwith syndrome
Microglossia	Hanhart's syndrome
Aglossia/ Hypoglossia	Hypodactilia syndrome
Fissured tongue	Melkerssons Rosenthal Syndrome
Glossitis	Chediak – Higashi Syndrome
Hypotomia Involving tongue	Floppy Infant syndrome
Multiple Hemangiomas	Maffucci's Syndrome
Atrophy of tongue	Parry- Romberg syndrome
Glossoptosis	Pierre Robin Syndrome

SYNDROMES ASSOCIATED WITH DEFORMATION IN PALATE[12]

FEATURES	SYNDROME
Hight palate vault	Apert's syndrome Crouzon syndrome Down's syndrome Marfan syndrome Teacher Collin syndrome
Cleft plate	Pierre robin syndrome Goltz gorlin syndrome
Pebbly lesion of palate	Cowden's syndrome

SYNDROMES ASSOCIATED WITH DEFORMATION IN GINGIVA[12]

FEATURES	SYNDROME
Severe gingivitis	Chediak- Higashi syndrome
Pebbly lesion of gingival	Cowden's syndrome
Gingival enlargement	Cross syndrome
Gingival Fibromatosis	Ramon Syndrome
Inflammatory gingival enlargement	Papillon- Lefevre syndrome
Congenital enlargement of gingival	Rutherfurd's syndrome
Massive growth of gingival	Sturge-Weber syndrome

SYNDROMES ASSOCIATED WITH DEFORMATION IN TEETH[12]

FEATURES	SYNDROME
Microdontia	Down's syndrome / Gardner's syndrome / Golts-Gorlin syndrome
Macrodontia	KBG syndrome
Hypodontia	Down's syndrome
Altered Eruption	Turner's syndrome
Congenital missing teeth	Curry-Hall syndrome / Bloch-Sulzberger syndrome
Absence of premolars and 3rd molars	Book's syndrome
Additional cusp	Ellis-Van Creveld syndrome
Peg or cone shaped teeth	Crouzen syndrome / Curry-Hall syndrome/ Trichondental syndrome
Supernumenary teeth	Down's syndrome/ Gardner's syndrome
Taurodontism	Trichodental syndrome
Talon's cusp	Rubinstein- Taybi Syndrome
Enamel hypoplasia	Ehlers-Danols syndrome
Development of crack during excessive occlusal force	Cracked tooth syndrome

SYNDROMES ASSOCIATED WITH SALIVARY GLAND[12]

FEATURES	SYNDROME
Decrease salivary secretion/ dry mouth	Sjogren's syndrome

SYNDROMES ASSOCIATED WITH JAW BONE [12]:

FEATURE	SYNDROME
Micrognathia	Pierr robin syndrome/ Turner's syndrome
Fibrous dysplasia	Albright's syndrome / Jaffe Lichenstein syndrome
Cortical thickening or hyperostosis	Caffe-Silverman syndrome
Mandibulofacial dysostosis	Treacher Collins Syndrome
Mandibular Hypolasia	Hutchinson Gilford syndrome
Maxillary hypoplasia	Reiger's syndrome
Cleft of Alveolar process	Orofacial Digital syndrome
Elongated styloid process	Eagles syndrome

Conclusion

There is a wide spectrum of syndromes that include dental, oral, and craniofacial abnormalities. Medical specialists like pediatricians and geneticists focus on general health issues related to diagnosis and prognosis of the condition, having a holistic view of the patient, whilethe dentist deal with (major) facial corrections where we primarily work with caries prevention, diagnosis, and treatment of structural tooth abnormalities, tooth size-shape discrepancies, deviations in tooth number and treatment of malocclusions or facial growth disturbances. In Contrary to the general and genetic diagnosis, most dental abnormalities can only be identified after the first years of life. This delays the dental and orofacial components of the syndromic diagnosis, which are, however, vital for the evaluation of prognostic factors and for the proper timing and management of oral function and, aesthetics as well as for social aspects. Therefore a clear knowledge about the syndromes helps in proper disgnosis .This makes early interventions and decisions concerning the type and timing of orthodontic treatment and maxillofacial surgery often difficult and critical, especially in patients with disrupted development in the craniofacial and dental structures.

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