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Syndromes of Head & Neck Region Associated with Teeth – A Review

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Abstract
The word syndrome is derived from the Greek synonym (together) and dromos (running) and refers to a "running together" or concurrence of symptoms.
Teeth are specialized structural components of the craniofacial skeleton. Developmental defects occur either alone or in combination with other birth defects. Focusing on dental aspects, we will review a selection of multiple congenital syndromes (MCA) associated with teeth number and/or shape anomalies, they may serve as a diagnostic indicator or help confirm a syndrome diagnosis.
Keywords: *Syndromes, head and neck, teeth, developmental defects.*

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I. 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 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 the orofacial region, and thorough knowledge of their manifestations and implications is pertinent for diagnosis and to provide optimal personalized care with an integral approach. [1]

Focusing on dental aspects, we will review a selection of multiple congenital anomaly (MCA) syndromes associated with teeth number and/or shape anomalies. We will consider successively syndromic forms of amelogenesis imperfecta or enamel defects, dentinogenesis imperfecta (i.e. osteogenesis imperfecta) and other dentin anomalies. A better knowledge of the dental phenotype may contribute to an earlier diagnosis of some MCA syndromes involving teeth anomalies. They may serve as a diagnostic indicator or help confirm a syndrome diagnosis.

AMELOGENESIS IMPERFECTA

Amelogenesis imperfecta is a genetic condition that causes teeth to be abnormally small or discoloured. Teeth are also likely to be pitted or grooved and more susceptible to being worn down and breaking.

There is a large group of inherited developmental defects in enamel collectively referred to as amelogenesis imperfecta. At least 14 phenotypes have been identified, and autosomal dominant, autosomal recessive, and X-linked inheritance have been reported. The condition is rare; only about 1 in 14,000 individuals have it. (2,3)

Three general categories of amelogenesis are recognized:

1) Hypoplastic type:

Inadequate formation of enamel matrix; both pitting and smooth types exist. Enamel may be reduced in quantity but is of normal hardness.

2) Hypomaturation type:

A defect in the crystal structure of enamel leads to a mottled enamel with white to brown to yellow colours. (2)

3) Hypocalcified type: A defect not in the quantity but in the quality of enamel. It is poorly mineralized, soft and chips and wears easily. (3)



Fig. 1: Affected teeth exhibit yellowish-brown discoloration. Corresponding radiograph demonstrating a distinct reduction in enamel radiopacity. Adapted from (14)

DENTINOGENESIS IMPERFECTA

Dentinogenesis imperfecta is a genetic disorder that interferes with normal tooth development. It affects approximately 1 in 6,000 to 8,000 people, according to the National Institute of Health. There are three types of dentinogenesis imperfecta.

Type I occurs in individuals who have another inherited disorder called osteogenesis imperfecta (causes brittle bones), whereas Type II and Type III occur in those without other genetic disorders. Some researchers believe Types II and III are part of a single disorder, along with another condition called dentin dysplasia Type II, which primarily affects baby teeth more than adult teeth.

General symptoms of dentinogenesis imperfecta include: Tooth discoloration (blue-gray or yellowish-brown),Teeth are translucent and weaker than normal teeth, making them prone to erosion, breakage, and loss. This is an autosomal dominant condition affecting both deciduous and permanent teeth. Affected teeth are grey to yellow-brown and have broad crowns with constriction of the cervical area, resulting in a "tulip" shape. Enamel is easily broken,

leading to exposure of dentin that undergoes accelerated attrition. The gene maps to chromosome #4. It encodes a protein called dentin sialophosphoprotein (DSPP). This protein constitutes about 50% of the non-collagenous component of the dentin matrix. (4)

The following classification has been proposed:

A) Dentinogenesis imperfecta Type I, with osteogenesis imperfecta.

B) Dentinogenesis imperfecta Type II, without osteogenesis imperfecta.

C) Dentinogenesis imperfecta Type III.

