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**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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1. Title: 19q13.32 microdeletion syndrome: Three new cases.
2. Citation: European Journal of Medical Genetics, November 2014, vol./is. 57/11-12(654-8), 1769-7212;1878-0849 (2014 Nov-Dec)
4. Language: English
Abstract: A previous report described a unique phenotype associated with an apparently de novo 732 kb 19q13.32 microdeletion, consisting of intellectual disability, facial asymmetry, ptosis, oculomotor abnormalities, orofacial clefts, cardiac defects, scoliosis and chronic constipation. We report three unrelated patients with development delay and dysmorphic features, who were all found to have interstitial 19q13.32 microdeletions of varying sizes. Both the previously reported patient and our Patient 1 with a larger, 1.3-Mb deletion have distinctive dysmorphic features and medical problems, allowing us to define a recognizable 19q13.32 microdeletion syndrome. Patient 1 was hypotonic and dysmorphic at birth, with aplasia of the posterior corpus callosum, bilateral ptosis, oculomotor paralysis, down-slanting palpebral fissures, facial asymmetry, submucosal cleft palate, micrognathia, wide-spaced nipples, right-sided aortic arch, hypospadias, bilateral inguinal hernias, double toenail of the left second toe, partial 2-3 toe syndactyly, kyphoscoliosis and colonic atony. Therefore, the common features of the 19q13.32 microdeletion syndrome include facial asymmetry, ptosis, oculomotor paralysis, orofacial clefting, micrognathia, kyphoscoliosis, aortic defects and colonic atony. These findings are probably related to a deletion of some combination of the 20-23 genes in common between these two patients, especially NPS1, NAPA, ARHGAP35, SLC8A2, DHX34, MEIS3, and ZNF541. These candidate genes are expressed in the brain parenchyma, glia, heart, gastrointestinal tract and musculoskeletal system and likely play a fundamental role in the expression of this phenotype. This report delineates the phenotypic spectrum associated with the haploinsufficiency of genes found in 19q13.32. Copyright 2014 Elsevier Masson SAS. All rights reserved.

Publication type: Journal Article
Source: MEDLINE

Citation: Annals of Plastic Surgery, December 2014, vol./is. 73 Suppl 2/(S130-5), 0148-7043;1536-3708 (2014 Dec)
Author(s): Basta MN, Silvestre J, Stransky C, Solot C, Cohen M, McDonald-McGinn D, Zackai E, Kirschner R, Low DW, Randall P, LaRossa D, Jackson OA
Language: English
Abstract: BACKGROUND: Associated comorbidities can put syndromic patients with cleft palate at risk for poor speech outcomes. Reported rates of velopharyngeal insufficiency (VPI) vary from 8% to 64%, and need for secondary VPI surgery from 23% to 64%, with few studies providing long-term follow-up. The purpose of this study was to describe our institutional long-term experience with syndromic patients undergoing cleft palatoplasty.METHODS: A retrospective review was conducted of all patients with syndromic diagnoses undergoing primary Furlow palatoplasty from 1975 to 2011. Outcomes included postoperative oronasal fistula (ONF) and need for secondary VPI surgery. Speech scores for verbal patients 5 years or older were collected via the Pittsburgh scale for speech assessment. Aggregate scores categorized the velopharyngeal mechanism as competent, borderline, or incompetent. Outcomes were analyzed by patient and operative factors.RESULTS: One hundred thirty-two patients were included with average age at repair of 20.7 months. Cleft type was 9% submucosal, 16% Veau class I, 50% class II, 12% class III, and 13% class IV. Forty-five syndromes were recorded, most commonly Stickler syndrome (n = 32) and 22q11.2 deletion syndrome [22q11.2DS (n = 19)]. Forty-four patients also had associated Pierre Robin sequence (PRS). The overall ONF rate was 4.5% and was highest in Veau class IV clefts (P = 0.048). Seventy-six patients were included in speech analysis, with an average age at last assessment of 10.4 years. Overall, 60.5% of patients had a competent velopharyngeal mechanism, 23.7% borderline, and 15.8% incompetent mechanism. Fifty percent of 22q11.2DS patients had borderline speech and none had competent speech, compared to 73.3% with Stickler syndrome (P = 0.01) and 71.4% of patients with associated PRS (P = 0.02). Secondary VPI surgery was performed in 11.3% of patients overall. Patients with PRS (13.6%) and with Stickler syndrome (15.6%) had secondary VPI surgery, compared to 31.6% of patients with 22q11.2DS (P = 0.01).CONCLUSIONS: This study demonstrates low rates of postoperative ONF after modified Furlow palatoplasty in syndromic patients. Speech outcomes were comparable to nonsyndromic patients at our institution, but patients with 22q11.2DS consistently had borderline-incompetent speech and a 3-fold higher incidence of secondary VPI surgery.
Publication type: Journal Article
Source: MEDLINE
Full text: Available Ovid at Annals of Plastic Surgery

3. Title: A Subcutaneous Alar Base (SCAB) Flap to Restore Symmetry of the Ala in Primary Cleft-Lip Nose Repair.
Citation: Annals of Plastic Surgery, December 2014, vol./is. 73/6(652-8), 0148-7043;1536-3708 (2014 Dec)
Author(s): Sherif MM
Language: English
Abstract: Primary surgical correction of cleft-lip nose deformity is now well accepted. Despite various surgical techniques proposed, perfect nasal symmetry has not been persistently attained. The main reason is that the maxillary deficiency is frequently overlooked. This paper presents 62 cases of unilateral cleft lip repaired over the last 20 years by using the author’s modification of the Millard primary lip nose repair. Each of these patients had a minimum follow-up of 1 year (range 1-12 years). The technique consists of raising a small subcutaneous alar base flap (SCAB) from the nasolabial region. The flap is turned over like a book page to add bulk to the deficient maxilla and to elevate the depressed ala. It also
controls alar rotation and permits its permanent fixation to the anterior nasal spine. The results show that this modification has improved the alar contour and symmetry in patients undergoing primary cleft-lip nasal repair. There was minimal increase in the operating time and the final scar is similar to the original Millard technique.

**Publication type:** Journal Article  
**Source:** MEDLINE  
**Full text:** Available Ovid at Annals of Plastic Surgery  
**Full text:** Available Ovid at Annals of Plastic Surgery

4. Title: A novel case of unbalanced translocation of chromosome 3 and 7  
**Citation:** Archives of Disease in Childhood, October 2014, vol./is. 99/(A368-A369), 0003-9888 (October 2014)  
**Author(s):** Rao S., Hussien E., Lees M., Shastri A.  
**Language:** English  
**Abstract:** Background It is estimated that 1 in 500 to 1 in 625 human newborns have a balanced reciprocal chromosomal translocations. Such individuals are usually healthy and do not have any specific features. We report an unusual case of unbalanced translocation of chromosome 3 and 7 and describe its features. Casereport A female baby born at 37 weeks gestation by a spontaneous vaginal delivery was admitted to our NICU with poor Apgar scores. Immediately after birth, she had a poor ventilatory effort requiring continuous positive airway pressure (CPAP) and further deterioration in her respiratory function led to intubation and surfactant administration. Echocardiogram revealed small perimembranous VSD. She had dysmorphic features including micrognathia, broad occiput, low set ears, single palmar crease, and large cleft palate. Subsequent genetic tests confirmed unbalanced translocation of chromosome 3 and 7. She was gradually weaned off ventilator support at 2 weeks and was discharged from NICU. A week following discharge she presented with bronchiolitis and has continued need for High flow (Vapotherm) support. She feeds on high energy formula via a nasogastric tube due to poor weight gain and remains on treatment for moderate to severe reflux. She is likely to need gastrostomy and cleft surgery. Conclusion Dysmorphic features as reported in this case report should raise suspicion of a chromosomal defect, which needs early genetic referral and microarray. Balanced translocations are common and usually do not have specific clinical features. However unbalanced translocations are uncommon but they may have significant clinical expressions.  
**Publication type:** Journal: Conference Abstract  
**Source:** EMBASE  
**Full text:** Available ARCHIVES OF DISEASE IN CHILDHOOD at Archives of disease in childhood  
**Full text:** Available ARCHIVES OF DISEASE IN CHILDHOOD at Salisbury District Hospital Healthcare Library

5. Title: A novel non invasive assessment of velopharyngeal insufficiency in children with treated cleft palate  
**Citation:** British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e47), 0266-4356 (October 2014)  
**Author(s):** Gardner A., Ray A., Russell C., Crampin L., Campbell L., Wynne D., Devlin M.  
**Language:** English  
**Abstract:** Introduction: Children with treated cleft palate may develop velopharyngeal insufficiency (VPI). This may result in problems with speech and nasal regurgitation of food and/or liquids. Early intervention is advisable to maximise the potential for normal speech development. Currently we assess the presence of VPI using methods which are either subjective and/or invasive (or both) eg speech assessment, lateral video fluoroscopy and nasendoscopy. Spirometry is a non invasive, well tolerated, quantitative and objective investigation that measures a range of values including peak flow and forced expiratory volume. These are influenced by velopharyngeal competence. Child friendly interfaces have been developed for the assessment of many respiratory conditions. The use of these as an indirect measure of nasal air escape in cleft patients undergoing assessment for palatal function is described in this paper. Method: The pre and post surgical spirometry of cleft patients undergoing secondary surgery was recorded and we present the preliminary results. Conclusion: Spirometry is a well established, non invasive investigation tool in the paediatric population which may provide valuable objective information in young children with treated cleft palate as to the presence of VPI. We present a novel use for spirometry in the cleft population in an attempt to attain an objective, quantitative outcome measure for VPI surgery.  
**Publication type:** Journal: Conference Abstract  
**Source:** EMBASE

6. Title: Airway Compromise following Palatoplasty in Robin Sequence: Improving Safety and Predictability.  
**Citation:** Plastic & Reconstructive Surgery, December 2014, vol./is. 134/6(937e-45e), 0007-1226;1529-4242 (2014 Dec)  
**Author(s):** Costa MA, Murage KP, Tholpady SS, Flores RL  
**Language:** English  
**Abstract:** BACKGROUND: Prior studies report a high incidence of airway complications in patients with Robin sequence following palatoplasty. The authors’ institution uses polysomnography to assess risk of airway compromise before palatoplasty in Robin sequence. This study compares airway complications in Robin sequence to cleft palate only using this screening airway protocol and identifies risk factors for airway complications after palatoplasty.

METHODS: A 12-year
retrospective review of patients with Robin sequence undergoing palatoplasty was performed. Robin sequence patients were divided into nonoperative management and mandibular distraction osteogenesis subgroups. Preoperative variables including comorbidities were recorded. The primary outcome was postoperative airway complication, defined as reintubation, emergency room visit, or hospital admission within 3 months of palatoplasty. RESULTS: One hundred thirteen patients met inclusion criteria: polysomnography, 34.5 percent; Robin sequence, 65.5 percent; and Robin sequence treated with mandibular distraction osteogenesis, 30.1 percent. Screening polysomnography was used to indicate patients for palatoplasty or other airway interventions. The total airway complication rate was 7.1 percent; this was similar in Robin sequence (5.8 percent) and cleft palate only (7.7 percent). In isolated Robin sequence, the reintubation rate was 0 percent. Lower airway anomalies were associated with airway complications (p = 0.03). Significant variables for reintubation were cardiac (p = 0.046), gastrointestinal (p = 0.04), and lower airway anomalies (p = 0.025) and syndromic diagnosis (p = 0.05). CONCLUSION: Screening polysomnography can control airway complications following palatoplasty in Robin sequence patients to a rate that is comparable to that of patients with cleft palate only. CLINICAL QUESTION/LEVEL OF EVIDENCE: Risk, III.

