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Cochrane Systematic Reviews

New Reviews – January 2015

Antibiotic prophylaxis for preventing infectious complications in orthognathic surgery

Cleft Palate-Craniofacial Journal – Latest Issue

Cleft Palate-Craniofacial Journal
ISSN: 1055-6656 Latest issue available from Allen Press in Journals@Ovid (Athens Authorization)

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Titles highlighted in green may be of particular interest to Speech and Language Therapists
Titles highlighted in orange may be of particular interest to Clinical Psychologists

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**Title:** "Three-unit" muscle reconstruction in secondary cleft lip repair

**Citation:** Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(88-95), 1055-6656;1545-1569 (01 Jan 2015)

**Author(s):** Fan Q., Li Y., Danning Z., Zhang B., Chen S., Wang J.

**Language:** English

**Abstract:** Background: Secondary deformities are common in people born with unilateral cleft lip and palate. In recent years, more and more attempts and efforts have been directed toward muscle reconstruction. The authors present a new method of orbicularis oris repair in correction of secondary cleft lip deformities. Methods: From April 2009 to April 2013, a total of 28 patients underwent this procedure in the authors’ department and had a follow-up with a minimum length of 1 year. Muscle reconstruction was divided into three units that deal with the nasal floor, white lip, and red lip. Common anatomical pathologies including a deviated columella, blunted alar-facial groove, lack of philtral column, "free border" deficiency, and unapparent vermilion tubercle can be corrected in a single operation. Results: The average follow-up period was 14.6 months (range, 12 to 24 months). Contractubex gel (Merz Pharma, Frankfurt, Germany) was used to treat prominent or reddish scars in 16 patients. No major complications occurred. All the patients were satisfied with their nasolabial appearance. Conclusions: "Three-unit" muscle repair was found to be effective and practical in secondary repair. Improved aesthetic and functional results can be achieved with this comprehensive procedure.

**Publication type:** Journal: Article

**Source:** EMBASE

**Full text:** Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

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**Title:** 2,3,7,8-tetrachlorodibenzo-p-dioxin delays palatal shelf elevation and suppresses Wnt5a and lymphoid enhancing-binding factor 1 signaling in developing palate

**Citation:** Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(54-61), 1055-6656;1545-1569 (01 Jan 2015)

**Author(s):** Hu X., Gao J.H., Liao Y.J., Tang S.J., Lu F.

**Language:** English

**Abstract:** Objective: 2,3,7,8-Tetrachlorodibenzo-p-dioxin contributes to cleft palate, but the cellular and molecular mechanisms responsible for the deleterious effect on the developing palate are unclear. Because Wnt signaling is associated with 2,3,7,8-tetrachlorodibenzo-p-dioxin in organ development, we wondered whether the malformation of the palate also results from altered Wnt signaling. Results: The 2,3,7,8-tetrachlorodibenzo-p-dioxin administration affected cell proliferation of the anteroposterior axis of the palatal shelf and delayed shelf elevation in mice. The activity of Wnt5a and lymphoid enhancing-binding factor 1 was inhibited by 2,3,7,8-tetrachlorodibenzo-p-dioxin in the developing palate. Conclusions: Downregulated Wnt5a and lymphoid enhancing-binding factor 1 are associated with 2,3,7,8-tetrachlorodibenzo-p-dioxin-induced cleft palate. Moreover, delayed shelf elevation by 2,3,7,8-tetrachlorodibenzo-p-dioxin is the crucial mechanism contributing to the high incidence of cleft palate. Our findings...
may help in elucidating the mechanisms of 2,3,7,8 tetrachlorodibenzo-p-dioxin-induced cleft palate.

**Publication type:** Journal: Article  
**Source:** EMBASE  
**Full text:** Available *The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association* at Cleft Palate-Craniofacial Journal

3.**Title:** A comparison of speech outcomes using radical intravelar veloplasty or furlow palatoplasty for the treatment of velopharyngeal insufficiency associated with occult submucous cleft palate.  
**Citation:** Annals of Plastic Surgery, February 2015, vol./is. 74/2(182-6), 0148-7043;1536-3708 (2015 Feb)  
**Author(s):** Afroz PN, Macsaac Z, Rottgers SA, Ford M, Grunwaldt LJ, Kumar AR  
**Language:** English  
**Abstract:** BACKGROUND: The safety, efficacy, and direct comparison of various surgical treatments for velopharyngeal insufficiency (VPI) associated with occult submucous cleft palate (OSMCP) are poorly characterized. The aim of this study was to report and analyze the safety and efficacy of Furlow palatoplasty (FP) versus radical intravelar veloplasty (IVV) for treatment of VPI associated with OSMCP. METHODS: A retrospective review of one institution’s experience treating VPI associated with OSMCP using IVV (group 1) or FP (group 2) during 24 months was performed. Statistical significance was determined by Wilcoxon matched-pair, Independent-Samples Mann-Whitney U, and analysis of variance (SPSS 20.0.0). RESULTS: In group 1 (IVV), 18 patients were identified from August 2010 to 2011 (12 male and 6 female patients; average age, 5.39 years). Seven patients were syndromic and 11 were nonsyndromic. In group 2 (FP), 17 patients were identified from August 2009 to 2011 (8 male and 9 female patients; average age, 8.37 years). Three patients were syndromic and 14 patients were nonsyndromic. There was statistical significance between the average pretreatment Pittsburgh Weighted Speech Score (PWSS) of the 2 groups (group 1 and 2 averages 19.06 and 11.05, respectively, P = 0.002), but there was no statistical significance postoperatively (group 1 and 2 averages 4.50 and 4.69, respectively, P = 0.405). One patient from each group required secondary speech surgery. Average operative time was greater for FP (140 minutes; range, 93-177 minutes) compared to IVV (95 minutes; range, 58-135 minutes), P < 0.001. Average hospital stay was 3.9 days for IVV (range, 2-9 days) and 3.2 days for FP (range, 2-6 days), with no significant difference (P = 0.116). There were no postsurgical wound infections, oral-nasal fistulas, postoperative bleeding complications, or mortalities. CONCLUSIONS: Nonsyndromic patients with hypernasal speech are treated effectively and safely with either IVV or FP. Intravelar veloplasty trended toward lower speech scores than FP (76% IVV, 58% FP PWSS absolute reduction). Syndromic patients with OSMCP may be more effectively treated with FP (72% IVV vs 79% FP PWSS absolute reduction). Intravelar veloplasty is associated with shorter operative times. Both techniques are associated with low morbidity, improved speech scores, and low reoperative rates.  
**Publication type:** Journal Article  
**Source:** MEDLINE  
**Full text:** Available Ovid at Annals of Plastic Surgery

4.**Title:** A complex Xp11.22 deletion in a patient with syndromic autism: Exploration of FAM120C as a positional candidate gene for autism  
**Citation:** American Journal of Medical Genetics, Part A, December 2014, vol./is. 164/12(3035-3041), 1552-4825;1552-4833 (01 Dec 2014)  
**Author(s):** De Wolf V., Crepel A., Schuit F., van Lommel L., Ceulemans B., Steyaert J., Seuntjens E., Peeters H., Devriendt K.  
**Language:** English  
**Abstract:** We present a male patient with sporadic Aarskog syndrome, cleft palate, mild intellectual disability, and autism spectrum disorder (ASD). A submicroscopic discontiguous deletion was detected on chromosome Xp11.2 encompassing FGD1, FAM120C, and PHF8. That the deletion encompassed FGD1 (exons 2-8) explains the Aarskog features while the deletion of PHF8 most likely explains the cleft palate and mild intellectual disability. We identify FAM120C as a novel X-linked candidate gene for autism for two reasons: first, a larger deletion encompassing FAM120C segregates with autism in a previously reported family and second, there is recent evidence that FAM120C interacts with CYFIP1, part of the FMRP (Fragile X Mental Retardation Protein) network. In the current study, resequencing of FAM120C in 87 Belgian male patients with autism spectrum disorder identified no novel mutations. Expression of Fam120c in mouse tissues showed enriched expression in pituitary, cerebellum, cortex, and pancreatic islets of Langerhans. Additionally, we found a cortical expression pattern of Fam120c similar to that of Fmr1. In conclusion, FAM120C is a novel candidate gene for autism spectrum disorder based on genetic evidence and the brain expression pattern. Thereby we highlight a role for FMRP network genes in ASD.
5. Title: A novel patient-controlled bidirectional palatal lift appliance
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(96-101), 1055-6656;1545-1569 (01 Jan 2015)
Author(s): Greene L.E., Wilson K., McIntyre G., Wilson J., Mehendale F.V.
Language: English
Abstract: Objective: Palatal lift appliances have a role in management of velopharyngeal dysfunction for immobile palates of adequate length where surgery is contraindicated. Conventional appliances involve acrylic/wire work adjustment over successive appointments until they can be tolerated without gagging. A novel appliance has been developed where the lifting plate is incrementally distalized by the patient and vertically adjusted to optimize soft palate positioning. Method: The design, construction, and utility of the appliance, which was developed in Dundee Dental Hospital, are described. Participants: The subject was a 12-year-old boy with a variant of Moebius syndrome and velopharyngeal dysfunction. Previous pharyngoplasty had been carried out and further surgery was contraindicated. Interventions: The appliance is constructed and fitted and the flexible spring arm is vertically adjusted to lift the soft palate. The screw is turned incrementally at home, extending the lifting plate posteriorly. Videofluoroscopy allows visualization of the appliance and soft palate positioning. Main Outcome Measures/Results: The procedure improved soft palate positioning, as demonstrated by videofluoroscopy, and objective speech outcomes. Conclusions: The appliance was well tolerated and led to improved speech outcomes for the patient. Adjustments were quick and easy for both clinician and patient. Further studies are needed to definitively determine the efficacy of the appliance.

6. Title: Are there bone dehiscences in maxillary canines orthodontically moved into the grafted alveolar cleft?
Citation: American Journal of Orthodontics & Dentofacial Orthopedics, February 2015, vol./is. 147/2(205-13), 0889-5406;1097-6752 (2015 Feb)
Author(s): Yatabe MS, Ozawa TO, Janson G, Faco RA, Garib DG
Language: English
Abstract: INTRODUCTION: The aim of this study was to assess the bone morphology of teeth mesialized into the grafted region in patients with unilateral alveolar cleft. METHODS: The sample comprised 30 patients with unilateral cleft lip and palate with a mean age of 20.5 years. High-resolution cone-beam computed tomography images of the maxilla were obtained 6 months to 2 years after comprehensive orthodontic treatment. The contralateral canines and lateral incisors were used as controls. Axial section was used to measure the bone thickness, and cross section was used to measure the alveolar crest height using the cementoenamel junction as a reference. Paired t tests and Wilcoxon tests were used to compare the cleft and noncleft sides (P <0.05). RESULTS: High individual variability was found. In general, the canines in the cleft side had statistically thinner buccal bone plates than the contralateral teeth. No differences between the cleft and noncleft sides were found for the lingual bone plate thickness. The canine on the cleft side showed a slightly greater distance between the lingual alveolar crest and the cementoenamel junction than the lateral incisor in the noncleft side. CONCLUSIONS: In patients with unilateral cleft lip and palate, mesial orthodontic movement of the maxillary canines into the grafted alveolar cleft results in acceptable buccal and lingual periodontal morphology. Copyright 2015 American Association of Orthodontists. Published by Elsevier Inc. All rights reserved.

