The Virtual Center for VCFS

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VCFS News: Your Information Resource

OUR ANNUAL APPEAL -- YOUR SUPPORT MATTERS

Your Support for the Virtual Center for VCFS Matters

As we approach the end of 2025, our registrants and friends will once again receive several emails from me asking for your support for the year ahead. As you know, whether someone speaks with our experts once or fifty times, we never charge for the help we provide. With nearly 2,000 registrants -- many of whom rely on us multiple times each year -- our services remain in high demand.

Although our operating costs are modest compared to many non-profit organizations, we still have essential expenses and financial commitments. This is why your support of the Virtual Center for VCFS is so vital: we simply cannot continue offering these services without a reliable annual budget.

Many of our registrants have benefited from our peer groups, and we are now engaged in potentially groundbreaking research with the potential to make a real difference for people with VCFS who suffer from mental disorders. While we are honored to offer these programs, we need the resources to sustain them.

We are a truly one-of-a-kind organization dedicated entirely to providing information and support to families navigating life with VCFS. Many registrants have reached out personally to express their gratitude for the help they receive—freely and without conditions.

As the year comes to a close, please consider supporting the Virtual Center.
We cannot do this important work without you.

Our Peer Group Leader, Raymond Cheng, speaking at the Retreat of 22q Texas, Inc. November 2025

Our highly successful peer groups would love to have YOU participate! Their only purpose is for the participants to make friends and socialize. Every session so far has been interesting and engaging, with a variety of topics to chat about, upcoming events and/or holidays to look forward to as well as online games. There have been laughs and deep connections. We would love for you to be part of this hour of fun and conversation! If you would like to join us, speak with me to find out more about the groups and how to join one.

Contact me by email at raymond.cheng@vcfscenter.org and we will set up a call to discuss your participation. I look forward to speaking to you!

Research: Why Do We Do It, and Why Aren't We All Living to Be 250 Years Old?

Robert J. Shprintzen, Ph.D.
Founder and Director
The Virtual Center for Velo-Cardio-Facial Syndrome, Inc.

Introduction

I am often asked the question, "What do you do?" I think the intent of the question is to ask about my professional background, my education, my expertise, etc. I get the same question when people must ask me the question, such as agents checking my passport when I leave or enter the country if I am traveling. I often need to scratch my head to think about what to say. I have degrees in psychology, biology and speech pathology (with a heavy emphasis on physiology), and I do use all those fields to define what I do, but my avocation since 1974 has been in human genetics. Initially, my colleagues referred to me as a "syndromologist," a term popular in the 1970s when my mentors, David Smith, M.D., Robert Gorlin, D.D.S. and M. Michael Cohen, Jr., D.M.D., Ph.D. were my idols. Syndromology morphed into "dysmorphology" in the 1980s, and then eventually became known as clinical genetics, although I also have dabbled in the field of molecular genetics in my research. I became very active in the field, publishing more than 260 articles in scholarly journals, more than 50 chapters in textbooks. I have published 7 textbooks all of which were about human anomalies, including diagnosis and treatment. I continue to learn since earning my Ph.D. in 1973. I have benefited from learned colleagues in many fields including medicine, psychology, dentistry, molecular and clinical genetics, and from research, both my own, and from people around the world who have published. In the field of biomedical research, there are between 1.5 million to 2 million articles published every year, and that number is increasing every year. My wife, Debby, who is an avid reader of both fiction and nonfiction books, speaks to me often of what she is reading, or what she would like to read. The range of her reading is enormous. She has asked me before why I don't read the kind of books that line the shelves of Barnes and Noble, Amazon, etc. The short answer is that I have no time. I read hundreds of pages every day, but it is all relevant to my fields of study and practice. The last book I read was Mel Brooks' autobiography which made me smile and laugh but caused me to fall behind in reading journal articles. In short, I am addicted to reading and writing research.

My reading of research and science has also led me to some adjunct interests in my career. I was the Editor in Chief of the Cleft Palate-Craniofacial Journal in the 1980s and not only read many hundreds of submitted research papers each year, but also was the deciding point for whether, or not the material would be published in the journal. I have also been a reader for many journals including the American Journal of Medical Genetics, The International Journal of Pediatric Otolaryngology, The European Journal of Human Genetics, Clinical Genetics, The Journal of Medical Genetics, The American Journal of Psychiatry, to name only a few. Therefore, I not only read published journal articles, I also read many articles that never get published because the quality of the research and the conclusion drawn from the research are not scientifically sound. Because the majority of articles submitted to scholarly journals are not published, the actual numbers of written articles are in the millions.