Publication type: Journal Article
Source: MEDLINE
Full text: Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery
Full text: Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery

7.Title: Are children with cleft palate in Greater Glasgow & Clyde receiving optimal audiology follow-up Citation: British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e53), 0266-4356 (October 2014)
Author(s): Boyd D., Cunning N., McSparron S., Wynne D., Russel C., Devlin M.
Language: English
Abstract: Aim: To determine if children with cleft palate are receiving appropriate audiology follow up and intervention by the cleft palate and audiology multidisciplinary team in Greater Glasgow and Clyde (GG&C). Methods: Data was extracted from logbooks, of children born from 2009 to 2011 with cleft anomalies. Children who did not receive audiology followup by GG&C were excluded leaving a cohort of 154. Results: 37% of the final sample size were not offered Visual Reinforcement Audiometry (VRA) or there is no record of the test outcomes in health records. 33/43 children have not had a third VRA test. 7 children had received 2 previous VRAs and the rest were lost to follow up at earlier stages. 6 children had an average range of hearing out with the NICE guideline and one child had a 4 monthly check up. * 36/43 of the entire cohort had no 4th VRA. * 40/43 of the entire cohort had no 5th VRA. * No child had a total of 6 VRA's on record. Children with Pierre Robin syndrome and the association of cleft lip and palate with socio-economic group was similar to previously published studies. Conclusion: Children in the GG&C are not receiving the best audiology services to meet their care needs. Children are lost to follow up, adequate treatment is not received and treatment bias occurs where those who present with symptoms may receive optimum audiology follow up, allowing less severe hearing loss to be missed. This study highlights the need for consistent audiological follow up.

Publication type: Journal: Conference Abstract
Source: EMBASE

8.Title: Cepstral analysis of voice in children with velopharyngeal insufficiency after cleft palate surgery.
Citation: Journal of Voice, November 2014, vol./is. 28/6(789-92), 0892-1997;1873-4588 (2014 Nov)
Author(s): Yang Z, Fan J, Tian J, Liu L, Gan C, Chen W, Yin Z
Language: English
Abstract: OBJECTIVES: The purpose of this study is to survey the cepstral peak prominence (CPP) of vowel sounds and to compare objective data obtained from patients with velopharyngeal insufficiency after cleft palate surgery with objective data of those with normal healthy controls using acoustic analyzer. METHODS: Participants were divided into a clinical group and a control group. Every participant was recorded phonating the sustained vowel /a/. Each participant in the clinical group was recorded before surgery, before and after speech therapy. All samples were subjected to acoustic analysis using Praat software. The vowels were analyzed acoustically by the measurement of smoothest cepstral peak prominence (CPPs). RESULTS: The results reveal lower values of CPPs in speakers with velopharyngeal insufficiency before and after the operation. And, the results also reveal that there is no significant difference across the control and the clinical groups after speech therapy. CONCLUSIONS: The results reveal lower values of CPPs in the clinical group before surgery and before speech therapy in comparison with the control group, which could be explained because of the body's compensation for the lack of normal introraoral pressure and habit of articulation. Copyright 2014 The Voice Foundation. Published by Elsevier Inc. All rights reserved.

Publication type: Journal Article
Source: MEDLINE

9.Title: Changing strategy and implementation of a new treatment protocol for cleft palate surgery in "Maria Sklodowska Curie" (MSC) Children's Hospital, Bucharest, Romania.
Citation: Journal of Plastic Surgery and Hand Surgery, December 2014, vol./is. 48/6(356-61), 2000-6764;2000-6764 (2014
evaluated in the authors’ craniofacial center. Average age at adoption was 30.5 months (range, 5.0 to 95.0 months).

RESULTS: Between May of 1993 and August of 2010, 83 adopted children with cleft deformities were evaluated in the authors’ craniofacial center. The surgical outcomes of children adopted with cleft deformities.

METHODS: The authors performed a retrospective review of children who were repaired abroad, adopted children who underwent repair performed by the two senior surgeons in the United States at various ages and states of repair. The operative and perioperative needs of these children are poorly understood. This study attempts to characterize the preadoption history, the postadoption course, and surgical outcomes of children adopted with cleft deformities.

RESULTS: One hundred and sixty-eight patients with cleft palate were admitted, 89 isolated cleft palate (ICP), 53 unilateral (UCLP) and 26 bilateral (BCLP). In these, 228 surgical interventions were performed. Soft Palate Repair (SPR) and Hard Palate Repair (HPR) were performed with the DHPC procedure. The transfer to this technique was successfully performed in three steps: one team visit to Gothenburg by a surgeon from MSC and two visits by surgeons from Gothenburg to the MSC. Patients with SPR and HPR were operated on without major complications and there were no differences in results between Gothenburg surgeons and MSC surgeons. The interventions with SPR and HPR technique were proven to be easy to teach and learn and successfully performed without major complications. For cleft patients at MSC hospital it has meant earlier surgery, less re-operations and complications. This report shows a successful change of strategy for palatal repair with improved outcome regarding surgery. In future, speech and growth will be followed on a regular basis and will be compared with results from the Gothenburg Cleft Team.

Abstract:

Language: English

Author(s): Spataru R, Mark H

Citation: Plastic & Reconstructive Surgery, December 2014, vol./is. 134/6(1279-84), 0007-1226;1529-4242 (2014 Dec)

Title: Cleft care in international adoption.

Language: English

Abstract: BACKGROUND: Standards of cleft care abroad differ from those in the United States, particularly in less developed countries, where international adoption rates are high. Children adopted from these countries present to plastic surgeons in the United States at various ages and states of repair. The operative and perioperative needs of these children are poorly understood. This study attempts to characterize the preadoption history, the postadoption course, and surgical outcomes of children adopted with cleft deformities.

METHODS: The authors performed a retrospective review of all adopted cleft lip-cleft palate patients presenting to an academic craniofacial referral center and compared outcomes among adopted children who were repaired abroad, adopted children who underwent repair performed by the two senior authors (C.R.D. and S.B.B.), and children born in the United States who underwent repair performed by one of the senior authors (S.B.B.).

RESULTS: Between May of 1993 and August of 2010, 83 adopted children with cleft deformities were evaluated in the authors’ craniofacial center. Average age at adoption was 30.5 months (range, 5.0 to 95.0 months).
Comparing outcomes among adopted children repaired abroad, adopted children repaired by the senior authors, and children born in the United States who underwent repair in the United States, the authors found no statistically significant differences in lip revision rates, fistula rates, or velopharyngeal insufficiency. CONCLUSIONS: Adopted cleft patients constitute a complex and variable population with high rates of revision and delayed presentation. Internationally adopted children with orofacial clefts fared no better or worse after undergoing primary cleft repair abroad or in the United States. CLINICAL QUESTION/LEVEL OF EVIDENCE: Risk, IV.

**Publication type:** Journal Article  
**Source:** MEDLINE  
**Full text:** Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery

12. **Title:** Cleft repair in the elderly population—the South Wales experience  
**Citation:** British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e96), 0266-4356 (October 2014)  
**Author(s):** Colbert S., Extence H., Drake D.  
**Language:** English  
**Abstract:** Introduction/Aims: Surgical repair of cleft in the elderly population is controversial. Some cleft surgeons are of the opinion that the outcomes of repairing cleft in elderly patients are poor and as such, this group of patients should not be treated surgically. However, there is very little evidence in the medical literature published to support this. We report the outcomes of surgical repair of cleft in a series of elderly patients with un repaired cleft lip and or palate. Materials/Methods: We review the South Wales experience of treating elderly patients with unoperated cleft lip and or palate. We document the presence of speech impairment, behavioral problems, depression and low self esteem due to teasing about their facial appearance and ability to communicate. Results/Statistics: our results confirm an improvement in speech, quality of life and aesthetic outcomes after surgical repair of cleft in this series of elderly patients with un repaired cleft lip and or palate. Conclusions/Clinical Relevance: We advise the cleft team to consider surgical repair of the untreated cleft lip and or palate in the elderly population. Further research is needed to objectively detect the primary aesthetic needs of elderly patients with cleft lip and or palate, which along with the subjective needs defined by the patient, should determine the aim of the planned treatment interventions in this patient group.  
**Publication type:** Journal: Conference Abstract  
**Source:** EMBASE

13. **Title:** Comparison of the rotation-advancement and philtral ridge techniques for unilateral cleft lip repair.  
**Citation:** Plastic & Reconstructive Surgery, December 2014, vol./is. 134/6(1269-78), 0007-1226;1529-4242 (2014 Dec)  
**Author(s):** Cline JM, Oyer SL, Javidnia H, Nguyen SA, Sykes JM, Kline RM, Patel KG  
**Language:** English  
**Abstract:** BACKGROUND: The Millard rotation-advancement flap has become the most widely used technique for unilateral cleft lip repair. The philtral ridge repair is a modified straight-line technique that was developed to further optimize the scar associated with the rotation-advancement flap. The purpose of this article is to introduce the philtral ridge repair and objectively compare the outcomes of these two techniques. METHODS: Two senior board-certified surgeons, who are active members of their respective craniofacial teams, use different surgical techniques for the unilateral cleft lip: the philtral ridge and rotation-advancement repairs. The authors retrospectively analyzed preoperative and postoperative photographs of consecutive patients who underwent repair performed by each surgeon between 2003 and 2009. Using Adobe Photoshop imaging software, facial points on the cleft and noncleft sides were measured, including height and symmetry of Cupid’s bow, width and height of the nasal vestibule, height of the vermilion, and alar base position. Ratios of cleft side to noncleft side measurements were calculated to standardize comparisons between patients. In addition, the symmetry of each lip repair was graded subjectively by health care professionals and the general public. RESULTS: There were no differences in preoperative ratios between the two techniques with the exception of a wider cleft nasal vestibule in the rotation-advancement group (p = 0.04). There were no statistically significant differences in postoperative measures or subjective analysis of symmetry between the groups. CONCLUSION: Both the rotation-advancement and philtral ridge techniques produced outcomes with a high degree of facial symmetry and are excellent options for unilateral cleft lip repair. CLINICAL QUESTION/LEVEL OF EVIDENCE: Therapeutic, III.  
**Publication type:** Journal Article  
**Source:** MEDLINE  
**Full text:** Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery

14. **Title:** Consent for surgery to cleft lip & palate; fit for purpose and who does it best?  
**Citation:** British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e96-e97), 0266-4356 (October 2014)  
**Author(s):** Katsarelis H., Clancy R., Bragg T.W.H., Goodacre T.  
**Language:** English  
**Abstract:** Introduction and aims: The aim of this study was to explore how informed people are, and how much information
they retain. The process of consent for cleft lip and palate surgery (CLAP) generally requires a third party to consent on behalf of a minor. Studies have shown patients generally retain low levels of information relating to their procedure.

Methods: Data was prospectively collected via a questionnaire from consecutive patients undergoing a spectrum of procedures related to the management of CLAP. Results: 32 participants were included. Participants reported high levels of satisfaction (97%) with the consenting process and showed good level of knowledge (mean 66.24%, median 68.87%). Approximately 1/5 respondents recorded scores of 50% or less; suggesting a subgroup of individuals require additional support. This group displayed a bias towards wanting the provision of information via a specialist nurse. Conclusions: This study, contrasting others on consent, indicates that despite the complexity of the procedures, those giving consent retained very good levels of detailed knowledge. We failed to identify any link with gender, educational attainment, age, procedure and time since operation. The study does identify a subgroup of patients, who rely upon input from the specialist nurse to achieve robust consent.

Publication type: Journal: Conference Abstract
Source: EMBASE

15. Title: Ear disease following cleft lip and palate surgery without tympanostomy tube placement

Citation: Otolaryngology - Head and Neck Surgery (United States), September 2014, vol./is. 151/1 SUPPL. 1(P102), 0194-5998 (September 2014)

Author(s): Markey J.D., Maine R.G., Daniels K., Corlew D.S., Gregory G., Palacio H.

