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CLEIDOCRANIAL DYSPLASIA : A PRELIMINARY REPORT

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A 9-year old boy suffering from cleidocranial dysplasia associated with impacted 4 supernumerary teeth and unerupted all permanent teeth is presented with his mother. The pedigree showed autosomal dominant pattern of inheritance, and the raiographic features of them were very similar in clavicle, skull, vertebrae, peivis and extremities. Although almost of the skeleton was involved with this syndrome, they did not recognize any other problem but except dental problem.

In mother, who was wearing removable partial dentures leaving 24 impacted teeth in her jaws, the radiographic abnormalities like cystic lesion were not detected. And in the son, who showed impacted 4 supernumerary and all permanent teeth, we have attempted surgical extraction of the supernumerary teeth and periodic surgical opening of the alveolar bone covering the permanent dentition to induce the eruption of permanent teeth at the proper position. Orthodontic treatment has also been combined to correct class III *malocclusion state.*

I. INTRODUCTION

Cleidocranial dysplasia is a syndrome of unknown etiology affecting the entire skeleton, which is often but not always hereditary. Both autosomal dominant and recessive patterns of inheritance have been reported by Gorlin et al. (1976)[1].

It is characterized by several skeletal abnormalities, the most frequent being partial or complete loss of clavicles, along with patent fontanelles and wormian bones in unclosed suture lines of the skull.

Oral manifestations include an underdeveloped maxilla(pseudoprognathism) with high and narrow arched palate, prolonged retention of primary teeth, and many impacted supernumerary teeth.

Although a number of surgical and prosthetic techniques have been suggested to deal with these oral deformities, the most promising treatment involves the surgical uncovering of unerupted permanent teeth with or without orthodontic intervention to induce eruption[2].

In this paper, we reviewed a 9 year old boy patient with cleidocranial dysplasia with his mother having shown very similar characteristics in clinical and radiographic examinations.

II. CASE REPORT

A 9 year old boy visited the Dept. of Dentistry, Inha General Hospital for evaluation of delayed eruption of permanent teeth with his mother on March, 1990.

According to his mother, he is the only child and born by cesarean section due to pelvic problem of her. He had pneumonia before age 1, and since then, he had had frequent upper respiratory infection until age 5. But his developmental milestones and intellectual status seemed to be normal.

Their family history disclosed that several members of the family had had same appearance as seen in the pedigree(Fig. 1), but she had not recognized any specific medical problems in them.

He demonstrated drooping shoulders and pronounced frontal head with depressed midportion. But he showed relatively straight profile when compared to his mother whose profile was concave. Their shoulders were hypermobile and both of them were able to appropriate their shoulders anteriorly(Fig. 2-7).

Oral manifestation of the son revealed prolonged retention of primary teeth except $\frac{D}{DC}\Big|\frac{}{D}$ with dental caries, delayed eruption of lst permanent molars, deep and high palatal vault, and class III malocclusion pattern with cross-bite on molar area.

Radiographically, unerupted all permanent teeth and impacted 4 supernumerary teeth on lower anterior portion were observed in his panoramic radio-

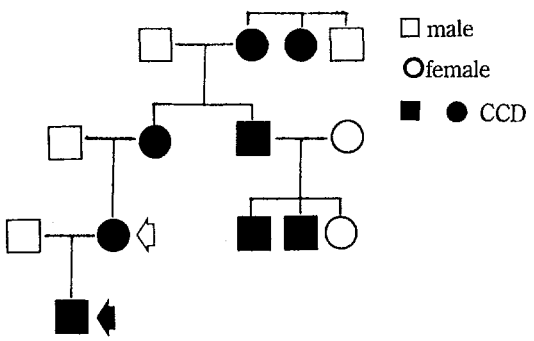

Fig. 1. Pedigree of the patient
(black arrow : son, white arrow : mother)