7. Title: Assessment of presurgical clefts and predicted surgical outcome in patients treated with and without nasoalveolar molding.
Citation: Journal of Craniofacial Surgery, January 2015, vol./is. 26/1(71-5), 1049-2275;1536-3732 (2015 Jan)
Author(s): Rubin MS, Clouston S, Ahmed MM, M Lowe K, Shetye PR, Broder HL, Warren SM, Grayson BH
Language: English
Abstract: Obtaining an esthetic and functional primary surgical repair in patients with complete cleft lip and palate (CLP) can be challenging because of tissue deficiencies and alveolar ridge displacement. This study aimed to describe surgeons' assessments of presurgical deformity and predicted surgical outcomes in patients with complete unilateral and bilateral CLP (UCLP and BCLP, respectively) treated with and without nasoalveolar molding (NAM). Cleft surgeon
members of the American Cleft Palate-Craniofacial Association completed online surveys to evaluate 20 presurgical photograph sets (frontal and basal views) of patients with UCLP (n = 10) and BCLP (n = 10) for severity of cleft deformity, quality of predicted surgical outcome, and likelihood of early surgical revision. Five patients in each group (UCLP and BCLP) received NAM, and 5 patients did not receive NAM. Surgeons were masked to patient group. Twenty-four percent (176/731) of surgeons with valid e-mail addresses responded to the survey. For patients with UCLP, surgeons reported that, for NAM-prepared patients, 53.3% had minimum severity clefts, 58.9% were anticipated to be among their best surgical outcomes, and 82.9% were unlikely to need revision surgery. For patients with BCLP, these percentages were 29.8%, 38.6%, and 59.9%, respectively. Comparing NAM-prepared with non-NAM-prepared patients showed statistically significant differences (P < 0.001), favoring NAM-prepared patients. This study suggests that cleft surgeons assess NAM-prepared patients as more likely to have less severe clefts, to be among the best of their surgical outcomes, and to be less likely to need revision surgery when compared with patients not prepared with NAM.

**Publication type:** Journal Article

**Source:** MEDLINE

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**8.Title:** Association between BMP4 rs17563 Polymorphism and NSCL/P Risk: A Meta-Analysis.

**Citation:** Disease Markers, 2015, vol./is. 2015/(763090), 0278-0240;1875-8630 (2015)

**Author(s):** Hu YY, Qin CQ, Deng MH, Niu YM, Long X

**Language:** English

**Abstract:** Objective. To investigate the association between bone morphogenetic protein 4 (BMP4) rs17563 polymorphism and nonsyndromic cleft lip with or without palate (NSCL/P) risk. Methods. Four online databases were researched and the related publications were collected. Odds ratio (OR) with 95% confidence interval (CI) was applied to assess the relationship; publication bias, metaregression, and sensitivity analysis were conducted to guarantee the strength of results. Results. Six published case-control studies were collected. Overall, no significant association between BMP4 rs17563 polymorphism and NSCL/P risk was found. It was notable that significant susceptibility on different ethnicity was observed in the stratified analysis. For Chinese population, the BMP4 rs17563 polymorphism was a significantly increased risk for NSCL/P (C versus T: OR = 1.52, 95% CI = 1.28-1.82, P < 0.01, I (2) = 0%; CC versus TT: OR = 2.58, 95% CI = 1.74-3.82, P < 0.01, I (2) = 0%; TC + CC versus TT: OR = 1.45, 95% CI = 1.14-1.84, P < 0.01, I (2) = 0%; CC versus TT + TC: OR=2.46, 95% CI = 1.46-4.14, P < 0.01, I (2) = 47.0%). On the contrary, significantly protective effects were found in Brazilian population (C versus T: OR = 0.69, 95% CI = 0.50-0.96, P = 0.03, I (2) = 68.5%; TC versus TT: OR = 0.52, 95% CI = 0.40-0.68, P < 0.01, I (2) = 0%; TC + CC versus TT: OR = 0.52, 95% CI = 0.35-0.78, P < 0.010, I (2) = 54.4%). Conclusion. This meta-analysis indicated that BMP4 rs17563 polymorphism could play a different role during the development of NSCL/P based on ethnicity diversity.

**Publication type:** Journal Article

**Source:** MEDLINE

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**9.Title:** Association of WNT9B gene polymorphisms with nonsyndromic cleft lip with or without cleft palate in Brazilian nuclear families

**Citation:** Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(44-48), 1055-6656;1545-1569 (01 Jan 2015)

**Author(s):** Fontoura C., Silva R.M., Granjeiro J.M., Letra A.

**Language:** English

**Abstract:** Objective: Nonsyndromic cleft lip with or without cleft palate (NSCL+/-P) is a common craniofacial anomaly of complex etiology in people. WNT pathway genes have important roles during craniofacial development, and an association of WNT genes with NSCL+/-P has been demonstrated in different populations. The aim of this study was to evaluate the association between polymorphisms in WNT3 and WNT9B genes and CL/P in Brazilian families. Patients: Seventy nuclear families composed of an affected child and the child's unaffected parents were examined clinically. Saliva samples were collected for molecular analyses. Design: Three single nucleotide polymorphisms (SNPs) in the WNT3 gene and two in WNT9B were investigated in real-time polymerase chain reaction using TaqMan chemistry. The Family Based Association Test and the transmission disequilibrium test were used to verify the association between each marker allele and NSCL6P. The level of significance was established at P < .01 after Bonferroni correction. Results: A positive association was detected between NSCL+/-P and SNP rs1530364 in the WNT9B gene. Haplotype analysis showed an association of WNT3 and WNT9B haplotypes. No association was detected between NSCL6P and individual SNPs in WNT3. Conclusion: Our study further supports the involvement of WNT9B as a cleft susceptibility gene in Brazilian families experiencing NSCL6P. Although additional studies are still necessary to unveil the exact mechanism by which WNT genes would contribute to NSCL6P, allelic polymorphisms in these genes and their interactions may partly explain the variance of individual susceptibility to NSCL6P.

**Publication type:** Journal: Article
10. Title: Bone tissue engineering for cleft lip and palate patients using non-invasive sources of stem cells
Citation: Tissue Engineering - Part A, December 2014, vol./is. 20/(S89-S90), 1937-3341 (December 2014)
Author(s): Bueno D.F., Tanikawa D., Rocha D.L., Pinheiro C., Andrade M.A., Reis L.L.
Language: English
Abstract: Cleft lip and palate (CLP), affects the alveolar bone in the great majority of the cases, and the reconstruction of this defect still represents a challenge in the rehabilitation of these patients. The gold standard in alveolar bone reconstruction is autogenous bone grafts. However, these surgical procedures may be subjected to complications such as donor area morbidity, post-surgical resorption and infections. To circumvent these problems, researchers have been focusing on the development of bone tissue engineering strategies that may offer alternative methods with no donor site morbidity for bone grafts. Therefore, in order to identify a non-invasive alternative source of stem cells with osteogenic potential, we have used Orbicular Oris Muscle (OOM) and Levator Palatine Muscle (LPM) fragments, which are regularly discarded during surgery repair of CLP patients. We also used dental pulp (DP) obtained from deciduous teeth of CLP patients. To obtain cells from these tissues we used previously described preplatting technique. These cells, through flow cytometry analysis and in vitro assays were characterized as mesenchymal stem cells and together with a biomaterial these cells lead to bone tissue reconstruction in animal model. In conclusion, we showed that cells from OOM, LPM and DP represent a promising source of stem cells for alveolar bone grafting treatment. Recently we obtained ethical permission to start the clinical trials using these mesenchymal stem cells from non-invasive sources to perform autogenous alveolar bone tissue engineering for CLP patients open new avenues to perform the treatment to them.
Publication type: Journal: Conference Abstract
Source: EMBASE

11. Title: Branchio-oculo-facial syndrome: a three generational family with markedly variable phenotype including neonatal lethality.
Citation: Clinical Dysmorphology, January 2015, vol./is. 24/1(13-6), 0962-8827;1473-5717 (2015 Jan)
Author(s): Titheradge HL, Patel C, Ragge NK
Language: English
Abstract: Branchio-oculo-facial syndrome (BOFS) is a rare autosomal dominant condition with variable expressivity, caused by mutations in the TFAP2A gene. We report a three generational family with four affected individuals. The consultand has typical features of BOFS including infra-auricular skin nodules, coloboma, lacrimal duct atresia, cleft lip, conductive hearing loss and typical facial appearance. She also exhibited a rare feature of preaxial polydactyly. Her brother had a lethal phenotype with multiorgan failure. We also report a novel variant in TFAP2A gene. This family highlights the variable severity of BOFS and, therefore, the importance of informed genetic counselling in families with BOFS.
Publication type: Journal Article
Source: MEDLINE

12. Title: CBS c.844ins68 polymorphism frequencies in control populations: Implications on nonsyndromic cleft lip with or without cleft palate
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(49-53), 1055-6656;1545-1569 (01 Jan 2015)
Author(s): Murthy J., Lakkakula S., Gurramkonda V.B., Pathapati R.M., Maram R., Lakkakula B.V.K.S.
Language: English
Abstract: Introduction: Nonsyndromic cleft lip with or without cleft palate (NSCLP) is a common birth defect with substantial clinical and social impact. Folate deficiency is one of the factors that have been associated with increased risk for NSCLP. Polymorphisms in folate and homocysteine pathway genes may act as susceptibility factors. Objective: The objective of this study was to evaluate prevalence estimates of cystathionine beta-synthase (CBS) insertion of 68-bp (c.844ins68) polymorphisms and their correlation with NSCLP. Material and Methods: A total of 236 unrelated individuals from seven Indian populations and an additional 355 cases with NSCLP and 357 controls without NSCLP were included in this study. We investigated the CBS c.844ins68 polymorphism in all samples. Genotyping was performed with polymerase chain reaction and electrophoresis. The data were statistically analyzed using the chi-square test. Results: The CBS c.844ins68 allele is present in six of the seven populations analyzed, and allele frequencies range from 1.5% in Balija to 9.1% in Sugali populations. The CBS c.844ins68 polymorphism showed a significant protective effect on NSCLP at both genotype (WW versus W): odds ratio [OR] = 0.54, 95% confidence
14. Title: Cleft lip and/or palate and auricular malformations  
**Citation:** Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(62-65), 1055-6656;1545-1569 (01 Jan 2015)  
**Author(s):** Suutarla S., Rautio J., Klockars T.  
**Language:** English  
**Abstract:** Objective: To study the relationship between cleft lip and/or palate and auricular malformations in Finnish patients with cleft. Design: Retrospective analysis of patients with an external ear malformation and either a cleft lip with or without a cleft palate (CL+/-P) or an isolated cleft palate (CP). Setting: Tertiary referral clinic. Patients: Review of hospital records of 100 patients from the register of 8200 patients with cleft in the Cleft and Craniofacial Centre at the Helsinki University Central Hospital. Main Outcome Measures: Proportions of variable auricular malformations among CL+/-P and CP patients. Results: Microtia is the most common auricular malformation among patients with cleft and is almost equally prevalent with both CL+/-P and CP. The prevalence of microtia increases as the severity of CL+/-P increases. The combination of microtia and CL+/-P or CP is frequently found with both oculo-auriculo-vertebral spectrum and Treacher Collins syndrome. Conclusions: Microtia seems to be the most common auricular malformation among patients with cleft. The prevalence of microtia seems to increase as the severity of CL6P increases, whereas in isolated CP microtia seems to occur independently.  
**Publication type:** Journal: Article  
**Source:** EMBASE  
**Full text:** Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

