Case Report

Management of Velopharyngeal Insufficiency in Velocardiofacial Syndrome†

Sriwimon Manochiopinig, Ph.D.*
Apirag Chuangsuwanich, M.D.**
Apisit Veeravuthipakorn, M.D.**

Abstract : **This report presents an analysis of the treatment of a patient with velocardiofacial syndrome. The patient had a cleft palate and an articulation disorder with hypernasal speech. The cleft palate repair was successful but the post-operative course was complicated by velopharyngeal insufficiency. Thus, further management including reassessment and treatment for better speech quality was required.**

Key words : velopharyngeal insufficiency, velocardiofacial syndrome

เรื่องย่อ : **การดูแลรักษาปัญหา Velopharyngeal Insufficiency ในผู้ป่วยกลุ่มอาการ Velocardiofacial Syndrome**

ศรีวิมล มโนเชี่ยวพินิจ Ph.D.*, อภิรักษ์ ช่วงสุวนิช พ.บ., อภิสิทธิ์ วีระวุฒิปกรณ์ พ.บ.****

†*นำเสนอในที่ประชุมวิชาการประจำปี ครั้งที่ 43 คณะแพทยศาสตร์ศิริราชพยาบาล, 3-7 มีนาคม 2546, คณะแพทยศาสตร์ศิริราชพยาบาล, กรุงเทพมหานคร 10700.*

**ภาควิชาเวชศาสตร์ฟื้นฟู, **ภาควิชาศัลยศาสตร์, คณะแพทยศาสตร์ศิริราชพยาบาล, มหาวิทยาลัยมหิดล, กรุงเทพมหานคร 10700.*

สารศิริราช 2546; 55: 25-30.

รายงานนี้นำเสนอการวิเคราะห์การดูแลรักษาผู้ป่วยกลุ่มอาการ velocardiofacial syndrome ที่มีปัญหาเพดานโหว่ พูดไม่ชัดและมีเสียงขึ้นจมูก การผ่าตัดแก้ไขเพดานโหว่ได้ผลดี แต่ผู้ป่วยยังคงมีปัญหาเสียงพูดผิดปกติที่แสดงว่าการทำงานของกล้ามเนื้อบริเวณช่องคอและเพดานอ่อนทำงานได้ไม่ดี จึงได้วิเคราะห์หาสาเหตุและประเมินปัญหาเพิ่มเติม และวางแผนการดูแลรักษาใหม่ให้ได้ผลการรักษาที่ดีที่สุดเพื่อช่วยให้ผู้ป่วยมีการพูดที่ดี

INTRODUCTION

Velocardiofacial syndrome (VCFS) was recognized in 1978 by Dr. RJ Shprintzen, Center for Craniofacial Disorders, Montefilore Medical Center, Bronx, New York. It is a hereditary autosomal dominant disorder characterized by cleft palate, heart defects and facial features as described by Shprintzen based on the observation of 12 cases. This syndrome is named after the physician as Shprintzen syndrome. It is also known as DiGeorge syndrome, craniofacial

†*Presented in the 43rd Siriraj Scientific Congress, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok 10700.*

**Department of Rehabilitation Medicine, **Department of Surgery, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok 10700.*

syndrome or conotruncal anomaly unusual face syndrome.

This genetic disorder is a congenital anomaly of unknown cause. It is due to a deletion in the long arm of chromosome 22q11 and 30 different problems can be found[1]. The physical manifestations of velocardiofacial syndrome include cleft plate, heart defects, characteristic facial appearance, learning problems, speech and feeding problems[2,3].

Cleft palate is the 4th most common birth defect with an incidence of 1/700 live births[4], meanwhile VCFS is found 5-8% of children born with a cleft palate[4]. It is estimated to occur over 130,000 individuals in the USA. Although cleft palate as part of the velocardiofacial syndrome is well recognized and identified clinically, it is rarely reported. In addition, there is no formal report regarding the VCFS population with cleft palate in Thailand. This report describes a case of velocardiofacial syndrome with nasal speech and the management for his velopharyngeal insufficiency.