Why Do People Publish Research?

There are many reasons why people publish research. Some of them are commendable and some are not as commendable as one would think. The main reason, I would like to believe, is to share science with the world and to advance their fields of endeavor. Journal articles do have that effect. When I published my first article in an attempt to describe "A New Syndrome Involving Cleft Palate, Cardiac Anomalies, Typical Facies, and Learning Disabilities: Velo-Cardio-Facial Syndrome," my motive was to increase recognition of a disorder that I thought had not been a recognized entity before that time. I was wrong, but I do believe that the paper was a strong impetus to increasing knowledge and recognition of the syndrome so more work could be done to improve treatment. I was wrong because cases with the syndrome had been published long before my submission to The Cleft Palate Journal. Angelo DiGeorge published cases of the disorder in 1968. Angelo was a wonderful and humble man and physician who worked at St. Christopher's Hospital in Philadelphia, and he focused on the endocrine, immunologic and cardiac disorders in infants with absent thymus. Also, a pediatric cardiologist, William Strong, published a family in 1968 with two generations affected with the syndrome indicating the probability of autosomal dominant transmission, but not ruling out X-linked dominant inheritance. But even before Angelo DiGeorge and William Strong, there was Eva Sedláčková, a in Czechoslovakian phoniatrist who published multiple cases of the syndrome in 1955 in a Czechoslovakian pediatric medical journal that received very little attention on the Western side of the iron curtain. Please note that my assumption of VCFS being a new Discovery was incorrect, but I tried to correct it in future publications on multiple occasions. Such corrections, unfortunately are rare in the medical literature.

People also do research to advance their careers. When scientists work in universities, medical schools or other academic institutions, their pay and their prestige is dependent on their academic level. In the USA, and most other countries, academic ranks include, from lowest to highest, Instructor, Assistant Professor, Associate Professor, and Professor (often called Full Professor). Salary increases with each promotion, and often, the work load is not as heavy and comes with benefits such as fewer classes to teach and teaching assistants who can fill in for them. In many countries, each department has only one Professor who is in charge of the department. In the USA, there can be multiple Professors in addition to Assistant and Associate Professors in a singe department. The Chairman in a department need not be at the rank of Professor in most USA institutions. Promotion requires three elements that influence the decision for promotion, often referred to as the "three-legged stool." The three elements are Grants, Publications, and Teaching excellence. When I worked at Montefiore/Einstein in New York City, I was put up for promotion by my chairman in 1985 and my credentials showed that I had a few grants that brought money into the Hospital and Medical School, many publications (more than 150 peer reviewed journal articles, five chapters in edited books, and one textbook).

I had lectured in many hospitals and universities in the USA, Europe, Australia, and Asia plus routinely lecturing at Grand Rounds at my own institution and had been interviewed on television and radio in New York City and nationally dozens of times which added to my teaching profile. In today's atmosphere in most places in the USA, I might not have been promoted to full professor at the young age of 39 years quite so easily because I did not have as many grants as might have been expected today. Today, grants are the most desirable component because it brings millions of dollars to universities with very little effort on the part of the institution. The point is that the difference in pay, perks, and position between being a Professor (senior faculty) and an Assistant Professor (junior faculty) is substantial. With grants, publications follow because it is an obligation associated with getting funded, especially with grants from a governmental agency such as the National Institutes of

Health (NIH). Last year, the NIH distributed 10.5 billion dollars in research funding for health-related research.

The previous paragraphs described some of the aspect of research that may not be known to most people and in many instances, the general public may be impressed by individuals who have a publication track record. But in the health sciences, I ask the question, with many millions of scientific publications, billions of dollars spent on grants, and so many professors among our ranks, why aren't we all living to 250 years and not having back pain at the age of 65? Average life span in the U.S.A. increased from 71 years in 1961 to 76 in 2010 according the to the U.S.A. Census. Over the same time span from 1910 to 1960, life expectancy in the USA went from 51 years to 69 years. Publications in the medical literature from 1961 to 2010 were increased by hundreds of percentage points over those from 1910 to 1960, but life span increase was more robust from 1910 to 1960. Why?