15. Title: Cleft lip-cleft palate in Zimbabwe: Estimating the distribution of the surgical burden of disease using...
16. Title: Cleft palate reconstruction using collagen and nanofiber scaffold incorporating bone morphogenetic protein in rats

Citation: Tissue Engineering - Part A, January 2015, vol./is. 21/1-2(85-95), 1937-3341;1937-335X (01 Jan 2015)

Author(s): Mostafa N.Z., Talwar R., Shahin M., Unsworth L.D., Major P.W., Doschak M.R.

Language: English

Abstract: Background: Absorbable collagen sponge (ACS) loaded with bone morphogenetic protein-2 (BMP-2) is approved for selected clinical applications; however, burst release limits its widespread use. Therefore, nanofiber (NF)-based scaffold with ACS backbone was developed to sustain release of loaded BMP-2 to improve the outcomes of bone grafting in a rodent model of cleft palate. Methods: BMP-2 was loaded on ACS scaffold and then NF hydrogel with different densities (1-2%) was added to sustain the BMP-2 release. The release profiles of BMP-2 from constructs with different NF densities were evaluated in vitro to explore the optimum NF density that could recapitulate physiological bone healing process. Subsequently, scaffold with the appropriate NF density was implanted into a rodent model of cleft palate. Wistar rats, with surgically induced maxillary cleft defects, were then assigned to one of the following groups (n=6/group): no scaffold (control), ACS, ACS+BMP-2, NF+ACS, and NF+ACS+BMP-2. Micro-computed tomography (muCT) was utilized to evaluate percent bone filling (%BF) at defect site as well as changes in anteroposterior and transverse dimensions of the maxilla at weeks 0, 4, and 8. Histological assessment of bone healing was performed at week 8. Results: In vitro release experiments showed that scaffolds containing 2% NF exhibited a release profile conducive to the natural stages of bone healing and, hence, it was utilized for subsequent in vivo studies. Bone healing occurred at the defect margins leaving a central bone void in the control, ACS, and NF+ACS groups over the 8-week study period. BMP-2-treated groups demonstrated higher %BF as compared with other groups at week 8 (p<0.05). Whereas the NF+ACS+BMP-2 group showed bone bridging of the defect as early as 4 weeks, which was not evident in ACS+BMP-2 group. In all groups, bone grafts did not disrupt anteroposterior and transverse growth of maxilla. Based on histological evaluations together with muCT data, NF+ACS+BMP-2 treatment resulted in clinically significant and consistent bone healing throughout the implanted scaffold when compared with the ACS+BMP-2 group. Conclusion: NF+ACS+BMP-2 constructs exhibited osteoinductive properties together with preparation simplicity, which makes it a novel approach for BMP-2 delivery for cleft palate reconstruction.

Publication type: Journal: Article

Source: EMBASE

17. Title: Co-occurrence of non-mosaic trisomy 22 and inherited balanced t(4;6)(q33;q23.3) in a liveborn female: Case report and review of the literature

Citation: Laryngoscope, February 2015, vol./is. 125 Suppl 1/(S1-S14), 0023-852X;1531-4995 (2015 Feb)

Author(s): Tollefson TT, Shaye D, Durbin-Johnson B, Mehdezadeh O, Mahomva L, Chidzonga M

Language: English

Abstract: OBJECTIVES/HYPOTHESIS: To evaluate the prevalence and unmet need for cleft lip-cleft palate reconstructive surgery by using incidence. Our hypotheses were that the age of presentation to screening clinics will decrease between 2006 and 2012, and the geospatial distribution of cases will expand to a more rural catchment area. STUDY DESIGN: Longitudinal cross-sectional/geospatial distribution study. METHODS: An online, secure database was created from intake forms for children with cleft lip-cleft palate (N=604) in Zimbabwe (2006-2012). Univariate analysis was completed. A linear regression model was fitted to test the time trend of a child's age at the time of presentation. Unique patient addresses (n=411) were matched. Maps presenting cleft diagnosis and presentation year were created with geographic information systems (GIS) software. RESULTS: The median age of presentation was greater for isolated cleft palate (4.2 years, n=106) than isolated cleft lip (1.5 years, n=251) and cleft lip-cleft palate (2.0 years, n=175). Cleft lip cases were mostly left sided with equal gender distribution. The overall age of presentation remained stable (P=.83). The age of children with isolated cleft palate decreased by 0.8 years per surgical trip (P=.01), suggesting the prevalence of unrepaired cleft palate is decreasing due to local and visiting surgeons. The catchment area extended to a less populous area, but clustered around Harare and Bulawayo. CONCLUSIONS: This study gives Zimbabwe-specific evidence that supports reports of the persistent burden of disease requiring attention. The GIS software provided data for the primary needs assessment, which will direct communication to healthcare providers and prospective patients outside of the current catchment area. LEVEL OF EVIDENCE: 3 Laryngoscope, 125:S1-S14, 2015. Copyright 2014 The American Laryngological, Rhinological and Otological Society, Inc.

Publication type: Journal Article

Source: MEDLINE
Title: Common mutations of the methylenetetrahydrofolate reductase (MTHFR) gene in non-syndromic cleft lips and palates children in North-West of Iran
Citation: Iranian Journal of Otorhinolaryngology, 2015, vol./is. 27/78(7-14), 2251-7251;2251-726X (2015)
Author(s): Abdollahi-Fakhim S., Estiar M.A., Varghaei P., Sharafi M.A., Sakhinia M., Sakhinia E.
Language: English
Abstract: Introduction: Cleft lips and cleft palates are common congenital abnormalities in children. Various chromosomal loci have been suggested to be responsible for the development of these abnormalities. The present study was carried out to investigate the association between the suspected genes (methylene tetrahydrofolate reductase [MTHFR] A1298C and C677T) that might contribute into the etiology of these disorders through application of molecular methods. Materials and Methods: This cross-sectional and explanatory study was carried out on a study population of 65 affected children, 130 respective parents and 50 healthy individuals between 2009 and 2012 at Tabriz University of Medical Sciences, IR Iran. After DNA extraction, amplification refractory mutation system-polymerase chain reaction (ARMS-PCR) and restriction fragment length polymorphism (RFLP)-PCR were used respectively to investigate the C677T and A1298C mutations for the MTHFR gene. Results: There was a significant difference in the rates of the C677T mutation when affected patients and their fathers were compared with the control group (odds ratio [OR]=0.44) (OR=0.64). However, there was no significant difference observed in the rate of this mutation between the patients’ mothers and the control group (OR=1.35). In addition, the abnormality rate was higher in patients with the A1298C mutation and their parents, when compared with the control group. This abnormality rate was higher for the affected children and their fathers in comparison with their mothers (Fathers, OR=0.26; Mothers, OR=0.65; Children, OR=0.55). No significant difference was seen in the rate of the polymorphism C677T in its CC, when the affected children and their parents were compared with the control group. However, there was a significant difference in the A1298C mutation. Conclusion: An association was seen between the A1298C mutation and cleft lip and cleft palate abnormalities in Iran. However, there seems to be a stronger relationship between the C677T mutation and these abnormalities in other countries, which could be explained by racial differences. Moreover, this association was more notable between the affected children and their fathers than their mothers. The findings in this study may be helpful in future studies and screening programs.
Publication type: Journal: Article
Source: EMBASE

18.Title: Compound heterozygosity of low-frequency promoter deletions and rare loss-of-function mutations in TXNL4A causes Burn-McKeown syndrome.
Citation: American Journal of Human Genetics, December 2014, vol./is. 95/6(698-707), 0002-9297;1537-6605 (2014 Dec 4)
Mutations in components of the major spliceosome have been described in disorders with craniofacial anomalies, e.g., Nager syndrome and mandibulofacial dysostosis type Guion-Almeida. The U5 spliceosomal complex of eight highly conserved proteins is critical for pre-mRNA splicing. We identified biallelic mutations in TXNL4A, a member of this complex, in individuals with Burn-McKeown syndrome (BMKS). This rare condition is characterized by bilateral choanal atresia, hearing loss, cleft lip and/or palate, and other craniofacial dysmorphisms. Mutations were found in 9 of 11 affected families. In 8 families, affected individuals carried a rare loss-of-function mutation (nonsense, frameshift, or microdeletion) on one allele and a low-frequency 34 bp deletion (allele frequency 0.76%) in the core promoter region on the other allele. In a single highly consanguineous family, formerly diagnosed as oculo-oto-facial dysplasia, the four affected individuals were homozygous for a 34 bp promoter deletion, which differed from the promoter deletion in the other families. Reporter gene and in vivo assays showed that the promoter deletions led to reduced expression of TXNL4A. Depletion of TXNL4A (Dib1) in yeast demonstrated reduced assembly of the tri-snRNP complex. Our results indicate that BMKS is an autosomal-recessive condition, which is frequently caused by compound heterozygosity of low-frequency promoter deletions in combination with very rare loss-of-function mutations. Copyright 2014 The American Society of Human Genetics. Published by Elsevier Inc. All rights reserved.
Many have wondered if anatomic differences are a cause or at least a contributor of this. In this sense, comparisons of sinus volumes of patients with different craniofacial clefts may be helpful to determine possible differences from normal. Thus, the present study aimed to evaluate and compare the maxillary sinus volume of patients with unilateral (UCLP) and bilateral (BCLP) cleft lip and palate to control, i.e. non-cleft patients, using cone beam computed tomography (CBCT) images. 

**Methods:** The sample consisted of 30 subjects with UCLP, 15 with BCLP and 15 control individuals (non-cleft). Each maxillary sinus was assessed three-dimensionally, segmented and its volume was calculated. The comparison between right and left sinuses was performed by Student t-test, and the differences between the control and cleft groups were calculated using ANOVA. 