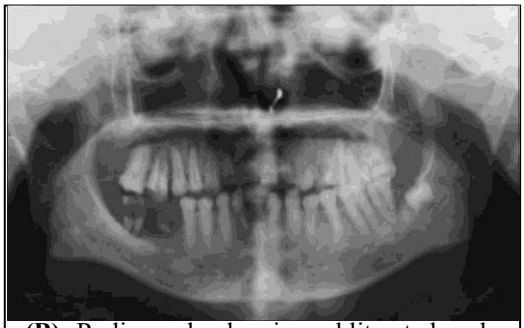
Dentinogenesis Type III is even more rare and paradoxically characterized by little dentin, resulting in "shell teeth."

Type III DI may be an allelic variant of Type II DI (a different mutation in the same gene). Both genes map to the same region on chromosome #4. (4,5)

Fig. 2: Adapted from (15)



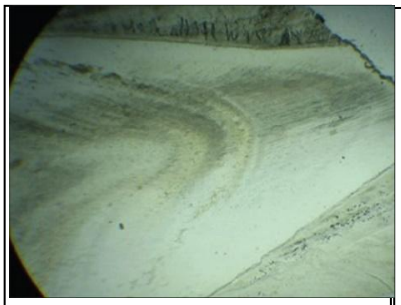
(A) Intraoral view with brownish discoloration and severe attrition



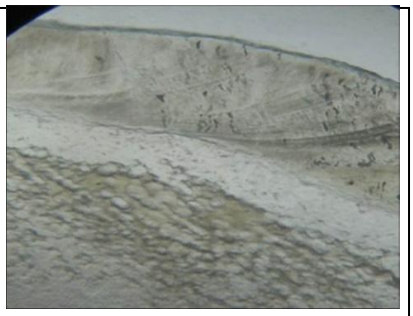
(B) Radiograph showing obliterated pulp chambers



(C) Photomicrograph showing irregular dentinal tubules



(D) Photomicrograph showing obliterated pulp chambers



(E) Photomicrograph showing increased interglobular dentin chambers

48,XXYY SYNDROME

A rare chromosomal condition that affects 1 in 18,000 to 50,000 males, 48,XXYY syndrome interferes with sexual development, causing reduced height, facial and body hair, increased risk of breast enlargement, infertility, progressive tremors, and other serious medical problems that develop increasingly later in life

Dental problems are also common. According to the NIH the delayed appearances of primary and secondary teeth, crowded and/or misaligned teeth; numerous cavities and thin tooth.

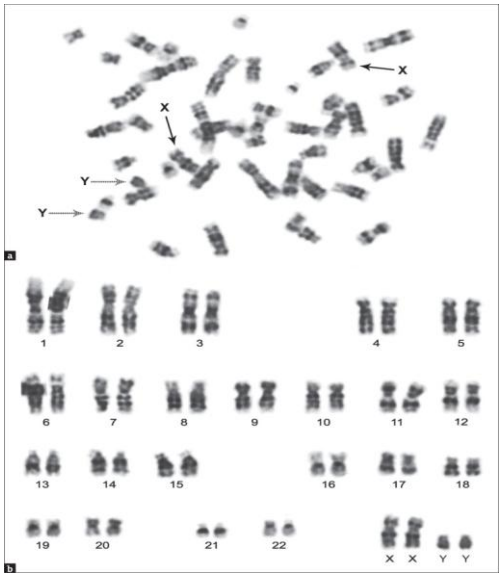


Fig. 3 : The Geimsa-Trypsin-Geimsa-banded 48, XXYY metaphase and karyotype of the patient, **(a)** 48, XXYY metaphase with two X (black arrow) and two Y (dotted arrow) chromosomes and **(b)** 48, XXYY karyotype prepared from the above metaphase sex chromosome aneuploidy. Adapted from (16)

HYPOHIDROTIC/ ANHIDROTIC ECTODERMAL DYSPLASIA.

Hypohidrotic ectodermal dysplasia is an inherited disorder affecting ectodermal structures, including the skin, nails, hair, sweat glands, and teeth. It occurs in approximately 1 in 17,000 individuals worldwide. The condition is characterized by variable involvement of these structures, with dental abnormalities being a prominent feature. According to the NIH, approximately 70% of carriers of the gene responsible for the condition, particularly those with only one mutated gene, may show manifestations such as missing or abnormal teeth, sparse hair, and abnormalities of the sweat glands.