Fig. 2

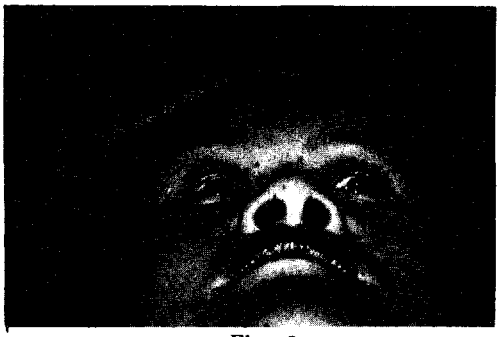

Fig. 3

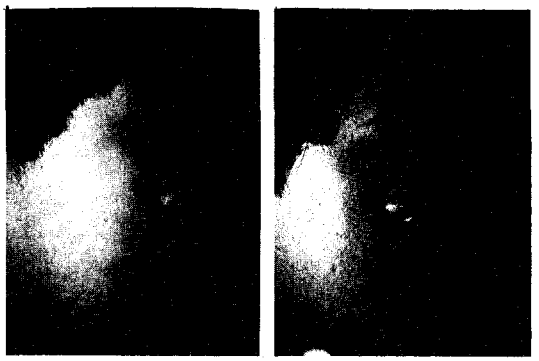

Fig. 4 & 5

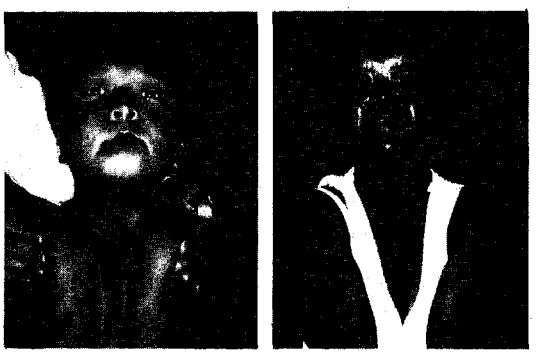

Fig. 6 & 7

graph(Fig. 8). And his mother's panoramic view revealed 24 impacted teeth(Fig. 9). They all showed round and obtuse mandibular angle due to absence of ante-gonial notch. The radiographs of chest revealed bony defects and pseudarthrosis at the lateral third of right clavicle in son and at the middle third of right clavicle in mother, and the short left clavicles were also noted(Fig. 10 & 11). In the skull series, the son and mother showed marked widening of metopic suture with multiple wormian bones along

Fig. 8. Panoramic View of Son

Fig. 9. Panoramic View of Mother

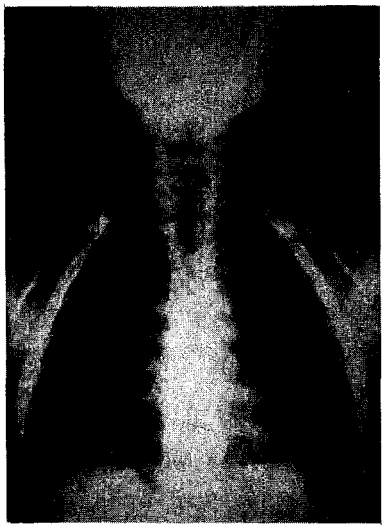

Fig. 10. Chest P-A View of Son
(white arrow : pseudarthrosis of Rt. clavicle
black arrow : neural arch defects)

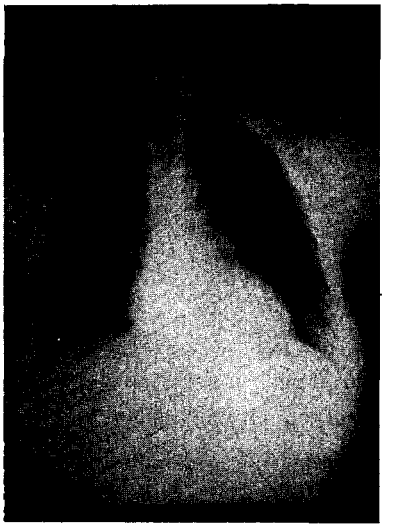

Fig. 11. Chest P-A View of Mother
(white arrow : pseudarthrosis of Rt. clavicle
black arrow : short Lt. clavicle)

