

Surgery for Speech in VCFS

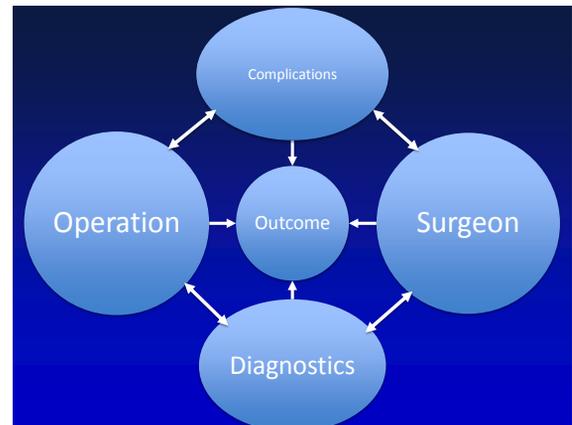
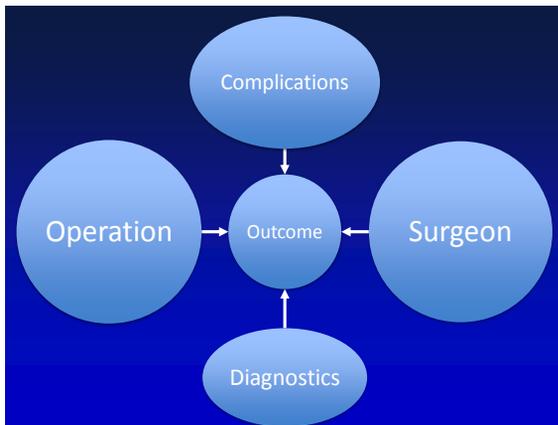
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Special, or the same as for other
 children with hypernasality?

Factors (variables)

- ✓ The operation
- ✓ The surgeon
- ✓ The diagnostics
- ✓ Complications
- ✓ Evil spirits ☺



Few (if any) studies have controlled for all of the variables or interactions between them.

Reports of results with specific operations would have you believe that the same operation by different surgeons will have the same outcome.

My wife's chocolate cake



My chocolate cake (same recipe)



Is it also true that a specific operation can have a different outcome based on primary diagnosis?

Example

Foot surgery in people with diabetes vs foot surgery in people without diabetes.

Surgery for VPI

Only 1 purpose: to stop air and sound from leaking out of the nose when it should not be coming out of the nose.

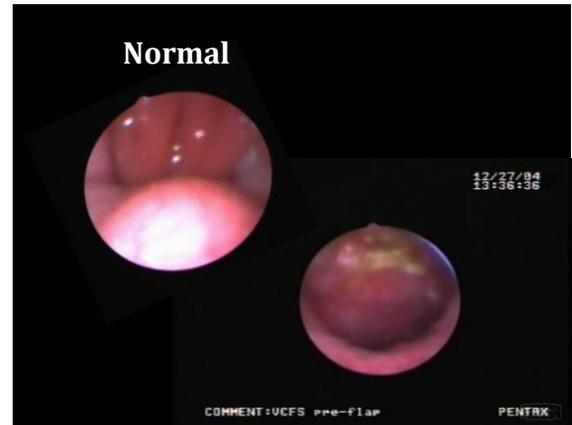
Are the reasons for abnormal nasality in VCFS different?

Reasons for VPI in VCFS

- ✓ Structural palate anomalies
- ✓ Hypotonia
- ✓ Cranial base anomalies
- ✓ Asymmetry
- ✓ Learning errors (including compensatory articulation)
- ✓ Evil spirits ☺

Structural palate anomalies

- ✓ Overt cleft palate
- ✓ Submucous cleft palate
- ✓ Occult submucous cleft
- ✓ Reduced muscle mass



Hypotonia

- ✓ Abnormal muscle fibers
- ✓ Fewer muscle fibers
- ✓ Central nervous system?

Cranial base anomalies

- ✓ Obtuse cranial base angle
 - ✓ Increased volume of the pharynx

Asymmetry

- ✓ Velar asymmetry
 - ✓ Structural and functional
- ✓ Pharyngeal asymmetry
 - ✓ Structural and functional

Learning errors

- ✓ Nasal fricatives
- ✓ Other nasal substitutions
- ✓ Habitual nasal resonance
- ✓ Compensatory articulation
- ✓ **THESE DO NOT REQUIRE OR RESPOND WELL TO SURGERY!**

Factors Unique to VCFS

- ✓ Palate and pharynx anatomy and physiology
- ✓ Risk
 - ✓ Surgical risk
 - ✓ Post-surgical risk

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Hypotonia

Zim S, 2002, Arch Facial Plast Surg

Abnormal distribution of type A and type B fibers in the pharynx in VCFS

Reduced fiber size

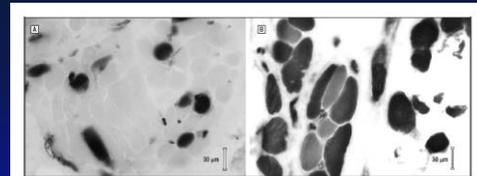


Figure 2. Cross sections from the superior pharyngeal constrictor muscle of a patient with velocardiofacial syndrome stained for myofibrillar adenosine triphosphatase (ATPhase) at pH 4.3 (A), where light-stained muscle fibers correspond to type 2 and dark-stained muscle fibers to type 1, and pH 9.4 (B), where light-stained muscle fibers correspond to type 1 and dark-stained muscle fibers to type 2 (original magnification $\times 20$).

