The Virtual Center for VCFS

Sunday **September 21, 2025** **Issue 11**

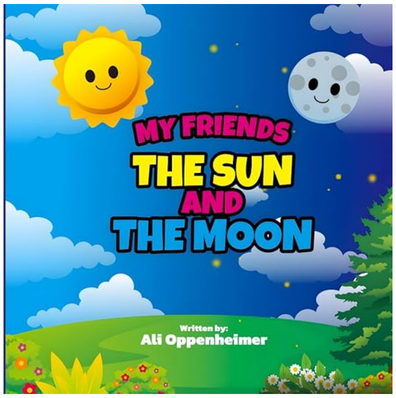
VCFS News: Your Information Resource

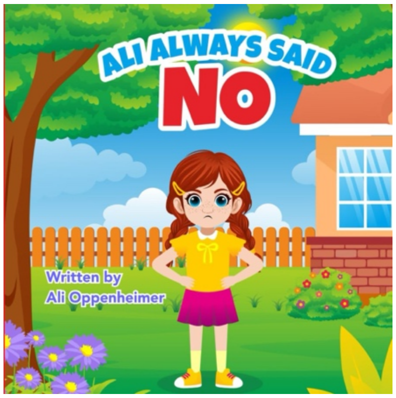
Issue 11 Contents: Q&A session with Gianni Faedda, M.D., Psychiatric Expert for the Virtual Center. Also, “Japan in a few bites.”

CALLING ALL VIRTUAL CENTER FOR VCFS REGISTRANTS!

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!





A generous donor of the Virtual Center for VCFS is still offering free copies of *Ali Always Said No* from our own Ali Oppenheimer. If you are interested in receiving a free copy, please contact Raymond Cheng with your name and mailing address at raymond.cheng@vcfscenter.org

Your Psychiatric Questions Answered:

The opinions expressed below are those of the staff of the Virtual Center for Velo-Cardio-Facial Syndrome and are often part of the information we provide to our registrants. We understand that these opinions may differ with those of your own health care providers or consultants. Our opinions are based on our experience, research, and personal opinions, as well as interactions with other professionals around the world. We also understand that such opinions also may change over time as our own research and that of others progresses and sheds new light on old subjects and knowledge. We are always happy to discuss these opinions and provide evidence for why we believe them to be valid. We do not intend to label anyone else offering opinions as being "wrong," nor do we believe that we are always right. We understand that there is no clinical application that is always right or always wrong. As is true of all good scientists and clinicians, we continuously seek the truth.

Q: I was told that my child with VCFS is autistic. Is this common in VCFS, and does it mean we treat him for autism in school and in therapy?

A: Some "so called" autistic traits are relatively common in children with VCFS, though a full autism spectrum disorder (ASD) diagnosis is very infrequent. Social communication difficulties, anxiety, and attention deficits can be mistaken for features of autism, such as social anxiety and shyness with poor or avoidant eye contact. These overlapping features, such as social anxiety, concrete thinking, and sensory sensitivities are simply clinical features and can occur in people who do not have either VCFS or autism, people who have other genetic disorders including Fragile X syndrome and Rett syndrome among others.

Clinical features are simply descriptions of an observation independent of a diagnosis. For example, sneezing is a clinical feature, but an observation of someone who is sneezing a lot could be an upper respiratory infection, allergic rhinitis, too much smoke in the air, dust, a strong aroma, and many more medical or environmental causes. Because the clinical features of people who have been labeled as autistic overlap with clinical features of VCFS does not mean that people with VCFS are autistic. What we do know is that there are treatment implications to making the assumption that the clinical features of so-called ASD (autism spectrum disorder) overlap with some clinical features of VCFS. We know what causes these clinical features in VCFS, but the cause of most cases of ASD is still a mystery in most occurrences.