Language: English

Abstract: Objectives: (1) Describe the current benefits and risks associated with perioperative prophylactic myringotomy during cleft lip/palate surgery. (2) Recognize potential predictive factors associated with middle ear disease following cleft lip/palate surgery. (3) Incorporate ethnic differences into treatment algorithms regarding tympanostomy tubes. Methods: A total of 241 children (129 Ecuadorian, 112 Chinese) underwent cleft lip/palate repair (2000-2009). Veau classification, age, history of ear infections, and cleft side were recorded. Average age was 2.4 years and 11.1 years for Ecuadorian and Chinese children, respectively. No patients underwent tympanostomy tube placement. Following surgical correction, serial otoacoustic emissions (OAE) testing, and tympanometry were performed, and a parental questionnaire was administered regarding behavioral hearing deficits and history of ear infections before and after surgery. Data were recorded and compared individually for the 2 populations and as a group to identify disease prevalence and correlative factors. Results: No association existed between Veau classification and deficits in tympanometry, OAE, or subjective hearing. Reported ear infections after surgery were fewer than before but were not significant (26% to 21%). Abnormal OAE testing was associated with abnormal tympanograms and subjective hearing deficits (P < .0001 and P = .004). Ecuadorian children had higher number of ear infections pre- and postoperatively (P = .043 and P < .001) and higher number of abnormal tympanograms (P = .003). No significant difference existed regarding OAE testing. Conclusions: Severity of the cleft lip/palate is not a predictive factor of middle ear disease and hearing impairment when no tympanostomy tube is placed. Ideal pressure equalization tube protocols should incorporate ethnic differences.

Publication type: Journal: Conference Abstract
Source: EMBASE

16. Title: Effects of retinoic acid on proliferation and gene expression of cleft and non-cleft palatal keratinocytes.

Citation: European Journal of Orthodontics, December 2014, vol./is. 36/6(727-34), 0141-5387;1460-2210 (2014 Dec)


Language: English

Abstract: SUMMARY BACKGROUND: Retinoic acid (RA) is a key regulator of embryonic development and linked to several birth defects including cleft lip and palate (CLP). The aim was to investigate the effects of RA on proliferation and gene expression of human palatal keratinocytes (KCs) in vitro. METHODS: KCs from children with and without CLP were cultured with 2 and 5 muM RA. Proliferation was measured by quantification of DNA after 2, 4, 6, and 8 days. In addition, we analysed the effects of RA on messenger RNA expression of genes for proliferation, differentiation, apoptosis, and RA receptors. RESULTS: RA similarly inhibited proliferation of palatal KC from cleft and non-cleft subjects. The proliferation of KCs from cleft subjects was reduced to 59.8+13.4% (2 muM) and 41.5+14.0% (5 muM, Day 6), while that of cells from age-matched non-cleft subjects was reduced to 66.9+12.1% (2 muM) and 33.9+10.1% (5 muM). RA treatment reduced the expression of several of the investigated genes; the proliferating cell nuclear antigen (PCNA) was reduced in CLP KCs only. Keratins 10 and 16 were downregulated in keratinocytes from both cleft and non-cleft subjects. P63, a master regulator for epithelial differentiation, was only downregulated in KCs from cleft subjects, as was the RXRa receptor. Two P63 target genes (GJB6 and DLX5) were strongly downregulated by RA in all cell lines. None of the apoptosis genes was affected. CONCLUSION: Overall, RA similarly inhibits proliferation of palatal KCs from cleft and non-cleft subjects and reduces the expression of specific genes. The Author 2014. Published by Oxford University Press on behalf of the European Orthodontic Society. All rights reserved. For permissions, please email: journals.permissions@oup.com.

Publication type: Journal Article
Source: MEDLINE

Full text: Available Oxford University Press NHS Pilot 2014 (NESLi2) at The European Journal of Orthodontics
17. Title: Evidence-based medicine: unilateral cleft lip and nose repair.
Citation: Plastic & Reconstructive Surgery, December 2014, vol./is. 134/6(1372-80), 0007-1226;1529-4242 (2014 Dec)
Author(s): Greives MR, Camison L, Losee JE
Language: English
Abstract: LEARNING OBJECTIVES: After reading this article, the participant should be able to: 1. Describe the anatomical malformations found in unilateral cleft lip deformity. 2. Discuss current methods of measuring the deformity and subsequent outcomes. 3. Discuss preoperative assessments, workup, and the use of early interventions before definitive cheiloplasty (e.g., preoperative orthopedics, lip adhesion). 4. Discuss the different techniques used for cheiloplasty and nasal repair. 5. Discuss the use of postoperative splints, taping, or molding. 6. Discuss the outcomes and evidence of cleft lip repairs and identify areas for future research.SUMMARY: The Maintenance of Certification module series is designed to help clinicians structure their individualized course of study to specific areas appropriate to their clinical practice. This article was prepared to accompany practice-based assessment of preoperative evaluation, anesthesia, surgical treatment plan, perioperative management, and outcomes. In this format, the clinician is invited to compare his or her methods of patient assessment and treatment, outcomes, and complications, with authoritative, information-based references. This information base is then used for self-assessment and benchmarking in parts II and IV of the Maintenance of Certification process of the American Board of Plastic Surgery. This article is not intended to be an exhaustive treatise on the subject. Rather, it is designed to serve as a reference point for further in-depth study by review of the reference articles presented.
Publication type: Journal Article
Source: MEDLINE
Full text: Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery
Full text: Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery

18. Title: Facial soft-tissue thickness in patients affected by bilateral cleft lip and palate: A retrospective cone-beam computed tomography study.
Citation: American Journal of Orthodontics & Dentofacial Orthopedics, November 2014, vol./is. 146/5(573-8), 0889-5406;1097-6752 (2014 Nov)
Author(s): Celikoglu M, Buyuk SK, Sekerci AE, Ersoz M, Celik S, Sisman Y
Language: English
Abstract: INTRODUCTION: The purposes of this study were to evaluate the facial soft-tissue thicknesses and craniomaxillofacial morphologies of patients affected by bilateral cleft lip and palate (BCLP) and to compare the findings with a well-matched control group without any clefts using cone-beam computed tomography.METHODS: The study sample consisted of 40 retrospectively and randomly selected patients divided into 2 groups: a BCLP group (20 patients; mean age, 13.78 + 3.20 years) and an age- and sex-matched control group without clefts (20 patients; mean age, 13.94 + 2.52 years). Craniofacial and facial soft-tissue thickness measurements were made with cone-beam computed tomography. The Student t test and multiple linear regression analyses were performed for the statistical evaluations.RESULTS: The BCLP group had an increased SN-MP angle (P = 0.003), a decreased Co-A (P = 0.000), and retruded maxillary (P = 0.000) and mandibular (P = 0.026) incisors. In addition, patients affected by BCLP had statistically significantly decreased thickness measurements for the variables subnasale (P = 0.005) and labrale superior (P = 0.026) compared with the controls. The most predictive variables were found at U1-SN (r = 0.417, P = 0.004), IMPA (r = 0.368, P = 0.010), and ANB (r = -0.297, P = 0.031) for subnasale and U1-SN (r = 0.284, P = 0.038) for labrale superior.CONCLUSIONS: The BCLP group showed greater vertical growth, greater retraction of the maxilla and the maxillary and mandibular incisors, and decreased subnasale and labrale superior thicknesses compared with the well-matched controls without clefts. These differences should be taken into account when planning orthodontic and orthognathic surgery treatment for those patients. Copyright 2014 American Association of Orthodontists. Published by Elsevier Inc. All rights reserved.
Publication type: Journal Article
Source: MEDLINE

19. Title: Fast and early mandibular osteodistraction (FEMOD) in severe Pierre Robin Sequence
Citation: Journal of Cranio-Maxillofacial Surgery, October 2014, vol./is. 42/7(1364-1370), 1010-5182;1878-4119 (01 Oct 2014)
Author(s): Cascone P., Papoff P., Arangio P., Vellone V., Calafati V., Silvestri A.
Language: English
Abstract: Pierre Robin Sequence (PRS) is a congenital abnormality characterized by mandibular hypoplasia, glossoptosis and often secondary palate cleft. It may be an isolated or part of a most complicated syndrome. The genetic syndrome that most frequently co-occurs is Stickler syndrome characterized by skeletal abnormalities, joint pain, congenital myopia and retinal detachment. The authors describe their fast and early mandibular osteodistraction (FEMOD) protocol in severe cases of PRS airway obstruction.
Publication type: Journal: Article
Source: EMBASE
20. Title: Fistula rate after primary repair of cleft palate: Novel exposed inverted horizontal mattress nasal closure
Citation: Otolaryngology - Head and Neck Surgery (United States), September 2014, vol./is. 151/1 SUPPL. 1(P140), 0194-5998 (September 2014)
Author(s): Hyde B.J., Steffen C.M., Miller J.J.
Language: English
Abstract: Objectives: Discuss options for midline nasal suture during 2-flap palatoplasty (2FPP). Compare effects of patient demographics, Veau class, associated syndromes, surgeon experience, and use of a previously unreported exposed inverted horizontal mattress suture for nasal mucoperiosteal closure on fistula rates and velopharyngeal insufficiency (VPI). Methods: Retrospective case series review of all children who underwent 2FPP by the attending surgeon (J.J.M.). Fiftyfive consecutive patients were analyzed over a 5.5-year period. Study variables were obtained from multidisciplinary craniofacial/cleft records. All patients underwent 2FPP with intravelar veloplasty using the suture technique described herein. No patients were excluded from primary review and statistical analysis. Results: Eighty-nine percent of patients were Veau class 2 or 3. Twenty-four (44%) patients had an associated syndrome. Patients with a lower Veau class were 3.6 times more likely to have an associated syndrome (P = .0001). Two (3.6%) patients developed a fistula. There was no association between Veau class, presence of an associated syndrome, or use of dermal allograft with the development of a fistula (P = 1.0, all). Postoperative VPI was noted in 11 (20%) patients, without association between Veau class, syndrome, or dermal allografting (P = 1.0, .74, and .45, respectively). Repairs in the first half of the patient sample had a 10-fold increased risk of postoperative VPI (P =.0024). Conclusion: Our use of an exposed inverted horizontal mattress suture for nasal mucoperiosteal closure during 2FPP appears safe and is comparable with the lowest published rates of postoperative fistula and VPI. Further standardized studies are needed in this area.
Publication type: Journal: Conference Abstract
Source: EMBASE

21. Title: Grommets for otitis media with effusion in children with cleft palate: A systematic review
Citation: Pediatrics, November 2014, vol./is. 134/5(983-994), 0031-4005;1098-4275 (01 Nov 2014)
Language: English
Abstract: BACKGROUND AND OBJECTIVE: No consensus has yet been reached with regard to the link between otitis media with effusion (OME), hearing loss, and language development in children with cleft palate. The objective of this study was to address the effectiveness of ventilation tube insertion (VTI) for OME in children with cleft palate. METHODS: A dual review process was used to assess eligible studies drawn from PubMed, Medline via Ovid, Cumulative Index to Nursing and Allied Health Literature, Cochrane Library, and reference lists between 1948 and November 2013. Potentially relevant papers were selected according to the full text of the articles. Relevant data were extracted onto a data extraction sheet. RESULTS: Nine high- or moderate-quality cohort studies were included in this study. VTI was administered in 38% to 53% of the OME cases, and more severe cases appeared more likely to undergo VTI. Compared with conservative forms of management (eg, watchful waiting), VTI has been shown to be beneficial to the recovery of hearing in children with cleft palate and OME. A growing body of evidence demonstrates the benefits of VTI in the development of speech and language in children with cleft palate and OME. These children face a higher risk of complications than those undergoing conservative treatments, the most common of which are eardrum retraction and tympanosclerosis, with an incidence of ~11% to 37%. CONCLUSIONS: This review provides evidence-based information related to the selection of treatment for OME in children with cleft palate. Additional randomized controlled trials are required to obtain bias-resistant evidence capable of reliably guiding treatment decisions. The conclusions in this review are based on underpowered cohort studies and very-low-strength evidence.
Publication type: Journal: Review
Source: EMBASE
Full text: Available EBSCOhost EJS at Pediatrics

22. Title: Identifying the effect of cleft type, deprivation and ethnicity on speech and dental outcomes in UK cleft patients: A multi-centred study.
Citation: Journal of Plastic, Reconstructive & Aesthetic Surgery: JPRAS, December 2014, vol./is. 67/12(1637-43), 1748-6815;1878-0539 (2014 Dec)
Language: English
Abstract: BACKGROUND: Outcome measures are increasingly important in the modern National Health Service. In the care of children born with cleft lip and/or palate there are many different outcomes to consider but only a few reliable, validated outcome measures exist. The dmft (decayed, missing and filled teeth) index and cleft speech characteristics (CSCs) are used regularly by cleft teams throughout the UK to assess outcomes in children with cleft lip and/or palate. We hypothesized that these two outcome measures might be significantly influenced by the demographics of the populations studied independent of the care provided.METHODS: A retrospective review of all patients aged between five and six
referred to three regional cleft centres during a twelve month period were included in the study. Speech and dental outcomes were compared with patient ethnicity, cleft type and level of deprivation as determined by the Carstairs score.