Most medical researchers will tell you that one reason for this discrepancy is the development of antibiotics that occurred with the invention of Penicillin in 1928 that also led to the development of other antibiotics in the years that followed. That makes sense. But shouldn't it also make sense that with the development of cures for cancer and other life-threatening illnesses that we see frequently these days that we should see a concomitant increase in life span boosting us a bit higher than seven years?

As a former Editor-in-Chief of a medical journal, I realize that much of the published literature in the field of health care is incorrect, misguided, or possibly even worse, made up. Moreover, what is thought to be true today, may not be so in 5 or 10 years. Researchers for the most part are honest and understand that their publications could have a major impact on people's well-being. It is also true that good quality research is not easy. There are many variables that need to be controlled in research and sometimes there are so many variables that it is impossible to control them all and some of these variables could alter the outcomes of the investigation. The following paragraph is a fictional example, but I have reviewed many papers with such problems.

> *A new vaccine is being developed to prevent people from being infected by the Frackle-Dackle protozoa that is often found in the reservoirs of two countries in the southern part of Europe, East Bippy and West Bippy. The name of the protozoa comes from the name of the reservoirs where the organism was originally found, the Frackle Reservoir being a mile from the eastern border of East Bippy and the Dackle reservoir one mile from the western border West Bippy. Both nations have been found to have large concentrations of the Frackle-Dackle protozoa, based on earlier research of two esteemed scientists, Brigitte DuFlambée in East Bippy and Angelo Fusillini from West Bippy.*
>
> *In analyzing the frequency of Frackle-Dackle infection, scientists were curious why many people in East Bippy suffered from the infection whereas very few people in West Bippy did, including people who lived on the shores of both bodies of water. Investigators concluded that because almost the entire population of East Bippy were ethnically derived from French ancestors, while almost the entire population of West Bippy was derived from Italian ancestors. They concluded that this must be a genetic effect based on slight genomic differences between the French who were descended from Gauls and Italians who were descended from Romans. Therefore, they were looking to develop a vaccine based on the "French gene."*

For decades other scientists researched the genomic differences between East Bippians and West Bippians but could never find one. The reason was found by a resident of North Bippy who on his vacation decided to take a three-week road trip through East Bippy and West Bippy where he ate at the finest restaurants in both countries. When he came home, he had lunch the next day with his good friend who wanted to hear about this buddy's trip. This friend was a microbiologist. The traveler said that his trip was wonderful and both East and West Bippians were wonderful, warm and cordial people, the hotels were nice, and the scenery beautiful. What he did not understand was that the cuisines of the two countries were quite different even though the countries bordered each other. In East Bippy, people at a lot of meat with delicious sauces, especially creamy sauces like Béarnaise sauce. In West Bippy, almost all meals were accompanied by pasta with acidic tomato-based sauces. His friend yelled "Eureka. Do you realize you may have just cured people in East Bippy from the scourge of Frackle-Dackle infections!" His friend explained that the Frackle-Dackle protozoa that was present in the water from both border reservoirs could not survive in a person's stomach when exposed to high acidity foods and gluten in combination. But the protozoa thrives on eggs which is a main ingredient in Béarnaise sauce causing the protozoa to replicate very rapidly causing the illness.

First, let me apologize to anyone from East Bippy, West Bippy, Italy or France. While I have not visited East or West Bippy (and obviously never will), I have many friends in both France and Italy and I have visited both countries many times. The example in italics is obviously fictional. But the example shows that it is almost impossible to control all variables when dealing with human subjects that often results in terms of data interpretation.

Two Major Types of Medical Research: Basic Science and Translational Research

There are many different types of research often dictated by the field of study of the researcher. The focus of this article is the broad field of medical research that by itself has many subcategories of investigations and literature. Research investigations can be related to outcome studies to see what treatments are successful and what are not. There are population studies that assess everything from the distribution of health care to birth and death statistics. This article is focused on two major fields of investigation: genomic research that is basic science investigations and translational research that has the purpose of bringing new treatments to patients as rapidly as possible.

