ACAPS Winter Retreat 2020

Joyce K. McIntyre, MD
### 2016

<table>
<thead>
<tr>
<th>Predictor</th>
<th>Volunteered % (n)</th>
<th>Did NOT Volunteer % (n)</th>
<th>OR</th>
<th>P-Value</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Gender</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>67.8% (394)</td>
<td>32.2% (187)</td>
<td>1.66</td>
<td>0.012</td>
</tr>
<tr>
<td>Female</td>
<td>56.0% (75)</td>
<td>44.0% (59)</td>
<td></td>
<td>reference</td>
</tr>
<tr>
<td><strong>Fellowship Training</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cosmetic</td>
<td>50.0% (43)</td>
<td>50.0% (43)</td>
<td>0.47</td>
<td>0.001</td>
</tr>
<tr>
<td>Craniofacial</td>
<td>84.1% (90)</td>
<td>15.9% (17)</td>
<td>3.10</td>
<td>&lt;0.001</td>
</tr>
</tbody>
</table>
# 2020

## Table 1: Cohort Demographics

<table>
<thead>
<tr>
<th>Training Level</th>
<th>LMIC Respondent</th>
<th>HIC Respondent</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Completed Training</td>
<td>75.5%</td>
<td>92.1%</td>
<td>0.01</td>
</tr>
<tr>
<td>Trainee</td>
<td>24.5%</td>
<td>7.9%</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Practice Location</th>
<th></th>
<th></th>
<th>0.05</th>
</tr>
</thead>
<tbody>
<tr>
<td>Urban</td>
<td>90.0%</td>
<td>72.6%</td>
<td></td>
</tr>
<tr>
<td>Mid Sized City</td>
<td>8.0%</td>
<td>21.0%</td>
<td></td>
</tr>
<tr>
<td>Rural</td>
<td>2.0%</td>
<td>6.5%</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Type of Practice</th>
<th></th>
<th></th>
<th>0.18</th>
</tr>
</thead>
<tbody>
<tr>
<td>Private Hospital or Clinic</td>
<td>50.0%</td>
<td>52.5%</td>
<td></td>
</tr>
<tr>
<td>Academic Hospital or Clinic</td>
<td>42.9%</td>
<td>32.8%</td>
<td></td>
</tr>
<tr>
<td>Public Hospital or Clinic</td>
<td>6.1%</td>
<td>11.5%</td>
<td></td>
</tr>
<tr>
<td>Non-government organization (NGO) Hospital or Clinic</td>
<td>8.2%</td>
<td>0.0%</td>
<td></td>
</tr>
<tr>
<td>Faith Based Hospital or Clinic</td>
<td>2.0%</td>
<td>3.3%</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Gender</th>
<th></th>
<th></th>
<th>0.55</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>85.7%</td>
<td>85.7%</td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>14.3%</td>
<td>14.3%</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Age</th>
<th></th>
<th></th>
<th>0.44</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;30 years old</td>
<td>2.0%</td>
<td>0.0%</td>
<td></td>
</tr>
<tr>
<td>30-45 years old</td>
<td>30.6%</td>
<td>20.6%</td>
<td></td>
</tr>
<tr>
<td>46-60 years old</td>
<td>49.0%</td>
<td>38.1%</td>
<td></td>
</tr>
<tr>
<td>61-75 years old</td>
<td>18.4%</td>
<td>41.3%</td>
<td></td>
</tr>
</tbody>
</table>
2016
Figure 3: Distribution of Origin Country of Visiting Surgeons
Diversity and Scientific Literature
Patterns of Orofacial Clefting in New York City From 1983 to 2010: Trends by Racial Background, Birthplace, and Public Health Strategies

Sydney C. Butts, MD¹, Simone Reynolds, MPH, PhD², Lyuba Gitman, MD³, Prayag Patel, MD³, and Michael Joseph, MPH, PhD²

The Birth Prevalence of Cleft Lip and/or Cleft Palate After the 2011 Tōhoku Earthquake and Tsunami

Yoshimichi Imai, MD, PhD¹, Takehiko Sanada, MD², and Masahiro Tachi, MD, PhD¹
Outcomes

Atypical Facial Clefts From Northcentral Nigeria, Review of 36 Cases

Seidu A. Bello, BDS, FMCDS Nig, FWACS1,2, Allwell Brown Ibikari, BDS1,3, Ifeoluwa Oketade, BDS1,3, and Saliu A. Balogun, BPT, MSc4

Abstract

Objective: This study aims to present the pattern and management of atypical facial clefts from a Nigerian population.