The dental manifestations range from hypodontia to anodontia. In severe cases, teeth may fail to develop completely. More commonly, most deciduous teeth are present, while only a few or no permanent teeth develop. The teeth that are present are typically small, peg-shaped, or conical.

The major features of anhidrotic ectodermal dysplasia include:

• Usually inherited as a sex-linked recessive trait.
• Hypodontia.
• Hypotrichosis (scanty hair).
• Anhidrosis (inability to sweat).

In cases of anodontia, the alveolar process fails to develop adequately because of the absence of teeth, resulting in insufficient bone to support implants. The resulting loss of vertical dimension may give the facial profile an aged appearance. The hair is characteristically fine and sparse, particularly in the tonsural region (2,3). The skin is smooth, shiny, and dry due to the absence of sweat glands, leading to poor tolerance to heat. The fingernails may also be defective. Treatment is primarily aimed at improving appearance and mastication, and dentures are generally well tolerated by children (5,11).

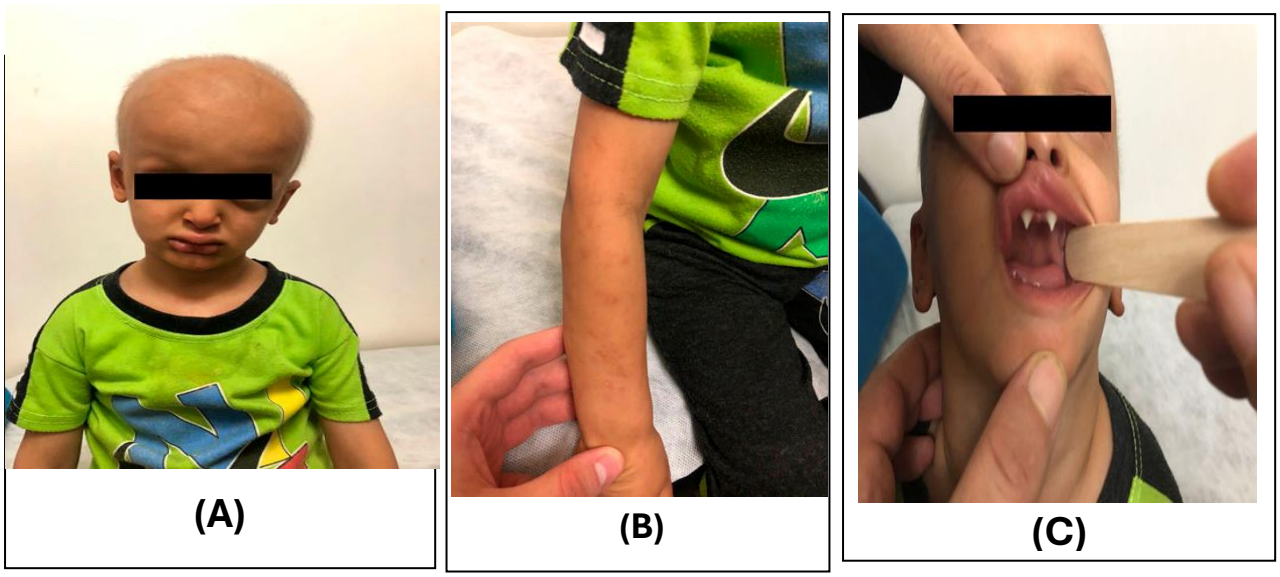


Fig. 4 : Showing **A)** sparse scalp hair, skimpy eyebrows and eyelashes, and fully everted lips. **(B)** Dry and excoriated skin. **(C)** Maxillary hypodontia with conical incisors. Adapted from (17)

OCULODENTODIGITAL DYSPLASIA.

Oculodentodigital dysplasia is an extremely rare genetic disease (with fewer than 1000 people diagnosed worldwide) that affect the eyes, fingers and teeth. Common tooth abnormalities include: small or missing teeth, numerous cavities, weak enamel and early tooth loss, according to the NIH. The condition can also lead to small eyes, vision loss, webbed skin and neurological problems. An autosomal dominant disorder, it develops when only one mutated gene is inherited from a parent.



Fig. 5: Showing Dental features: Hypodontia, microdontia, dental caries and partial anodontia. Adapted from (18)

RECOMBINANT 8 SYNDROME

Recombinant 8 Syndrome is a rare disease of unknown incidence primarily affecting an Hispanic population descending from the San Luis Valley of Colorado and Northern New Mexico.