RESULTS: The data of 287 patients were used. Speech was significantly affected by cleft type (p < 0.03), whereas dentition was significantly affected by ethnicity (p = 0.002) and deprivation (p = 0.012). CONCLUSIONS: This study demonstrates that the demographics of cleft populations can significantly affect the measures of outcome used to assess the quality of care provided by cleft teams. It has also demonstrated that these demographics are not evenly distributed across the country and that some cleft teams will have a more 'at risk' population than others. LEVEL OF EVIDENCE: Risk, level II. Copyright 2014 British Association of Plastic, Reconstructive and Aesthetic Surgeons. Published by Elsevier Ltd. All rights reserved.

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

**23. Title:** Immediate fat grafting in primary cleft lip repair.  
**Citation:** Journal of Plastic, Reconstructive & Aesthetic Surgery: JPRAS, December 2014, vol./is. 67/12(1644-50), 1748-6815;1878-0539 (2014 Dec)  
**Author(s):** Balkin DM, Samra S, Steinbacher DM  
**Language:** English  
**Abstract:** BACKGROUND: Successful cleft lip repair creates symmetric nasolabial morphology with minimal scar. Fat grafting is used in cosmetic and reconstructive settings to provide contour, condition tissue and aid healing. This study employs immediate fat grafting concurrent with primary cleft nasolabial repair. We hypothesize that simultaneous fat transfer is safe and may optimize the result. METHODS: This retrospective analysis included a series of consecutive infants who underwent primary cleft lip repair with immediate fat grafting. Demographic and peri-operative details were recorded. Post-operative photographs were analyzed by three blinded reviewers (Al-Omari et al. and Asher-McDade et al.). Kappa statistics were employed to assess inter-rater reliability (Randolph and Watkins MW). RESULTS: 30 children, 37 sides (13 left, 10 right, 7 bilateral; 62% complete, 38% incomplete) who underwent cleft lip repair at Yale were included. 20 underwent nasolabial repair with simultaneous fat grafting. Mean age of repair was 3.5 mo (range 1.5-6.4). Fat was hand suctioned from the thighs (15 left; 2 right; 3 both) with mean yield of 2.1 cc (range 1.5-5 cc). An average of 1.4 cc (range 0.5-2.5 cc) was injected to the philtrum, vermillion, piriform and ala. No complications were experienced with lip repair, fat harvest or graft injection. Mean follow-up was 24.7 months (range 12.4-60.2 months). Postoperative photographic assessment revealed minimal residual cleft stigmata with inter-rater reliability. Each ordinal score was statistically significant compared fat grafted repairs to those without fat grafting (p < 0.05). CONCLUSIONS: Simultaneous fat grafting and cleft lip repair can be performed safely. The augmentation and modulation of scar formation may optimize results. Prospective comparison is necessary to further corroborate our findings. LEVEL OF EVIDENCE: Therapeutic (Level IV). Copyright 2014 British Association of Plastic, Reconstructive and Aesthetic Surgeons. Published by Elsevier Ltd. All rights reserved.

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

**24. Title:** Incidence of palatal fistula formation after primary palatoplasty in northern Finland.  
**Citation:** Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, December 2014, vol./is. 118/6(632-6), 2212-4411 (2014 Dec)  
**Author(s):** Lithovius RH, Ylikontiola LP, Sandor GK  
**Language:** English  
**Abstract:** OBJECTIVE: The purpose of this retrospective study was to determine the incidence of palatal fistulas after primary cleft palate repair. STUDY DESIGN: The study included 136 patients who were treated at the Oulu University Hospital cleft lip and palate center between 1998 and 2011. All patients were treated by the same surgeons with 1-stage palatoplasty closing the hard and soft palate concurrently. RESULTS: The overall frequency of postoperative fistula was 9.6% of patients. Patients with cleft lip and palate (20.0%) were more likely to develop postoperative palatal fistulas than patients with cleft palate (6.6%). Surgical technique and cleft severity were not significant factors for the development of palatal fistulas. CONCLUSIONS: The majority of patients undergoing primary palatal repair do not develop palatal fistulas. Copyright 2014 Elsevier Inc. All rights reserved.  
**Publication type:** Journal Article
Citation: Annals of Plastic Surgery, December 2014, vol./is. 73 Suppl 2/($126-9), 0148-7043;1536-3708 (2014 Dec)
Author(s): Albert MG, Babchenko OO, Lalikos JF, Rothkopf DM
Language: English
Abstract: BACKGROUND: The lifetime cost of a child with an orofacial cleft is estimated at $101,000, which amounts to $697 million total for those born each year with orofacial clefts. There has been a trend toward outpatient procedures for cleft lip repair (CLR) and alveolar bone grafting (ABG), and studies have shown no disparities in safety or outcome between inpatient and ambulatory treatment. The financial implications of outpatient versus inpatient procedures have not been compared.METHODS: Financial data were collected for outpatient (n = 33) and inpatient (n = 2) CLR, as well as outpatient (n = 7) and inpatient (n = 5) ABG during a 5-year period at our institution. We examined hospital charges and reimbursement for these procedures by private insurance plans and Medicaid Managed Care (MMC) plans. RESULTS: The average total reimbursements for inpatient and outpatient CLR were similar at $6848 and $5557, respectively. Average facility reimbursement for CLR was greater for inpatient ($5344) than outpatient ($4291) procedures. Average professional reimbursement was similar between inpatient ($1504) and outpatient ($1266) CLR. For ABG, the average total inpatient reimbursement was $14573, whereas outpatient was $8877. Average facility reimbursements were greater for inpatient ($12398) than outpatient ($7183) ABG. Average professional reimbursement was similar between inpatient ($2175) and outpatient ($1693) ABG, with 35% and 31% of charges reimbursed, respectively. A substantial difference existed between reimbursements based on insurance types for both outpatient CLR and outpatient ABG. On average for CLR, commercial payers reimbursed 52% ($7344) of overall charges, whereas Medicaid and MMC reimbursed 9% ($1447). For ABG, commercial payers reimbursed an average of 78% ($11950) of overall charges, whereas Medicaid and MMC reimbursed 10% ($1192). CONCLUSIONS: Fewer patients’ insurance companies are reimbursing for inpatient stays; in many cases, even patients who remain hospitalized up to 48 hours are treated as “day surgery” from a reimbursement perspective. For outpatient surgery, a greater percentage of CLR and ABG charges were successfully recouped compared to inpatient surgery. Awareness of higher payment for inpatient surgery and potential savings through use of the outpatient setting is crucial for hospitals and the US health care system as a whole.
Publication type: Journal Article
Source: MEDLINE
Full text: Available Ovid at Annals of Plastic Surgery

26. Title: Intragenic duplication--A novel causative mechanism for SATB2-associated syndrome.
Citation: American Journal of Medical Genetics. Part A, December 2014, vol./is. 164/12(3083-7), 1552-4825;1552-4833 (2014 Dec)
Author(s): Lieden A, Kvarnun g M, Nilssson D, Sahlin E, Lundberg ES
Language: English
Abstract: Previous studies have shown that genetic aberrations involving the special AT-rich sequence-binding protein 2 (SATB2) gene result in a variable phenotype of syndromic intellectual disability. Although only a small number of patients have been described, there is already considerable variation in regard to the underlying molecular mechanism spanning from structural variation to point mutations. We here describe a male patient with intellectual disability, speech and language impairment, cleft palate, malformed teeth, and oligodontia. Array CGH analysis identified a small intragenic duplication in the SATB2 gene that included three coding exons. The result was confirmed by multiplex ligation-dependent probe amplification and low coverage whole genome mate pair sequencing. WGS breakpoint analysis directly confirmed the duplication as intragenic. This is the first reported patient with an intragenic duplication in SATB2 in combination with a phenotype that is highly similar to previously described patients with small deletions or point mutations of the same gene. Our findings expand the spectra of SATB2 mutations and confirm the presence of a distinct SATB2-phenotype with severe ID and speech impairment, cleft palate and/or high arched palate, and abnormalities of the teeth. For patients that present with this clinical picture, a high-resolution exon targeted array CGH and/or WGS, in addition to sequencing of SATB2, should be considered. 2014 Wiley Periodicals, Inc. 2014 Wiley Periodicals, Inc.
Publication type: Journal Article
Source: MEDLINE

27. Title: Lingual hamartoma associated with a cleft palate in a newborn.
Citation: Ear, Nose, & Throat Journal, October 2014, vol./is. 93/10-11(E9-E11), 0145-5613;1942-7522 (2014 Oct-Nov)
Author(s): Daramola OO, Suchi M, Chun RH
Language: English
Abstract: A hamartoma is a benign malformation of native tissue that may occur in any area of the body. Hamartoma of the tongue is a rare developmental lesion. We describe the case of a pendulant lingual hamartoma in a 2-day-old girl that had not been identified on prenatal ultrasonography. We also review the utility of prenatal imaging options, the role of
preoperative imaging, the mechanical relationship between lingual hamartoma and cleft palate, the histopathology of this tumor, surgical treatment, and emergency airway management.