Design: This is a retrospective study of patients seen during series of surgical outreach programs in North Central Nigeria, between 2011 and 2017. All cases of atypical facial clefts encountered were evaluated for the pattern of presentation using Tessier classification system. All of them were surgically repaired and outcome was determined using a Likert scale. Data were presented using descriptive statistics.

Setting: Outreach programs in North Central Nigeria by Cleft & Facial Deformity Foundation.

Results: Thirty-six patients, 19 (52.8%) males and 17 (47.2%) females, were studied. The age range was 1 week to 34 years, with mean (standard deviation) of 11.2 (11.8) years. Fifty-eight cleft cases were recorded, with Tessier 1 being the commonest (n = 14; 24.1%) cleft type. There were 35 (60.3%) cases of middle cleft, 14 (24.2%) cases of oblique cleft, and 9 (15.5%) cases of lateral cleft. Typical cleft lip and palate coexisted with atypical facial cleft in 2 (5.6%) patients. Cleft was found to be median in 12 (33.3%) patients and right sided in 9 (25%) patients. Majority (n = 24, 66.7%) of the cases were repaired by simple excision plus direct closure with successful outcome.

Conclusions: Numerical classification by Paul Tessier has reduced the inconsistencies of nomenclature among practitioners. Majority of atypical facial clefts could be treated by simple techniques with satisfactory outcomes. However, the more complex cases will require multistaged surgical approach.

Keywords

atypical facial clefts, Northcentral Nigeria, cleft repair
Outcomes

Evaluating Nasalance Values Among Bilingual Mandarin–English Speakers

Eshan Pua, MS¹, Yolanda Holt, PhD¹, Lakshmi Kollara, PhD², Balaji Rangarathnam, PhD³, Xiangming Fang, PhD³, and Jamie L. Perry, PhD¹

Abstract

Objective: The goals of this research are (1) to establish normative nasalance values for bilingual Mandarin–English speakers and compare values to those of previously reported monolingual Mandarin speakers, and (2) to examine whether sex, age, dialect, and language proficiency affect levels of nasalance among Mandarin–English speakers in both English and Mandarin.

Design: All participants recorded the speech stimuli, constructed to include oral sentences, nasal sentences, oronasal sentences, and vowels /a, i, u/ in Mandarin and English. Nasalance measurements were recorded using the Nasometer II 6450.

Participants and Setting: A total of 45 (20 males and 25 females) native Mandarin speakers between 20 and 54 years of age from mainland China participated in the study.

Results: Mean nasalance scores of the Mandarin oral sentence (Mean [M] = 17.64, standard deviation [SD] = 7.33), oronasal sentence (M = 54.62, SD = 7.81), and nasal sentence (M = 68.73, SD = 8.09) are reported. Mean nasalance scores of the English oral sentence (M = 20.02, SD = 7.83), oronasal sentence (M = 58.71, SD = 7.59), and nasal sentence (M = 65.27, SD = 7.45) are reported. A repeated measures analysis of variance showed significant sex difference in nasalance scores for English stimuli (P = .031) and Mandarin stimuli (P = .040). There was no significant effects of age, dialect, and language proficiency on Mandarin or English stimuli.

Conclusions: This is the first study to report normative values for Mandarin–English speakers using the Nasometer II. Values reported can be used for objective assessment of bilingual speakers.

Keywords
Mandarin, Chinese, English, nasalance, nasometer, resonance
Patient-Perceived Barriers to Accessing Cleft Care at a Tertiary Referral Center in São Paulo, Brazil

Ananda Ise, MD¹, Camila Menezes¹, Joao Batista Neto¹, Saurab Saluja, MD, MPP², Julia R. Amundson, MD, MPH², Hillary Jenny, MD², Ben Massenburg, MD², Isabelle Citron, BmBCh², and Nivaldo Alonso, MD, PhD¹

Abstract
https://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1007501
Association Studies Between Regulatory Regions of IRF6/TP63 Genes and Nonsyndromic Oral Clefts