CASE REPORT

A 21-year-old male patient with a cleft palate consulted a plastic surgeon for appropriate management. The patient presented with an elongated face, almond-shaped eyes (Figure 1), cyanosis and tapered fingers (Figure 2). Physical manifestation, examination/investigations, findings and manage-ment are shown in table 1 and 2. The patient had received primary care with conservative and/or intervention of the multiple defects and problems.

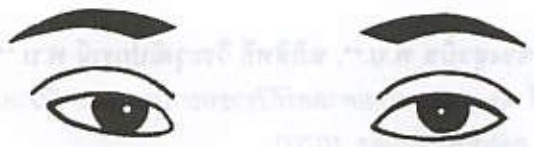


Figure 1. The almond-shaped eyes of the patient with velocardiofacial syndrome.

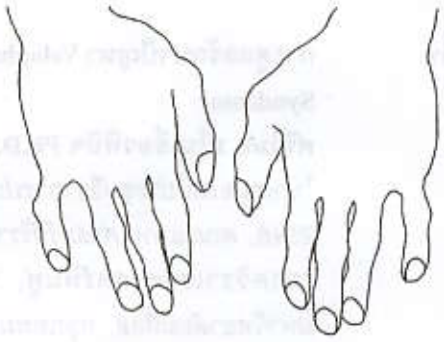


Figure 2. The tapered fingers of the patient with velocardiofacial syndrome.

Table 1. Physical manifestations of the patient.

Organ	Characteristic
Eye	1. Upward slant palpable fissure 2. Narrow palpable fissure 3. Epicanthic fold 4. Telecanthus 5. Almond shaped eyes
Ear	Abnormal ear shape
Nose	Bulbous nose
Face	Elongated face
Mouth	Incomplete cleft palate, malocclusion
Hand	Clubbed, cyanosed and tapered fingers
Motor	General weakness

Table 2. Investigations/examinations, findings and management of the defects/problems.

Investigation / Examination	Finding	Management
Echocardiography and Cardiac catheterization	Heart defects: 1. Tetralogy of Fallot 2. Patent foramen ovale 3. Right-sided aortic arch	1. Right modified B-T shunt 2. Medication (ASA) 3. Plan for total correction
Serum parathyroid hormone, T cell function study	Hypoparathyroidism	Medication (thyroid hormone)
Blood for DNA and chromosome study and FISH test	Microdeletion of 22q11-13	Genetic counseling
Eye examination	Eye defects: 1. Epicanthus with mild degree of vitreous degeneration 2. Refrection error	Observation (no medical treatment)
Hearing assessment (audiometry)	1. Otitis media 2. Conductive hearing loss (Rt = 70 dB, Lt = 45 dB)	1. Myringotomy with tympanostomy tube insertion 2. Medication (antihistamine and antibiotic)
Nose examination	Chronic rhinitis	Medication (antihistamine and antibiotic)
Orofacial examination	Incomplete cleft palate	Repaired cleft palate : 2-flap palatoplasty with intravelar veloplasty
Intellectual assessment (IQ Test)	IQ = 51 (revised WISC) (Thai version)	Counseling and informal education
Language assessment (informal functional test)	1. Poor speech intelligibility (50% understandable for unfamiliar conversational partner) 2. Borderline listening ability (understand most daily conversation) 3. Limited reading and writing abilities (common words, mostly at imitation level) due to mental deficiency, no formal education	Language intervention by therapist and informal education by mother
Speech assessment (the Thai articulation test)	Articulation defect: substitution, omission, distortion.	Intensive speech correction: once weekly, home program, educated and train mother to carry out home pro gram
Resonation test (nasometer)	Abnormal nasality speech: 1. hyponasality with nasalance scores of the nasal sound = 54.47% (normal = 63.29%) 2. hypernasality with nasalance scores of the oral sound = 49.89% (normal = 13.63%) 3. hypernasality with nasalance of the mixed sound = 90.92% (normal = 39.43%)	Intensive speech training for 6 months. (improving velopharyngeal function)

Table 2. (Cont.)

