

The Virtual Center for  
Velo-Cardio-Facial Syndrome



Issue 8

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## VCFS News: Your Information Resource

### Peer Groups

The VCFS peer group program has the sole purpose of bringing together people with VCFS in a social setting. This program launched just a few weeks ago but the response has been tremendous, with participants eagerly awaiting the next session. Our participants range in age from late teens to 40's and live in various locations throughout the USA and several international locations. The groups are very casual and a lot of fun as we discuss everything from cooking to music to the weather where each of us lives, school and work and hobbies. We are purposely keeping the peer groups small so anyone can participate in the conversations. We also create break-out rooms in Zoom to keep the groups small. We would love for you to participate in these fun, engaging peer groups! If you are interested, please contact Raymond Cheng, Director of Peer Relations at [raymond.cheng@vcfscenter.org](mailto:raymond.cheng@vcfscenter.org). We look forward to seeing you!

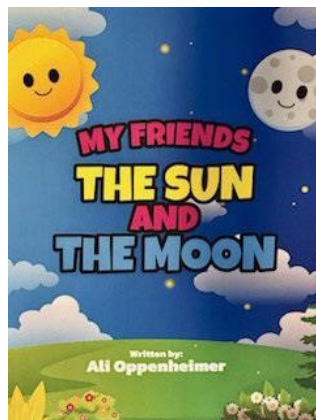
### Issue #8 Content

VPI (VeloPharyngeal Insufficiency): Q&A with Dr. Antonio Ysunza beginning on page 2

#### FREE OFFER!

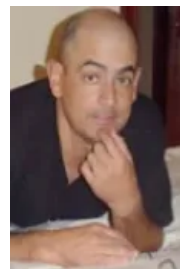


Author, Ali Oppenheimer



As we announced in previous Newsletters, we have made a limited, but substantial supply of a wonderful children's book available to anyone who wants one while the supply lasts. The book, written by Ali Oppenheimer shown here, is a wonderful story reflecting the wonderful smile that always occupies Ali's face. To obtain a copy of the book, simply send a request by email to Jodi Bloom at [jodi.bloom@vcfscenter.org](mailto:jodi.bloom@vcfscenter.org) or Raymond Cheng at [Raymond.cheng@vcfscenter.org](mailto:Raymond.cheng@vcfscenter.org). If you are interested, Ali's personal story is highlighted in issue #5 of the Newsletter, and it is a story about someone who overcame the diagnosis of VCFS in a remarkable way.

## About Tony Ysunza



**Antonio Ysunza, M.D., Ph.D.** is a graduate of the Medical School of the National University of Mexico in 1978. He completed a 3-year medical residency in Audiology, Speech Pathology and Neuro-Otology at the National Institute of Communication in Mexico City in 1984. After residency, Dr. Ysunza completed a fellowship at the Albert Einstein College of Medicine in New York City. During this fellowship he met and worked with often Dr. Shprintzen who had just recently published the first paper describing velo-cardio-facial syndrome. After returning to Mexico City, Dr. Ysunza received a M.S. degree with emphasis in Speech and Hearing Sciences from the National University of Mexico in 1987.

He then earned his Ph. D. degree with emphasis in Neuroscience from the National University of Mexico in 1992. Dr. Ysunza was a medical researcher at the Hospital Gea Gonzalez in Mexico City until 2012. At the present time he is a member of the biomedical staff of the Speech-Language Pathology department at William Beaumont Hospital in Royal Oak, Michigan. Dr. Ysunza has published over 100 scientific papers in peer-reviewed international journals, mainly focusing on craniofacial anomalies and has lectured all over the world, including the recent VCFS symposium in Trieste, Italy that is featured on the home page of the Virtual Center's website.

He has been especially interested in the diagnosis and treatment of resonance and speech disorders in patients with VCFS. He is fluent in English and Spanish. He also understands French, Italian and Portuguese. He was the Regional Director for Latin America for the Velo-Cardio-Facial Syndrome Educational Foundation, Inc. from 2003 until the Foundation completed operations in 2014. He has been an Associate Editor of the International Journal of Pediatric Otolaryngology for decades. According to Dr. Shprintzen, "Dr. Ysunza is truly the world's leading expert and practitioner in the application of advanced techniques for diagnosing VPI (velopharyngeal insufficiency). I am proud to have had him as a Fellow more than 40 years ago, and to watch him become a super expert unrivaled anywhere in the world."