**Publication type:** Journal Article  
**Source:** MEDLINE  
**Full text:** Available **ENT: Ear, Nose & Throat Journal** at **Ear, Nose and Throat Journal**

28. **Title:** Long-Term Comparison of the Results of Four Techniques Used for Bilateral Cleft Nose Repair: A Single Surgeon’s Experience.  
**Citation:** Plastic & Reconstructive Surgery, December 2014, vol./is. 134/6(926e-36e), 0007-1226;1529-4242 (2014 Dec)  
**Author(s):** Chang CS, Liao YF, Wallace CG, Chan FC, Liou EJ, Chen PK, Noordhoff MS  
**Language:** English  
**Abstract:** BACKGROUND: The purpose of this study was to evaluate progressive changes in surgical techniques and results, aiming for improved nasal shape in primary bilateral cleft rhinoplasty.METHODS: This is an institutional review board-approved retrospective study. Ninety-one consecutive patients with bilateral complete cleft lip underwent primary cheiloplasty with four different techniques of nasal reconstruction from 1992 to 2007 as follows: group I, primary rhinoplasty alone; group II, nasoalveolar molding alone; group III, nasoalveolar molding plus primary rhinoplasty; group IV, nasoalveolar molding plus primary rhinoplasty with overcorrection; and group V, patients without cleft lip. The surgical results were analyzed using photographic records obtained at age 3 years. Four measurements and one angle measurement were obtained. A panel assessment was obtained to grade the appearance of the surgical results.RESULTS: The results are expressed in order from groups I through V. The nostril height-to-width ratio was 0.49, 0.59, 0.62, 0.78, and 0.82, respectively. The nasal tip height-to-nasal width ratio was 0.29, 0.39, 0.49, 0.57, and 0.60. The columella height-to-nasal width ratio was 0.11, 0.18, 0.22, 0.27, and 0.28. The dome-to-columella ratio was 1.88, 1.25, 1.26, 1.14, and 1.10. The nostril area ratio was 1.2, 1.17, 1.13, 1.11, and 1.07. The nasolabial angle was 144.95, 143.98, 121.98, 120.99, and 100.88. Finally, group IV had the best panel assessment.CONCLUSIONS: The results revealed that group IV had the best overall result. Presurgical nasoalveolar molding followed by primary rhinoplasty with overcorrection resulted in a nasal appearance that was closer to the patients without cleft lip.CLINICAL QUESTION/LEVEL OF EVIDENCE: Therapeutic, III.  
**Publication type:** Journal Article  
**Source:** MEDLINE  
**Full text:** Available **Journal of reconstructive microsurgery** at **Plastic and Reconstructive Surgery**  
**Full text:** Available **Journal of reconstructive microsurgery** at **Plastic and Reconstructive Surgery**

29. **Title:** Morphologic variability of nonsyndromic operated patients affected by cleft lip and palate: A geometric morphometric study  
**Citation:** American Journal of Orthodontics and Dentofacial Orthopedics, September 2014, vol./is. 146/3(346-354), 0889-5406 (September 2014)  
**Author(s):** Toro-Ibáñez V., Cortes Araya J., Díaz Munoz A., Manriquez Soto G.  
**Language:** English  
**Abstract:** Introduction In this study, we compared patterns of morphologic variations of the craniofacial skeleton between patients affected by clefts who were operated on and unaffected subjects, aiming to discuss possible morpho-functional consequences of treatment in craniofacial development. Methods The lateral cephalograms of 76 subjects, comprising patients with operated unilateral cleft lip and palate (OpC) and a group matched for sex and age without cleft, were used. Thirteen landmarks were used as variables in geometric morphometric tests quantifying and describing overall shape variation, differences between group means, allometry, and upper-lower face covariation. Results The OpC group showed broader shape variations including noncleft group characteristics, but mainly a retrognathic maxilla, a vertically elongated face, a more open mandibular angle, and a more closed basicranial angle. Group means differed mainly in the maxillomandibular relationships. Allometry differed between groups, with the smallest OpC patients showing the most altered morphology. Upper and lower face covariation was stronger in the OpC group, showing mainly vertical changes in the anterior face. Conclusions Operated patients affected by clefts achieve a broad range of morphologies; the most altered were found in those with skeletal Class III and small size. Furthermore, their strongest upper and lower face shape covariation suggests that a harmonic dental occlusion could be a key factor in achieving “normal” craniofacial morphology. Copyright 2014 by the American Association of Orthodontists.  
**Publication type:** Journal: Article  
**Source:** EMBASE

30. **Title:** Normal emotion regulation in adults with cleft lip and palate: An exploratory study  
**Citation:** Journal of Cranio-Maxillofacial Surgery, October 2014, vol./is. 42/7(1271-1276), 1010-5182;1878-4119 (01 Oct 2014)  
**Author(s):** Gassling V., Kessler H., Klein M.O., Detjen A.-M., Koos B., Limbrecht-Ecklundt K., Traue H.C., Wiltfang J., Gerber W.-D.  
**Language:** English
Abstract: Introduction Cleft lip and palate (CLP) represent the most common congenital malformations of the midfacial region. Although these patients show differences in their facial appearance, we hypothesize that CLP-affected individuals do not show an alteration in their emotion regulation abilities compared to unaffected individuals. This is because of the strong biological basis of facial emotion and expression that is inherent and receives little influence from external factors.

Material and methods The present study evaluated various aspects of emotion regulation in 25 adults with CLP and an equally sized control group of unaffected volunteers. The study was divided into three parts. First, we investigated emotion regulation strategies. Here, each participant was asked to complete the Emotion Regulation Questionnaire (ERQ) and Ambivalence over Emotional Expressiveness Questionnaire G 18 (AEQ-G18). Second, we examined the recognition of facially expressed basic emotions (FEEL test). Third, we evaluated the expression of an emotion induced by an odor sample. Results Habitual emotion regulation, measured by ERQ and AEQ-G18, was not different between CLP and controls subjects for all of the sub-scales. Recognition of facially expressed basic emotions was also the same for both groups. Facial emotion encoding did not differ for both groups. Conclusions To summarize, the findings suggest that individuals with an orofacial cleft show undisturbed emotion regulation and recognition. This may be explained by the strong biological basis of facial emotion recognition and regulation as well as by the healthy emotional resilience and social functioning of CLP patients.

Publication type: Journal: Article
Source: EMBASE

31. Title: Novel interstitial deletion of 10q24.3-25.1 associated with multiple congenital anomalies including lobar holoprosencephaly, cleft lip and palate, and hypoplastic kidneys.
Citation: American Journal of Medical Genetics. Part A, December 2014, vol./is. 164/12(3132-6), 1552-4825;1552-4833 (2014 Dec)
Author(s): Peltekova IT, Hurteau-Millar J, Armour CM
Language: English
Abstract: Chromosome 10q deletions are rare and phenotypically diverse. Such deletions differ in length and occur in numerous regions on the long arm of chromosome 10, accounting for the wide clinical variability. Commonly reported findings include dysmorphic facial features, microcephaly, developmental delay, and genitourinary abnormalities. Here, we report on a female patient with a novel interstitial 5.54Mb deletion at 10q24.31-q25.1. This patient had findings in common with a previously reported patient with an overlapping deletion, including renal anomalies and an orofacial cleft, but also demonstrated lobar holoprosencephaly and a Dandy-Walker malformation, features which have not been previously reported with 10q deletions. An analysis of the region deleted in our patient showed numerous genes, such as KAZALD1, PAX2, SEMA4G, ACTRA1, INA, and FGF8, whose putative functions may have played a role in the phenotype seen in our patient. 2014 Wiley Periodicals, Inc. 2014 Wiley Periodicals, Inc.

Publication type: Journal Article
Source: MEDLINE

32. Title: Partial and complete trisomy 14 mosaicism: Clinical follow-up, cytogenetic and molecular analysis
Citation: Molecular Cytogenetics, September 2014, vol./is. 7/1, 1755-8166 (25 Sep 2014)
Author(s): Salas-Labadia C., Lieberman E., Cruz-Alcivar R., Navarrete-Meneses P., Gomez S., Cantu-Reyna C., Buiting K., Duran-McKinster C., Perez-Vera P.
Language: English
Abstract: Background: Trisomy 14 mosaicism is a rare chromosomal abnormality. It is associated with multiple congenital anomalies. We report a 15 year-old female with an unusual karyotype with three cell lines: 47,XX,+mar/47,XX,+14/46,XX. At six months old she had short stature, cleft palate, hyperpigmented linear spots in arms and legs and developmental delay. At present, she has mild facial dysmorphism and moderate mental retardation. Methods: Cytogenetic analysis was performed in peripheral blood lymphocytes and in the light and dark skin following standard methods. DNAarray-Oligo 180 k was carried out using Agilent Technologies and FISH analysis was accomplished using DNA BACs probes to confirm the result obtained by DNAarray. Methylation-Specific PCR (MS-PCR) of the MEG3 promoter and microsatellite analysis were performed. Results: Microarray analysis confirmed partial trisomy 14 mosaicism; the marker chromosome was found to be from chromosome 14, the result was confirmed with FISH. Methylation (14q32.3) and microsatellite (14q11-14q32.33) analysis were carried out and UPD was discarded. The global result was: mos 47,XX,+del(14)(q11.2)[45]/47,XX,+14 [10]/46,XX[45]. Conclusions: This is a unique case because of the coexistence of two abnormal cell lines, including one with +14 and another with +del(14)(q11.2). To our knowledge, only three patients have been reported with trisomy 14 and another abnormal cell line. The array analysis identified the marker chromosome and characterized the breakpoint. The del(14)(q11.2) does not seem to be related to any particular phenotypic characteristic of the patient; the clinical features of our patient observed until now, can be attributed to trisomy 14 mosaicism. Nevertheless, we cannot discard the manifestation of new symptoms related to her karyotype in the future.
Publication type: Journal: Article
Source: EMBASE
Full text: Available ProQuest at Molecular Cytogenetics
33. Title: Patient satisfaction with orthognathic surgery within a centralised hospital service model  
Citation: British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e82-e83), 0266-4356 (October 2014)  
Author(s): Tercan N., Shah D., Adali N., Von Arx D.  
Language: English  
Abstract: Aim: To investigate patient satisfaction with orthognathic surgery within centralised hospital service model  
Method: This was a retrospective questionnaire based audit. The gold standard was an overall satisfaction score of 95%. Ethical approval was granted and a pilot study using the abridged validated BOS Orthognathic patient satisfaction questionnaire was performed. For the main study, the exclusion criteria included osteotomy without orthodontics, patients with cleft lip and palate and craniomaxillofacial deformities. 213 consecutive patients who underwent combined orthognathic surgery within a centralised hospital network over a 3 year period from 07/10/2010 to 19/09/2013 were included. The patients had operations at the 'hub' unit, Luton and Dunstable Hospital, and the orthodontic component at 'spoke' units. Four Surgeons and six Consultant Orthodontists were involved. An introductory letter was sent. Four weeks later, the abridged questionnaire was posted. Descriptive and inferential statistical analysis was used to assess the data obtained. Results: The patient's were 92 males and 121 female. All surgeries were bimaxillary osteotomy or single jaw surgery. All had completed active orthodontics. There was high satisfaction with patient-clinician relationship and continuity with the centralised model but concerns with access and waiting times. There was no difference between surgeons (P > 0.05). The overall score was over 90%. Conclusions: There were high levels of satisfaction with different aspects of orthognathic treatment pathway and outcome and gold standard achieved. The centralised model of care was not highlighted as a concern. The study is limited by recall bias due to the retrospective design and should be repeated prospectively.  
Publication type: Journal: Conference Abstract  
Source: EMBASE

34. Title: Pioneer steps in correcting secondary cleft lip and palate deformities: My philosophy and procedures  
Citation: Journal of Cranio-Maxillofacial Surgery, October 2014, vol./is. 42/7(1023-1047), 1010-5182;1878-4119 (01 Oct 2014)  
Author(s): Obwegeser H.L.  
Language: English  
Publication type: Journal: Review  
Source: EMBASE

35. Title: Psychosocial issues faced by children after cleft surgery: A study in a selected hospital in Kathmandu, Nepal  
Citation: British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e97), 0266-4356 (October 2014)  
Author(s): King A.  
Language: English  
Abstract: Introduction: Cleft lip and palate is a congenital condition which can cause adverse psychosocial effects, including social exclusion, negative emotions, and low confidence. Previous research in developing countries has not concentrated on post-surgical experiences. Therefore, it is unclear whether these adverse psychosocial effects continue after repair, or whether this is able to counteract the negative experiences, increasing psychosocial well-being. Aim: To discover which psychosocial issues are faced by children living in Nepal after cleft surgery. Methods: This was a qualitative interview-based study with a sample group of six children and five guardians. Thematic content analysis was used to uncover themes. Results: Pre-surgery experiences were similar to those in current literature. The expectations were for a brighter future in which the cleft had no effect. Expectations were met to a degree, but the operations did not provide a complete change - some negative effects remain. The post-surgical experiences were mostly positive, with children finding it easier to join in with social activities and feeling happier. There were still a few issues, such as continued teasing and unhappiness with scar appearance. The participants discussed community education as a method to reduce negative social experiences. Conclusion: The post-surgical psychosocial experiences of these children are more positive than before repair. However, the results are still not entirely positive and some residual problems remain. Four recommendations have been made for further research and changes in practice.  
Publication type: Journal: Conference Abstract  
Source: EMBASE

36. Title: RFC1 and non-syndromic cleft lip with or without cleft palate: An association based study in Italy  
Citation: Journal of Cranio-Maxillofacial Surgery, October 2014, vol./is. 42/7(1503-1505), 1010-5182;1878-4119 (01 Oct 2014)  
Author(s): Girardi A., Martinelli M., Cura F., Palmieri A., Carinci F., Sesenna E., Scapoli L.  
Language: English
The molecular basis of orofacial development is largely unknown and needs to be unravelled. Non-syndromic cleft lip with or without cleft palate (NSCL/P) is the most common craniofacial malformation, with an incidence of about 1/700 live births, although variable according to ethnicity. Being a multifactorial disease, it arises as a result of an interplay between genetic and environmental factors. Several approaches have been developed to identify susceptibility genes. Genes belonging to the folate/homocysteine pathway are attracting increasing interest because folate supplementation before and during early pregnancy can reduce the risk of NSCL/P. We performed a family based association study in order to assess if a genetic variant of RFC1 could be involved in NSCL/P onset. We genotyped 404 unrelated probands and their relatives for three biallelic polymorphic variants (rs1051266, rs4818789 and rs3788205), that were selected because they produced conflicting results on previous investigations. Evidence of association was found between the investigated polymorphisms and NSCL/P in our sample of the Italian population, albeit with weak significance levels. Results from this investigation provided a support of previous studies suggesting a role of RFC1 in NSCL/P aetiology, reinforcing the concept that genetic predisposition to NSCL/P varies enormously within different ethnic groups.