Fig. 12. Skull Towne's View of Son
(large white arrow : widely open post. fontanelle
small white arrow : wormian bones
black arrow : widely open ant. fontanelle)

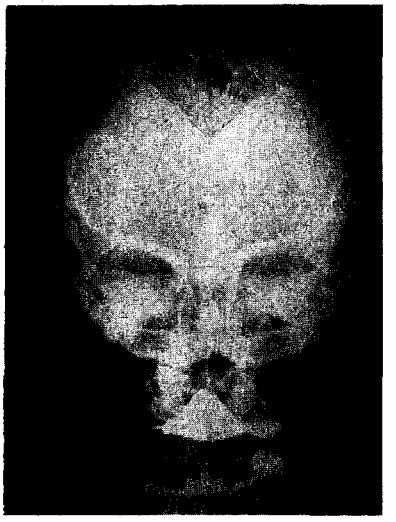

Fig. 13. Skull P-A View of Mother
(arrow : widening of sagittal suture)

Fig. 14. Skull Lateral View of Son
(arrow : wormian bones along the lambdoidal suture)

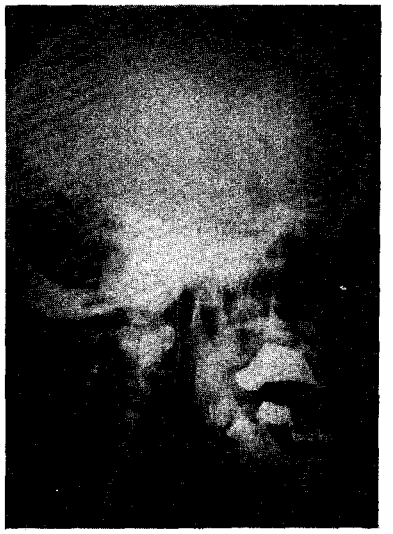

Fig. 15. Skull Lateral View of Mother
(arrow : same as Fig. 14.)

the sagittal and lambdoidal sutures, and especiallly in the son, the anterior and posterior fontanelles were open widely. Frontal, parietal and occipital bossing was also found in the radiograph of mother with maxillary underdevelopment(Fig. 12-15). Other radiographic abnormalities of the boy included incomplete closure of neural arches of cervical and thoracic spines and irregular metaphyses and cone shaped epiphyses of both 2nd and 5th middle phalanges and metacarpal bones, in addition to large cone shaped epiphyses of proximal phalanges in both feet. And in his mother, incomplete closure of neural arch of cervical spine, posterior wedging deformities of number 8 and 9 thoracic vertebrae and shortening of 4th metatarsal bone of right fooot were demonstrated.

III. DISCUSSION

Cleidocranial dysplasia was first reported by Martin in 1765, and Marie and Sainton recorded the hereditary clavicular, cranial, and dental abnormalities that led to the term cleidocranial dysplasia in their subsequent reports in 1898[3]. Since then over 700 cases of this syndrome have been described in the liter ature.

According to Gorlin et al. (1976), this syndrome is inherited by both autosomal dominant and recessive patterns[1,4] with the sporadic cases being due to the frequent occurrence of dominant mutation[5].

In our case, as the trait appears in every generation without skipping, and the transmission of the trait are not influenced by sex[6], it seems to be an autosomal dominant type of inheritance.

In cleidocranial dysplasia, the abnormalities occur during the 6th to 8th week of gestation, when ossification of the clavicles and cranial vault begin. Later, the lateral arches of the spine, the pelvis and the bones of the hand and feet fail to ossify, and involvement of these structures suggests a genetic mesodermal anomaly[5].

As the clavicles are frequently partially or completely absent, with complete absence occurring in 10% of cases, the characteristic appearance of drooping shoulders, an elongated neck, and the ability to approximate the shoulders anteriorly is represented by this clavicular abnormality and associated soft tissue loss[7,8].