## QUESTIONS AND ANSWERS FROM TONY

1. **What is VPI or VPD (another term some people use)? How do you know when to test to see if my child has velopharyngeal insufficiency (VPI)?**

VPI, an abbreviation for velopharyngeal insufficiency, occurs when the velopharyngeal sphincter cannot perform its function properly. The velopharyngeal sphincter is a valve

that helps to direct air or block air that goes through the vocal tract during speech, and it also prevents food and liquid from going into the nasal cavity during swallowing. In humans, the main function of the vocal tract is to produce speech. The tract starts at its lowest part at the vocal cords and ends on the lips and for some sounds, the nose. Besides the vocal cords and the lips which work as valves, there are other structures which function as valves producing speech.

One of these valves is the velopharyngeal sphincter which includes the soft palate, the posterior wall in the back of the throat, and the lateral walls of the throat on the sides. The function of this valve is to block the passage between the throat and the nose. When the sphincter is not working correctly, the air flow escapes through the valve creating an excessive nasal resonance, and sometimes nasal emission or audible airflow through the nose during speech sounds that require a high oral air pressure, like p, t, s, z, sh, j, tsh, dj, f, v, k, and g. The result is nasal speech and nasal emissions of air when there isn't supposed to be any.

VPI is a resonance disorder. The most common cause of VPI is the structural anomaly of cleft palate, but there are also other causes including disorders of the nerves and muscles of the throat, such as strokes causing paralysis of the muscles of speech, the presence of excessively large tonsils that prevent the structures of the throat from moving, trauma to the throat or palate, and congenital anomalies of the brain such as cerebral palsy. A speech pathologist or a physician can know that a patient presents with VPI by a thorough examination of the nose, oral cavity, throat, and most importantly speech.

However, it is necessary to have a trained ear to detect the abnormal nasal resonance and nasal emission. There are procedures for the evaluation of VPI including endoscopy and X-ray procedures (radiography), but a complete clinical history and thorough examination are the most crucial elements for the diagnosis of VPI.

## **2. Can speech therapy cure VPI?**

Speech therapy cannot correct VPI that is caused by structural problems such as those caused by VCFS. A surgical procedure is necessary in nearly all cases of VCFS who have VPI. However, the treatment of VPI in some rare cases of non-structural disorders can sometimes be treated with medications (as in myasthenia gravis) and some inconsistent cases of hypernasal speech can resolve by themselves, as in strokes if there is a recovery from a temporary paralysis.

There is sometimes a type of hypernasality, or nasal air escape called phoneme specific VPI, also called snorting or a posterior nasal fricative, that is a sound substitution that is learned, and which is easily corrected with speech therapy. Phoneme-specific nasal escape can occur in VCFS, but most VPI in VCFS occurs on all normally non-nasal sounds. VPI is

found in about 70% of people with VCFS and is related to both structural problems of the palate and movement disorders of the muscles of the throat. Essentially all these cases will require surgery.

Although a surgical procedure is necessary to correct VPI, the presence of VPI at birth can cause compensatory articulation disorders (CAD) that require intensive speech therapy. Moreover, speech therapy may be necessary to be continued following the surgical correction. CAD is a learned disorder caused by the child trying to make pressure consonant sounds like those discussed in question #1. Because air escapes through the nose when children attempt to make these sounds, they are not heard as the correct production of a “p” or “s” sound and the child substitutes a sound produced by the vocal cords. This sound is called a glottal stop. To hear this sound, click the link: <https://vimeo.com/1020731885glottal stop+>.

In this clip, you can hear that all the initial consonant sounds are glottal stops and sound identical, but incorrect and unintelligible. The surgical treatment of VPI does not correct compensatory speech disorders. The surgical procedure restores or supplements the function of the velopharyngeal sphincter, but it does not correct the compensatory speech disorders. Glottal stops will persist, and speech intelligibility will only increase when speech therapy corrects the articulation impairment.

**3. In your opinion, what is the best way to determine exactly how to treat velopharyngeal insufficiency (VPI)? Are there any special diagnostic tests that need to be done, and why?**

A complete clinical history and thorough examination of the oral cavity, the throat, and the nose are essential to determine the cause of VPI. If VPI is secondary to a structural anomaly such as cleft palate, VPI requires a surgical procedure or a prosthetic appliance. The surgical procedure is always the best option, and surgery is applied to almost all cases of VCFS unless their heart anomalies are so severe as to make them poor candidates for surgery which is exceptionally rare.