Educational and therapeutic plans should always be individualized. Even without an ASD diagnosis, social skills training, structured learning supports, and sensory accommodations can be helpful. Treatment should be based on needs, not just diagnostic labels. Some people accept the diagnosis of ASD because it opens up funding resources for people diagnosed as autistic. But what if those resources are not applicable to people with VCFS, or even harmful to them?

Q: Our daughter has had learning disabilities throughout school, but we still think it would be great if she can go to college to see what kind of career she might have. We had her tested and her IQ was 71, that she had a severe anxiety disorder, and that she also had ADHD. Do you think sending her to college is a good idea?

A: An IQ of 71 is in the borderline intellectual functioning range, and severe anxiety plus ADHD may make traditional college environments stressful without substantial supports. However, success is possible if:

- She attends a program with strong academic accommodations (e.g., reduced course load, tutoring).
- Vocational or community college options with practical skill training are considered.
- Anxiety and ADHD are managed with therapy, coaching, and possibly medication.

Focusing on her strengths, interests, and learning style would be essential. A career assessment and a structured transition plan can help clarify if post-secondary education is realistic and safe for her mental health. It would be important to focus strongly on living arrangements, especially if roommates are involved. It may be that living independently in a college environment would be overwhelming and cause severe anxiety. College environments can also expose students to drug use and other social activities that would be harmful to people with VCFS and cause serious anxiety leading to more severe mental disorders. For many people with VCFS, independent living skills are not well developed and we find that in some cases, people take advantage of that innocence. In many cases, if college is being considered, a lighter course load and living at home near a local college makes more sense.

Q: Our son, who is 17 years old, seems to have developed a seizure disorder recently. His eyes roll back, he becomes stiff, his hands tremble, and after about 5 or 10 minutes, he stops. The second time it happened, we brought him to the emergency room, and they did an EEG and said his brain waves were normal, but they gave him a seizure medication anyway. While it was happening, we thought he was unconscious, but he told us he was awake the whole time. Does this happen to people with VCFS a lot?

A: VCFS increases the risk for seizures, but your son's description—maintained awareness, prolonged duration, normal EEG—may suggest a non-epileptic event (e.g., psychogenic nonepileptic seizure, often called PNES.

Recommendation:

- Referral to a neurologist familiar with VCFS for repeat or extended EEG/video-EEG.
- Consider metabolic and medication review. Many psychoactive medications are of little benefit to people with VCFS and can actually add to anxiety that might increase the risk of have PNES.
- Monitor for anxiety, sleep deprivation, or psychiatric triggers.

  Seizure-like episodes in VCFS need careful workup to differentiate epileptic events from non-epileptic events in order to specify the course of treatment.

Q: We have a 22-year-old daughter who is doing OK and does not have too many problems. She does tend to be very shy, and she doesn't really have many, if any, friends. She is afraid to do a lot of things that most people do, like going to a restaurant or a movie. We were told that most people with VCFS who have psychosis usually have it when they are teenagers. Should we believe that she will not develop psychosis because she is older than that? Is she still at risk?

A: While many cases of psychosis in VCFS have onsets in adolescence or early adulthood, late-onset cases do occur. The lifetime risk for psychotic disorders in VCFS is elevated well into adulthood. Shyness, avoidance, and anxiety are not necessarily predictive of psychosis, but any changes in thinking, perception, or functioning should be monitored.

Recommendation: Maintain regular psychiatric follow-up and encourage gradual social engagement to reduce isolation and support early detection. The Virtual Center for VCFS is running a highly successful peer group that allows people with VCFS to get together online weekly in a purely social, fun environment so you may want to consider getting her involved in the peer groups.

Q. We are parents to a beautiful little girl, now 2 years old. She does not have congenital heart problems and medically, she is doing very well. Is there any way for us to tell if she might grow up to have mental illness? Is there anything we can do to avoid it?