In our case, the mother and son all showed excessive adduction due to the bony defects and pseudoarthrosis in right clavicles which found typically in this syndrome. And especially in the son, the left clavicle

was shortened.

The radiographic abnormalities are very numerous, but the most notable and characteristic abnormalities are found in the skull with pronounced frontal, parietal and occipital bossing. Broad cranial sutures and large and persistent fontanelles with multiple wormian bones are common. Hypertelorism and underdeveloped paranasal sinuses also are frequently observed[8].

The skull series of our cases revealed multiple wormian bones along the sagittal and lambdoidal suture lines, and especially in the son, marked widening of metopic suture and anterior and posterior fontanelles were noticed. The skull radiograph of mother showed focal thickening of occipital bone.

Other skeletal abnormalities include vertebral and pelvic dysplasia in addition to the abnormality of bone in extremities. The vertebral abnormalities include scoliosis, kyphosis, lordosis, vertebral synostosis, delayed mineralization of the neural arches, and posterior wedging of thoracic vertebral bodies. In the pelvis, underdevelopment and narrowing of pelvis and delayed fusion of pubic symphysis are usually noted. And in the extremities, pseudoepiphyses in the small tubular bones of the hand and foot, coxa valga, coxa vara and small scapulae are common[9, 10, 11, 12].

In 1979, Short reported a case of vascular complication in subclavian artery, and according to him, Lasker(1946), in a carefully documented survey of over 500 cases collected from the world literature, found 2 examples of cases in which signficant neurologic or vascular symptoms had been present[13]. And in 1987, Dore and his associates reported syringomyelia in cleidocranial dysplasia patient and stressed that in a patient with any neurologic symptoms, progressive scoliosis after skeletal maturation, the presence of syringomyelia should be considered and early diagnosis and operative therapy were important to prevent permanent neurologic problems[7]. Another medical problem may include deafness secondary to malformation of the cartilaginous structures and membranous bones in the middle ear[14].

In our cases, neither of them, the son and mother, showed distinct abnormalities in general appearance of vertebrae and extremities. But, in reviewing of the radiographs of the body, the son revealed neural arch defects in vertebrae, widely open symphysis pubis and abnormalities of both hands and feet in comparison with his mother who showed neural arch defects in cervical spine, wedging deformities of number 8 and 9 thoracic vertebrae, narrowed pelvis, and minor deformities of both feet.

Cleidocranial dysplasia is originally believed to involve only bones of membranous origin. The mandible is an intramembranously derived bone for most of its bulk, at least during the development of the tooth germs. The appearance of the secondary cartilage of the condylar and coronoid process is a later development. In skulll of the children, endochondral growth is mainly found at the mandibular condyle. On the other hand, the maxilla including calvarium develops intramembranously. This difference of bone development between both jaws makes the premaxilla to be hypoplastic, yet the growth of mandible is usually normal[15].

In the cephalometric analysis, patients with cleidocranial dysplasia have a large brachycephalic skull. And disharmony between the jaws due to retursion of the middle face and mandibular prognathism is common. However, Järvinen(1981), who reported cephalometric findings in three cases of cleidocranial dysplasia, found that all the cases were characterized by real maxillary prominence in addition to mandibular prognathism[16]. And about the abnormalities of the cranial base in this syndrome, Kreiborg(1981) reported that all of the 17 patients presented significant short anterior and posterior cranial bases[17].

The results of cephalometric analysis of our patients were listed in table I in comparison to normal ranges. Both of them revealed prominent maxillas, prognathic mandibles, and short cranial base length. And they are in consistency with the results of Järvines and Kreiborg. In relation to the skeletal development of the face, it may be considered that the present patients have atypical rather than typical cases

Table 1

patient(age)	SNA	SNB	S-N length	S-Ar length
son(9)	87.5	87.5	62.0	36.0
normal	81±3	78±3	68±3	34±3
mother(40)	91.0	93..0	59.0	29.0
normal	81±3	79±3	69±3	37±3

* reference : skeletal analysis used in the Dept. of Orthodontics, College of Dentistry, Yonseil University

(S ; sella, N ; nasion, A ; A point, B ; B point, Ar ; articulare)

of cleidocranial dysplasia.