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**Publication type:** Journal: Article

**Source:** EMBASE

37. **Title:** Screening for obstructive sleep apnea in children with syndromic cleft lip and/or palate.

**Citation:** Journal of Plastic, Reconstructive & Aesthetic Surgery: JPRAS, November 2014, vol./is. 67/11(1475-80), 1748-6815;1878-0539 (2014 Nov)

**Author(s):** Silvestre J, Tahiri Y, Paliga JT, Taylor JA

**Language:** English

**Abstract:** BACKGROUND: Craniofacial malformations including cleft lip and/or palate (CL/P) increase risk for obstructive sleep apnea (OSA). While 30% of CL/P occurs in the context of underlying genetic syndromes, few studies have investigated the prevalence of OSA in this high-risk group. This study aims to determine the incidence and risk factors of positive screening for OSA in this complex patient population. METHODS: The Pediatric Sleep Questionnaire (PSQ) was prospectively administered to all patients cared for by the cleft lip and palate clinic at the Children's Hospital of Philadelphia between January 2011 and August 2013. The PSQ is a 22-item, validated screening tool for OSA with a sensitivity and specificity of 0.83 and 0.87 in detecting an apnea-hypopnea index (AHI) >5/hour in healthy children. The Fisher exact and Chi-square tests were used for purposes of comparison. RESULTS: 178 patients with syndromic CL/P completed the PSQ. Mean cohort age was 8.1 ± 4.4 years. Patients were predominately female (53.9%), Caucasian (78.1%), and had Veau Class II cleft (50.6%). Craniofacial syndromes included isolated Pierre Robin Sequence (PRS) (29.8%), 22q11.2 deletion syndrome (14.6%), Van der Woude syndrome (6.7%), and other rare genetic abnormalities (28.8%). The overall incidence of positive OSA screening was 32.0%. Males were at increased risk for positive OSA screening (P = 0.030), as were non-Caucasians (P = 0.044). Symptoms with the highest positive predictive value for OSA were "others comment on child appearing sleepy" (76.2%) and "stops breathing during the night" (75.0%). Notably, patients with 22q11.2 deletion syndrome were at highest risk for positive screens (50.0%, P = 0.042). CONCLUSIONS: Nearly a third of our patients with syndromic CL/P screened positively for OSA (32.0%), highlighting the importance of screening in this at-risk population. Future work will correlate screening results with polysomnograms to help validate these findings. CLINICAL QUESTION/LEVEL OF EVIDENCE: Diagnostic, III. Copyright 2014 British Association of Plastic, Reconstructive and Aesthetic Surgeons. Published by Elsevier Ltd. All rights reserved.

**Publication type:** Journal Article

**Source:** MEDLINE

38. **Title:** Secondary cleft rhinoplasty: impact on self-esteem and quality of life.

**Citation:** Plastic & Reconstructive Surgery, December 2014, vol./is. 134/6(1285-92), 0007-1226;1529-4242 (2014 Dec)

**Author(s):** Roosenboom J, Hellings PW, Picavet VA, Prokopakis EP, Antonis Y, Schoenaers J, Poorten VV, Claes P, Hens G

**Language:** English

**Abstract:** BACKGROUND: Rhinoplasty is one of the most challenging facial plastic procedures. Although patient satisfaction is the real outcome parameter in rhinoplasty, most authors have studied objective outcomes evaluated by professionals. The purpose of this study was to determine patient satisfaction after rhinoplasty in patients born with a cleft lip compared with outcome assessment by professionals, and to assess the impact of the procedure on appearance-related distress and generic quality of life. METHODS: Patient evaluation of the nose was performed before and 1 year after secondary cleft rhinoplasty (n = 33) using a visual analogue scale for nasal function and shape, and the Rhinoplasty Outcome Evaluation. General sinonasal complaints were evaluated using the Sino-Nasal Outcome Test. Appearance-related psychological distress was measured using the Derriford Appearance Scale. The Sheehan Disability Scale evaluated quality of life. Aesthetic outcome was evaluated by scoring of preoperative and postoperative photographs by two independent surgeons. RESULTS: One year postoperatively, patients showed significantly higher visual analogue scale scores for nasal...
shape (p < 0.0001) and function (p = 0.005) and higher Rhinoplasty Outcome Evaluation (p < 0.0001) scores. Correspondingly, Sino-Nasal Outcome Test scores were lower (p = 0.006). The appearance-related psychological distress was lower (p < 0.0001), and the generic quality of life was increased after rhinoplasty (p = 0.01). No correlation was found between patient outcome evaluation and surgeons’ scores.

CONCLUSION: There is high patient satisfaction at 12 months after secondary cleft rhinoplasty, resulting in a significant improvement of self-esteem and generic quality of life.

CLINICAL QUESTION/LEVEL OF EVIDENCE: Therapeutic, IV.

Publication type: Journal Article
Source: MEDLINE
Full text: Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery

Full text: Available Journal of reconstructive microsurgery at Plastic and Reconstructive Surgery

39. Title: Siamese twins with craniofacial duplication and bilateral cleft lip/palate in a ceramic representation of the chimu culture (Peru): A comparative analysis with a current case
Citation: Twin Research and Human Genetics, 2014, vol./is. 17/3(211-214), 1832-4274;1839-2628 (2014)
Author(s): Pachajao H., Hernandez-Amaris M.F., Porras-Hurtado G.L., Rodriguez C.A.
Language: English
Abstract: Craniofacial duplication or diprosopus is a very rare malformation that is present in approximately 0.4% of conjoined twins. Here is presented a case of craniofacial duplication in association with bilateral cleft lip/palate in both heads found in a ceramic representation from the early Chimu culture from Peru. A comparative analysis is made with a current case of a 28-week-old fetus with similar characteristics. After reviewing the medical literature on conjoined twins, very few reports of facial cleft in both twins were found, with no reports at all of bilateral cleft lip/palate. This ceramic crock is considered one of the first representations suggestive of craniofacial duplication, and probably the first reporting it in association with facial cleft.

Publication type: Journal: Article
Source: EMBASE

40. Title: Significant association of MTHFD1 1958G>A single nucleotide polymorphism with nonsyndromic cleft lip and palate in Indian population
Citation: Medicina Oral, Patologia Oral y Cirugia Bucal, November 2014, vol./is. 19/6(e616-e621), 1698-4447;1698-6946 (01 Nov 2014)
Author(s): Murthy J., Gurramkonda V.-B., Lakkakula B.V.K.S.
Language: English
Abstract: Objectives: Nonsyndromic cleft lip and palate (NSCLP) is genetically distinct from those with syndromic clefts, and accounts for ~70% of cases with Oral clefts. Folate, or vitamin B9, is an essential nutrient in our diet. Allelic variants in genes involved in the folate pathway might be expected to have an impact on risk of oral clefts. Given the key role of methylenetetrahydrofolate dehydrogenase 1 (MTHFD1) in folate metabolism, it would be of significant interest to assess its role in NSCLP etiology. Study Design: The present study aims at examining the association between MTHFD1 1958G>A polymorphism and NSCLP risk by conducting a case-control study in south Indian population. Our sample comprised of 142 cases with nonsyndromic clefts and 141 controls without clefts or family history of clefting. The MTHFD1 1958G>A polymorphism was genotyped using PCR-RFLP. Results: An increased risk was found for the heterozygous 1958GA (OR=2.44; P=0.020) and homozygous 1958AA (OR=2.45; P=0.012) genotypes in the children. When the dominant model (AG+AA vs GG) was applied the risk remained the same as co-dominant model, but the level of significance increased (OR=2.44; P=0.002). Conclusions: The results indicated the MTHFD1 1958G>A polymorphism to be one of the important genetic determinants of NSCLP risk in South Indian subjects.

Publication type: Journal: Article
Source: EMBASE

41. Title: Single versus segmental maxillary osteotomies and long-term stability in unilateral cleft lip and palate related malocclusion.
Citation: Journal of Oral & Maxillofacial Surgery, December 2014, vol./is. 72/12(2514-21), 0278-2391;1531-5053 (2014 Dec)
Author(s): Watts GD, Antonarakis GS, Forrest CR, Tompson BD, Phillips JH
Language: English
Abstract: PURPOSE: To investigate the stability of single-piece versus segmental (2-piece) maxillary advancement in patients with unilateral cleft lip and palate (UCLP) treated using conventional Le Fort I orthognathic surgery.

PATIENTS AND METHODS: A retrospective study was undertaken in 30 patients with nonsyndromic UCLP treated with the same surgical and orthodontic protocol from 2002 through 2011. Standard lateral cephalometric radiographs were taken preoperatively, immediately postoperatively, and at least 1 year postoperatively. Patients were divided into single-piece and segmental Le Fort I groups based on planned surgical movement. Postoperative movements were compared between groups using repeated measures analysis of variance. RESULTS: The mean skeletal horizontal advancement was 7.3 and 7.5 mm in the
single-piece and segmental groups, respectively. The skeletal horizontal relapse was 1.3 mm (18%) for the single-piece group and 1.9 mm (25%) for the segmental group. The skeletal surgical extrusion was 2.7 mm for the 2 groups. The skeletal vertical relapse was 0.6 mm (22%) and 1.5 mm (56%) for the single-piece and segmental groups, respectively. The mean dental horizontal postoperative movement was an advancement of 0.4 mm for the single-piece group and a relapse of 0.2 mm (3%) for the segmental group. The mean dental vertical relapse was 0.1 mm (4%) for the single-piece group and 0.3 mm (11%) for the segmental group. There was no statistically significant difference in relapse between the single-piece and segmental groups for all movements (P > .05).

CONCLUSION: Skeletal and dental relapse was similar between single-piece and segmental maxillary advancements using conventional Le Fort I orthognathic surgery in patients with UCLP.

