BIFID UVULA AND SUBMUCOUS CLEFT PALATE IN CORNELIA DE LANGE SYNDROME

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Abstract

Cornelia de Lange syndrome is a rare congenital disease characterized by growth and psychomotor retardation, peculiar facial feature as skeletal and craniofacial deformities, gastrointestinal and cardiac problems and malformation of the upper limb. The prevalence is estimated around 0.6/100000 in the population[1].

The diagnosis is based on clinical findings and the etiology is still unclear. We present a case of a 17-year-old patient, who came to our attention for dental pain. After an oral examination carried out under general anesthesia the patients presented most of the charactericts described in the literature as micrognathia , high arched palte, delayed aruption, missing of some teeth. The most peculiar findings were the bifid uvula and the submucous cleft palate. The entity of clefting can be determined only with a Magnetic Resonance Imaging which should be carried out under general anesthesia .

Caries and periodontal disease were present and the entire dental treatment has been carried out in one sitting without any anestesiologic problems[2-5].

Case report* (*J Int Dent Med Res 2011; 4: (2), pp. 74-76)

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Introduction

Cornelia De Lange Syndrome (CdLs) is an autosomal dominant disorder[6], a rare syndrome of multiple congenital anomalies and multisystemic disease, also called Brachmann-de Lange syndrome.

Brachmann had firstly described a child with similar features in 1916[7].

CdLs is classically characterized by typical features, such as microbrachycephaly, mental deficiency, abnormal speech development, seizures and hypotonia. Behavioral problem as regurgitation, projectile vomit, difficulties in chewing and swallowing are reported. The facial phenotype overrides racial characteristics.

Eyebrows are often confluent. Micrognatia is a very common feature. Delayed tooth eruption and microdontia, wide spaced teeth and cleft palate are other features reported in Literature[8].

Usually the hands and feet are small. Hirsutism is generalised, nipples and humbelicus are often hypoplastic.

Around 20% of the affected patients have a congenital heart defect[3].

Diagnosing classic cases of Cornelia de Lange syndrome is usually straightforward, however, diagnosing mild cases may be challenging, even for an experienced clinician[7].

We report a case of an affected patient in which we made an intraoperatory identification and diagnosis of bifid uvula and submucous cleft palate.

The diagnose of CdLs was made at the age of 4 years old based upon clinical findings and multi-specialistic examination.

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Case Report

We report a case of a 17-year-old girl who came to our attention , in the unit of Pediatric Dentistry and Maxillo Facial Surgery of our Institute because of dental pain.

She presented low posterior hairline, long eyelashes, thin lips, downturned angle of the mouth, small hands, feet with short digits, hirsutism, small nipples. The neck was short and thick.

The oral examination has been impossible to carry out in the dental office because of poor collaboration.

Radiographic examination, intraoral and orthopantomography could not be carry out due to lack of cooperation of the patient. The dental treatment has been carried out under general anesthesia (Figure 1).

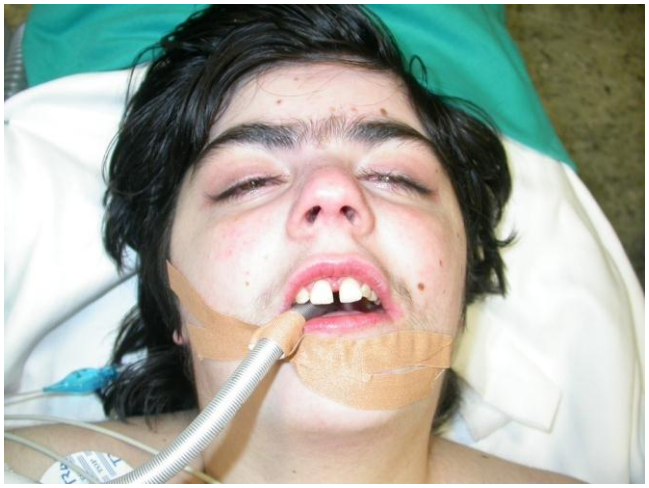

Figure 1. The patient has been intubutaded endotracheally.

After a total disinfection of the operatory field, an oral examination was made , which revealed missing of some teeth probably because of non-erupted anchilosed teeth or in ectopic position with still chance for a delayed eruption .

Old amalgam and composite restaurations were present due to a previous operation carried out under general anesthesia, but more important bifid uvula and a submucous cleft palate was identified, not so severely significant for a surgical operation and correction of the submucous clefting, along with the consideartion of the difficulty of devices which requires any kind of impressions[9].

Severe gingivitis was present, therefore an accurate ultrasound scaling ablation has been carried out, polishing of old fillings, and extraction of still present primary canines and the permanent maxillary left canine was done and detersion of the socket, presenting granular infective tissue allowing us to eradicate the cause of the referred oral pain by parents.

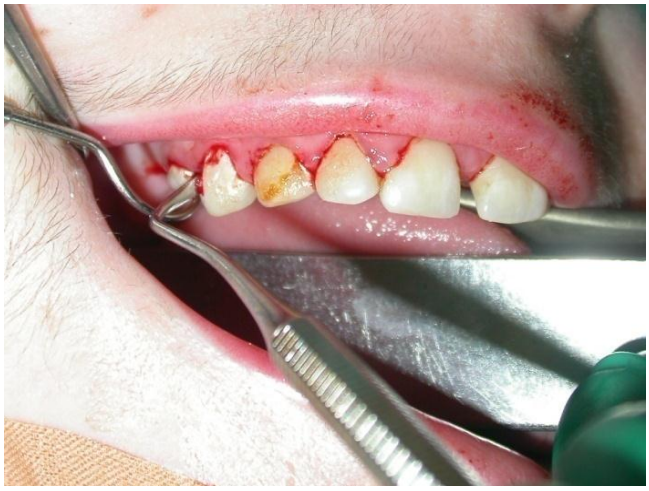

Figure 2. Dental status revaeling severe gingivitis.

