Oral Manifestations of Pediatric Population with Congenital Anomalies

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ABSTRACT

Congenital anomalies can be defined as structural or functional anomalies that occur during the intrauterine life and can be identified prenatally, at birth, or sometimes may not be detected later in infancy, such as hearing defects. In simple terms “congenital” refers to the existence at or before birth. According to WHO, the congenital anomalies may be the result of genetic, infectious, nutritional, or environmental factors. But it is often difficult to identify the exact cause of these abnormalities. Some of them can be prevented with vaccinations, vitamin intake and other antenatal care. However, congenital anomalies are an important cause of infant and childhood deaths, chronic illness, and disability.

Most common congenital disorders affecting children with orofacial manifestations include, Zika virus congenital syndrome, Myelomeningocele, cleft lip and palate, Soft tissue lesions of oral cavity, Amelogenesis imperfecta, Dentinogenesis imperfecta, Hemangioma, Kabuki syndrome, Ellis Van-Creveld syndrome, Congenital heart defects etc. A comprehensive study and their manifestations are illustrated below.

Keywords: Congenital heart defects, Congenital anomalies, Oral manifestations, Pediatrics, Childhood deaths

INTRODUCTION

Congenital anomalies can be defined as structural or functional anomalies that occur during the intrauterine life and can be identified prenatally, at birth, or sometimes may not be detected later in infancy, such as hearing defects. In simple terms “congenital” refers to the existence at or before birth. According to WHO, the congenital anomalies may be the result of genetic, infectious, nutritional, or environmental factors. But it is often difficult to identify the exact cause of these abnormalities. Some of them can be prevented with vaccinations, vitamin intake and other antenatal care. However, congenital anomalies are an important cause of infant and childhood deaths, chronic illness, and disability.

Zika virus congenital syndrome (ZVCS)

Zika Virus Congenital Syndrome (ZVCS) is a condition that causes congenital defects to the fetus in women infected with the Zika virus during pregnancy. Brazil is a country reporting the highest number of infections by the Zika virus [1]. Some characteristics are present, among them the occurrence of severe microcephaly, reduction in brain tissue, excess muscle tone and limited mobility, among other alterations. Since microcephaly is a recent condition, there are few studies that describe the implications for the stomatognathic system. Alterations in the oral cavity include hypotonia or labial hypertonia and alterations in the lingual mobility that can interfere in suctioning, swallowing and lip seal, contributing for children with this condition to become mouth breathers, causing difficulties when breastfeeding or bottle feeding [2].
Specifically, in relation to the eruption of deciduous teeth, it has been observed that children present numerous signs and symptoms closely related to this physiological process, such as increased salivation, occurrence of gingival itching, episodes of diarrhea, irritability, gingival pruritus, among other conditions [3,4]. Recent research with Brazilian children with microcephaly revealed that there is a slight delay in dental eruption, with the first teeth erupting at around 12.3 months of age [3].

The presence of manifestations resulting from the eruption of deciduous teeth may directly interfere in the child’s behavior and consequently in his / her adherence to the proposed therapy [4]. Therefore, the treatment of signs and symptoms resulting from the eruptive process may include the use of oral analgesics and oral teether.

Congenital Zika syndrome may present delayed chronology of eruption, ankyloglossia, ogival-shaped palate, and enamel hypoplasia, requiring dental follow-up aimed at prevention, promotion, and rehabilitation of the health of these children.

Another extremely relevant aspect to which the pediatrician should be aware is the fact that, due to the presence of multiple comorbidities, many children with microcephaly manifest, from an early age, episodes of epilepsy and frequent seizures, being therefore habitual users of continuous use drugs. The literature has shown that many pediatric drugs include sucrose in their composition [5,6]. Since dental caries is a biofilm-dependent disease and sucrose is an extremely important component for its development, it is essential that mothers are correctly oriented regarding the correct oral hygiene of their children. In addition, they should be informed about the benefits of the rational use of fluoride and the proper control of dental biofilm through regular visits to the dentist.

Myelomeningocele (MMC)

Myelomeningocele (MMC) is a congenital malformation of the neural tube that occurs in the first weeks of pregnancy. This malformation refers to the caudal non-closure of the neural tube and neural tissue exposure, which lead to neurological problems, such as hydrocephalus, motor disability, genitourinary tract and skeletal abnormalities and mental retardation.

Regarding oral health, children with Neural tube defects are at high caries risk due to a deficient diet, poor oral hygiene and prolonged use of sugar-containing oral medicines. MMC patients need assistance to do most of their everyday tasks. The physical disabilities, limitations and medical problems of these patients are so demanding that, sometimes, oral health care is excusably not regarded as a priority. Dental care of special needs patients is usually complicated due to a combination of factors that might include uncontrolled involuntary body movements, lack of motor ability, intellectual deficit and difficult to open the mouth or inability to maintain a sufficient interincisal space to permit adequate hygiene or treatment [7]. During dental treatment, additional care should be taken because MMC patients have acknowledged predisposition to latex allergy, with prevalence ranging from 28 to 67%.