Inherited as an autosomal dominant pattern, recombinant 8 syndrome causes distinctive facial abnormalities, moderate to severe intellectual disability and heart and urinary tract problems.

Abnormal teeth, an overgrowth of gums, small chin, thin upper lip and downturned mouth are all characteristic features.



Fig. 6 : Illustrates age progression of hypertelorism, thin upper lip, anteverted nares, wide face, infraorbital creases, low set ears, downturned mouth, micrognathia, depressed nasal bridge, thick lower lip, frontal upsweep characteristic of recombinant 8 syndrome.
Adapted from (19)

TRICHO-DENTO-OSSEOUS SYNDROME

The tricho-dento-osseous syndrome is characterized by kinky, coarse and curly, fair coloured hairs, present at birth in 80% of cases. Half of the patients retain this phenotype beyond infancy.More than 90% of the individuals with TDO have an increased cranial thickness, obliterated diploe and absence of mastoid pneumatization.

Teeth show uniformly thin or pitted hard enamel, enlarged pulp chambers, and taurodontism. The degree of taurodontism and the disturbed distribution of affected teeth are highly variable.In some individuals there is severe taurodontism of all the posterior teeth while the anterior teeth show only enlarged pulp chambers. Both primary and permanent teeth are typically affected.

Dental anomalies in TDO are similar to those observed in AI hypomaturation-hypoplastic type with taurodontism, which is inherited as a highly penetrant autosomal dominant trait. AIHHT shows no alteration in hair or bone.

The clinical pattern of expression of DLX3 mutations depends on the altered functional domain of the DLX3 protein; AI and taurodontism is a constant feature but hair and bone anomalies are observed with mutations outside the homeodomain.

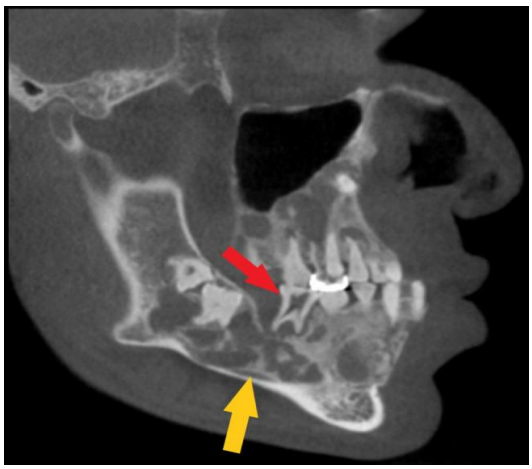


Fig. 7 : There is elongated pulp chamber, apically positioned furcation, shortened roots are noted in tooth #19 and molars on right side, consistent with taurodontism (red arrow). Adapted from (20)

VITAMIN D-DEPENDENT RICKETS

Vitamin D-dependent rickets, type I is an AR defect in vitamin D metabolism. Patients with VDDR1 have mutations of the 1-alpha-hydroxylase gene (located at 12q13; 1eq13.3) resulting in decreased levels of $1,25(OH)_2$ vitamin D_3.

Clinical features include growth failure, hypotonia, rachitic bones and enamel hypoplasia. Zambrano et al. described a patient with hypoplastic, yellow- to brownish enamel on all permanent teeth, and periodontal disease. Dental radiographs showed large quadrangular pulp chambers and short roots. Ultrastructural examination showed anomalies affecting both enamel & dentin**.**

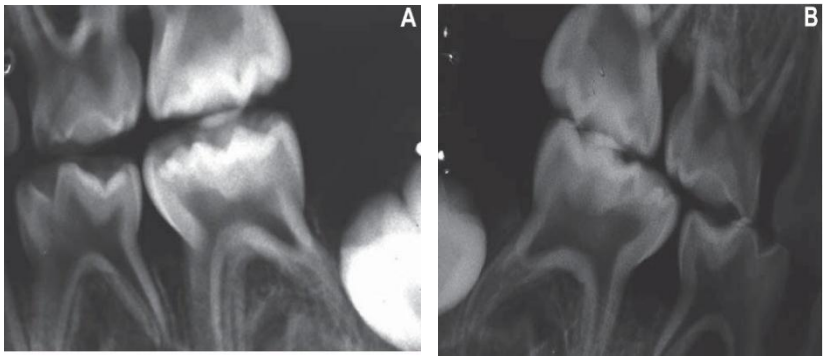


Fig. 8: Periapical radiography showed pulp chambers enlarged with pulp horns extending to the dentino-enamel junction. Adapted from (21)