Investigation / Examination	Finding	Management
Velopharyngeal closure evaluation (nasoendoscopy)	Velopharyngeal insufficiency (incomplete and inconsistent clusure during speech; complete closure during swallowing	Intensive training of velopharyngeal function for speaking skill for 6 months.

Routine follow up was conducted after the first week and the first month post-operation. It was found that the patient had good wound healing and recovery without any complication after palatoplasty and intravelar veloplasty. During the first two visits the flap looked good. When seen in the out-patient clinic (surgery section) the patient was making a good recovery, and the wound was fully healed by the second follow up session. Velopharyngeal closure following the operation was adequate for deglutition, but not for speech. The patient had been enrolled in a speech-language program once a week for one month pre- and post-operation. Speech disorders were still present with both moderate-severe articulation defects and resonation problems after palatoplasty. It was felt that the potential for functional recovery of velopharyngeal incompetence was so poor that further management and treatment were indicated. Reassessment was conducted in an attempt to improve his verbal communication.

Therefore, reassessment at 3 months post-operation by various investigation/tests was performed. These included perceptual assessment using an articulation test, resonation test by nasometer and direct functional velopharyngeal closure evaluation by nasoendoscopy. Test results were assessed. Intensive speech-language treatment was given for 6 months to improve velopharyngeal function as well as articulation.

DISCUSSION

VCFS is a genetic defect. It is a rare disorder affecting approximately 1/ 700 live births. It can be inherited from either a parent who has syndrome, or can arise from a fresh mutation. According to the chromosome study, the mode of inheritance of this case reported here is autosomal dominant sporadic. Thus, genetic counseling plays an important role in treatment. The patient is primarily characterized by specific malformations[3,8,9]. Apart from the more obvious major features including various heart defects, facial malformations and learning difficulty, extreme hypernasality and speech deformities create several problems for the patient's interaction with his environment, preventing normal intellectual development and generating psychological problems as severe as those caused by multiple problems as seen in the case of cleft palate patient with velopharyngeal insufficiency, hearing loss and mental retardation. The deformities seen here are complex[3,6-10].

Ideally, treatment of VCFS begins with the correct diagnosis, followed by identification of the patient's individual problems and needs, and the appropriate facilities to administer what is needed[11]. A multidisciplinary approach is the best arrangement. A Cranio-Maxillo-Facial team may consist of a plastic surgeon, neurosurgeon, ENT, pediatrician, genetic counselor, audiologist, speech-language pathologist, ophthalmologist, psychologist, dentist, orthodontist, social worker, and nurse. The team approach is used to determine the best collaborative corrective plan for the deficiencies of the patient. Cleft palate repair results vary a lot depending on the complexity of the problems. Because of the rarity of these conditions, effective use of resources and good results are obtained in a specialized centre with a multidisciplinary team. Between mid July 1995 and June 2002, only 1 case with a definite diagnosis of velocardiofacial syndrome was referred to the authors for repair of the cleft palate. The timing at which the various surgeries and treatments for other defects and problems are performed as diverse; however, there has been a trend towards earlier intervention in order to obtain good functional speech development before the age of two[11-13].

Postoperation, the patient did not demonstrate affected postpalatoplasty auditory perceptual symptoms of velopharyngeal incompetence. This is in agreement with some previous studies[14]. For example, preliminary results have been reported from a prospective, alternated, single-institution study of the effect of intravelar veloplasty upon postpalatoplasty velopharyngeal incompetence[14]. Their findings suggested that there was no beneficial effect of intravelar veloplasty upon velopharyngeal competence.

Articulation and resonation disorders appeared to be unaffected by the palatoplasty. Not only did the patient show no reduction in overall hypernasal resonance and nasal emission, but he also developed nasal turbulence. The defect persisted postoperatively, and might require secondary surgery for elimination of hypernasality[15] and speech training to correct the articulation defect.