A: It is not possible to predict with certainty if she will develop a mental illness. In general, the risk is higher for anxiety, distractibility, and learning difficulties. Protective factors include:

- Early developmental surveillance.
- Enriching, supportive environments.
- Good sleep, healthy routines, and positive social experiences.

While early intervention services (speech, occupational therapy, social skills support) can improve long-term outcomes, it would be important to make sure that the proper therapies are provided because treatments are often syndrome specific. It is probable that many therapies that are effective in the general population are not well-suited to children and adolescents with VCFS. It may be difficult to find people who are knowledgeable about VCFS, and not all people who claim to be knowledgeable can back up that claim, so contacting the Virtual Center to determine if you are on the right track might be a good idea. We should add that many people with the syndrome have escaped severe psychiatric disorders and difficulties in life. We have followed many cases where a parent of a recently diagnosed child was found to have the deletion, but no one suspected it or diagnosed it. We should always keep in mind that health care professionals only see people who are referred with problems. We have no way of knowing with accuracy how many people with the deletion are leading normal lives because they have no reason to see us.

Q. My son was a good kid during his childhood, but when he was 16 in high school, we thought he had started talking to himself, but it soon became clear that he was talking to people who weren’t there, and then one night he started yelling because he thought he saw a bear in his room. Everything went downhill from there and I brought him to a psychiatrist who thought he had a psychotic break and was possibly schizophrenic. He wasn’t sleeping well and one night he ran out of the house without his clothes on during a rainstorm and was found by the police who brought him to a hospital which they said they had to do. They kept him in the emergency room and gave him some sedatives, I think it was valium, and eventually he fell asleep. They kept him in the hospital for a few days, and he saw a psychiatrist after a couple of days who started him on Abilify. When that did not calm him down, they added some other medications to calm him and help him sleep. They discharged him after a week and now he is home, but he is very psychotic, not talking to us, sometimes throwing things, talking to the voices in his head, and not taking care of himself. We don’t know what to do, and the medicine is obviously not helping. What should we do?

A. This description of clinical findings suggests severe, treatment-resistant psychosis in VCFS, which can be challenging to manage. Common steps include:

- Comprehensive re-evaluation to confirm diagnosis (rule out seizure disorders, metabolic problems, medication side effects).
- Try to find as much information as you can about treatments that have been shown to be effective in people with VCFS, including both medications and behavioral therapy.
- Combine medication with behavioral strategies, structured daily routine, and career support.
- Coordination between psychiatry, neurology, and genetics teams.
- If psychosis is severe and self-care is impaired, hospital-based stabilization or specialty referral is warranted. Treatment choices for VCFS are most often different than those for people who do not have VCFS. Finding a good information resource in terms of treatment protocols would be very important, such as the experts at the Virtual Center for VCFS. Keep in mind that what is treated in cases with a confirmed deletion is the syndrome, not the list of clinical presentations. Psychiatric diagnoses and medications recommended for those specific diagnoses are specifically tested or approved for use in people with VCFS. In other words, no clinical trial has ever been completed for any psychoactive medication and therefore, the use of any drug is experimental.

Q. (Same person as in the previous questions): I contacted you about my son who had a psychotic break and was hospitalized when he was 16. He has been seen by a local psychiatrist, and he has been on at least eight different antipsychotics and many other medications, but nothing has had any effect on his psychosis. He is also taking medications given to him to get rid of some of the side effects he has, like tremors, incontinence, and aggressive behaviors. Are there any medicines that work well for VCFS cases? Nothing seems to help.