Often, these two types are studying the same things with different methods and, also different scopes. The substance of this article is obviously my own interpretation that is derived from being a researcher for more than 52 years. In my own situation, I have done both basic science and translational research that in years past was labeled as clinical research. My concern about most basic science research is that much of it provides specific information that adds little or no information that can be translated to benefit care of an individual. This type of basic science research will often lead to additional research that can be shown to be useful in treat…sort of a building block for determining if there is more to pursue in terms of coming up with better treatments. When this is the case, it will mean that treatments for an illness or medical problem will be years away. With translational research, the information that is gained will tell you that the results will directly lead to the application of a technique, a medication, or approach to a problem that will prove to be successful.

Perhaps one the best examples would be the research involved in the elimination of polio in the United States and ultimately most of the world. It was preceded by important work in the laboratory…basic science…that allowed Jonas Salk to develop what he thought would be a method to create a vaccine that would prevent people from developing polio if exposed to the polio virus by using a killed polio virus to stimulate a person's immune system to produce anti-polio antibodies. His first step, which is the translational portion after his lab work, was to test his vaccine by injecting volunteers with the vaccine to see if they would produce the anti-poliomyelitis antibodies. The volunteers included himself, his wife, and his children. With this translational evidence Salk, Salk was able to get funding from the National Foundation for Infantile Paralysis which later became known as the March of Dimes because the money for the trial was financed largely by small donations of dimes gathered from small tin cans placed in every store, restaurant, and office. In 1954, when I was in third grade, a consent form for participation in a clinical trial was sent to every family in my school, the Henry Barnard Elementary School in the New Rochelle, NY public school system, and my parents signed it. One million children participated in the study that was placebo controlled. In other words, some got the vaccine, some were injected with sterile water. One year later, the results were released…the vaccine worked to prevent polio. Those, including myself, who were given the actual vaccine, were called Polio Pioneers. We received a small pin to wear on our shirts and a card that I still have today, 72 years later.

The bold steps taken by Salk don't happen often, but I find myself asking, "Couldn't we take such brave steps to better the lives of people with VCFS?" At this point in time, we know that treatments for the structural issues in VCFS can be managed very well if one understands the unique nature of the syndrome. Heart surgery, surgery for hypernasality, the treatment of hypocalcemia, the treatment of thyroid disorders, the management of immune disorders, scoliosis, and more are managed well in VCFS with outcomes that are as consistently as good as they are in the general population. My friend Bruno Marino in Rome, Italy who has a wealth of experience with VCFS and cardiac surgery has reported in multiple studies that for the most part, surgical outcomes for congenital heart disease in VCFS is essentially the same as it is for people with similar heart malformation who do not have VCFS. My experience with the treatment of velopharyngeal insufficiency has shown that our success with surgical correction is approximately 95% and our experts based in the Detroit area, Drs. Ysunza and Rontal, have published a similar 95% success rate in eliminating VPI. Where treatment has not been as good in VCFS is primarily in the area of behavioral and psychiatric disorders. In both the instances of Drs. Ysunza and Rontal, and my own experience in two different institutions with different surgeons, but research done to assess how the diagnostic procedures influence the surgical outcomes, the success rate was essentially identical. This conformation of translational research has dramatically improved surgical management of VPI so that nearly all people with VCFS can have normal speech.

Where are We Now and What is Next?

There have literally been thousands of research papers published in medical and scientific journals about VCFS. In the early years, many of those papers were descriptions of the syndrome meant to familiarize readers with the clinical features so that recognition would become more common. As more cases were described, individual anomalies of the syndrome were studied in more detail in order to understand the association of the anomalies with specific genes. Studies included the use of

an animal model, usually mice, because mice had nearly all of the same genes on mouse chromosome 16.
These studies were valuable in terms of understanding the mechanism of gene effects but did not reveal much information about treatment. Research in laboratories continues but the application of those studies to treatment has been rare. Why?

I have been following several thousand individuals for 50 years and established interdisciplinary programs for VCFS first at Montefiore Medical Center in the Bronx, then at Upstate Medical University in Syracuse. One of the things I learned was that the thing my patients and their families worried about the most was the chance that they would develop psychosis. In 1992, I published a report in *The American Journal of Medical Genetics* of psychosis being a common clinical feature of the syndrome. My colleagues and I diverted much of our research to mental illness in the syndrome because of the concern expressed by our patients and the frequency of the finding with poor treatment outcomes. In that 1992 publication, my colleagues and I reported that many of the cases had been diagnosed by their community doctors as having schizophrenia. I did not say that we thought our patients were schizophrenic. In subsequent publications, investigators believed that their VCFS patients were bipolar. We at the Virtual Center don't believe that either.