**Results:** No statistical differences were found when the sides were compared (p> 0.05). In relation to the assessment among groups, all comparisons had statistically significant differences (p<0.05), with the UCLP group presenting the lowest sinus volume. 

**Conclusion:** UCLP individuals present maxillary sinuses with smaller volumes, without differences found between the cleft and non-cleft side. BCLP subjects also present a reduction in the volume when compared to a control sample, but the average sinus volume is larger than in UCLP patients.

**Publication type:** Journal Article

**Source:** MEDLINE

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**23. Title:** Do orofacial clefts represent different genetic entities?

**Citation:** Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(115-120), 1055-6656;1545-1569 (01 Jan 2015)

**Author(s):** Reiter R., Brosch S., Ludeke M., Fischbein E., Rinckleb A., Haase S., Schwandt A., Pickhard A., Maier C., Hogel J., Vogel W.

**Language:** English

**Abstract:** Objective: To contribute to the understanding of potential genetic differences between different cleft types. Method: Analysis of family history concerning cleft type and search for cleft-type-specific associations in candidate genes performed in 98 individuals from 98 families. Results: In a given family, the cleft type of a second case was more often identical to the index case than expected by chance. Each type of cleft (cleft lip [CL], cleft lip and palate [CLP], cleft palate only [CP], and submucous cleft palate only [SMCP]) was associated with different genes. Conclusion: Family history indicates some specificity of cleft types. The observed phenotype-genotype associations were compatible with this interpretation in that significant associations occurred with disjoint sets of genes in each cleft type. These observations indicate that CL, CLP, CP, and SMCP might represent genetically different entities.

**Publication type:** Journal: Article

**Source:** EMBASE

**Full text:** Available [The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3996433/) at Cleft Palate-Craniofacial Journal

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**24. Title:** Does the use of particulate bone graft increase the incidence of postoperative infection in surgery for craniosynostosis?

**Citation:** Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(e14-e17), 1055-6656;1545-1569 (01 Jan 2015)

**Author(s):** Muzaffar A.R., Nguyen T.B., Baker L., Warren A.J.

**Language:** English

**Abstract:** Objective: The use of particulate bone graft (PBG) has become an accepted technique for filling cranial defects created during cranial vault expansion for craniosynostosis. However, the use of PBG may be a risk factor for postoperative infection. The aim of this study was to compare the rate of postoperative infection in patients who received particulate bone graft (PBG+) with that in patients who did not (PBG-). Design: An Institutional Review Board-approved, retrospective, cohort study of consecutive patients was performed. Twenty-seven consecutive patients in the PBG-group were compared with 21 consecutive patients in the PBG+group. The two cohorts were assessed for incidence of surgical-site infection. Results: Statistical analysis was performed using the Fisher exact probability test. Surgical site infection occurred in none of the PBG-patients (0%) versus one of the PBG+ patients (4.76%). This difference in infection rates between the two cohorts was not statistically significant (P = .4375). Conclusions: Although there may be concern that PBG could serve as a facilitative medium for bacterial growth, this study demonstrates no statistically significant increase in infection rates with its use. Particulate bone grafting of cranial defects resulting from cranial vault expansion in craniosynostosis remains a useful and valuable technique.

**Publication type:** Journal: Article

**Source:** EMBASE

**Full text:** Available [The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3996433/) at Cleft Palate-Craniofacial Journal
25. Title: Effects of nasal port area on perception of nasality and measures of nasalance based on computational modeling
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(110-114), 1055-6656;1545-1569 (01 Jan 2015)
Author(s): Bunton K.
Language: English
Abstract: Objective: This study examined the relation between nasal port area, nasalance, and perceptual ratings of nasality for three English corner vowels, /i/, /u/, and /a/. Design: Samples were simulated using a computational model that allowed for exact control of nasal port size and direct measures of nasalance. Perceptual ratings were obtained using a paired stimulus presentation. Participants: Four experienced listeners. Main Outcome Measures: Nasalance and perceptual ratings of nasality. Results: Findings show that perceptual ratings of nasality and nasalance increased for samples generated with nasal port areas up to and including 0.16 cm² but plateaued in samples generated with larger nasal port areas. No vowel differences were noted for perceptual ratings. Conclusions: This work extends previously published work by including nasal port areas representative of those reported in the literature for clinical populations. Continued work using samples with varied phonetic context and varying suprasegmental and temporal characteristics are needed.
Publication type: Journal: Article
Source: EMBASE
Full text: Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

26. Title: Evidence of olfactory deficits as part of the phenotypic spectrum of nonsyndromic orofacial clefting.
Citation: Journal of Craniofacial Surgery, January 2015, vol./is. 26/1(84-6), 1049-2275;1536-3732 (2015 Jan)
Author(s): May MA, Sanchez CA, Deleyiannis FW, Marazita ML, Weinberg SM
Language: English
Abstract: Improved understanding of the phenotypic spectrum associated with nonsyndromic orofacial clefting (OFC) has the potential to inform efforts to uncover the etiology of this complex trait. Prior studies report that individuals with OFC are characterized by impaired olfactory ability. In this study, we test whether olfactory dysfunction extends to the unaffected parents of children with OFC. The University of Pennsylvania Smell Identification Test was used to measure olfactory ability in a sample of 60 unaffected mothers and fathers with cleft-affected children. The proportion of deficit was compared with reference data obtained from published sex- and age-specific norms on more than 2700 individuals. The proportion of deficit was significantly higher in unaffected parents compared with baseline control subjects (41.7% vs 12.6%; P < 0.001). Of unaffected fathers, 41.7% displayed evidence of deficit compared with 15.1% of male control subjects (P = 0.001), whereas 41.7% of mothers exhibited deficits compared with 10.4% of female control subjects (P < 0.001). Olfactory deficits are present at a high proportion in the unaffected parents of individuals with OFC. This suggests that the deficits observed in affected cases may not simply be a secondary consequence of surgical repair and may instead be an informative phenotype reflecting underlying etiology.
Publication type: Journal Article
Source: MEDLINE

27. Title: Fracture of the vomero-premaxillary junction in a repaired bilateral cleft lip and palate patient
Citation: Craniomaxillofacial Trauma and Reconstruction, December 2014, vol./is. 7/4(302-305), 1943-3875;1943-3883 (25 Dec 2014)
Author(s): Zwahlen R.A., Jayaratne Y.S.N., Htun S.Y., Butow K.-W.
Language: English
Abstract: Although dental trauma is common in bilateral cleft lip and palate (BCLP), patients' reports on bony fractures of the vomero-premaxillary junction cannot be found. The aim of this report is to illustrate clinical findings and the technique of fracture fixation in a child suffering from a fractured vomero-premaxillary junction as well as subsequent columella lengthening. A 4-year-old girl with a repaired BCLP presented with an open mucosal laceration and fractured vomeropremaxillary junction. Open reduction and fixation of the dislocated premaxilla was performed under general anesthesia. Fractured bone pieces of the vomero-premaxillary junction were removed and sharp bone edges at the vomer and the premaxilla were grinded. The repositioned premaxilla was fixed to the lateral alveolar arches with two mucoperiosteal sutures on each side. Additional columella lengthening was performed 2 years later. All family members were very happy about the new aesthetics of the girl. Although rare, fractures of the vomero-premaxillary junction present several challenges to clinicians related to anatomical, physiological, and psychological issues. Immediate and minimal invasive treatment strategies are recommended when managing such cases.
Publication type: Journal: Article
is associated with malignant hyperthermia.

are the first two cases where both patients were also diagnosed with congenital myopathy, and one developed malignant hyperthermia. These patients were characterized by camptodactyly, cleft palate, and talipes equinovarus. We report two exceptional cases of Gordon syndrome (GS), a rare syndrome documented to have an autosomal dominant inheritance pattern or to occur sporadically. The aim of this article is to publish a literature review and report on two new cases of Gordon syndrome (GS).

Abstract: Objective: To determine the frequency of oro-nasal fistula in patients undergoing complete cleft palate repair by two flaps. Study Design: Case series. Place and Duration of Study: Department of Plastic Surgery, Services Hospital, Lahore, from January to December 2013. Methodology: Patients admitted to the study place for repair of cleft palate after informed consent obtained were included. Cleft palate was repaired by two flap palatoplasty, using Bardach technique. Patients were discharged on the second postoperative day and followed-up at third week postoperatively. During follow-up visits, fistulae formation and their sites were recorded on pre-designed proforma. Results: Among the total 90 patients, 40 patients (44.4%) were male and 50 patients (55.6%) were female. The mean age was 6.4 +/- 5.7 years ranging from 9 months to 20 years. At third week follow-up, 5 patients (5.6%) had fistulae formation. Four patients (80%) had anterior fistulae and one patient (20%) had posterior fistula. Conclusion: With two-flap palatoplasty Bardach procedure for repair of cleft palate, the complication of fistula formation was uncommon at 5.6%, provided the repair was tension free and multi-layered.

Publication type: Journal: Article
Source: EMBASE

28.Title: GAD65/GAD67 double knockout mice exhibit intermediate severity in both cleft palate and omphalocele compared with GAD67 knockout and VGAT knockout mice

Citation: Neuroscience, March 2015, vol./is. 288/(86-93), 0306-4522;1873-7544 (March 02, 2015)
Author(s): Kakizaki T., Oriuchi N., Yanagawa Y.
Language: English
Abstract: Inhibitory neurotransmitters, -aminobutyric acid (GABA) and glycine, are transported into synaptic vesicles by the vesicular GABA transporter (VGAT). Glutamate decarboxylase (GAD) is a GABA-synthesizing enzyme and two isoforms of GAD, GAD65 and GAD67 are encoded by two independent genes. There was virtually no GABA content in GAD65/GAD67 double knockout (GADs DKO) mouse brains. Neither GABAergic nor glycinergetic inhibitory postsynaptic currents were almost detected in VGAT knockout (KO) mouse cultured neurons and spinal cords. GAD67 KO and VGAT KO mice displayed developmental abnormalities, cleft palate and omphalocele, suggesting that GABAergic transmission is involved in palate and abdominal wall formations. However, the incidence and severity of both failures in GAD67 KO mice were lower and less than those in VGAT KO mice. These results raise the possibility that GABAergic transmission mediated by GAD65-produced GABA and/or glycinergetic transmission contributed to both palate and abdominal wall formations. However, it still remains unclear whether GABAergic transmission mediated by GAD65 and glycinergetic transmission contribute to those formations. Here, to answer these questions, we generated GADs DKO mice and compared the phenotypes of GADs DKO mice with those of GAD67 KO and VGAT KO mice. Our anatomical analyses demonstrated that the incidence of cleft palate and omphalocele in GAD67 KO mice was 65.8% and 58.9%, respectively, but the incidence of both phenotypes in GADs DKO and VGAT KO mice was 100%. The severity of cleft palate and omphalocele was evaluated by elevation of palate shelves and size and liver inclusion of omphalocele, respectively. We observed that the phenotypes of cleft palate and omphalocele in GADs DKO mice were more and less severe than those in GAD67 KO and VGAT KO mice, respectively. These results indicate the significant contribution of not only GAD65-mediated GABAergic but also glycinergetic transmissions to both palate and abdominal wall formations.