A. Some individuals with VCFS and psychosis respond poorly to typical antipsychotics. In some cases, clozapine has benefited some treatment-resistant cases, but it requires careful monitoring for side effects (including seizures and agranulocytosis). Lower doses may be needed due to increased sensitivity. Adjunctive mood stabilizers, antidepressants, or anti-anxiety medications may help target specific symptoms. There have been reported cases of a good response to dopamine depleting medications like metyrosine and methyldopa. Elevated levels of dopamine in the brain of people with VCFS have been demonstrated and related to one of the deleted genes in the syndrome. Medications that can prevent the formation of dopamine can lower those levels to a more normal level which has been shown to be effective in many cases. We have seen many very good responses to these medications and some people have been using them for many years, some over two decades. For more information on this approach, go to the Virtual Center web site and view the video from a meeting that was held in Trieste, Italy at the Burlo Children's Hospital in 2022. The entire video can be accessed on the Home Page of our web site at www.vcfscenter.org. The video is nearly 6 hours long, but is in two sections. The first half, running approximately 2 hours 48 minutes, is devoted to the diagnosis and surgical treatment of hypernasal speech. The second half is devoted to psychiatric and psychological with three speakers.

Q. Is the treatment of schizophrenia in VCFS different than it is in people who have schizophrenia but do not have VCFS?

A. First, although the psychotic behaviors seen in VCFS are found in people who have VCFS, it is not schizophrenia. It is VCFS-related psychosis. There is a unique pattern of the illness in VCFS, including the lack of positive response to treatments typically applied to people who do not have the syndrome. The cause of the psychiatric illness in VCFS is unique to VCFS…a deletion of genes that have been identified as contributing to the illness. Core treatment principles are similar to "schizophrenia" without VCFS, but important differences include:

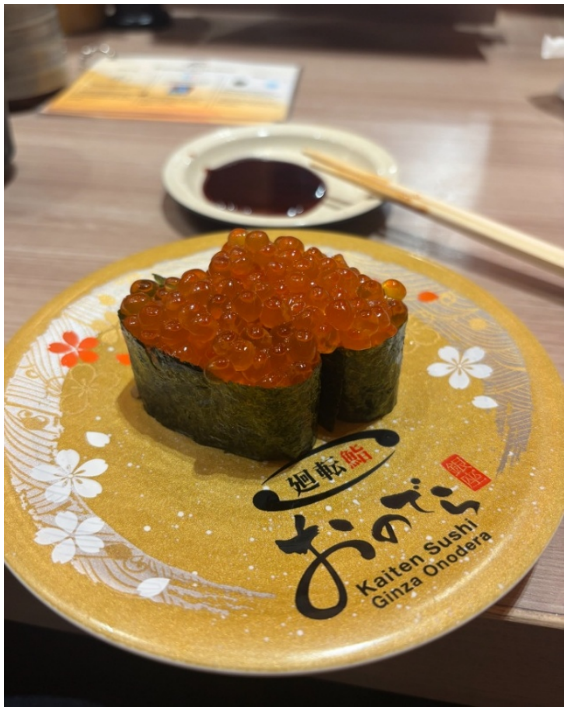
- Greater medication sensitivity which may result in negative side effects. If commonly used medications are implemented, start low, go slow.
- Higher risk of side effects, especially movement disorders, seizures, and metabolic issues.
- Increased need for psychosocial rehabilitation and cognitive remediation due to learning differences.
- Close coordination between psychiatric and medical/genetic teams.
- Treatments aimed at reducing dopamine levels in the brain have provided benefits to VCFS patients with psychotic symptoms. This is an off-label use of such medications that may reduce the ability to obtain them. These effective medications do not focus on the uptake of dopamine by working on the dopamine receptor. Rather, they act on nullifying the formation of dopamine so that levels of dopamine in the brain are lower. Excessive dopamine levels have been known to cause psychosis and other neurogenic disorders, and one copy of the gene that controls that function, catechol-O-methyltransferase is deleted in people with VCFS. Therefore, the focus in people with VCFS is the genome, not the "diagnosis" of so-called schizophrenia.