However, if one reads the literature from many authors, the authors often label the psychiatric problem as "schizophrenia." The result of this label is that people with VCFS and psychosis are labeled as schizophrenic and treated with medications used to treat schizophrenia in the general population. Those cases labeled as being bipolar were being treated with mood stabilizers and bipolar medications. Reviewing our registrants, the positive response rate to these medications is generally poor.

So, what is the mental disorder in people with VCFS? It is simply VCFS-related mental illness, or VCFS-related psychosis if psychotic behaviors are present. The classifications of mental illness found in the DSM 5 (Diagnostic and Statistical Manual of Mental Disorders, 5th Edition) refer to labels such as schizophrenia, schizoaffective, bipolar, etc. as brain-related conditions and disorders. They are not diseases. There is no blood test or X-ray or laboratory test that can detect any of those conditions. Really what they are is observations or descriptions of things that people are doing or saying. In VCFS, we know what is causing the mental illness seen in the syndrome…THE DELETION. In determining a treatment for the problem, we have the advantage of knowing the cause because all of the genes in the deletion have been identified and their contribution to mental illness has been hypothesized…the question (hypothesis) is, which gene is it and can we treat what the deletion of that gene does. Can we prove it? Can this information be translated to treatment? The answer to the question is "yes."

Enter William D. Graf, M.D., a child neurologist who, I am proud to say, was a resident who rotated through my service as an elective during his residence in pediatrics at Montefiore Medical Center in 1985-86. During his rotation, he saw many cases with the syndrome. Following a Fellowship in neurodevelopmental disorders at the University of Washington he was on the faculty at the University of Washington in Seattle where he planned and implemented a study based on this hypothesis:

> *The commonly deleted region in 22q11.2 deletion syndrome spans more than 30 genes, one of which is the gene for catechol-O-methyltransferase (COMT). COMT inactivates catecholamine neurotransmitters (dopamine [DA], epinephrine [EPI], norepinephrine [NE]) by O-methylation. A deficiency in COMT could result in higher concentrations of catecholamines or abnormal ratios of O-methylated to deaminated catecholamine*

metabolites and could contribute to the neuropsychiatric manifestations of the 22q11.2 deletion syndrome.

Very interesting. Psychiatrists have long thought that dopamine (an essential neural transmitter that modulates pleasure, movement, mood, attention, stress response, blood vessel function, focus, learning, and more) if present in excess in the brain causes abnormal responses by neurons (brain cells) causing dysfunction and mental illness. Because people with VCFS have one copy of the *COMT* gene rather than 2 copies, and for most of our functions, it takes 2 copies of a gene to perform a task correctly, the elimination of dopamine from the brain after it has done its job will be impaired so that too much dopamine stay in the brain causing the neurons to malfunction. To test this hypothesis, Dr. Graf measured the amount of dopamine used in the central nervous system to confirm the excess and then treated 5 people with VCFS and neuropsychiatric/behavioral dysfunction. He treated them with a drug, metyrosine, which has the job of preventing the production of dopamine by inhibiting tyrosine hydroxylase, the enzyme that is the first step in the production of dopamine. Four of the five cases in the study had marked improvement in behavior and a reduction in psychotic behaviors and anxiety. All cases in the study who had improvement continued to take metyrosine after the study was completed. Dr. Graf, I should mention, is not only on our team of superb clinicians at the Virtual Center, he also serves on our Board of Directors.

Metyrosine is an FDA approved medication for the treatment of hypertension in patients who have a rare adrenal tumor, pheochromocytoma. It reduces blood pressure by reducing the stiffness of the blood vessels which is largely regulated by dopamine. It has not been approved for use in VCFS (but neither has any other drug been approved for the use in VCFS), but it has been used to treat a substantial number of people who have VCFS-related psychosis. We are in the process of preparing a paper reporting our experience with dozens of cases with a success rate consistent with the report published by Dr. Graf. This is a perfect example of translational research. The research started in the laboratory and finished with the treatment of patients. We continue this research with much larger numbers of cases and, also trying other approaches to perform the same task. Expect to hear more about these efforts in the coming year. We are very excited with these advancements.