Publication type: Journal: Article
Source: EMBASE

29.Title: Gordon syndrome: Literature review and a report of two cases

Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(e18-e22), 1055-6656;1545-1569 (01 Jan 2015)
Author(s): Botha S.J.P., Kloppers H.P., Butow K.-W.
Language: English
Abstract: The aim of this article is to publish a literature review and report on two new cases of Gordon syndrome (GS), a rare syndrome documented to have an autosomal dominant inheritance pattern or to occur sporadically; it is characterized by camptodactyly, cleft palate, and talipes equinovarus. We report two exceptional cases of GS where both patients were also diagnosed with congenital myopathy, and one developed malignant hyperthermia. These are the first two cases reported where patients were diagnosed with both GS and congenital myopathy or where GS is associated with malignant hyperthermia.
31. Title: High-resolution array comparative genomic hybridization utility in polish newborns with isolated cleft lip and palate.

Citation: Neonatology, 2015, vol./is. 107/3(173-8), 1661-7800;1661-7819 (2015)


Language: English

Abstract: Cleft lip with or without cleft palate is one of the most common birth defects of unknown etiology. A fraction of its genetic causes is attributable to copy number variations detected by array comparative genomic hybridization. The value of array comparative genomic hybridization screening as a first-tier test in the newborn population with multiple congenital anomalies has now been accepted. Due to unspecific clinical picture at this age, it can also be applied to neonates with isolated anomalies. Our purpose was to assess utility of array comparative genomic hybridization in the population of newborns with isolated cleft lip and palate. We conducted the study in a group of 52 Polish newborns with apparently isolated cleft lip and palate. In the study group, we found 8 rearrangements. Of these, 2 de novo events have been noted that potentially explain the phenotype. In addition, 2 novel candidate genes for cleft lip and palate, CHN2 and CDH19, are suggested. Given the high number of inherited potentially benign changes, we question the clinical utility of array comparative genomic hybridization in the newborn population with isolated cleft lip and palate, at the same time pointing to the need of skilled professional's clinical assessment at a later age. However, the value of this technology in searching for the cause of isolated anomalies cannot be underestimated. 2015 S. Karger AG, Basel.

Publication type: Journal Article

Source: MEDLINE

32. Title: Higher risk of orofacial clefts in children born to mothers with angina pectoris: A population-based case-control study

Citation: Congenital Anomalies, February 2015, vol./is. 55/1(49-54), 0914-3505;1741-4520 (01 Feb 2015)

Author(s): Czeizel A.E., Vereczkey A., Banhidy F.

Language: English

Abstract: Previously an unexpected association of maternal angina pectoris (MAP) during pregnancy with a higher risk of orofacial clefts in their children was found. There were three objectives of this study: (i) to evaluate the validity of MAP-diagnoses in the previous study and the recent history of mothers with MAP in a follow-up study; (ii) to estimate the prevalence of other congenital abnormalities in the offspring of mothers with MAP; and (iii) to analyze the possible effect of confounders for the risk of orofacial clefts. The large dataset of population-based Hungarian Case-Control Surveillance System of Congenital Abnormalities, 1980-1996 was evaluated including 22843 cases with congenital abnormalities and 38151 controls without any defect. Twenty-two cases (0.10%) and 12 controls (0.03%) were born to mothers with medically recorded MAP (odds ratio [OR] with 95% confidence interval [CI]: 3.7, 1.8-7.3). Of 22 cases, six had isolated cleft lip+/-palate (OR with 95% CI: 13.3, 4.9-35.9) and two were affected with isolated cleft palate (OR with 95% CI: 10.5, 2.3-47.6). The diagnosis of MAP was confirmed in seven women visited at home in 2009-2010, two had recent myocardial infarction and five were smokers. There was no higher risk for other congenital abnormalities. In conclusion the higher risk of orofacial clefts was confirmed in the children of mothers with MAP and smoking may trigger the genetic predisposition of both MAP and orofacial clefts. However, the number of cases was limited and therefore further studies are needed to confirm or reject this theoretically and practically important observation.

Publication type: Journal: Article

Source: MEDLINE

33. Title: Hypodontia and supernumerary and impacted teeth in children with various types of clefts.

Citation: American Journal of Orthodontics & Dentofacial Orthopedics, February 2015, vol./is. 147/2(221-5), 0889-5406;1097-6752 (2015 Feb)

Author(s): Jamilian A, Jamilian M, Darnahal A, Hamedi R, Mollaei M, Toopchi S

Language: English

Abstract: INTRODUCTION: The purpose of this study was to determine the prevalence of hypodontia, hyperdontia, and impacted teeth in children with various types of clefts.METHODS: This study sample consisted of 201 cleft
patients including 131 male subjects with a mean age of 12.3 +/- 4 years and 70 female subjects with a mean age of 12.6 +/- 3.9 years. Charts, models, radiographs, and intraoral photographs were used for the study. t tests, chi-square tests, and binomial tests were used for assessment of the data.RESULTS AND CONCLUSIONS: Hypodontia was found in 129 subjects (64.1%). The chi-square test showed no statistically significant difference between the type of cleft and hypodontia (P <0.319). The binomial test showed that the frequencies of subjects with hypodontia were significantly higher in both unilateral and bilateral cleft lip and palate patients (P <0.015 and P <0.001, respectively). Hyperdontia and impacted teeth were also found to occur mostly in the maxillary arch, and maxillary canines were the most commonly impacted teeth in both unilateral and bilateral cleft lip and palate patients.

**Publication type:** Journal Article

**Source:** MEDLINE

34. **Title:** Important points for primary cleft palate repair for speech derived from speech outcome after three different types of palatoplasty.

**Citation:** International Journal of Pediatric Otorhinolaryngology, December 2014, vol./is. 78/12(2127-31), 0165-5876;1872-8464 (2014 Dec)

**Author(s):** Funayama E, Yamamoto Y, Nishizawa N, Mikoya T, Okamoto T, Imai S, Murao N, Furukawa H, Hayashi T, Oyama A

**Language:** English

**Abstract:** OBJECTIVE: This study was performed to investigate speech outcomes after three different types of palatoplasty for the same cleft type. The objective of this study was to investigate the surgical techniques that are essential for normal speech on the basis of each surgical characteristic.

**METHODS:** Thirty-eight consecutive nonsyndromic patients with unilateral complete cleft of the lip, alveolus, and palate were enrolled in this study. Speech outcomes, i.e., nasal emission, velopharyngeal insufficiency, and malarticulation after one-stage pushback (PB), one-stage modified Furlow (MF), or conventional two-stage MF palatoplasty, were evaluated at 4 (before intensive speech therapy) and 8 (after closure of oronasal fistula/unclosed hard palate) years of age.

**RESULTS:** Velopharyngeal insufficiency at 4 (and 8) years of age was present in 5.9% (0.0%), 0.0% (0.0%), and 10.0% (10.0%) of patients who underwent one-stage PB, one-stage MF, or two-stage MF palatoplasty, respectively. No significant differences in velopharyngeal function were found among these three groups at 4 and 8 years of age.

Malarticulation at 4 years of age was found in 35.3%, 10.0%, and 63.6% of patients who underwent one-stage PB, one-stage MF, and two-stage MF palatoplasty, respectively. Malarticulation at 4 years of age was significantly related to the presence of a fistula/unclosed hard palate (P<0.01). One-stage MF palatoplasty that was not associated with postoperative oronasal fistula (ONF) showed significantly better results than two-stage MF (P<0.01). Although the incidences of malarticulation at 8 years of age were decreased in each group compared to at 4 years of age, the incidence was still high in patients treated with two-stage MF (45.5%). On the whole, there was a significant correlation between ONF/unclosed hard palate at 4 years of age and malarticulation at 8 years of age (P<0.05).

**CONCLUSION:** Appropriate muscle sling formation can compensate for a lack of retropositioning of the palate for adequate velopharyngeal closure. Early closure of the whole palate and the absence of a palatal fistula were confirmed to be essential for normal speech. To avoid fistula formation, multilayer repair of the whole palate may be critical.

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**Publication type:** Journal Article

**Source:** MEDLINE

35. **Title:** Investigation of genetic factors underlying typical orofacial clefts: mutational screening and copy number variation.

**Citation:** Journal of Human Genetics, January 2015, vol./is. 60/1(17-25), 1434-5161;1435-232X (2015 Jan)

**Author(s):** Simioni M, Araujo TK, Monlleo IL, Maurer-Morelli CV, Gil-da-Silva-Lopes VL

**Language:** English

**Abstract:** Typical orofacial clefts (OFCs) comprise cleft lip, cleft palate and cleft lip and palate. The complex etiology has been postulated to involve chromosome rearrangements, gene mutations and environmental factors. A group of genes including IRF6, FOXL1, GLI2, MSX2, SKI, SATB2, MSX1 and FGF has been implicated in the etiology of OFCs. Recently, the role of the copy number variations (CNVs) has been studied in genetic defects and diseases. CNVs act by modifying gene expression, disrupting gene sequence or altering gene dosage. The aims of this study were to screen the above-mentioned genes and to investigate CNVs in patients with OFCs. The sample was composed of 23 unrelated individuals who were grouped according to phenotype (associated with other anomalies or isolated) and familial recurrence. New sequence variants in GLI2, MSX1 and FGF8 were detected in patients, but not in their parents, as well as in 200 control chromosomes, indicating that these were rare variants. CNV screening identified
new genes that can influence OFC pathogenesis, particularly highlighting TCEB3 and KIF7, that could be further analyzed. The findings of the present study suggest that the mechanism underlying CNV associated with sequence variants may play a role in the etiology of OFC.