Due to the extremely hypernasality and speech disorder of this patient postoperatively, clinical reassessment and multidisciplinary management of the velopharyngeal insufficiency problem were reviewed and rearranged. Conventionally, the primary treatment for the cleft palate condition usually includes surgery in the first year of life to correct the structural defect[11-13,15]. This is performed first to insure there is adequate soft tissue to close the velopharynx and enhance normal speech development during this period of life. These procedures may need to be done in one or two stages depending on the degree of deformity. However, there are differences of opinion among speech-language pathologists on various management issues related to velopharygeal incompetence[16-19].

For this case, we felt that the potential for functional recovery of velopharyngeal incompetence was so poor that management and treatment should be reconsidered. More specifically, the function of the velopharyngeal closure following the operation was normal for deglutition, but not for speech. It might be argued that secondary surgery should have been reconstruction because the presence of good velopharyngeal closure in the patient (during swallowing) and with the poor prognosis for the syndrome (inconsistency during speaking) were not definite indications for secondary surgery. Therefore, further related indications for secondary surgery were reviewed. By interviewing the patient and his parent (mother), we were able to assess their satisfaction with appearance, speech ability and intelligibility. Both of them were concerned about his appearance and speech. The level of concern was moderate, although the problems disturbed the patient's quality of life. This perceptual aspect is in agreement with many studies[19,20], such as those of Strauss and colleagues in 1988. As a result, we suggest that intensive speech-language treatment should continue with a home program and significant family member (mother) should be involved, in spite of secondary surgery. We do believe that this technique is particularly appropriate in this case. Although there are some successful methods of surgery, it is desirable not to conduct secondary surgery but still able to adjust velopharyngeal closure. Therefore, a course of intensive speech-language treatment for 6 months to improve velopharyngeal closure and articulation function was arranged.

Nasoendoscopy has become a popular clinical tool for assessing velopharyngeal function[21-27]. The combined auditory and visual perceptual evaluation inherent in nasoendoscopy appears to be the most significant asset for both clinical and research applications[24]. In addition, nasoendoscopy has also been suggested as a visual feedback therapy tool to establish and promote consistent closure of the velopharyngeal port during connected speech after pharyngeal flap surgery. As reported by Witzel, Tobe and Salyer in 1989[28], nasoendoscopy biofeedback treatment is useful in helping motivated adults with velopharyngeal incompetence to achieve better velopharyngeal closure during connected discourse after pharyngeal flap surgery or revision. Nasoendoscopy revealed inconsistent closure.

In conclusion, this paper illustrates the values of combined auditory speech assessment and nasoendoscopic examination of velopharyngeal function in the re-examination of speech. Use of these combined measures enables clinicians to identify accurately velopharyngeal function. Furthermore, this paper points out the need for systematic investigation into the effects of surgery on speech.