For more information about this success please access our web site and on the home page, click on 2. Treatment of psychiatric disorders in VCFS. You will be able to access a meeting held in Trieste, Italy in 2022 at the Burlo Garofolo Children's Hospital. The psychiatric material begins at 2 hours 50 minutes into the meeting. Our psychiatric expert, Dr. Gianni Faedda, our psychologist, Bronwyn Glaser, and I present out information relative to our experience. We will, of course, speak with anyone interested in our findings and experience. We ask you to register with us and there is no charge for this and our web site can be accessed by anyone, anywhere in the world.

The beautiful artwork of Amy Isenberg of Prairie Village, Kansas

Our artist, Amy, showing one of her recent artworks in her studio. A talented artist, she has sold some of her excellent works that have been displayed in prominent Kansas City locations.

New Professionals Joining Our Center

We are pleased to announce the addition of two new members of our team, with extensive knowledge of VCFS: **Robert Marion, M.D. and Michael Mars**, D. Orth., Ph.D.

Dr. Marion is a clinical geneticist, first introduced to Dr. Shprintzen when he was a medical student at the Albert Einstein College of Medicine. Dr. Marion is recently retired from most of his many duties at Montefiore Medical Center and the Albert Einstein College of Medicine. He was Executive Director of the Children's Evaluation and Rehabilitation Center and the University Center of Excellence in Developmental Disabilities at the Rose F. Kennedy Center. He was Chief of the Divisions of Genetics and of Development Medicine at the Children's Hospital at Montefiore and Director of the Center for Congenital Disorders. A faculty member at Einstein since 1984, Dr. Marion's interests include the natural history and genetic basis of multiple malformation syndromes.

At The Children's Hospital at Montefiore he served as Medical Director of the Spina Bifida Center for more than 20 years, was the founder and Medical Director of the Williams Syndrome Center, and he helped organize the Center for CardioGenetics, the Neurofibromatosis Center, and the Center for Excellence in Autism. He has published extensively in the medical literature in these areas and, in addition, is the author of seven books. Dr. Marion is the recipient of Albert Einstein College of Medicine's Samuel Rosen Award for Excellence in Medical Student Teaching (selected by medical students) and the Alumni Association's Lifetime Service Award. He is also the winner of the Lewis Fraad Award for Residency Education and the Obrinsky Award for excellence in medical student teaching in the Department of Pediatrics. Dr. Marion also received the Zella Bronfman Butler Change Agent Award, given by the UJA-Federation of New York.

Dr. Michael Mars of the United Kingdom is an orthodontist who has been one of the leading dental specialists in the field of cleft palate and craniofacial disorders. He was the Lead Consultant Orthodontist for the Cleft Lip and Palate Centre at The Hospital for Children at Great Ormond Street (often referred to as GOSH) in London from 1983 to 2013. He was also the Director of Special Surgery at GOSH for Maxillofacial, ENT, and Plastic Surgery. He held the position of Honorary Senior Lecturer in the Dept of Developmental Biology at the Institute of Child Health. He has been and continues as a prolific author of research literature and is the creator of the Goslon Yardstick which is used internationally to assess facial growth outcome in children with cleft lip and palate.

He has traveled and lectured extensively internationally and is an Honorary Fellow of the Sri Lankan College of Paediatricians and is also an Honorary Fellow of the Royal College of Speech and Language Therapists in the United Kingdom. His extensive work in Sri Lanka is well-known and is a Visiting Professor on the Faculty of Medicine at Peradeniya, Sri Lanka, and at the University of Malaya in Kuala Lumpur, Malaysia. He is the Founder of the Sri Lankan Cleft Lip and Palate Project and directed that organization from 1984-2009. He was also a Founder and Chairman of Trustees of CLAPA (Cleft Lip And Palate Association) (1979-2005) and Founder and Chairman of Trustees of MAGE (Medical Aid to Galle Sri Lanka) 2004-2011 post tsunami.