Dental problems associated with cleidocranial dysplasia include delayed loss of deciduous teeth, failure of permanent dentition to erupt, and presence of unerupted supernumerary teeth[18]. The roots of the permanent teeth tend to be short, thin and deformed with absence or paucity of cellular cementum on their root surfaces[19,20].

The cause of unerupted teeth is said to be a disturbance of bone resorption, a lack of cellular cementum, or a lack of union between the dental follicle and the mucosa due to interposed fibrous tissue acting as barriers to eruption. In addition, Yamamoto and his associates(1989) reported a case with more than 60 unerupted teeth, and considered that the mechanical obstruction by these unerupted teeth was the chief factor contributing to failure of eruption[15]. About the timing of development of supernumerary teeth, Frame et al. (1989) reported progressive development of 9 more supernumerary teeth in a 9 year old boy suffering from cleidocranial dysplasia during a 4 year observation and treatment period, and concluded that development of more teeth would be possible in the future[21].

To deal with these oral deformities, several techniques have been suggested by many authors. Some have recommended the removal of some or all erupted or unerupted teeth, followed by construction of complete or overlay dentures. But this therapy may result in extensive alveolar bone loss. Construction of dentures over the alveolar ridge remaining the unerupted teeth has also been refuted on the basis of possible cyst formation and denture discomfort. According to Ohman and Ohman(1980), the body attempts to heal a surgically created cystic cavity by filling it can induce teeth eruption[22]. On the basis of this hypothesis, early surgical management with serial uncovering of the permanent teeth has been attempted, and as the teeth erupt, good alveolar bone formation and the development of straight root is anticipated[2,18,19,21]. Sometimes an orthodontic intervention in combination with this surgical management may be necessary.

The mother wearing partial dentures of both jaws had 24 unerupted teeth, but we could not identify what is the normal tooth that should have been erupted. Althaugh she had multiple impacted teeth in both jaws, cystic lesion was not revealed in the panoramic radiograph. And she did not recognized any discomfort in relation to her old partial dentures. So we did not attempt to induce the eruption of the impacted teeth. The treatment plan of the son was listed in the summary.

Ⅳ. SUMMARY

A 9 year old boy suffering from cleidocranial dysplasia with multiple unerupted permanent and impacted supernumerary teeth is presented with his mother. They showed very similar radiographic features of cleidocranial dysplasia, and the child seemed to be involved more skeleton than mother. But, besides of the dental problems of delayed dentition, they did not recognize any specific medical problem.

To induce the eruption of permanent teeth at the proper position, we have attempted surgical intervention for removal of 4 supernumerary teeth and the alveolar bone covering the permanent dentition which acts as a mechanical barrier of preventing eruption of permanent teeth. And in the maxilla with high and narrow palatal vault, we have tried to expand the narrow palate with removable orthodontic appliance. Periodic recall check up with radiograph and surgical and orthodontic treatment will be followed to improve the malocclusion.

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국문초록

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본 증례는 다발성의 영구치 맹출 지연을 주소로 내원한 9세 남아와 그의 어머니에서, 상염색체에 의해 우성으로 유전된 쇄골 두개골 이골증의 증례로, 모자는 모두 쇄골, 두개골, 척추, 골반 및 사지 등에서 서로 유사한 방사선학적 소견을 나타내었으나 특별한 의학적인 문제점은 보이지 않았으며, 치과적인 관점에서, 어머니는 24개의 매복치를 보존한 상태로 가철성 보철물을 장착하고 있었지만 방사선학적 검사상 낭종등의 이상 소견은 관찰되지 않았으나, 아들에서는 4개의 과잉치와 모든 영구치들이 매복되어있었던 바, 저자등은 과잉치의 발거 및 주기적인 외과적 개방술로 영구치의 맹출을 유도함과 아울러 3급 부정교합 상태의 개선을 위해 교정치료를 병행 중에 있다.