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**Publication type:** Journal Article

**Source:** MEDLINE

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**42. Title:** Speech outcome in unilateral complete cleft lip and palate patients: a descriptive study

**Citation:** European journal of paediatric dentistry : official journal of European Academy of Paediatric Dentistry, September 2014, vol./is. 15/3(293-296), 1591-996X (Sep 2014)

**Author(s):** Rullo R., Di Maggio D., Addabbo F., Rullo F., Festa V.M., Perillo L.

**Language:** English

**Abstract:** In this study, resonance and articulation disorders were examined in a group of patients surgically treated for cleft lip and palate, considering family social background, and children's ability of self monitoring their speech output while speaking. Fifty children (32 males and 18 females) mean age 6.5 + 1.6 years, affected by non-syndromic complete unilateral cleft of the lip and palate underwent the same surgical protocol. The speech level was evaluated using the Accordi's speech assessment protocol that focuses on intelligibility, nasality, nasal air escape, pharyngeal friction, and glottal stop. Pearson product-moment correlation analysis was used to detect significant associations between analysed parameters. A total of 16% (8 children) of the sample had severe to moderate degree of nasality and nasal air escape, presence of pharyngeal friction and glottal stop, which obviously compromise speech intelligibility. Ten children (10%) showed a barely acceptable phonological outcome: nasality and nasal air escape were mild to moderate, but the intelligibility remained poor. Thirty-two children (64%) had normal speech. Statistical analysis revealed a significant correlation between the severity of nasal resonance and nasal air escape (p < 0.05). No statistical significant correlation was found between the final intelligibility and the patient social background, neither between the final intelligibility nor the age of the patients. The differences in speech outcome could be explained with a specific, subjective, and inborn ability, different for each child, in self-monitoring their speech output.

**Publication type:** Journal: Article

**Source:** EMBASE

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**43. Title:** Spread of X inactivation on chromosome 15 is associated with a more severe phenotype in a girl with an unbalanced t(X; 15) translocation

**Citation:** American Journal of Medical Genetics, Part A, 2014, vol./is. 164/10(2521-2528), 1552-4825;1552-4833 (2014)


**Language:** English

**Abstract:** Wereport on a baby girl with multiple congenital abnormalities, including cleft palate, intrauterine growth restriction, and double outlet right ventricle (DORV) with ventricular septal defect. She had an unbalanced chromosome translocation t (X;15) resulting in monosomy 15pter -> p10 and trisomy Xq13.1 -> q28. All three copies of Xq encompass the XIST gene. It is known that X chromosome inactivation could spread to the autosome part of an unbalanced translocation involving chromosome X and an autosome. To confirm the spread of X chromosome inactivation on chromosome 15, we evaluate the methylation change by the HumanMethylation450 BeadChip, a whole genome DNA methylation micorarray that includes 15,259 probes spanning 717 genes on chromosome 15. Results showed there was gain in DNA methylation of more than 20% in 586 CpG sites spanning the long arm of chromosome 15. We further examined the hypermethylated CpG sites located in CpG-island promoter, because genes subjected to X chromosome inactivation will have an increase in DNA methylation level in this region. A total of 75 sites representing 24 genes were hypermethylated. Nearly all of these probes are located in region proximal to the breakpoint, from 15q11.2 to 15q21.3 (35Mb) suggesting that X inactivation was spread to the proximal region of 15q. Gain of DNA methylation, especially in the CpG-island promoter, can result in functional inactivation of genes, and therefore could potentially worsen the phenotype of our patient.

**Publication type:** Journal: Article

**Source:** EMBASE

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**44. Title:** Submucous cleft palate with bifid uvula

**Citation:** Journal of Pediatrics, October 2014, vol./is. 165/4(872), 0022-3476;1097-6833 (01 Oct 2014)

**Author(s):** Hasan A., Gardner A., Devlin M., Russell C.
45. Title: Surface area scanning of cleft palate dimension-a predictor of outcome
Citation: British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e44-e45), 0266-4356 (October 2014)
Author(s): Colbert S., Williams S., Extence H., Drake D.
Language: English
Abstract: Introduction/Aims: studies investigating risk factors for cleft speech characteristics (CSCs) among patients with cleft palates have been reported. Most focus on surgical technique, demographic factors, or type of palatal cleft. We sought to examine the impact of cleft width on the development of CSCs among patients with cleft palate by scanning the dimension of the cleft palate. Materials/Methods: We scanned the models of cleft palates repaired in the South Wales Cleft Unit in 2005 and 2006. The ratio of the cleft palate to the surface area of the palate anterior to the inter-tubercular line was calculated. The Cleft Audit Protocol for Speech - augmented (CAPSA) speech outcomes were recorded at the 5yr audit outcomes on all patients. We investigated for a relationship between speech outcome and cleft palate surface area. Results/Statistics: the relationship between cleft palate surface area and speech outcomes is reported. A critical ratio between cleft palate surface area to palatal surface area is calculated where speech outcome scores were found to change significantly. Conclusions/Clinical Relevance: this study is the first attempt to specifically quantify cleft width as a risk factor for postoperative speech outcomes. Our novel method of scanning models of cleft palates can be used to predict speech outcomes of palatal surgery, and as such, allow the surgeon to counsel the parents regarding the likely outcome of their child's surgery.
Publication type: Journal: Conference Abstract
Source: EMBASE

46. Title: The Northern Cleft Foundation: A safety survey of anaesthetic adverse incidents during a 10 day cleft lip and palate camp in India
Citation: Anaesthesia, October 2014, vol./is. 69/(75), 0003-2409 (October 2014)
Author(s): Sharma N., Gleeson S., Shorrock P., Girotra V., Kelgiri N.
Language: English
Abstract: In the UK, 1 child in 800 is born with cleft lip or palate, with similar incidence in India; however access to early surgical repair is lacking in the latter [1]. A team of 40 personnel visited Mure Memorial Hospital, India to conduct the Northern Cleft Foundation’s 14th camp. Conditions were challenging and equipment sub-standard to the UK. We operated on 134 patients over 10 days. Having reviewed the literature we identified a lack of data on anaesthetic adverse incidents during cleft surgery both in the developed and developing world and therefore focused on this in order to introduce improvement strategies. Methods We collected data daily on all patients. We defined anaesthetic adverse events using the RCOA's definition of a critical incident, 'an event that led to harm or could have led to harm had it been allowed to progress.' [2] Data was entered on an Excel spreadsheet and analysed. Results We operated on similar numbers of patients based on gender. The mean age of our patients was 4.5 years, the oldest 37 years and youngest, 3 months. 48 patients were associated with adverse incidents, with 53 incidents occurring within this group. Environmental incidents that occurred included power failure, oxygen supply failure, N2O leak, the presence of non-medical volunteers and communication errors. Five cases were cancelled; 3 after induction of anaesthesia, 2 preoperatively. These cancellations could have been decided in the preoperative assessment clinic. (Table Presented) Discussion Preoperatively we would recommend: Robust consultant led preoperative assessment which in potential difficult cases leads to these consultants conducting the patient's operation, professional interpreters, decreased fasting time and when unavoidable, the use of preload drink, greater focus on patient ID band/lanyard, patient unit number on all documentation (ideally 3 identifiers), greater communication when order of theatre lists is changed and use of WHO checklist. Intraoperatively: End tidal agent monitoring, BM check at start and end of the case, standardised temperature monitoring/patient warming, throat pack stickers and tranexamic acid as standard for high risk cases eg. Vomer flap. Postoperatively: Team debrief with discussion of adverse events. In general, an induction pack and group introduction is needed with briefing for non-medical volunteers. We also recommend that operating begins on day 2 of the trip not the day of arrival. These recommendations will be presented to the NCF committee and an induction pack is being produced before the next camp.
Publication type: Journal: Conference Abstract
Source: EMBASE

47. Title: The effect of maxillary advancement on speech outcomes in patients with cleft lip and palate
Citation: British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e50), 0266-4356 (October 2014)
Author(s): Colbert S., McDaid J., Williams S., Extence H., Sugar A., Drake D.
Language: English
Abstract: Introduction/Aims: Orthognathic surgery can alter symptoms of velopharyngeal insufficiency in cleft patients. The goal of this study was to evaluate how advancing the maxilla would affect the speech and articulation disorders of
these patients. Materials/Methods: This was a retrospective study in which we evaluated the speech scores on 50 cleft lip and palate patients who underwent maxillary advancement at our unit. The following variables were recorded from both preoperative and postoperative speech evaluations: presence of a pharyngoplasty, nasality, velopharyngeal function assessment, and overall speech score. Preoperative and postoperative changes in the data were analyzed. Results/Statistics: we report on the competency of velopharyngeal function mechanisms postoperatively in patients with pharyngoplasties and in cases of borderline incompetence preoperatively. Speech scores after surgery are reported using the CAPS - A scoring system in patients with preoperative hypernasality. Conclusions/Clinical Relevance: our study is compared to previous findings that patients with clefts of the lip and palate or palate alone are predisposed to velopharyngeal function alteration after maxillary advancement, particularly patients with borderline function and/or pharyngoplasty preoperatively.

**Publication type:** Journal: Conference Abstract

**Source:** EMBASE

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**48. Title:** The impact of a cleft interface training on unit outcomes in primary palate surgery: Post repair fistula rates

**Citation:** British Journal of Oral and Maxillofacial Surgery, October 2014, vol./is. 52/8(e51), 0266-4356 (October 2014)

**Author(s):** Devlin M., Holmes K., Russell C., Ray A., Handley T.

**Language:** English

**Abstract:** Introduction: The aim of this study was to assess the impact of a training interface group cleft fellow on the outcomes of palate surgery within their training unit. Fistula rate was the measured parameter. Method: Retrospective case note reviwevover an 18 month period from the start of the fellowship. Patients with at least a 6 week review were grouped into either isolated cleft palate (group 1) or cleft lip and palate (Group 2). Primary surgery only was considered. Various factors were assessed including the Scottish index of multiple deprivation, type of cleft, syndromic involvement, type of repair, surgeon and complications. The end point observed was whether an oronasal fistula had developed at 6 weeks. Results: Total of 66 patients. 42 male and 24 female (ratio 7:4). 40 operations were performed by the fellow, 19 by trainer 1 and 7 by trainer 2. Due to the small number of operations performed, trainer 2 was excluded from analysis. Fistula rate of fellow 12.5%, vs trainer 1 5.3%. Fisher exact test p= 0.653 Conclusion: The fellow demonstrated an acceptable fistula rate as compared to the literature. 4 out of 5 fistulae occurred during the first 5 months of training. This suggested a higher rate at the commencement of training with a gradual improvement over time. We conclude that training a cleft fellow does not result in an unacceptable fistula rate for the training unit.

**Publication type:** Journal: Conference Abstract

**Source:** EMBASE

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**49. Title:** The role and function of the VEGF pathway during palate development

**Citation:** Otolaryngology - Head and Neck Surgery (United States), September 2014, vol./is. 151/1 SUPPL. 1(P108), 0194-5998 (September 2014)

**Author(s):** Jacobs B.H., Goudy S.L., Hill C.A.

**Language:** English

**Abstract:** Objectives: (1) Define the evolution of palatal vascularization during palatal shelf formation and effects of vascular endothelial growth factor (Vegf) deletion in the cranial neural crest. (2) Determine downstream mediators of Vegf signaling. Methods: Conditional deletion of Vegf using Wnt1-Cre; Vegf F/F mice (VegfCKO) led to a cleft palate phenotype. PECAM staining was used to determine vascular patterning in the VegfCKO developing palate daily from E13.5 to E16.5 compared to controls. The VegfCKO palate shelves at E13.5 and E14.5 were analyzed using quantitative PCR (qPCR) to assess mediators and targets of Vegf signaling. Results: VegfCKO had aberrant vascular branching via PECAM staining in comparison to controls. In E13.5 VegfCKO mice, there were no significant differences in mediators and targets of Vegf signaling measured by qPCR. However, at E14.5, VegfCKO palates demonstrated significant reductions in pyruvate dehydrogenase kinase (PDK4) (P = .045), which is a glycolytic gene, and stromal cell derived factor 1 (SDF1) (P = .036), which induces endothelial migration. Conclusions: Vegf was required for palatal vascularization and palatal elongation. During palatal development Vegf was upstream of SDF1, a cytokine necessary for endothelial progenitor cell recruitment and migration. Vegf also modulates glycolysis through variations in PDK4, a protein involved in glucose metabolism.