The medical literature has extensively reported that experienced clinicians and surgeons should customize the diagnosis and treatment individually in every case. There is no “one size (i.e. type of operation) fits all” for treating VPI in every case. The surgical procedure must be “tailor made” according to the specific characteristics of the velopharyngeal sphincter. These characteristics require imaging procedures for an adequate analysis including endoscopy (nasopharyngoscopy, also called nasendoscopy) and dynamic X-ray procedures or video-fluoroscopy in multiple views, usually referred to as multi-view videofluoroscopy or multi-planar videofluoroscopy. These procedures are not options; they are both required in every case.

The rationale is that each procedure provides valuable data for customizing the surgical procedure. The surgeon should never perform a procedure for correcting VPI without a careful examination of the velopharyngeal structures by endoscopy and fluoroscopy during speech. The surgeon should plan each procedure according to the imaging findings provided by the preoperative tests.

**4. Is there anything special about VCFS that requires additional testing than, for example, VPI but in someone who does not have VCFS?**

VPI in cases of VCFS can occur because of a cleft palate. However, the cleft palate is most often a submucous cleft (separation of the muscles in the palate that is under the skin of the palate) or often an occult submucous cleft palate. Occult submucous cleft palate means that the anomaly is not visible by an oral examination of the palate and can only be determined by endoscopic examination and even then, requires an experienced and knowledgeable examiner. The word occult translates as “mysterious”, indicating it takes a bit of detective work to recognize it. The medical definition of “occult” is “a problem not accompanied by easily discernible signs or symptoms.” VPI can also occur in cases of VCFS without any palate malformation. There are other causes in cases of VCFS including low muscle tone or hypotonia that can affect throat muscles, the angle of the skull base, and very large tonsils that interfere with the movement of the palate or lateral pharyngeal walls. Because VPI in cases of VCFS is different than VPI caused by cleft palate in the general population, the diagnosis and surgical treatment should be specific and comprehensive.

Surgical procedures which can be effective in cases of VPI not associated with a syndrome, are usually ineffective in cases of VCFS. Another issue is that VCFS can be frequently associated with a malformation of the internal carotid arteries. This represents a substantial risk of bleeding when the surgeon operates in the throat. Thus, an imaging assessment of the arteries is indispensable before planning a surgical procedure for correcting VPI in cases of VCFS.

Magnetic Resonance Imaging of the blood vessels (Magnetic Resonance Angiography or MRA) or a Soft Tissue Neck CT or CT angiography scan can provide adequate assessment of the position and anatomical course of the carotid arteries. Tonsils and adenoid (lymphoid tissue like the tonsil but situated behind the nose) are almost always removed (tonsillectomy and adenoidectomy, or T&A) before surgical procedures for correcting VPI. This is to prevent postoperative obstructive sleep apnea which is a more prevalent risk factor in people with VCFS because of their hypotonia of the throat muscles.

Published data has confirmed that T&A prior to reconstructive surgery for speech dramatically reduces the postoperative risk of airway obstruction. In short, treating VPI in people with VCFS is different than it would be for non-VCFS cases of VPI.

**5. Is there anything dangerous about the testing that you are recommending?**

There is nothing dangerous about the tests for assessing VPI. However, experienced clinicians should be the ones performing these tests. What can be extremely dangerous is to perform surgical procedures for correcting VPI without previous imaging procedures – tests and careful planning resulting in failure and additional operations.

**6. Do all clinicians who treat VPI do these tests? If not, why not?**

Concerning correction of VPI, very few clinicians perform the necessary tests to achieve the best outcome in cases of VCFS. The main reason is that clinicians may not be aware of the specific characteristics of VPI in cases of VCFS. The result of not performing adequate preoperative testing and planning is a high number of failures. Inexperienced clinicians and surgeons can think that they do not need the tests to perform the surgical procedure for correcting VPI, but this is because they do not have the experience and training in these cases. Speech pathologists in training in many universities do not have access to advanced courses in VPI. The issue is usually included in a course on anatomy, and discussed in a very limited manner, typically by people who would not be considered experts in that subject. Because few graduate and postgraduate programs in universities and medical schools teach or even know of the procedures, they have not been taught to more than a few clinicians in their professional education. We are very proud that within the Virtual Center, we have three experts who have been performing and advocating for these procedures around the world for more than 40 years. They have published scientific reports that demonstrate the markedly improved outcomes from the outcomes of surgery when they are used. We stress that the success of surgery for the treatment of VCFS is both surgical skill, but also a careful and thorough diagnostic evaluation.