In-office dental visits should start even before the eruption of the first teeth and the parents/caregivers should be well instructed and trained on at-home dental care. Toothbrushing is usually challenged by several conditions including the patients’ inability to rinse/spit and keep their mouth open, voluntary/involuntary movements and vomiting reflexes during brushing. Parents/caregivers should be enlightened and trained on the use of auxiliary resources as mouth openers and physical restraint strips, if necessary. The use of electric, adapted, or custom-made toothbrushes should also be stimulated whenever possible. In more severe cases, prescription of antimicrobial agents and scheduling of more frequent professional care sessions are essential approaches in addition to home-based oral hygiene measures [8].

Cleft lip and palate

The most severe of congenital anomalies which affect the mouth and related structures. When the tissues that form the upper lip fail to join up in the middle of the face, a gap occurs in the lip. Usually, a single gap occurs below one or other nostril is the Unilateral cleft lip. Sometimes there are two gaps in the upper lip, each below a nostril is Bilateral cleft lip. Cleft lip may be associated with syndromes like Down syndrome, Vander Woude's syndrome and Pierre Robin syndrome etc.

The incidence of cleft lip and palate varies from
eruption hematomas when the cyst fluid is mixed with blood. No treatment is needed; eruption cysts resolve spontaneously as the tooth erupts through the lesion. If the tooth does not erupt within two weeks, the child should be reexamined to evaluate other causes [11,12].

**Epstein pearls**

These are epithelial remnants entombed along the fusion line of the palatal halves. Their incidence is 7.3/1000 live born male newborn babies. They are smooth, white, keratin filled 1-4 mm papules. Epstein pearls resolve in the first 3 months hence treatment is not needed [13,14].

**Gingival/dental lamina cyst of neonates**

These are remnants of the dental lamina. Asymptomatic, multiple, 1-3 mm, nodular, creamy white lesions present bilaterally on the anterior aspect of dental ridges. Histopathology shows keratin-filled true epithelial cysts. These lesions are present at birth and are self-resolving. Therefore, do not require any intervention [15-17].

**Bohn's nodules**

Bohn’s nodules are remnants of minor salivary gland epithelium. This is asymptomatic, smooth, whitish keratin filled nodules or papules ranging from 1-3 millimeter arise on the buccal and lingual aspects of the ridge away from the midline. They resolve in the first 3 months of life and have an Incidence of 47.4% [16-18].

**Congenital epulis of the newborn**

Congenital Epulis is a rare benign tumor of uncertain histogenesis in newborn infants. It arises as a protuberant gingival mucosal mass on the anterior maxillary ridge. The etiology is unclear hence the lesion is perhaps hormone-related, degenerative, or reactive. It is single and firm with a regular surface. It may be multilobed, sessile or pedunculated, pink or red lesion does not tender to palpation. The diameter varies from a few millimeters to over 7 cm. Larger lesions may lead to mechanical obstruction in respiration and feeding. Diagnosis is confirmed by site of origin, prenatal/natal Ultrasonography (USG), Computed Tomography (CT)/Magnetic Resonance Imaging (MRI), and histopathology showing scattered odontogenic epithelium, absence of interstitial cells, angulate bodies, and vessels. Surgical excision is the treatment of choice [19].

0.5 to 3.63 per 1000 live births. The factors responsible for development of cleft lip and palate are genetic, environmental, and geno-environmental interactions. Clefts of the palate only are more common in girls while clefts of the lip, with or without palatal involvement, are more common in boys. It is interesting to know that the left side is more often than the right [9]. Children with a cleft of palate are prone to upper respiratory tract infections, and as a result there is a high incidence of middle ear problems and resultant defects in hearing.

Numerous tooth defects are commonly seen in cleft lip and palate cases. Clefts of the lip and palate give rise to problems related to actual structures involved in a cleft. In general, clefts of the lip give rise to aesthetic problems, clefts of alveolus give rise to dental problems and clefts of palate give rise to speech problems. Children with the cleft lip or palate deformity tend to present a poor gingival state, often a high caries rate and tendency to neglect the general care of their teeth. Oral manifestations that includes Congenitally missing teeth, Supernumerary teeth, Malformed teeth, Fistulas may be obturated, Ectopic eruption of primary maxillary anterior dentition. Over 90%, of cleft lip and palate children develop normal speech, a minority requiring the help of a speech therapist [10].

