

## Clinical Manifestation of Blepharochelodontic (BCD) Syndrome: A Case Review (Manifestasi Klinikal Sindrom Belfarokeladontik (BCD): Satu Tinjauan Kes)

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### ABSTRACT

*BCD syndrome is a rare birth syndrome characterised by cleft lip and palate, euryblepharon with/without limb anomalies, ectropion of lower eyelids and hypertelorism. The dental anomalies include cone shaped microdontic teeth and oligodontia. Different combinations of these signs have been found sporadically in patients. This syndrome is also known as Elschnig's syndrome. By at large, patients with this syndrome have no potentially fatal anomalies. Their growth and development were usually normal. This case review described the clinical manifestation and oral findings of a patient with BCD syndrome. The dental need of the patient was outlined.*

*Keywords: Ectropion; Elsching syndrome; euryblepharon; lagophthalmia; oligodontia*

### ABSTRAK

*Sindrom BCD adalah satu sindrom kelahiran yang jarang dijumpai dan mempunyai ciri-ciri seperti rekahan bibir dan lelangit, 'euryblepharon' dengan/tanpa anomali pada lengan tangan dan kaki, ektropion kelopak mata bawah dan hipertelorisme. Anomali pergigian termasuk kehadiran gigi-gigi mikrodontik yang berbentuk kon dan oligodontia. Akan tetapi, kombinasi berlainan daripada ciri-ciri yang dinyatakan boleh juga dijumpai pada segelintir pesakit secara sporadik. Sindrom ini juga dikenali sebagai sindrom Elschnig. Kebiasaannya, pesakit sindrom ini jarang memiliki anomali serius yang boleh menyebabkan kematian. Tumbesaran dan perkembangan mereka juga adalah normal. Tinjauan ini melaporkan manifestasi klinikal serta penemuan oral pada seorang pesakit sindrom BCD. Keperluan pergigian untuk pesakit ini juga dibincangkan.*

*Kata kunci: Ectropion; euryblepharon; lagophthalmia; oligodontia; sindrom Elschnig*

### INTRODUCTION

Blepharochelodontic (BCD) syndrome is a rare birth syndrome characterized by cleft lip and palate (most often bilateral), cone shaped microdontic teeth, hypodontia or oligodontia, ectropion of the lower eyelid, lagophthalmia, hypertelorism, distichiasis and euryblepharon. Limb anomalies such as clinidactyly and syndactyly may also be seen. In 1912, Elschnig described complications with cleft lip and palate, ectropion of lower eyelid and euryblepharon but it took some time for this to be recognized as a syndrome. Gorlin et al. (1996) identified this syndrome as an autosomal dominant disorder. Eyelid anomalies distinguishes blepharochelodontic syndrome from other syndromes that also manifests with features like cleft lip and palate with oligodontia or microdontia. The clinical manifestation and oral findings of a patient with BCD syndrome will be discussed in this review. The dental needs of this patient are also outlined.

### CASE REVIEW

A 5 year old girl with multiple missing teeth and diagnosed with BCD syndrome was referred to us by the Paediatric Department of the National University of Malaysia Medical Centre (UKMMC). Her maternal grandmother complained of

dissatisfaction regarding her dental appearance. The patient was in a depressed state and was very shy of attending school due to her facial and dental appearance. The patient also had a very low self-esteem. She is of Chinese descent whose parents are not related. Her mother had an uneventful pregnancy and delivery. Her other siblings and family members are normal. There is no history of facial/dental anomalies/syndrome running in the family. She was born with bilateral cleft lip and palate. Extraorally, the patient presented with bilateral eye anomalies that include ectropion of the lower eyelids, lagophthalmos, hypertelorism, distichiasis and euryblepharon (Figure 1). Her scalp hair was sparse and straight. Neither syndactyly nor hypoplastic nail was detected. Repaired bilateral cleft lip was observed. She was in a Class III skeletal relationship with a hypoplastic maxilla. Intraorally, there is a fistula associated with the primary palate due to eventful healing of the surgical repair (Figure 2). Also, the patient presented with microstomia and has oligodontia and microdontia with conical shaped teeth (Figures 3 and 4). Radiographically, an orthopantomogram of this patient revealed many missing permanent tooth buds with a reduced alveolar bone level and a hypoplastic maxilla (Figure 5). The initial management involved preventive measures including fluoride application and oral hygiene instructions. We



FIGURE 1. Appearance of the patient



FIGURE 4. Intraoral view of the lower arch



FIGURE 2. Intraoral view of the palate showing the fistula



FIGURE 5. Orthopantomogram view of the patient



FIGURE 3. Intraoral view of the upper arch

decided to construct a pair of partial dentures for this patient since this was her main concern as well as in the prospect of improving her aesthetics, speech and function so that she could build up her confidence level. Our patient was extremely pleased with her new appearance since the new set of partial dentures had boosted her self confidence apart from improving her aesthetics, speech and function (Figure 6). Long term management for this patient would probably demand for bone grafting and implants.