Not having the resources for performing the tests is never a justification for doing surgery without them. The risk of failure and even potentially fatal consequences is too high. A patient with VCFS and VPI should be under the care of experienced clinicians in an institution with all the necessary resources to perform adequate preoperative testing and surgical treatment. Both nasopharyngoscopy and multi-view videofluoroscopy were first described in the medical literature more than 50 years ago. Although there have been some refinements in the procedures and better equipment developed to perform them, the basic techniques are unchanged.

In 2021, the procedures were discussed at a symposium at the Burlo Garofolo Children's Hospital in Trieste, Italy where they are now being implemented successfully. You can see the presentations from that meeting by accessing the video in either English or Italian on the homepage of the Virtual Center website at [www.vcfscenter.org](http://www.vcfscenter.org).

**7. At what age should these tests be done? Please explain why.**

A thorough examination of speech and resonance requires adequate or at least acceptable speech and language development. Thus, it is necessary that the patient can produce speech appropriately before the test to plan the speech therapy and surgical procedure. In most cases, this means waiting to implement the tests until four years of age in most children, although in some rare cases, successful examinations can be done at slightly younger ages. This allows resolution of VPI just before school age and children being old enough to cooperate with the procedures. There is no upper limit for age.

**8. Does the language that someone speaks (English, French, German, Spanish, Japanese, Polish, Mandarin, etc.) make a difference in how you would evaluate or treat VPI?**

The native language is not crucial for performing or interpreting the tests. But the clinician performing the procedures should be fluent in the specific language spoken by the patient or there should be a clinician, preferably a Speech Pathologist who is fluent in the specific language to provide assistance during the tests. The Speech Pathologists assisting for the tests does not necessarily have to be familiar with the procedures, but it is important that a person with knowledge of how the velopharyngeal sphincter works during speech is essential.

Speech pathologists assisting in the procedure and interacting with the patient, especially young children, are as important as an experienced radiologist for a successful examination, and it is important that the speech pathologist be able to communicate with the patient in their native language. If that is not possible, someone who can translate instructions and responses from the patient should be available.

**9. How long does each test take and are they painful?**

None of the tests are painful when done properly. Nasopharyngoscopy may cause some mild discomfort in young children, but it is not painful. Most children easily tolerate fluoroscopy after 3 years of age. An adequate preparation for the tests enhances the best compliance during the procedure. The endoscopy takes about 5 minutes. Fluoroscopy should not take more than 40 seconds to avoid excessive exposure to X rays. Thus, both procedures, but most especially fluoroscopy should be analyzed on the recording not during the performance of the test. Compliance with videofluoroscopy is close to 100% as long as young children are told exactly what will be happening and what they will be asked to do and say.

Having a prepared speech sample that can be rehearsed with the child should be a part of

the preparation. The speech sample should include all consonant sounds in the child's native language and should include phrases that the child can repeat and that they are capable of saying. If the child's language is not sufficiently developed, then the procedures should be deferred until it is sufficient to repeat the speech sample. Isolated vowels should not be a part of the speech sample used, such as ooo, ahhh, or eee because velopharyngeal closure does not happen in most people who do not have VCFS during isolated vowel production and this may be misinterpreted as VPI.

**10. Do these tests affect the outcome of surgery to eliminate VPI? Is there research that proves the importance of the tests?**

Preoperative testing is paramount for achieving the best outcome of the surgical procedure for correcting VPI. There are clinical research papers in high impact scientific journals describing the importance of these tests. When the surgical procedure is planned according to an adequate clinical assessment and imaging findings, the success rate has been reported to be well over 90 percent.

**11. Do any of the tests involve radiation from X-rays? Should I worry about this?**

Fluoroscopy involves radiation (X-rays). That is why the test should be performed by experienced clinicians, and a standard protocol should be strictly followed. If the test is performed according to a carefully designed protocol, the American Association of Physicists in Medicine reports that the dose of radiation represents no risk. It should be emphasized that the analysis of fluoroscopy must always be performed on the recording of the test, not during the performance of the procedure.

**12. What specialists perform these tests? Should they be reviewed by more than one person?**

An interdisciplinary team should be involved in performing and analyzing the test, but as mentioned in previous newsletters, a "transdisciplinary" team is best. This means that the team members not only perform and discuss the results of the tests, but they know what the other members do and how they do it. The team must include at least the Craniofacial Surgeon, the Speech Pathologist, the Radiologist, and the Otolaryngologist. Each member performs their specific tests, but each case should be carefully discussed and planned individually considering all aspects.