(FAMILIAL ADENOMATOUS POLYPOSIS)

Gardner's syndrome, is inherited as an autosomal dominant disorder, it is characterized by intestinal polyposis, multiple osteomas, fibromas of the skin, epidermal and trichilemmal cysts, impacted permanent and supernumerary teeth and odontomas. The genetic defect is found in a small region on the long arm of chromosome 5 (5q21), where the familial adenomatous polyposis (APC) gene resides.

The majority of patients with Gardner's syndrome do not exhibit the complete spectrum of clinical disease expression.

Osteomas associated with syndrome may be found in the jaws (especially the mandibular angle) and in facial and long bones. The intestinal polyps associated with Gardner's syndrome are commonly located in the colon and rectum. Significantly, the polyps, found microscopically to be adenomas, exhibit a very high rate of malignant transformation to invasive colorectal carcinoma.

Gardner's Syndrome comprises multiple osteomas of the jaw, polyposis coli with a high malignant potential, and often other abnormalities such as dental defects and epidermal cysts or fibromas. It is inherited as an autosomal dominant trait but penetrance is weak.

Early recognition of these oral features should prompt bowel radiography or endoscopy and possibly prophylactic colectomy. Gardner's syndrome is characterised by multiple osteomas, especially of the jaws, colonic polyps and skin tumours.

The majority of patients have dental abnormalities. These include impacted teeth other than third molars, supernumerary or missing teeth and abnormal root formation. Both familial adenomatous polyposis and Gardner's syndrome are caused by mutations in the same gene, the APC tumour suppressor gene, but only some patients develop the extra-colonic manifestations such as the dental anomalies.
The colonic polyps have a 100% frequency of malignant change by middle age, and the mortality is high. The dental abnormalities can be detected in childhood or adolescence and recognition of this syndrome may be life-saving by suggesting the need for colectomy. (2,6)



Fig. 9: Showing homogenous radiopaque masses over the right and left inferior aspect of the mandible suggestive of osteomas (large arrows). Also multiple small, homogeneous radiopaque masses surrounded by a radiolucent halo could be seen throughout the maxilla and mandible suggestive of complex odontomas (small arrows).Diffuse sclerotic masses are present throughout the body of mandible giving it a mottled appearance. Adapted from (22)

FAMILIAL HYPOPHOSPHATEMIC VITAMIN D-RESISTANT RICKETS

Familial hypophosphatemic vitamin D-resistant rickets or X-linked dominant hypophosphatemia (XLH) presents with growth retardation, rachitic and osteomalacic bone disease, hypophosphatemia, renal defects in phosphate reabsorption and vit D metabolism, and dental findings.
The patients often have multiple periodontal abscesses, but no evidence of dental caries, trauma or periodontal disease on the corresponding teeth. Radiographic examination of teeth shows root dysplasia and enlarged pulp chambers. The histological examination shows marked globular dentin and increased predentin width.
Various syndromes with dentin anomalies are associated with primordial dwarfism; this term is used to describe patients with severe intrauterine and postnatal growth retardation, and they can be subclassified into three main types: Seckel syndrome, microcephalic osteodysplastic primordial dwarfism (MOPD) type I/III & MOPD type II. Variants of MOPD or Seckel-like syndrome have been described.
(4,7)



Fig. 11: **A)** Clinical aspect. **B)** Teeth 21 with chromatic alteration compatible with pulp necrosis. **C)** X-ray shows enlarged pulp chambers extending from the pulp horns. Adapted from (23)

CLEIDOCRANIAL DYSPLASIA

In this rare familial disorder there is defective formation of the clavicles, delayed closure of fontanelles and sometimes protrusion of the maxilla. Partial or complete absence of clavicles allows the patient to bring the shoulders together in front of the chest.