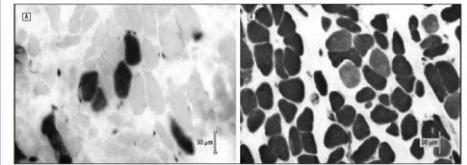


Figure 3. Cross sections from the superior pharyngeal constrictor muscle of an adult patient without velocardiofacial syndrome stained for myofibrillar adenosine triphosphatase (ATPhase) at pH 4.3 (A), where light-stained muscle fibers correspond to type 2 and dark-stained muscle fibers to type 1, and pH 9.4 (B), where light-stained muscle fibers correspond to type 1 and dark-stained muscle fibers to type 2 (original magnification $\times 20$).

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Upper airway asymmetry in velo-cardio-facial syndrome

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Internal Carotid Anomalies

Plast Reconstr Surg, Volume 97(5) Supplement, April 1996, pp 908-919

The Use of Magnetic Resonance Angiography prior to Pharyngeal Flap Surgery in Patients with Velocardiofacial Syndrome

Robin J. Mitnick, M.D., Jacqueline A. Bello, M.D., Karen J. Golding-Kushner, Ph.D., Ravelo V. Argamaso, M.D., and Robert J. Shprintzen, Ph.D.

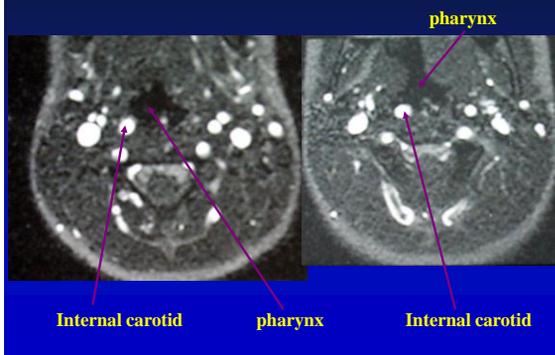
Int J Pediatr Otorhinolaryngol. 2010 June ; 74(6): 619-625. doi:10.1016/j.ijporl.2010.03.006.

Cervical Vascular and Upper Airway Asymmetry in Velo-Cardio-Facial Syndrome: Correlation of Nasopharyngoscopy with MRA

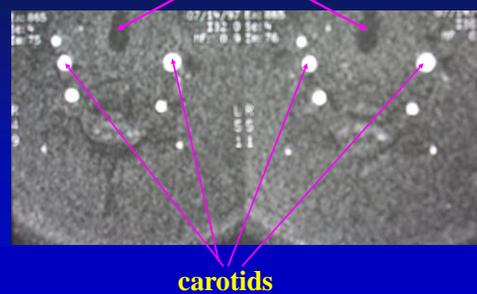
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Ectopic Internal Carotids



Normal Carotid Placement



MRA should be done immediately before surgery to the posterior pharyngeal wall.(Oppenheimer et al, 2010, Mitnick et al., 1996)

Adenotonsillectomy should be done before pharyngeal flap surgery (Shprintzen et al., 1992, Shprintzen, 1988)

Now for some data

- ❖ **Sample population: 562 people with VCFS who had VPI surgery**
- ❖ **348 evaluated recommended for specific surgery by RJS**
- ❖ **46 of these failed previous surgery elsewhere**
- ❖ **26 sphincter, 10 flaps, 10 palatoplasties**

- ❖ **214 cases with surgery elsewhere**
 - ❖ **Referral unrelated to speech**
 - ❖ **159 failures**
 - ❖ **55 elimination of VPI**
 - ❖ **51 pharyngeal flaps**
 - ❖ **4 sphincter pharyngoplasties**
 - ❖ **0 palatoplasties**

Criterion for success

Normal resonance for non-nasal speech

Factors Unique to VCFS

- ✓ **Palate and pharynx anatomy and physiology**
- ✓ **Risk**
 - ✓ **Surgical risk**
 - ✓ **Post-surgical risk**

Obstructive Sleep Apnea

- ✓ **Risk of OSA essentially the same for pharyngeal flap as sphincter pharyngoplasty (Witt et al., 1996), perhaps even slightly worse.**

Obstructive Sleep Apnea

- ✓ **Risk of OSA reduced from approximately 10% to 1 - 2% with adenotonsilleomy done pre-operatively.**
- ✓ **In this series, frequency of OSA was 2% for all cases, but 1% for cases with pre-operative T&A.**