**Publication type:** Journal Article  
**Source:** MEDLINE

**36. Title:** Limited incision with thorough elevation palatoplasty: technical evolution for superior results in cleft repair of the secondary palate.  
**Citation:** Annals of Plastic Surgery, February 2015, vol./is. 74/2(187-90), 0148-7043;1536-3708 (2015 Feb)  
**Author(s):** Baek RM, Koo YT, Kim BK  
**Language:** English  
**Abstract:** BACKGROUND: All kinds of palatoplasty emphasize elongating the soft palate and reconstructing the velar musculature without complication. We present the limited incision with thorough elevation (LITE) palatoplasty that leaves the anterior margin of the hard palate intact, achieving a fully movable bipedicled flap for complete closure and an adequate functioning velar muscular sling.METHODS: Fifty-six patients consecutively underwent the LITE palatoplasty. The patients were diagnosed with varying degrees of cleft of the secondary palate. The length of the soft palate was measured, preoperatively and postoperatively, to quantify the lengthening effect of the surgical procedure. The LITE palatoplasty lengthens the soft palate by full mobilization of the velar musculature and reconstruction of the muscles. The LITE palatoplasty also completely repairs the hard palate and leaves no raw surfaces, which can be disadvantageous to the maxillary growth.RESULTS: The average length of soft palate was 18.5 +/- 3.1 mm preoperatively, and the increased length of the soft palate was 5.06 +/- 2.41 mm (27.3 +/- 17.4%). There were no complications including fistula formation, hematoma, or wound problems. After 2 years of operation, only 2 patients who had multiple congenital problems showed grade 1 hypernasality in speech assessment.CONCLUSIONS: The LITE palatoplasty gives satisfactory results in elongating the soft palate and reconstructing a functional velar sling without leaving any raw surfaces that can be detrimental to healing and facial growth. And there was a better speech outcome without complications.  
**Publication type:** Journal Article  
**Source:** MEDLINE  
**Full text:** Available Ovid at Annals of Plastic Surgery

**37. Title:** Loss-of-function mutation in the X-linked TBX22 promoter disrupts an ETS-1 binding site and leads to cleft palate.  
**Citation:** Human Genetics, February 2015, vol./is. 134/2(147-58), 0340-6717;1432-1203 (2015 Feb)  
**Author(s):** Fu X, Cheng Y, Yuan J, Huang C, Cheng H, Zhou R  
**Language:** English  
**Abstract:** The cleft palate only (CPO) is a common congenital defect with complex etiology in humans. The molecular etiology of the CPO remains unknown. Here, we report a loss-of-function mutation in X-linked TBX22 gene (T-box 22) in a six-generation family of the CPO with obvious phenotypes of both cleft palate and hyper-nasal speech. We identify a functional -73G>A mutation in the promoter of TBX22, which is located at the core-binding site of transcription factor ETS-1 (v-ets avian erythroblastosis virus E26 oncogene homolog 1). Phylogenetic analysis showed that the sequence around the -73G>A mutation site is specific in primates. The mutation was detected in all five affected male members cosegregating with the affected phenotype and heterozygote occurred only in some unaffected females of the family, suggesting an X-linked transmission of the mutation in the family. The -73G>A variant is a novel single nucleotide mutation. Cell co-transfections indicated that ETS-1 could activate the TBX22 promoter. Moreover, EMSA and ChIP assays demonstrated that the allele A disrupts the binding site of ETS-1, thus markedly decreases the activity of the TBX22 promoter, which is likely to lead to the birth defect of the CPO without ankyloglossia. These results suggest that a loss-of-function mutation in the X-linked TBX22 promoter may cause the cleft palate through disruption of TBX22-ETS-1 pathway.  
**Publication type:** Journal Article  
**Source:** MEDLINE

**38. Title:** Objective measurements for grading the nasal esthetics on basal view in individuals with secondary cleft nasal deformity  
**Citation:** Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(66-69), 1055-6656;1545-1569 (01 Jan 2015)  
**Author(s):** He X., Li H., Shao Y., Shi B.  
**Language:** English
Abstract: Objective: The purpose of this study is to ascertain objective nasal measurements from the basal view that are predictive of nasal esthetics in individuals with secondary cleft nasal deformity. Design: Thirty-three patients who had undergone unilateral cleft lip repair were retrospectively reviewed in this study. The degree of nasal deformity was subjectively ranked by seven surgeons using standardized basal-view measurements. Nine physical objective parameters including angles and ratios were measured. Correlations and regressions between these objective and subjective measurements were then analyzed. Results: There was high concordance in subjective measurements by different surgeons (Kendall's harmonious coefficient=W=.825, P=.006). The strongest predictive factors for nasal aesthetics were the ratio of length of nasal alar (r=.370, P=.034) and the degree of deviation of the columnar axis (r = .451, P = .008). The columellar angle had a more powerful effect in rating nasal esthetics. Conclusion: There was reliable concordance in subjective ranking of nasal esthetics by surgeons. Measurement of the columnar angle may serve as an independent, objective predictor of esthetics of the nose.

Publication type: Journal: Article
Source: EMBASE
Full text: Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

39. Title: Otitis media with effusion: Experiences of children with cleft palate and their parents
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(23-30), 1055-6656;1545-1569 (01 Jan 2015)
Author(s): Tierney S., O'Brien K., Harman N.L., Sharma R.K., Madden C., Callery P.
Language: English
Abstract: Objective: To explore the views of children with cleft palate and their parents about daily life with otitis media with effusion and associated hearing loss. Design: A qualitative study. Semi-structured interviews were used to collect data from parents. Participatory techniques, including activities on a tablet computer, were used to collect data from children. Framework analysis was applied to interview transcripts. Setting: Two English cleft units. Participants: A purposive sample of parents of 37 children aged 0 to 11 years with experience of otitis media with effusion. Their children also took part if aged 6 to 11 years (n = 22). Results: Themes related to the following: (1) emotions (frustration, anger, sadness, happiness, anxiety), (2) educational experiences (struggling at school, having to sit at the front of the class, requiring extra support, missing lessons for appointments or due to ear infections), (3) social interactions (isolation, communication, reliance on siblings, participation in activities). Conclusions: A number of areas of interviewees' everyday life were affected by the presence of otitis media with effusion. Parents may need to be forewarned about the possible ongoing nature of this condition and its impact on a child's social and emotional experiences. Children may also benefit from age-appropriate information about otitis media with effusion and its treatment, including information on hearing tests, to help reduce any anxiety.

Publication type: Journal: Article
Source: EMBASE
Full text: Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

40. Title: Parenting a child with a cleft: The father's perspective
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(31-43), 1055-6656;1545-1569 (01 Jan 2015)
Author(s): Stock N.M., Rumsey N.
Language: English
Abstract: Objective: To explore the impact of having a child born with a cleft lip and/or palate from the father's perspective. Design: Individual qualitative telephone interviews. Participants: A total of 15 fathers of children born with cleft lip and/or palate were recruited throughout the U.K. via advertisements. Results: Supported by a number of subthemes, four overarching themes were identified: variations in care and support; appraisals of the cleft; perceptions of treatment; and looking back and moving forward. Conclusions: Fathers reported experiences comparable to those previously reported by mothers, in addition to a number of further support and information needs. Participants played a key role in supporting their families through the treatment process, yet fathers are under-represented in the research literature. Recommendations are made for the adequate inclusion of fathers in future research and in relation to methods of support for fathers through their children's diagnosis and treatment.

Publication type: Journal: Article
Source: EMBASE
Full text: Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

41. Title: Perceptions of team members working in cleft services in the United Kingdom: A pilot study
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1[e1-e7], 1055-6656;1545-1569 (01 Jan 2015)
Language: English
Abstract: Background: Cleft care provision in the United Kingdom has been centralized over the past 15 years to improve outcomes for children born with cleft lip and palate. However, to date, there have been no investigations to examine how well these multidisciplinary teams are performing. Methods: In this pilot study, a cross-sectional questionnaire surveyed members of all health care specialties working to provide cleft care in 11 services across the United Kingdom. Team members were asked to complete the Team Work Assessment (TWA) to investigate perceptions of team working in cleft services. The TWA comprises 55 items measuring seven constructs: team foundation, function, performance and skills, team climate and atmosphere, team leadership, and team identity; individual constructs were also aggregated to provide an overall TWA score. Items were measured using five-point Likert-type scales and were converted into percentage agreement for analysis. Results: Responses were received from members of every cleft team. Ninety-nine of 138 cleft team questionnaires (71.7%) were returned and analyzed. The median (interquartile range) percentage of maximum possible score across teams was 75.5% (70.8, 88.2) for the sum of all items. Team performance and team identity were viewed most positively, with 82.0% (75.0, 88.2) and 88.4% (82.2, 91.4), respectively. Team foundation and leadership were viewed least positively with 79.0% (72.6, 84.6) and 76.6% (70.6, 85.4), respectively. Conclusions: Cleft team members perceive that their teams work well, but there are variations in response according to construct.
Publication type: Journal: Article
Source: EMBASE
Full text: Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

42. Title: Piezosurgery: A new and safe technique for distraction osteogenesis in Pierre Robin sequence review of the literature and case report.
Citation: International Journal of Surgery Case Reports, 2015, vol./is. 6C/(269-72), 2210-2612;2210-2612 (2015)
Author(s): Galie M, Candotto V, Elia G, Clauser LC
Language: English
Abstract: INTRODUCTION: Pierre Robin sequence (PRS) is characterized by microgenia and retrognathia. Cleft palate and glossoptosis are frequently associated with airway obstruction and difficulty in swallowing. Distraction osteogenesis with micro-distractors has recently been considered as a surgical option during the neonatal age.CASE PRESENTATION: A 6-week-old female with PRS underwent mandibular lengthening in neonatal age. Mandibular osteotomies were performed with the piezoelectric scalpel.DISCUSSION: Piezosurgery represents an innovative technique as it offers the maxillofacial surgeon the opportunity to make precise bone cuts without damaging the soft tissue, minimizing the invasiveness of the surgical procedure, and the opportunity of working in a field which is almost totally blood free.CONCLUSION: The use of a piezoelectric device to perform this kind of surgery provides clinical and surgical results which would be difficult with traditional instruments, not only for the patient’s benefit but also for the surgeon’s. Preservation of the original bony structure, especially of the cancellous bone, will benefit the bone healing process due to its high estrogenic potential.Copyright 2014 The Authors. Published by Elsevier Ltd.. All rights reserved.
Publication type: Journal Article
Source: MEDLINE

43. Title: Presurgical nasoalveolar remodeling - an experience in the journey of cleft lip and palate.
Citation: Clinical Cosmetic & Investigational Dentistry, 2015, vol./is. 7/(1-7), 1179-1357;1179-1357 (2015)
Author(s): Mandwe RS, Puri S, Shingane S, Pawar G, Kolhe VR, Alsi A
Language: English
Abstract: AIMS AND OBJECTIVES: To assess the effect of presurgical nasoalveolar molding (PNAM) therapy in the management of patients with nonsyndromic unilateral cleft lip and palate (UCLP).MATERIAL AND METHOD: Ten patients with UCLP treated from 2009 to 2012. The initiation for PNAM treatment was 7 days and the average time of the treatment was 175 days. Measurements on patients and of casts were made, and statistical analysis was used to evaluate the changes in pre- and posttreatment measurements.RESULTS: Subsequent to PNAM treatment, there was a statistically considerable rise in cleft nostril height and columellar width. There was reduction in both intraoral cleft width and columellar deviation, which was significant statistically.CONCLUSION: PNAM treatment reduces alveolar cleft width. It enhances symmetry of the nose by changing columellar angulation, preserving alar width bilaterally, gaining height of the nostril on the affected side, and increasing columellar length.
Publication type: Journal Article
44. Title: Prevalence of non-syndromic orofacial clefts among jews and arabs, By type, Site, Gender and geography: A multi-center study in israel

Citation: Israel Medical Association Journal, December 2014, vol./is. 16/12(759-763), 1565-1088 (01 Dec 2014)

Author(s): Shapira Y., Shpack N., Blum I., Haklai Z., Amitai Y.