#### DISCUSSION

As of 2006, only 32 cases of BCD syndrome have been reported worldwide (Iida et al. 2006). It is also known as clefting-ectropion-conical teeth disease since it describes the main clinical features of this disease. Growth and development are normal in all the patients that have been studied.

From the literature review, it was found that there was no gender predilection in terms of this syndrome (Iida et al. 2006). However, there was a pattern of familial cases, suggesting an autosomal dominant inheritance (Allanson & McGillivray 1985). Lopes et al. (2003) had proposed the possibility of involvement of p63 and Interferon



FIGURE 6. Completed partial dentures

Regulatory Factor 6 (IRF6) genes in this condition. It has also been shown that patients with this syndrome do not have any fatal anomalies. Our patient exhibited normal growth and development for her age. There is no history of the syndrome inheritance in her family. The possible involvement of a particular gene cannot be ruled out. However, there was no evidence of this reporting in her medical records. Probably the occurrence of this case may be of sporadic in nature.

Basically, patients with this syndrome will manifest mainly eye anomalies, dental anomalies and limb anomalies. Eye anomalies include euryblepharon, ectropion, distichiasis, hypertelorism and lagophthalmia. Dental anomalies are cleft lip and palate (most often bilateral), conical teeth, hypodontia or oligodontia and microdontia. Oligodontia has been found in three quarter of the cases reported whereas conical teeth have been noted in less than half of these patients. Iida et al. (2006) reviewed detailed reports about oligodontia among BCD patients in the literature and found that it varied from a mild form to anodontia. The sites of missing teeth in mild oligodontia cases were usually found adjacent to the cleft of the alveolus (Guion-Almeida et al. 1998). Iida et al. (1996) also noted that 2 cases had severe oligodontia, and all teeth except permanent molars were symmetrically missing (Guion Almeida et al. 1998; Martinez et al. 1987). Limb anomalies including clinidactyly and syndactyly are not always seen unlike eye anomalies and dental anomalies which is a must to diagnose this condition. Other sporadic symptoms that have been reported in patients with this disorder are hypothyroidism and imperforate anus (Lopes et al. 2003). Studies have also shown that patients with this syndrome may present with sparse hair (Iida et al. 2006). Our patient manifested both oligodontia and conical teeth. Five teeth were noted in each arch by clinical and radiographic assessments. She has bilateral cleft lip and palate which concurs with majority of the cases reported in the literature. Her hair was also sparse and straight. However, limb anomalies were not found in this patient. Another feature found in

our patient was microstomia with thinning of both lips. Whether this was a true presentation of this syndrome or an effect due to the surgical repair of cleft lip was difficult to ascertain.

Besides the clinical presentation, our patient presented with a complex aesthetic problem that needed an urgent attention. Since patient is a school going child, she is quite aware of her aesthetically unpleasing appearance mainly due to her eye anomalies, hypoplastic maxilla and multiple missing teeth. As a result, she was teased and seen in a less than friendly view by her friends at school. She has started to compare her appearance with others around her. This has led her to being depressed and often shies away from school. Her low self-esteem furthermore affected her studies as she is unable to concentrate completely at school apart from being absent from school. Not only that, but patient has problem chewing food and is unable to eat varieties of food due to the multiple missing teeth. Most of the time, she is on a soft diet. In terms of speech, she has problem pronouncing certain alphabets. It was therefore decided to construct dentures for this patient in view of her psychological and functional status. Denture construction will be an intermediate treatment for this patient since bone grafting and implant placement may be carried out as a long term management in the future. During construction of the denture for this patient, we had to perform a few careful manipulations such as using the fluoride tray during the primary impression taking and also constructing the lower denture first in order to obtain the bite registration for the construction of the upper denture. This was done due to the inability of the patient to open the mouth widely because of microstomia.

#### CONCLUSION

As in many syndromic cases, the orodental problems were often overshadowed by the major syndromic manifestations. As dentists, we shouldn't be alarmed with the mere appearance of these patients and shy away from them. We must be able to appreciate their special dental needs and provide the appropriate remedy to their problem. In the current case, we not only improved her appearance, but also psychologically had made her more confident to live life. However, she will still need to continue monitoring and review appointments into adulthood.

#### ACKNOWLEDGEMENT

The authors would like to thank the patient and her guardian for their cooperation in allowing us to use the clinical pictures for this review.

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Received: 19 December 2011

Accepted: 18 May 2012