**Publication type:** Journal: Conference Abstract

**Source:** EMBASE

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**50. Title:** Therapeutic options for conductive hearing loss in children with cleft palate

**Citation:** Archives of Disease in Childhood, October 2014, vol./is. 99/(A549), 0003-9888 (October 2014)

**Author(s):** Franchella S., Barillari M.R., Bovo R., Martini A., Franchella A.

**Language:** English

**Abstract:** Background and aim Cleft lip and palate is a common congenital malformation correlated with otological disorders like Eustachian tube dysfunction, otitis media with effusion and conductive hearing loss correlated with speech disorders. Methods In our study we evaluate the therapeutic options for the conductive hearing loss in these children, such as: middle ear ventilation tube insertion, hearing aids and adenoidectomy. We conducted a retrospective chart
review on 19 patients who underwent last follow-up during 2013, aged between 1 and 16, affected by cleft palate. We
identified 3 cases of Treacher-Collins syndrome, 2 cases of CHARGE syndrome, 2 case sof Pierre Robin, 1 case of Goldhenar
and 1 case of Di George syndrome. In 9 cases the cleft palate was isolated. In 14 cases we conducted phoniatric and
logopaedic evaluations and 10 of these showed a speech disorder. Results We found a conductive hearing loss in 12 of the
19 children.2 of these patients used hearing aids with an improvement of speech performance; 1 patient underwent
adenoidectomy for the appearance of sleep apnea but he modified his quality of voice with hypernasality and nasal
emission after surgery; finally 9 children had spontaneous resolution of otitis media with effusion. Conclusions In
conclusion we found that the more effective way to resolve the problem of conductive hearing loss and the resulting
speech disorder is to use the hearing aids until the resolution of the hearing loss, which normally occurs around 7 years
old.

Publication type: Journal: Conference Abstract
Source: EMBASE
Full text: Available ARCHIVES OF DISEASE IN CHILDHOOD at Archives of disease in childhood
Full text: Available ARCHIVES OF DISEASE IN CHILDHOOD at Salisbury District Hospital Healthcare Library

51. Title: Use of human recombinant bone morphogenetic protein is associated with increased hospital charges in
children with cleft lip and palate having bone graft procedures.
Citation: Journal of Oral & Maxillofacial Surgery, December 2014, vol./is. 72/12(2531-8), 0278-2391;1531-5053 (2014 Dec)
Author(s): Allareddy V
Language: English
Abstract: PURPOSE: There is wide variation in the timing of secondary alveolar bone grafting in patients with cleft lip and
 palate. The objective of the present study was to examine the timing and hospitalization outcomes of bone grafting
 procedures in a cohort of 5- to 13-year-old patients with a diagnosis of cleft lip and palate.MATERIALS AND METHODS: The
 Nationwide Inpatient Sample for 2004 to 2010 was used. Outcomes examined included complications, disposition status,
hospital charges, and length of stay. Multivariable linear regression analysis was used to examine the association between
hospital charges and a group of heterogenous patient-level (age, gender, race, comorbid burden, insurance status, and
concomitant procedures) and hospital-level variables.RESULTS: In total, 3,478 patients underwent bone grafting
 procedures. Excision of bone for grafting was obtained from pelvic bones in 79.4% of cases. Recombinant bone
morphogenetic protein (rhBMP) was inserted in 1% of all cases. Nearly 99% were discharged routinely. The overall
complication rate was 1.5%. The mean length of hospital stay was 1.3 days and the mean hospitalization charge was
$23,852 per case. Those who had insertion of rhBMP had an excess of $14,695 compared with those who did not have
rhBMP after adjustment for all other variables (P < .0001).CONCLUSIONS: Bone grafting procedures are safe when
performed in patients 5 to 13 years old. Insertion of rhBMP was performed in nearly 1% of patients undergoing bone
grafting procedures. Use of rhBMP was associated with a considerable increase in hospital charges. Copyright 2014
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Publication type: Journal Article
Source: MEDLINE

52. Title: Velopharyngeal incompetence in patients with cleft palate, flexible video pharyngoscopy and perceptual
speech assessment: a correlational pilot study.
Citation: Journal of Laryngology & Otology, November 2014, vol./is. 128/11(986-90), 0022-2151;1748-5460 (2014 Nov)
Author(s): Rajan S, Kurien M, Gupta AK, Mathews SS, Albert RR, Tylicus D
Language: English
Abstract: OBJECTIVES: To assess the role of video endoscopy in evaluating velopharyngeal incompetence and investigate
a possible relationship between velopharyngeal incompetence type and speech defect in cleft palate patients.METHODS: A
prospective study of 28 pre- or post-operative cleft palate patients with speech defects who attended Plastic Surgery-Cleft
Palate and ENT out-patient clinics was performed. The velar defect type was determined using a flexible endoscope and
findings were video recorded. Speech pathology was assessed using the cleft palate audit protocol for speech.RESULTS: A
significant, clinically relevant relationship was noted between the perceived characteristics of hypernasality and
velopharyngeal insufficiency type. Hypernasal speech was a definite clinical indicator of velopharyngeal incompetence,
and the type 1 velopharyngeal defect was most common. Type 1 velopharyngeal coronal-type dysfunction was strongly
associated with hypernasality (p < 0.05). When speech substitution was noted, type 2 velopharyngeal (or sagittal)
incompetence could be predicted (p < 0.05).CONCLUSION: In the management of cleft palate patients, it is important that
surgical correction of the defect and achieving velopharyngeal competency for speech are performed simultaneously.
Pre-operative velopharyngeal endoscopy with speech assessment will define the anatomical and functional bases for
velopharyngeal correction and assist in planning and tailoring the pharyngeal flap.

Publication type: Journal Article
Source: MEDLINE
Full text: Available Journal of Laryngology & Otology at Journal of Laryngology and Otology
Full text: Available Journal of Laryngology & Otology at Journal of Laryngology and Otology, The
53. Title: Versatility of the buccinator myomucosal flap in atypical palate reconstructions
Citation: Journal of Cranio-Maxillofacial Surgery, October 2014, vol./is. 42/7(1310-1314), 1010-5182;1878-4119 (01 Oct 2014)
Author(s): Franco D., Rocha D., Arnaut M., Freitas R., Alonso N.
Language: English
Abstract: Initially described for the treatment of cleft palate, the anatomical bases of the buccinator myomucosal flap were described by Bozola et al. (1989). A meticulous search found several reports of its use for the correction of post-palatoplasty oronasal fistulas, with only a few reports of its use for other palate-related pathologies. A retrospective analysis was undertaken of patients treated by the Plastic Surgery Units at the Rio de Janeiro Federal University Hospital (HU-UFRJ) and the Sao Paulo University Hospital (HC-USP), suffering from palatal lesions not associated with a cleft palate and treated through the use of buccinator myomucosal flaps. The average age was 47 years, with 70% of the patients being male. Assorted aetiologies were noted for palatal defects. When there was significant damage to the soft palate, a superior base pharyngeal flap was used. Of this total, in 71% of the cases only the buccinator myomucosal flap was used. In all cases, the flaps were unilateral, adequately covering the defects in question. The buccinator myomucosal flap is a good option for reconstructing medium to large palate defects, as it is a flap with good vascularization and dimension, in addition to an ample arc of rotation, with primary closure of the donor site, without adding significant morbidity.
Publication type: Journal: Article
Source: EMBASE

54. Title: What factors are associated with impacted canines in cleft patients?
Citation: Journal of Oral and Maxillofacial Surgery, November 2014, vol./is. 72/11(2109-2114), 0278-2391;1531-5053 (01 Nov 2014)
Author(s): Westerlund A., Sjostrom M., Bjornstrom L., Ransjo M.
Language: English
Abstract: Purpose It is important to predict and prevent the impaction of canines. The aim of this study was to estimate the prevalence of impacted canines in patients with unilateral cleft lip and palate (UCLP) and to identify factors associated with impaction. Materials and Methods This retrospective cohort study included patients with nonsyndromic UCLP. The predictors were pre-eruptive inclination angle, deviation in tooth number (agenesis or supernumerary lateral incisors), and reoperation of bone transplant. The outcome variable was impacted and surgically exposed canines. Results The prevalence of impacted and surgically exposed canines in the 68 consecutive patients with UCLP was 20.6%. The pre-eruptive inclination angle was significantly larger (34.4\(^\text{degree}\)) for the impacted canines on the cleft side compared with the spontaneously erupted canines on the cleft and non-cleft sides (25.5\(^\text{degree}\) vs 15.4; \(P < .05\)). Reoperation of the bone transplant significantly increased canine impaction (50%; \(P < .05\)). Conclusion The eruption of maxillary canines needs to be supervised carefully in patients with UCLP, because the prevalence of impaction is 10 times higher compared with the general population. Factors associated with canine impaction are a pre-eruptive inclination larger than 30\(^\text{degree}\) and reoperation of the bone transplant.
Publication type: Journal: Article
Source: EMBASE

55. Title: [Cleft rhinoplasty, from primary to secondary surgery] [French] Rhinoplastie de fente primaire et secondaire.
Citation: Annales de Chirurgie Plastique et Esthetique, December 2014, vol./is. 59/6(555-84), 0294-1260;1768-319X (2014 Dec)
Author(s): Talmant JC, Talmant JC
Language: French
Abstract: Despite fifty years of statistics, congresses, publications, the cleft nose remains an enigma to the great majority of cleft specialists. Most of the published papers give recipes to camouflage the cleft deformity, very few are concerned by the functional anatomy and its relation with facial growth. The complexity of the matter, the results frequently disappointing, the lack of awareness of the necessity of early nasal breathing, and the academic condemnation of any imperfect attempt to correct the nose at the time of the first operation have led to resignation. For the last forty years, we have been involved in a careful and obstinate research about the early correction of the cleft nose deformity. We wish to present our conclusions in this chapter with at least 17 years of follow-up. They are as following: in cleft patients the nasal cartilages are only deformed. We can achieve sub periosseal and sub periocdndral dissections by 6 months of age without being harmful for facial and nasal growth. Repositioning accurately the nasal structures is enough if we are able to control the healing process and prevent endonasal wound contraction. We have not to do any compromise and favor one function with regard to the others, nasal ventilation being the most important for a good facial growth. In a word, nasal pediatric surgery is necessary at the time of the first operation from 6 months of age and should be carried on with a double demand, aesthetic and functional. To achieve this goal, we must have a sound knowledge of the cleft nose deformity, of the adequate surgical techniques and of the logic chronology to reach the best result. The nose repair cannot be limited to the nasal cartilages. The whole nasal structure is concerned especially its bony framework, the width
of which at the level of the piriform orifice and the nasal floor depends on the outcomes of any surgical step that it would relate to the lip, palate or alveolar closure. Interaction of all these factors calls for an appropriate answer in adequation with the diagnosis of the deformity and a coherent answer as we know that any local action may induce an unfavorable chain reaction and should integrate a global and logic project. After the primary surgery, additional correction for aesthetic or functional purpose as well, may be useful during the period of growth. For cleft teenagers or adults, the rhinoplasty can simply be indicated for harmonization after a good primary nasal correction and optimal facial growth. On the contrary, the rhinoplasty may be more or less a complex operation for the usual and severe deformities. In the last case, the diagnosis must take into account all the residual deformities, even the labial and alveolar ones, and the treatment plan integrate all the principles and techniques of the primary surgery. What has not been done at the time of the primary surgery, should be done secondarily: all the structures are present, only deformed and embedded in scarred tissues. Primary or secondary cleft rhinoplasty must be undertaken by surgeons accustomed to cleft patients, but also trained in the other fields of nasal surgery, aesthetic and reconstructive. Copyright 2014. Published by Elsevier Masson SAS.

Publication type: English Abstract, Journal Article
Source: MEDLINE