This disorder is one of the few recognisable causes of delayed eruption of the permanent dentition. Many permanent teeth may remain embedded in the jaw and frequently become enveloped in dentigerous cyst.

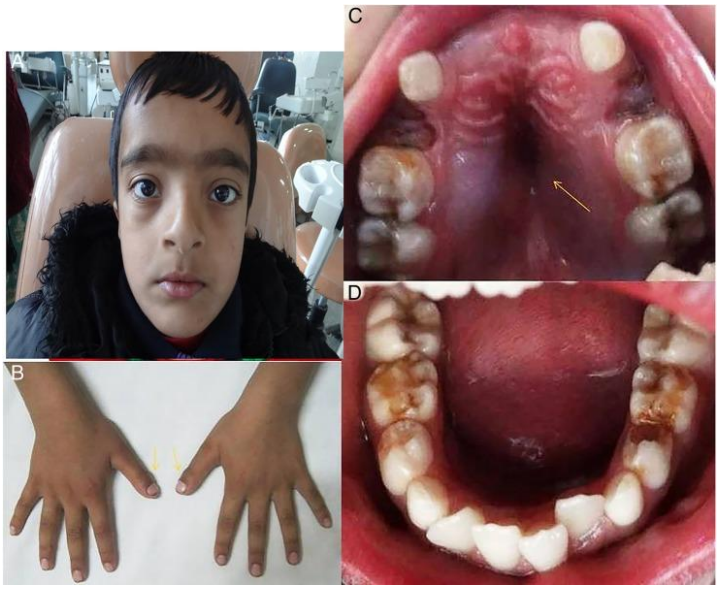


Fig. 12: (A) Broad head, frontal depression on the forehead, hypertelorism, broad and flat nasal bridge. **(B)** brachydactyly, tapering fingers and broad thumbs. **(C)** High arched palate and tooth size—arch size discrepancy. **(D)** Occlusal view of mandible. Adapted from (24)

Cleidocranial dysplasia is an AD disorder affecting bone & teeth development & is caused by mutations in RUNX2, a gene encoding an osteoblast-specific transcription factor which maps to 6p21. The symptoms include hypoplastic calvarial bones and clavicles. Dental abnormalities include supernumerary teeth (sometimes leading to a "third dentition"), delayed eruption, failure of exfoliation of the primary dentition, and malocclusion. (8,9,10)

DOWN SYNDROME

Congenital absence of teeth has been reported in 69.8% of females & 90.7% of the males with Down syndrome, in a Danish population.Lateral maxillary incisors, lower central incisors and second premolars are the most commonly missing teeth, one or both primary upper lateral incisors were missing in more than 10% of the patients, and peg-shaped maxillary lateral incisors are seen in 10%. Shapira et al. studied dental anomalies in 34 individuals with Down syndrome in Israel excluding third molars, 59% had missing teeth and 25% of the individuals had small or peg-shaped upper lateral incisors. (5)

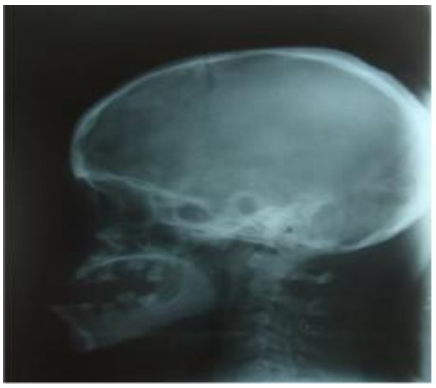


(A) **(B)** **(C)**

Fig. 14: Maxilla **(A)** & Mandible **(B)** showing grossely decayed teeth **(C)** Macrogossia



(D)



(E)

Fig. 15: (D) OPG showing missing teeth **(E)** Lateral Cephalogram showing maxillary hypoplasia. Adapted from (25)

Wolf-Hirschhorn Syndrome

The Wolfheirschhorn Syndrome (WHS) is caused by deletions of a subterminal region (WHCR) of chromosome 4p. Reported dental abnormalities are delayed eruption, fusion of incisors and oligodontia. A single case with congenital tooth agenesis has been described. Oligodontia is probably due to the loss of one copy of MSX1, which is located at 4p16. (4)



Fig. 15: Oligodontia in a patient with Wolf-Hirschhorn syndrome.