Language: English

Abstract: Background: Orofacial clefts are the most common craniofacial congenital malformations, with significant anatomic, ethnic, racial and gender differences. Objectives: To investigate the prevalence, distribution and characteristic features of various types of non-syndromic clefts among Israeli Jews and Arabs. Methods: We conducted a retrospective multi-center survey in 13 major hospitals in Israel for the period 1993-2005. To obtain the true prevalence and detailed clinical characteristics, data on liveborn infants with non-syndromic clefts were obtained from the Ministry of Health's National Birth Defect Registry and completed by chart reviews in the 13 surveyed hospitals. Results: Of 976, 578 liveborn infants, 684 presented unilateral or bilateral clefts, with a prevalence of 7.00/10,000 live births; 479 were Jews and 205 were Arabs. The prevalence was higher among Arabs compared to Jews (11.12 and 6.22 per 10,000 live births in Arabs and Jews, respectively, P < 0.00001). Males had higher cleft rates than females (7.69/10,000 and 6.17/10,000 live births, respectively, P = 0.05). Males had more cleft lips (P < 0.05) and cleft lips with cleft palate (P < 0.001). There was left-side predominance. Newborns of younger mothers (age < 20 years) and of older mothers (age > 45 years) had higher cleft rates than those with mothers in the 20-44 year bracket (P < 0.009). Children born at or above the 5th birth order had a higher cleft rate (P < 0.001). Conclusions: The prevalence of non-syndromic clefts was 7.00/10,000 live births. The markedly higher rate in Arabs is related to the high rate of consanguinity. Both very young and old maternal age represents a higher risk of clefts in their offspring.

Publication type: Journal: Article
Source: EMBASE

45. Title: Psychiatric problems in mothers of children with cleft lip and palate(CLP) using standarized interviews

Citation: Indian Journal of Psychiatry, January 2015, vol./is. 57/5 SUPPL. 1(S49-S50), 0019-5545 (January 2015)

Author(s): Gajarao H.M., Podaralla R.

Language: English

Abstract: Background and Aims: The present study was conducted with an aim to determine the Psychological status of mothers of Cleft lip and palate children and to study the change in Quality of Life across different sociodemographic variables using standardized interviews. The subjects were randomly assigned and after initial interview, sociodemographic data, education of mothers, employment status, living situations, social support, sex and age of the child, order of the child, type of pregnancy and Family History of CLP were collected using an intake proforma. Settings and Design: Plastic Surgery OPD in Owaisi Hospital and Research Centre affiliated to Deccan College of Medical Sciences, Kanchanbagh, Hyderabad. Materials and Methods: In total 60 subjects were included and interviewed using intake proforma and structured questionnaires (GHQ, HAM-A, HAM-D and WHOQOL Brief). The data was analysed and following conclusions were drawn. Results and Conclusions: 55% of the mothers in the study sample have SEVERE Anxiety and 35% have MODERATE Anxiety and 10%(n=6) were having MILD Anxiety. 36.7%(n=22) of mothers in the study have VERY SEVERE Depression, 18.3% (n=11) of mothers have SEVERE Depression and 30% (n=18) of them have MODERATE Depression and 15%(n=9) have MILD Depression. Depression was higher among the mothers those who are unemployed when compared to that of the employed mothers. Order of the child has a significant impact on the Psychological status of mothers. Higher Anxiety and Depression was found in the 1st order children when compared to that of the 2nd and further order children. Anxiety and depression was higher in mothers if the CLP child is a female when compared to that of the male CLP child. Depression and Anxiety was higher in mothers if the child's age is less than 3months when compared to older children.

Publication type: Journal: Conference Abstract
Source: EMBASE

Full text: Available ProQuest at Indian Journal of Psychiatry

46. Title: Spectral features and perceptual judgment of place of affricate in Putonghua-speaking pre-adolescents with normal and cleft palate.

Citation: International Journal of Pediatric Otorhinolaryngology, February 2015, vol./is. 79/2(179-85), 0165-5876;1872-8464 (2015 Feb)

Author(s): Jiang C, Whitehill TL, McPherson B, Ng ML

Language: English
Abstract: PURPOSE: The aim of the study was to examine the relationship between the spectral features and perceptual judgments of places of affricate in Putonghua (standard Mandarin Chinese), and to explore the possible contribution of different spectral moments on correct perception of place of articulation information by typical pre-adolescent speakers and those with cleft palate.METHOD: A total of 139 affricates produced by 10 normal pre-adolescent speakers and 14 speakers with repaired cleft palate (distorted articulation) were presented to 12 listeners tasked with making a judgment of the accuracy of place of articulation using a visual analog scale.RESULTS: Statistical analysis showed a significant relationship between the third spectral moment (L3) and listeners' perceptual judgment of typical alveolar and retroflex affricates. For productions by pre-adolescents with cleft palate, the first spectral moment (M1) was significantly correlated with listener perceptual scaling of retroflex affricates, but not for the alveolar affricates.CONCLUSION: Spectral features were associated with the perception of place of affricate in Putonghua. Different spectral moment characteristics might be used by listeners when perceiving speech from individuals with cleft palate.Copyright 2014 Elsevier Ireland Ltd. All rights reserved.

Publication type: Journal Article
Source: MEDLINE

47. Title: Spelling processes of children with nonsyndromic cleft lip and/or palate: A preliminary study
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(70-81), 1055-6656;1545-1569 (01 Jan 2015)
Author(s): Lee K.S.M., Young S.E.-L., Liow S.J.R., Purcell A.A.
Language: English
Abstract: Objective: To compare the cognitive-linguistic processes underlying spelling performance of children with cleft lip and/or palate with those of typically developing children. Design: An assessment battery including tests of hearing, articulation, verbal short-term and working memory, and phonological awareness, as well as word and nonword spelling, was administered to both groups. Participants: A total of 15 children with nonsyndromic cleft lip and/or palate were casematched by age and sex to 15 typically developing children. The children were aged between 6 and 8 years and were bilingual, with English the dominant language. Results: Wilcoxon signed-rank tests revealed that the performance of children with cleft lip and/or palate was significantly poorer on phoneme deletion and nonword spelling (P < .05) compared with typically developing children. Spearman correlation analyses revealed different relationships between the cognitive-linguistic and spelling measures for the cleft lip and/or palate and typically developing groups. Conclusions: Children with cleft lip and/or palate underachieve in phonological awareness and spelling skills. To facilitate early intervention for literacy problems, speech-language pathologists should routinely assess the cognitive-linguistic processing of children with cleft lip and/or palate, especially phonological awareness, as part of their case management protocols.
Publication type: Journal: Article
Source: EMBASE
Full text: Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

48. Title: Syngnathia and obstructive apnea in a case of popliteal pterygium syndrome.
Citation: European Journal of Pediatrics, December 2014, vol./is. 173/12(1741-4), 0340-6199;1432-1076 (2014 Dec)
Author(s): Posey JE, Dariya V, Edmonds JL, Lee EI, Probst FJ, Premkumar MH
Language: English
Abstract: UNLABELLED: We describe an infant with popliteal pterygia, syngnathia, cleft lip and palate, and retrognathia diagnosed with popliteal pterygium syndrome (PPS). The neonatal course was complicated by severe obstructive apnea necessitating tracheostomy.CONCLUSION: This report illustrates the potential for airway compromise in PPS patients and the need for thorough neonatal airway assessment.
Publication type: Journal Article, Research Support, N.I.H., Extramural
Source: MEDLINE

49. Title: Tak1, Smad4 and Trim33 redundantly mediate TGF-beta3 signaling during palate development.
Citation: Developmental Biology, February 2015, vol./is. 398/2(231-41), 0012-1606;1095-564X (2015 Feb 15)
Author(s): Lane J, Yumoto K, Azhar M, Ninomiya-Tsuji J, Inagaki M, Hu Y, Deng CX, Kim J, Mishina Y, Kaartinen V
Language: English
Abstract: Transforming growth factor-beta3 (TGF-beta3) plays a critical role in palatal epithelial cells by inducing palatal epithelial fusion, failure of which results in cleft palate, one of the most common birth defects in humans. Recent studies have shown that Smad-dependent and Smad-independent pathways work redundantly to transduce TGF-beta3 signaling in palatal epithelial cells. However, detailed mechanisms by which this signaling is mediated still remain to be elucidated. Here we show that TGF-beta-activated kinase-1 (Tak1) and Smad4 interact genetically in
palatal epithelial fusion. While simultaneous abrogation of both Tak1 and Smad4 in palatal epithelial cells resulted in characteristic defects in the anterior and posterior secondary palate, these phenotypes were less severe than those seen in the corresponding Tgfb3 mutants. Moreover, our results demonstrate that Trim33, a novel chromatin reader and regulator of TGF-beta signaling, cooperates with Smad4 during palatogenesis. Unlike the epithelium-specific Smad4 mutants, epithelium-specific Tak1:Smad4- and Trim33:Smad4-double mutants display reduced expression of Mmp13 in palatal medial edge epithelial cells, suggesting that both of these redundant mechanisms are required for appropriate TGF-beta signal transduction. Moreover, we show that inactivation of Tak1 in Trim33:Smad4 double conditional knockouts leads to the palatal phenotypes which are identical to those seen in epithelium-specific Tgfb3 mutants. To conclude, our data reveal added complexity in TGF-beta signaling during palatogenesis and demonstrate that functionally redundant pathways involving Smad4, Tak1 and Trim33 regulate palatal epithelial fusion. Copyright 2014 Elsevier Inc. All rights reserved.

50. Title: Targeted mutations of genes reveal important roles in palatal development in mice. 
Citation: Annals of Plastic Surgery, February 2015, vol./is. 74/2(263-8), 0148-7043;1536-3708 (2015 Feb)
Author(s): Ma L, Shi B, Zheng Q
Language: English
Abstract: The process of palatal development is regulated by growth factors, extracellular matrix (ECM) protein, and cell adhesion molecules, of which disturbance may result in cleft palate. Knockout mice are important animal models for studying the role of genes during palatal development. Therefore, in this review, we will describe genes knockout in mice to reveal the biological mechanisms of these genes in the formation of the cleft palate.