Dr Mars is also Past President of the Craniofacial Society of UK and Ireland, Hunterian Professor of the Royal College of Surgeons of England and has been a Lecturer and course organizer in UK, America, Mexico, Japan, China, Thailand, Malaysia, Israel, S Africa, Sri Lanka, India, Russia, Sweden, Italy, Denmark, Holland, Belgium, Brazil and Australia. Besides his many peer-reviewed papers in the medical literature, he was senior co-editor of the textbook, Management of Cleft Lip and Palate in the Developing World.

In Memorium: Dr. Alan Shanske

It is with a heavy heart and much sadness that I am reporting on the passing of one of our team's experts, Dr. Alan L. Shanske, of blessed memory.

Alan and I worked together for many years, initially at Montefiore Medical Center and the Albert Einstein College of Medicine in the 1990s, until he passed away on October 31, a month and a half ago.

I first interacted with Alan in the 1970s when he was on the faculty of Schneider Children's Hospital at Long Island Jewish Hospital where he was the Director of the Craniofacial Center, and subsequently at Montefiore Medical Center and the Albert Einstein College of Medicine where he was Professor of Pediatrics, Pathology, Obstetrics and Reproductive Medicine, and Dentistry. A native of New York City, Dr. Shanske eventually settled in New Rochelle, NY where coincidentally I grew up, as did Dr. Golding-Kushner, our speech pathology expert.

Alan served his country as a medical officer in the Air Force. He was a board-certified pediatrician, clinical geneticist, and cytogeneticist. As a researcher, Dr. Shanske published 127 peer reviewed scientific papers and many chapters in scholarly textbooks. His name lives on for his work in the identification of a genetic syndrome, Levy-Shanske syndrome. I worked closely with him when he joined the team of the Velo-Cardio-Facial Syndrome Institute at Montefiore Medical Center in the Bronx in 1995, where we saw patients together every Tuesday and learned from each other continuously.

When I left Montefiore in 1997 to take a position at Upstate Medical University, Dr. Shanske took over my former position as Director of the Center for Craniofacial Disorders at Montefiore. We remained close friends and colleagues until his passing. I shared with him the passion for publishing a paper about anything he found that was new and exciting. He was also a teacher to the medical students, residents and Fellows he took under his wing, as well as the patients he counseled.

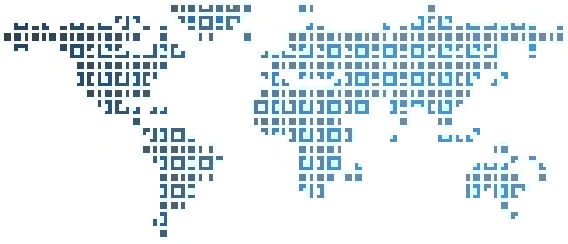
Among his many qualifications, he loved clinical genetics the best, and he particularly enjoyed speaking to the patients he counseled. He was an original member of our team at the Virtual Center and he loved his interactions online, even after he was confined to a wheelchair. On a more personal level, Alan's professional life was important to him, but most important was his devotion to his family and friends. Alan and his wife, Sara, were married for 55 years and their three children -- Darien, Alisa and Uri -- were more important to them than anything else.

Alan adored his grandchildren. At his 80th birthday celebration, a gigantic ballroom was filled with what looked like hundreds of friends and relatives who recognized Alan's kindness, humor and dedication. I will miss Alan terribly, but I am a better person for having been a close friend. His battle with the cancer that took his life after battling it for decades showed me a man with an

amazing love of life, family, and knowledge. I will no longer hear his stories about fishing, playing handball in the Bronx, traveling the world, how much he liked the tongue sandwich at Liebman's Kosher Deli, and more recently, his newest granddaughter who was "so beautiful."

Alan and I had very similar backgrounds, complementary senses of humor, and a thirst for knowledge. He was my brother from a different mother. I was privileged to know him, and I will miss our weekly telephone calls that continued to his last week of life. I know that someday Debby and I will be sitting with Sara or Uri and will share laughter over things that Alan did or said, or look at photos of Alan in front of a fighter jet or in the jungles of Costa Rica, but after the laughter dies down, it will be followed by a few minutes of solemn silence as we wish he were sitting with us.

Robert J. Shprintzen, Ph.D.
President and Director
The Virtual Center for Velo-Cardio-Facial Syndrome, Inc.



The Virtual Center for VCFS