REFERENCES

1. Dyce O, McDonald-McGinn D, Kirschner RE, Zackai E, Young K, Jacobs IN. Otolaryngologic manifestations of the 22q11.2 deletion syndrome. Arch Otolaryngol Head Neck Surg 2002; **128:** 1408-12.
2. Shprintzen RJ. Velo-cardio-facial syndrome. In :Dover MA. Birth Defects Encyclopedia. Center for Birth Defects Information Services, 1990: 1744-45.
3. Shprintzen RJ, Goldberg RB, Young D, Wolford L. The velo-cardio-facial syndrome : a clinical and genetic analysis. Pediatrics 1981; **67:** 167-72.
4. Cohen MM, Gorlin RJ, Fraser FC. Craniofacial disorders. In : Emery AEH. Rimoin D's Principles and Practice of Medical Genetics, 3rd ed. New York: Churchchill Livingstone, 1997: 1121-47.
5. Shprintzen RJ, Siegel-Sadewitz VL, Amato J, Goldberg RB. Anomalies associated with cleft lip, cleft palate, or both. Am J Med Genet 1985; **20:** 585-95.
6. Golding-Kushner KJ, Weller G, Shprintzen RJ. Velo-cardio-facial syndrome : Language and psychological profiles. J Craniofac Genet 1985; **5:** 259-66.
7. Shprintzen RJ, Goldberg R, Golding-Kushner KJ, Marion R. Late-onset psychosis in the velo-cardio-facial syndrome. Am J Med Genet 1992; **42:** 141-42.
8. Goldberg R, Motzkin B, Marion R, Scambler PJ, Shprintzen RJ. Velo-cardio-facial syndrome : a review of 120 cases. Am J Med Genet 1992; **45:** 313-19.
9. Witzel MA. Communicative impairment associated with clefting. In : Shprintzen RJ, Bardach J, eds. Cleft palate speech management : a multidisciplinary approach. St. Louis : Mosby, 1995: 137-66.
10. MacKenzie-Stepner K, Witzel MA, Stringer DA, Lindsay WK, Munro IR, Hughs H. Abnormal carotid arteries in the velocardiofacial syndrome : a report of three cases. Plast Reconstr Surg 1987; **80:** 347-51.
11. Shprintzen RJ, Bardach J. Cleft palate speech management: A multidisciplinary approach. St. Louis : Mosby, 1995.
12. Dorf D, Curtin JW. Early cleft palate repair and speech outcome. Plas Reconstr Surg 1982; **70:** 74-79.
13. Riski JE, De Long E. articulation development in children with cleft lip / palate. Cleft Palate J 1984; **21:** 57-64.
14. Marsh JL, Grames LM, Holtman B. Intravelar veloplasty : a prospective study. Cleft Palate J 1989; **26:** 46-50.
15. Witt PD, D'Antonio LL. Velopharyngeal insufficiency and secondary palatal management : a new look at an old problem. Clin Plas Surg 1993; **20:** 707-21.
16. Pannbacker M, Laww NJ, Stout BM. Speech-language pathologists' opinions on the management of VPI. Cleft Palate J 1990; **27:** 68-70.
17. Dalson RM, Warren DW. The diagnosis of velopharyngeal inadequacy. Clin Plas Surg 1985; **12:** 685-95.
18. D'Antonio LL. Evaluation and management of velopharyngeal dysfunction. A speech pathologist's viewpoint. Cleft Palate Surg 1992; **2:** 86-111.
19. Endriga MC, Kapp-Simon KA. Psychological issues in craniofacial care : state of the art. Cleft Palate Craniofac J 1999; **36:** 3-11.
20. Strauss RP, Broder H, Helms RW. Perceptions of appearance and speech by adolescent patients with cleft lip and palate and by their parents. Cleft Palate J 1988; **25:** 335-42.
21. Chuangsuwanich A, Manochiopinig S. Nasoendoscopy: Clinical implications. Siriraj Hosp Gaz 2002; **54:** 757-71.
22. Croft CB, Shprintzen RJ, Rakoff SJ. Patterns of velopharyngeal valving in normal and cleft palate subjects: A multiview videofluoroscopic and nasoendoscopic study. Laryngeaoscope 1981; **91:** 265-71.
23. Miyazaki T, Matsuya T, Yamaoka M. Fiberscopic methods for assessment of velopharyngeal closure during various activities. Cleft Palate J 1975; **12:** 107-14.
24. D'Antonio LL, Marsh JL, Province MA, Muntz HR, Phillips CJ. Reliability of flexible fiberoptic nasopharyngoscopy for evaluation of velopharyngeal function in a clinical population. Cleft Palate J 1989; **26:** 217-25.
25. D'Antonio LL, Muntz HR, Marsh JL, Marty-Grames M, Backensto-Marsh R. Practical application of flexible fiberoptic nasopharyngoscope for evaluating velopharyngeal function. Plast Reconstr Surg 1988; **82:** 611-18.
26. Witt PD, D'Antonio L, Chait D, Lotz W, Netsell R. Pediatric videonasoendoscopy for speech and voice evaluation. Otolaryngol Head Neck Surg 1986; **94:** 578-83.
27. Matsuya T, Yamaoka M, Miyasaki T. A fiberscopic study of velopharyngeal closure in patients with operated cleft palates. Plas Reconstr Surg 1979; **63:** 497-500.
28. Witzel MA, Tobe J, Salyer KE. The use of videonasopharyngoscopy for biofeedback therapy in adults after pharyngeal flaps surgery. Cleft Palate J 1989; **26:** 126-34.