**Soft tissue lesions of oral cavity**

Soft tissue lesions in children may be normal/developmental findings. Children and adolescents exhibit a wide spectrum of oral lesions including hard and soft tissue lesions of the oral maxillofacial region. The prevalence of congenital pediatric oral lesions is scanty but in the United States, the prevalence rate is 4-10% excluding infants. Neonates displaying intraoral lesions need detailed assessment, diagnosis, management, and parental counseling in conjunction with reassurance. This aid timely diagnosis of both usual and rare oral tissue presentation in neonates. Some are discussed below.

**Eruption cyst/Hematoma**

Eruption cysts are dome-shaped soft tissue lesions associated with the eruption of primary or permanent teeth. They are caused by fluid accumulation within the follicular space of the erupting tooth. Eruption cysts are called
**Epidermoid and dermoid cysts**
This slow-growing, asymptomatic cyst arises in the floor of the mouth and the submental region. These soft, cystic lesions are nodular with a sessile base and lined with squamous epithelium. Clinical diagnosis is via enlargement leading to respiratory distress and feeding difficulties and tests including Magnetic Resonance Imaging (MRI)/Computed Tomography (CT), prenatal/natal Ultrasonography (USG), Fine Needle Aspiration Biopsy (FNAB), and histopathology. Surgical enucleation is the treatment of choice and recurrence is rare. The prevalence in head and neck patients is 7% and in the oral cavity is 1.6% [20,21].

**Amelogenesis imperfecta**
Amelogenesis imperfecta (AI) is a developmental disturbance that interferes with normal enamel formation in the absence of a systemic disorder. In general, it affects all or nearly all of the teeth in both the primary and permanent dentitions. The most widely accepted classification of AI includes four types: hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplastic with taurodontism.

Children with AI can exhibit accelerated tooth eruption compared to the normal population or have a late eruption. Other clinical implications of AI include low caries susceptibility, rapid attrition, excessive calculus deposition, and gingival hyperplasia. Pathologies associated with AI are enlarged follicles, impacted permanent teeth, ectopic eruption, congenitally missing teeth, crown and/or root resorption, and pulp calcification. Agenesis of second molars also has been observed. Although uncommon in AI, enamel resorption and ankylosis has been reported.

Differential diagnosis: Other forms of enamel demineralization will exhibit a pattern based upon the time of insult, thus affecting the enamel forming at the time. In contrast, AI will affect all teeth similarly and can have a familial history. Fluorosis can mimic AI, but usually, the teeth are not affected uniformly, often sparing the premolars and second permanent molars. History of fluoride intake can aid in the diagnosis [22-26].

**Dentinogénesis Imperfecta**
Dentinogenesis imperfecta (DI) is a hereditary developmental disturbance of the dentin originating during the histodifferentiation stage of tooth development. DI may be seen alone or in conjunction with the systemic hereditary disorder of the bone, osteogenesis imperfecta (OI). The diverse mutations associated with the COL1A1 and COL1A2 genes can cause the DI.

Clinical manifestation: In all three DI types, the teeth have a variable blue-gray to yellow-brown discoloration that appears opalescent due to the defective, abnormally-colored dentin shining through the translucent enamel. Due to the lack of support of the poorly mineralized dentin, enamel frequently fractures from the teeth leading to rapid wear and attrition of the teeth. The severity of discoloration and enamel fracturing in all DI types is highly variable, even within the same family. If left untreated, it is not uncommon to see the entire dentition is affected. Shields Type I occurs with osteogenesis imperfecta. All teeth in both dentitions are affected. Primary teeth are affected most severely, followed by the permanent incisors and first molars, with the second and third molars being the least altered.

Radiographically, the teeth have bulbous crowns, cervical constriction, thin roots, and early obliteration of the root canal and pulp chambers due to excessive dentin production. Periapical radiolucencies and root fractures are evident. Amber translucent tooth color is common. Shields Type II is also known as hereditary opalescent dentin. Both primary and permanent dentitions are equally affected, and the characteristics previously described for Type I are the same. Radiographically, pulp chamber obliteration can begin prior to tooth eruption. Shields Type III is rare; its predominant characteristic is bell-shaped crowns, especially in the permanent dentition. Unlike Types I and II, Type III involves teeth with a shell-like appearance and multiple pulp exposures. Shell teeth demonstrate normal-thickness enamel in association with extremely thin dentin and dramatically enlarged pulps. The thin dentin may involve the entire tooth or be isolated to the root [27,28].

**Hemangioma**
This is a benign vascular neoplasm emerging as a macule on birth but may appear a few weeks after and regresses into spotted pigments. The course of disease follows a rapid proliferating phase (0-1 yr.), an involuting phase (1-5 yr.), and...
an involuted phase (5-10 yr.). It appears on the neck and head, trunk, extremities, lips, tongue, buccal mucosa, palate, and uvula. Predisposing factors include infantile age, infant birth weight, childbearing age, gestational hypertension, Kasabach-Merritt syndrome. Diagnosis is made by history, Fine-Needle Aspiration Cytology (FNAC), MRI, and/color doppler USG, histopathology, and immunohistochemistry ruling out other vascular malformations. Stage-specific treatment drugs (α-interferon, propranolol, corticosteroids), surgery and lasers (CO2, flash lamp pulsed dye, diode) are the treatment modalities. Some cases resolve completely but some show permanent skin such as hypopigmentation, telangiectasias, anetoderma stippled scarring, and fibro-fatty residues. Incidence is 4 to 5% [29].