The operation has been carried out in an hour and 45 minutes, and after that, the patient who was staying in the hospital under DH regime has been placed in the department of Pediatric Surgery.

Instruction for good oral hygiene was given to the parents, and post operatory control and follow up established. After 6 months we observe an improoved dental status health of the patient affected by CdLs, confirmed after 12 months consultation.

To our knowledge this is a particular and special case which allowed the dental and maxillo-facial staff to carry out a complete dental treatment leading to a clinical identification and diagnosis of bifid uvula and submucous cleft palate after an accurate inspection, noticing the difficulty of the approach which always require general anesthesia for any examiantion or clinical investigation in patients affected by CdLs.

Discussion

CdLs is a rare disease which requires a multisciplinary careful approach. Fetal Alcohol Syndrome and tetrasomy 18p[10] should be considered for differential diagnosis.

CdLs is a genetical and usually sporadic disease. In the past few years it has been shown that CdLs is caused by gene mutations affecting proteins involved in sister chromatid cohesion. Studies in model organisms, and more recently in human cells, have revealed, somewhat unexpectedly, that the developmental deficits in CdLS likely arise from changes in gene expression[11].

Aitken DA et al. reported that second-trimester maternal serum pregnancy associated plasma protein-A measurements may be of value as an adjunct to ultrasonography in the prenatal diagnosis of Cornelia de Lange syndrome[12]. As reported in the literature caries and periodontal disease are typical dental features in these patients.

Bifid uvula and not severe submucous clefting were not unusual finding still rare, along with the rarity of the disease. Cleft palate is present in 20% of the diagnosed cases[13]. This case is particular for the accuracy of the examination which led to the identification of the bifid uvula and submucous cleft palate and the complete dental treatment.

Phonation, speech , mastication might be compromised because of clefting, still, its mild manifestation do not require an immediate maxillo-facial and plastic surgeon correction, especially not prior a strumental examination as a CT (Computer Tomography) or MRI (Magnetic Resonace Imaging) to carry out absolutely under general anesthesia, which can reveal the entity of the submucous cleft until now only clinically diagnosed.

Conclusions

Thorough oral examination evaluation by dental health professionals the diagnosis of Cornelia de Lange syndrome is based on clinical findings and the etiology is still unclear.

The most peculiar findings are the bifid uvula and the submucous cleft palate. Health care workers must be able to recognize the disease and treatment way.

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Declaration of Interest

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References

1. Liu J, Baynam G. Cornelia de Lange syndrome. Adv Exp Med Biol. 2010; 685: 111-23.
2. Washington V, Kaye AD. Anesthetic management in a patient with Cornelia de Lange syndrome. Middle East J Anesthesiol. 2010; 20(6): 773-8.
3. Robert J. Gorlin, M. Michael Cohen Jr., Raoul C.M. Hennekam Syndromes of The Head and Neck 4th Edition Int J Oral Maxillofac Surg. 1993; 22(3): 171-2.
4. Takeshita T, Akita S, Kawahara M. Anesthetic management of a patient with Cornelia De Lange syndrome. Anesth Prog. 1987; 34(2): 63-5.
5. O'Donnell D, Davis PJ, King NM Management problems associated with Cornelia de Lange syndrome. Spec Care Dentist. 1985; 5(4): 160-3.
6. Liu J, Baynam G. Cornelia de Lange syndrome. Adv Exp Med Biol. 2010; 685: 111-23.
7. Brachmann W. Ein Fall von symmetrischer Monodaktylie durch Ulnadefekt, mit symmetrischer Flughautbildung in den Ellenbeugen, sowie anderen Abnormalitaten. Jahr Kinderheilkunde. 1916; 84: 225-35.
8. Toker AS, Ay S, Yeler H, Sezgin I. Dental findings in Cornelia de Lange syndrome. Yonsei Med J. 2009; 50(2): 289-92.
9. Chate RA. Respiratory arrest during an orthodontic impression of a cleft palate, in a baby with Brachmann-de Lange syndrome. J R Coll Surg Edinb. 1994; 39(2): 121-3.
10. G Borck, R Redon, D Sanlaville, M Rio, M Prieur, S Lyonnet, M Vekemans, N P Carter, A Munnich, NIPBL mutations and genetic heterogeneity in Cornelia de Lange syndrome. L Colleaux, V Cormier-Dair. J Med Genet 2004; 41: 128.
11. Dorsett D, Krantz ID. On the molecular etiology of Cornelia de Lange syndrome. Ann N Y Acad Sci. 2009; 1151: 22-37.
12. Aitken DA, Ireland M, Berry E, Crossley JA, Macri JN, Burn J, Connor JM. Second-trimester pregnancy associated plasma protein-A levels are reduced in Cornelia de Lange syndrome pregnancies. Prenat Diagn. 1999; 19: 706-10.
13. Yamamoto K, Horiuchi K, Uemura K, Shohara E, Okada Y, Sugimura M, Yoshioka A. Cornelia de Lange syndrome with cleft palate. Int J Oral Maxillofac Surg. 1987; 16(4): 484-91.