51. Title: Tfap2a-dependent changes in mouse facial morphology result in clefting that can be ameliorated by a reduction in Fgf8 gene dosage
Citation: DMM Disease Models and Mechanisms, January 2015, vol./is. 8/1(31-43), 1754-8403;1754-8411 (01 Jan 2015)
Language: English
Abstract: Failure of facial prominence fusion causes cleft lip and palate (CL/P), a common human birth defect. Several potential mechanisms can be envisioned that would result in CL/P, including failure of prominence growth and/or alignment as well as a failure of fusion of the juxtaposed epithelial seams. Here, using geometric morphometrics, we analyzed facial outgrowth and shape change over time in a novel mouse model exhibiting fully penetrant bilateral CL/P. This robust model is based upon mutations in Tfap2a, the gene encoding transcription factor AP-2alpha, which has been implicated in both syndromic and non-syndromic human CL/P. Our findings indicate that aberrant morphology and subsequent misalignment of the facial prominences underlies the inability of the mutant prominences to fuse. Exencephaly also occurred in some of the Tfap2a mutants and we observed additional morphometric differences that indicate an influence of neural tube closure defects on facial shape. Molecular analysis of the CL/P model indicates that Fgf signaling is misregulated in the face, and that reducing Fgf8 gene dosage can attenuate the clefting pathology by generating compensatory changes. Furthermore, mutations in either Tfap2a or Fgf8 increase variance in facial shape, but the combination of these mutations restores variance to normal levels. The alterations in variance provide a potential mechanistic link between clefting and the evolution and diversity of facial morphology. Overall, our findings suggest that CL/P can result from small gene-expression changes that alter the shape of the facial prominences and uncouple their coordinated morphogenesis, which is necessary for normal fusion.

52. Title: The differential influence of vowels and palatal covering on nasalance scores
Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(82-87), 1055-6656;1545-1569 (01 Jan 2015)
53. Title: The furlow palatoplasty for velopharyngeal dysfunction: Velopharyngeal changes, speech improvements, and where they intersect

Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(12-22), 1055-6656;1545-1569 (01 Jan 2015)

Author(s): Pet M.A., Marty-Grames L., Blount-Stahl M., Saltzman B.S., Molter D.W., Woo A.S.

Abstract: Objective: We investigated how Furlow palatoplasty changes velopharyngeal morphology and speech characteristics, as well as how the anatomical and clinical results might be related. We hypothesized that Furlow palatoplasty would result in measurable velar elongation, tightening of the genu angle, and retropositioning of the levator sling and that the achievement of these modifications might be associated with clinical speech improvement. Design: Retrospective analysis of preoperative and postoperative videofluoroscopic and speech data. Setting: Tertiary care center. Patients/Participants: A total of 29 patients with velopharyngeal insufficiency in the setting of previous cleft palate repair or submucous cleft palate. Interventions: Furlow palatoplasty for treatment of velopharyngeal insufficiency. Outcome Measures: Lateral videofluoroscopy and perceptual speech examination were conducted preoperatively and postoperatively in order to measure velopharyngeal dimensions and speech quality. We describe anatomical and speech changes associated with the Furlow palatoplasty and undertake an exploratory analysis of the relationship between surgical changes to the velopharynx and clinical outcomes. Results: Furlow palatoplasty results in significant velar elongation, increased acuity of the genu angle, and retropositioning of the levator sling. Postoperative speech improvement was identified on the three subscales of resonance, nasal emission, and stops/plosives. Speech improvement and the absence of need for reoperation were most consistently associated with tightening of the genu angle. Conclusions: Furlow palatoplasty lengthens the palate, while both tightening and retropositioning the levator sling. These changes reflect transverse recruitment of lateral velar tissues, along with transverse tightening and anterior release of the muscle fibers, respectively. Levator tightening is most consistently associated with improved speech outcomes.

Publication type: Journal: Article
Source: EMBASE
Full text: Available The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association at Cleft Palate-Craniofacial Journal

54. Title: The need for orthognathic surgery in nonsyndromic patients with repaired isolated cleft palate

Citation: Cleft Palate-Craniofacial Journal, January 2015, vol./is. 52/1(e8-e13), 1055-6656;1545-1569 (01 Jan 2015)

Author(s): Antonarakis G.S., Watts G., Daskalogiannakis J.

Abstract: Objective: To determine the frequency of need for orthognathic surgery among nonsyndromic patients with isolated cleft palate repaired during infancy at The Hospital for Sick Children in Toronto, Canada. Design: Retrospective cohort study. Patients: Patients with nonsyndromic isolated cleft palate born between 1970 and 1997 with available records including a lateral cephalometric radiograph taken at 15 years of age. Methods: Patients who had undergone or were being prepared for orthognathic surgery were automatically counted as requiring surgery. For the remaining patients, lateral cephalometric radiographs were traced and analyzed. Arbitrarily set
Tongue-palate contact during selected vowels in children with speech sound disorders.

Lee, Alice, Gibbon, Fiona E., Kearney, Elaine, Murphy, Doris

English

Abstract: There is evidence that complete tongue-palate contact across the palate during production of vowels can be observed in some children with speech disorders associated with cleft palate in the English-speaking and Japanese-speaking populations. Although it has been shown that this is not a feature of typical vowel articulation in English-speaking adults, tongue-palate contact during vowel production in typical children and English-speaking children with speech sound disorders (SSD) have not been reported in detail. Therefore, this study sought to determine whether complete tongue-palate contact occurs during production of five selected vowels in 10 children with SSD and eight typically-developing children. The results showed that none of the typical children had complete contact across the palate during any of the vowels. However, of the 119 vowels produced by the children with SSD, 24% showed complete contact across the palate during at least a portion of the vowel segment. The results from the typically-developing children suggest that complete tongue-palate contact is an atypical articulatory feature. However, the evidence suggests that this pattern occurs relatively frequently in children with SSD. Further research is needed to determine the prevalence, cause, and perceptual consequence of complete tongue-palate contact.
57. Title: Use of the buccal fat pad as free graft for closure of oronasal fistula in a cleft palate patient.
Citation: Journal of Craniofacial Surgery, January 2015, vol./is. 26/1(e14-6), 1049-2275;1536-3732 (2015 Jan)
Author(s): de Castro CH, Souza LN, Fernandes Santos Melo M
Language: English
Abstract: Oronasal fistulas are frequent complications after cleft lip and palate surgery, with difficult treatment because of the presence of fibrotic and scarred tissue as well as the absence of local virgin tissue, representing a challenge in oral and maxillofacial surgery. The size of the fistula, its location, and the cause of the defect are important factors to determine the type of treatment and surgical technique. The use of pedicled buccal fat pad (BFP) for the repair of cleft palate has shown promising results, becoming a safe and effective method. On the other hand, the use of BFP as a free graft for oral defects has been rarely described in the literature. The current study is the first case report that shows the use of free graft of BFP in oronasal fistula after cleft lip and palate surgery and aimed to discuss the promising results of this surgical technique, suggesting it as a treatment option for anterior maxillary defects, when properly indicated.
Publication type: Journal Article
Source: MEDLINE

58. Title: Using electronic tablet as a teaching tool for marking cleft lip repairs
Citation: Journal of Plastic, Reconstructive and Aesthetic Surgery, January 2015, vol./is. 68/1(122-123), 1748-6815;1878-0539 (01 Jan 2015)
Author(s): Deshpande G., Schonmeyr B.
Language: English
Publication type: Journal: Letter
Source: EMBASE
Full text: Available JOURNAL OF PLASTIC, RECONSTRUCTIVE & AESTHETIC SURGERY (formerly BRITISH JOURNAL OF PLASTIC SURGERY) at Journal of Plastic, Reconstructive and Aesthetic Surgery
Full text: Available JOURNAL OF PLASTIC, RECONSTRUCTIVE & AESTHETIC SURGERY (formerly BRITISH JOURNAL OF PLASTIC SURGERY) at Salisbury District Hospital Healthcare Library

59. Title: Verbal competence in narrative retelling in 5-year-olds with unilateral cleft lip and palate.
Citation: International Journal of Language & Communication Disorders, January 2015, vol./is. 50/1(119-28), 1368-2822;1460-6984 (2015 Jan)
Author(s): Klinto K, Salameh EK, Lohmander A
Language: English
Abstract: BACKGROUND: Research regarding expressive language performance in children born with cleft palate is sparse. The relationship between articulation/phonology and expressive language skills also needs to be further explored.AIMS: To investigate verbal competence in narrative retelling in 5-year-old children born with unilateral cleft lip and palate (UCLP) and its possible relationship with articulation/phonology at 3 and 5 years of age.METHODS & PROCEDURES: A total of 49 children, 29 with UCLP treated according to three different procedures for primary palatal surgery and a comparison group of 20 children (COMP), were included. Longitudinally recorded audio files were used for analysis. At ages 3 and 5, the children were presented with a single-word test of word naming and at age 5 also the Bus Story Test (BST). The BST was assessed according to a test manual. The single-word test was phonetically transcribed and the percentage of consonants correct adjusted for age (PCC-A) was calculated. Differences regarding the BST results within the UCLP group were analysed. The results were compared with the results of the COMP group, and also with norm values. In addition, the relationship between the results of the BST and the PCC-A scores at ages 3 and 5 years was analysed.OUTCOMES & RESULTS: No significant group differences or correlations were found. However, 65.5% of the children in the UCLP group had an information score below 1 standard deviation from the norm value compared with 30% in the COMP group.CONCLUSIONS: A larger proportion of children in the UCLP group than in the COMP group displayed problems with retelling but the differences between the two groups were not significant. There was no association between the BST results in the children with UCLP and previous or present articulatory/phonological competence. Since group size was small in both groups, the findings need to be verified in a larger study.Copyright 2014 Royal College of Speech and Language Therapists.
Publication type: Journal Article
Source: MEDLINE

60. Title: Verification of the therapeutic process in cleft patients.
Citation: Codas, December 2014, vol./is. 26/6(457-63), 2317-1782;2317-1782 (2014 Dec)
Author(s): Bautzer AP, Guedes ZC
Language: English, Portuguese

Abstract: PURPOSE: This study was conducted to verify the origin of a longer or even failed therapeutic process in patients with cleft lip and palate as to its difficulty. METHODS: Eighteen children undergoing therapeutic process were observed for at least 6 months and divided into two groups: presenting isolated cleft lip and palate (group I) and having been diagnosed by a Speech-Language Pathologist with reading and writing disorders, with manifestation of phonological awareness deficit (group II). Two tests were applied for the evaluation of speech and language: ABFW Language Test for Young Children (phonology) and Phonological Awareness: Instrument of Sequential Assessment (CONFIAS). RESULTS: Group I presented higher percentages in ABFW test than group II, except in the "simplification of consonant cluster" and "plosive devoicing" variables. It was also observed that, in the process of omission, group I hardly omits the vibrant consonant, as observed in group II. At the syllable level of CONFIAS, the percentages observed in group I tended to be higher than in group II, with the exception of the following tasks: "medial syllable," "production of rhyme," and "exclusion." At the phoneme level, the percentages observed in group II tended to be higher than in group I, with the exception of the following tasks: "starts with given sound," "exclusion," "synthesis," and "segmentation." No significant difference was observed between percentage distributions in groups I and II (p>0.118). CONCLUSIONS: The differences found between groups I and II, although not statistically significant, may suggest that the presence of malformation hinders speech and language acquisition and development and prolongs the therapeutic process if directive interventions are not carried out, including phonological awareness therapy.
Publication type: Journal Article
Source: MEDLINE

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