Kabuki syndrome
Kabuki syndrome is a rare, multiple congenital anomaly/mental retardation syndrome first described in 1981. Initially reported in Japanese children, this syndrome may occur in many ethnic groups. The most striking aspects of the disorder are the unique facial features, including long palpebral fissures, arched eyebrows, short columella, and prominent ears. Other findings include microcephaly, hypotonia, seizures, hearing loss, ptosis, strabismus, congenital heart defects, renal anomalies, growth-hormone deficiency, skeletal abnormalities, and immune deficiencies. Dental and craniofacial findings include a trapezoidal-shaped philtrum, cleft palate or cleft lip/palate, lip pits, hypodontia, microdontia, a small dental arch, maxillary recession, and midfacial hypoplasia. Mental retardation is considered a cardinal manifestation of the disorder. Approximately 300 cases have been reported in the literature. There is no standard testing for this syndrome, and the diagnosis is made from phenotypic characteristics. Clinicians should be aware of the possibility of self-injurious behavior of the patient when treating a Kabuki syndrome child [30,31].

Ellis van-creveld syndrome
Ellis-van Creveld (EVC) syndrome is an uncommon genetic disease that can be diagnosed at any age. It is also called dysplasia chondroectodermal dysplasia. It is an autosomal recessive disorder caused by a genetic anomaly located on chromosome 4p16. There is no predilection for sex; however, history of consanguineous marriage is present in 30% cases. The intelligence of affected patients is usually normal. Nearly half of these patients die during childhood as a result of cardiopulmonary malformations. For this reason, the life expectancy of patients with EVC is determined by the severity of their congenital heart disease. EVC can be diagnosed by ultrasound from the 18th week of pregnancy onward in the prenatal period and by clinical examination (by a tetrad of symptoms: chondroectodermal dysplasia, polydactyly, cardiac malformations, congenital dental hypoplasia) in the postnatal period.

The oral manifestations are diverse and affect not only the soft tissues but also the number, shape, and structure of the teeth. The most frequent manifestation is represented by the labiogingival adhesions, which result in an absence of the labial buccal vestibule. The anterior part of the lower alveolar ridge is often serrated, and the presence of multiple labial frenula is noted. The teeth tend to be small and tapered. Molars may exhibit abnormal or additional cusps and sometimes enamel hypoplasia. Congenital oligodontia in temporary and permanent teeth, the presence of supernumerary teeth, dysmorphic natal and neonatal roots, and late dental eruptions have also been reported. Dental care depends on each case and requires a multidisciplinary medical team of geneticists, speech therapists, orthopedic surgeons, cardiologists, surgical doctors, pediatricians, and dental specialists [32-35].

Congenital heart defects
Children with Congenital Heart Defects (CDH) present defects in the walls of the heart, valves of the heart and/or the blood vessels near the heart. The defects will be present at birth. Children with CDH have a higher rate of cavities due to intake of sweetened medications. Children with CDH may have Microdontia (teeth that appear abnormally small). Enamel Hypoplasia (softening of the outermost layer of teeth) may occur in children. Prevention is essential in managing the dental health of the child with CHD. Scheduling the child for early professional dental examinations is recommended [36].

Tetralogy of fallot
Tetralogy of Fallot (TOF) is the most common
cause of cyanotic heart disease. The anatomic defects comprising TOF lead to the systemic circulation of oxygen-poor (desaturated) blood, resulting in symptoms of cyanosis, polycythemia, and hypoxia. Untreated, most patients with this disorder die during childhood. Children with systemic diseases such as Tetralogy of Fallot have a higher rate of caries, poor oral hygiene, high susceptibility to other infections and bacterial endocarditis, cyanotic mucous membranes, and enamel hypoplasia. Primary prevention is critical, proper dental hygiene, regular dental check-ups and the use of antibiotic prophylaxis are particularly important, especially in high-risk patients [36].

CONCLUSION

It can be concluded that most common congenital disorders affecting children with orofacial manifestations include, Zika virus congenital syndrome, Myelomeningocele, cleft lip and palate, Soft tissue lesions of oral cavity, Amelogenesis imperfecta, Dentinogenesis imperfecta, Hemangioma, Kabuki syndrome, Ellis Van-Creveld syndrome, Congenital heart defects etc and they have different oral manifestations.

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