Yah-Huei Wu-Chou, PhD\textsuperscript{1,2}, Yi-Chieh Lu, MS\textsuperscript{1,2}, Kuo-Ting Philip Chen, MD\textsuperscript{2,3}, Hsien-Fang Chang, MS\textsuperscript{4}, Yin-Ting Lin, BS\textsuperscript{1,2}, and Lun-Jou Lo, MD\textsuperscript{2,3,5}
Target Capture/Next-Generation Sequencing for Nonsyndromic Cleft Lip and Palate in the Japanese Population

Masayasu Shibano, DDS¹, Akira Watanabe, DDS, PhD¹, Nobuo Takano, DDS, PhD², Hiroyuki Mishima, DDS, PhD³, Akira Kinoshita, PhD³, Koh-ichiro Yoshiura, MD, PhD³, and Takahiko Shibahara, DDS, PhD¹

Abstract

Objective: The pathogenesis of nonsyndromic cleft lip with or without cleft palate (NSCL ± P) and nonsyndromic cleft palate only (NSCP) may be associated with genetic factors. Although some predisposing genes/loci have been reported, their attributable risk is too small to be clinically meaningful. To clarify the genetic causes and mechanisms of NSCL±P or NSCP, we conducted mutation analysis of target genes using a next-generation sequencing (NGS) approach.

Methods: The target genes, IRF6, WNT5A, WNT9B, TP63, MSX1, TFAP2A, PAX9, DLX3, DLX4, and MN1, were selected based on previous reports of potential associations with the development of NSCL±P or NSCP from genome-wide association studies and candidate gene analyses. Mutation analysis was conducted using NGS on 74 Japanese trios (patient and parents) and 18 Japanese patients only families.

Results: We detected single-nucleotide variants (SNVs) for 7 genes: IRF6, DLX4, WNT5A, TFAP2A, WNT9B, TP63, and PAX9. The SNVs found on IRF6 and DLX4 were missense mutations, whereas those identified on WNT5A, TFAP2A, WNT9B, TP63, and PAX9 were rare variants in the noncoding region; no de novo mutation was identified in the trio samples. The amino acid change on DLX4 was detected within the highly conserved homeodomain and was predicted to have a deleterious impact on the protein function by in silico analysis.

Conclusions: The DLX4 missense mutation c.359C>T (Pro120Leu) was found in 1 Japanese patient with NSCL±P and was located in the homeodomain region. This mutation likely plays a role in the development of NSCL±P in the Japanese population.

Keywords
mutation analysis, next-generation sequencing, cleft lip/palate, SNV, Japanese
SMAD6 Genotype Predicts Neurodevelopment in Nonsyndromic Craniosynostosis

Robin T. Wu, M.D.
Andrew T. Timberlake, M.D., Ph.D.
Paul F. Abraham, B.S.
Kyle S. Gabrick, M.D.
Xiaona Lu, M.D.
Connor J. Peck, B.S.
Rajendra F. Sawh-Martinez, M.D., M.H.S.
Derek M. Steinbacher, M.D., D.M.D.
Michael A. Alperovich, M.D., M.Sc.
John A. Persing, M.D.

New Haven, Conn.

**Background:** De novo or rare transmitted mutations in the SMAD6 gene affect 7 percent of midline nonsyndromic synostosis patients. This study aimed to determine the neurocognitive sequelae of SMAD6 synostosis.

**Methods:** Nonsyndromic synostosis patients 6 years or older with SMAD6 mutations and non-SMAD6 nonsyndromic synostosis controls were recruited. All patients completed a double-blinded neurodevelopmental battery (i.e., Wechsler Fundamentals, Wechsler Abbreviated Scale of Intelligence, Beery-Buktenica Developmental test), and parents/guardians completed behavioral surveys (Behavior Rating Inventory of Executive Function and Behavior Rating System for Children).

**Results:** Twenty-eight patients participated: 10 known SMAD6 patients (average age, 10 years; 1 female; eight metopic and two sagittal; nine treated with cranial vault remodeling and one treated with strip craniectomy) and 18 non-SMAD6 controls (age, 9.5 years; three female; 12 metopic and six sagittal; 17 treated with cranial vault remodeling and one treated with strip craniectomy). There were no differences between any demographics. Testing age, surgical...
Surgical Correction of Craniofacial Microsomnia: Evaluation of Interventions in 565 Patients at Three Major Craniofacial Units