Adapted from (26)

HOLOPROSENCEPHALY

Holoprosencephaly (HPE) is a developmental field in which midline cleavage of the forebrain and craniofacial structures is impaired. HPE is etiologically heterogeneous, caused by teratogens or genetic factors. It occurs in 1/16,000 newborns, half of whom have chromosomal anomalies. Mutations and deletions in these genes are observed in roughly 20% of non-chromosomal cases.

The sonic hedgehog gene (SHH) at 7q36 (HPE3 locus) is the most frequently involved. An extreme intrafamilial phenotypic variability is observed, ranging from the classical phenotype with alobar HPE and typical severe craniofacial abnormalities to very mild clinical signs of choanal stenosis or solitary median maxillary central incisor. (5,11)



Fig. 16: Fetal face: presence of a proboscis above a single orbit with one eyeball. Adapted from (27)

KALLMANN SYNDROME

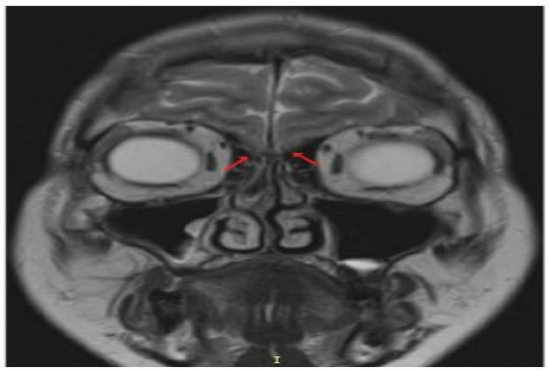
Kallmann syndrome is a developmental disease that combines hypogonadotropic hypogonadism and anosmia/hyposmia.The prevalence is estimated at 1/10,000 in males and at 1/50,000 in females, with sporadic cases more frequent than familial cases. Kallmann syndrome is genetically heterogeneous.

A rare autosomal recessive form is reported for which no causal mutation has been identified. (KAL1 and KAL2 mutations account for only 20% of Kallmann syndrome).

To date, dental agenesis is described only in KAL2 and may or may not be associated with cleft palate or lip. The type of dental anomalies varies from a single central maxillary incisor to lateral maxillary incisor agenesis, and premolar agenesis. In some cases the first four permanent molars and mandibular canine are missing. (4,7)



Fig. 17(A): Cleft Palate in Kalmann syndrome patient Adapted from (28)



(B): T2-weighted coronal MRI image showing olfactory bulbs hypoplasia. Adapted from (29)

II. Summary and Conclusion

Orofacial malformations have been recorded since time immemorial. While observational studies have assisted in the recognition of syndromes, little light has been shed on the causal mechanisms which interfere with orofacial development. The great advantage of dental analyses compared to all other analyses performed on human tissue is that deviations in the hard tissue persist and remain stable during the developmental course. As dental tissues do not reorganize, they are easy to analyse and use in analyses of fields. Therefore, a dental approach to orofacial syndromes by analysis of developmental fields contributes to an understanding of the pathogenesis of orofacial syndromes.

A problem raised by Hennekam in 2007 is which clinical condition can be called a syndrome and which cannot. There are arguments in favour for using aetiology and pathogenesis as the core issue, but Hennekam (12) also states that there are arguments to make the patient's phenotype decide the syndrome definition.

Another problem that should be solved in the orofacial analyses is the genetics behind the orofacial field. (13)

Accumulated on normal growth and development and the pathogenesis of orofacial syndromes, little is really known about the determinants of an individual's facial growth pattern.

It is essential to measure and assess many more markers of anomalous development in order to understand the complex patterns of growth that one finds in orofacial deformity.

The successful rehabilitation of the patient with abnormal development of the cranium, face and mouth is contingent on accurate diagnosis and an understanding of the aetiopathogenesis of the basic defect.

As the scientific basis of malformation is clarified it will be easier for the surgeon and the orthodontist to improve the quality and stability of treatment. Research efforts in both basic and clinical spheres will eventually provide the answer to many of the problems of abnormal development of the maxillofacial region; problems which this paper has merely served to emphasize rather than explain.

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