Britt I. Pluijmens, M.D.,
D.M.D.
Cornelia J. J. M. Caron,
M.D., D.M.D.
Lara S. van de Lande, M.D.
Sonje Schaal, M.D.
Irene M. Mathijssen, M.D.,
Ph.D.
Eppo B. Wolvius, D.D.S.,
M.D., Ph.D.
Neil Bulstrode, M.B.B.S.,
M.D.
Robert D. Evans, M.Sc.D.,
F.D.S.R.C.S.(Eng. and Ed.),
M.Oreh., R.C.S.(Ed.)
Bonnie L. Padwa, D.M.D.,
M.D.
Maarten J. Koudstaal, M.D.,
D.M.D., Ph.D.
David J. Dunaway, C.B.E.,
F.D.S.R.C.S., F.R.C.S.(Plast.)
Rotterdam, The Netherlands; Boston,
Mass.; and London, United Kingdom

**Background:** Craniofacial microsomnia is characterized by an asymmetric hypoplasia of derivatives of the first and second pharyngeal arch, leading to a variety of phenotypic presentations. Studies on surgical correction of patients with craniofacial microsomnia have small cohorts, leaving controversial opinions on the optimal treatment modality, the indication for surgery, and the optimal timing of surgery. The purpose of this study was to evaluate the types of, timing of, and total number of surgical corrections performed and the number of surgical procedures in correlation to the severity of the phenotype.

**Methods:** A retrospective chart study was conducted including patients diagnosed with craniofacial microsomnia from three large craniofacial units. Demographic, radiographic, and clinical information was obtained, including type and number of surgical procedures and age at the time of surgery.

**Results:** A total of 565 patients were included. In total, 445 (78.4 percent) of all patients underwent some form of surgery during their life, varying from skin tag removal to major craniofacial operations. The number of surgical interventions was higher with increasing severity of phenotype, bilateral presentation, and a younger age at the first intervention.

**Conclusions:** Multiple surgical corrections are frequently seen in patients with a more severe or bilateral presentation. Furthermore, those who are treated earlier in life for correction of asymmetry of the mandible will undergo significantly more surgical procedures to correct the asymmetry later on, independent of the Pritzansky-Kaban type mandible. A prospective international multicenter study is designed with a uniform registration and outcome measurement tool to identify the optimal treatment strategy. ([Plast. Reconstr. Surg. 143: 1467, 2019.](#))
Discussion: *Surgical Correction* of *Craniofacial Microsomia*: Evaluation of Interventions in 565 Patients at Three Major *Craniofacial* Units
Molina, Fernando M.D.
Facial and Nasolabial Aesthetics of Complete UCLP Submitted to 2-Stage Palate Repair With Vomer Flap

Terumi Okada Ozawa, DDS, MSc, PhD¹, Luciana Lais Savero Reis, DDS¹, Renata Mayumi Kato, DDS¹, Diógenes Laercio Rocha, PhD¹, Renata Sathler, DDS, MSc, PhD¹, and Daniela Gamba Garib, DDS, MSc, PhD²

Abstract

Objective: To evaluate the aesthetics of nasolabial appearance and facial profile of children with unilateral cleft lip and palate (UCLP) submitted to 2-stage palate repair with vomerine flap.

Design: Retrospective.

Setting: Single center.

Patients: Forty patients with UCLP, mean age of 7.81 years of both sexes, rehabilitated at a single center by 1 plastic surgeon.

Interventions: Lip and anterior palatal repair with nasal alar repositioning was performed at 3 to 6 months of age by Millard technique and vomer flap, respectively. Posterior palate was repaired at 18 months by Von Langenbeck technique.

Main Outcome Measure(s): Four cropped digital facial photographs of each patient were evaluated by 3 orthodontists to score the nasolabial aesthetics and profile. Frequencies of each score as well means and medians were calculated. Kappa test was used for evaluating inter- and intrarater reproducibility.

Results: The nasal form and deviation was scored as good/very good in 70%, fair in 22.5%, and poor in 7.5% of the sample. The nasal–subnasal aesthetic was considered good/very good in 55%, fair in 30%, and poor in 15% of the sample. The lip vermilion border and the white part of surgical scar aesthetics were good/very good in 77.5% and 80%, fair in 17.5% for both categories, and poor in 5% and 2.5% of the cases, respectively. In all, 67.5% showed convex facial profile, 20% was straight, and 12.5% was concave profile.

Conclusions: Two-stage palatoplasty presented an adequate aesthetical results for the majority of patients with UCLP in the mixed dentition.