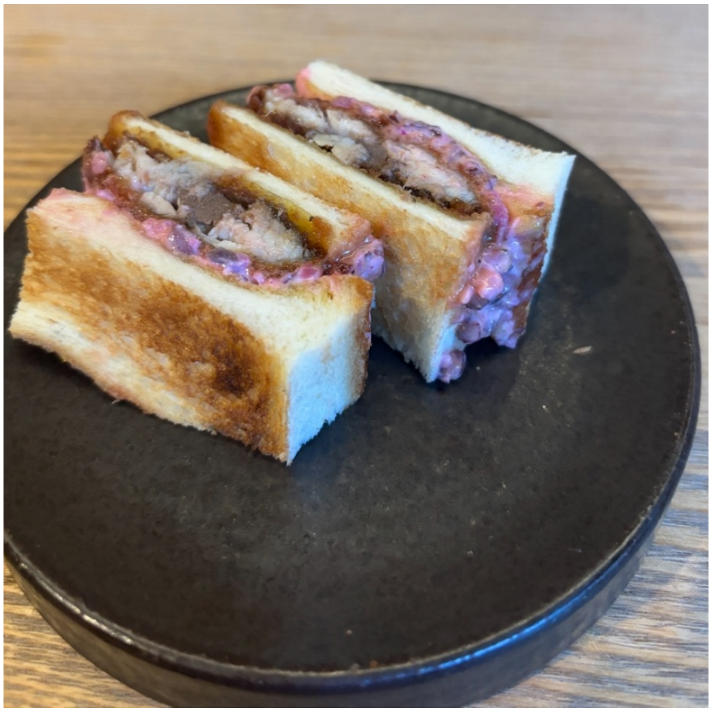
FOR A MORE DETAILED DESCRIPTION OF THE APPROACH DESCRIBED BY DR. FAEDDA, VISIT OUR WEB SITE, AND ON THE HOME PAGE, FIND THE LINK TO A MEETING HELD IN TRIESTE, ITALY IN 2023, OR USE THE LINK SHOWN BELOW. THE MEETING'S RECORDING IS ABOUT 6 HOURS LONG, SO FOR THE PSYCHIATRIC PORTION OF THE MEETING, FAST FORWARD THE RECORDING TO 2 HOURS 38

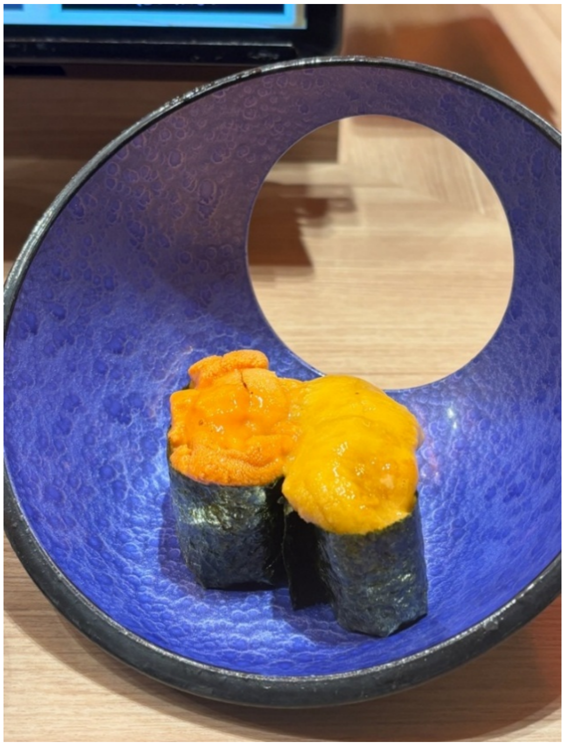
MINUTES AND YOU WILL BE ABLE TO ACCESS SESSIONS BY DR. SHPRINTZEN, DR. FAEDDA, AND DR. GLASER. THE FIRST TWO HOURS AND 48 MINUTES ADDRESS THE ISSUES OF HYPERNASAL SPEECH AND SURGICAL MANAGEMENT.
https://www.youtube.com/watch?v=zjpNvu7Y-0A&t=11427s

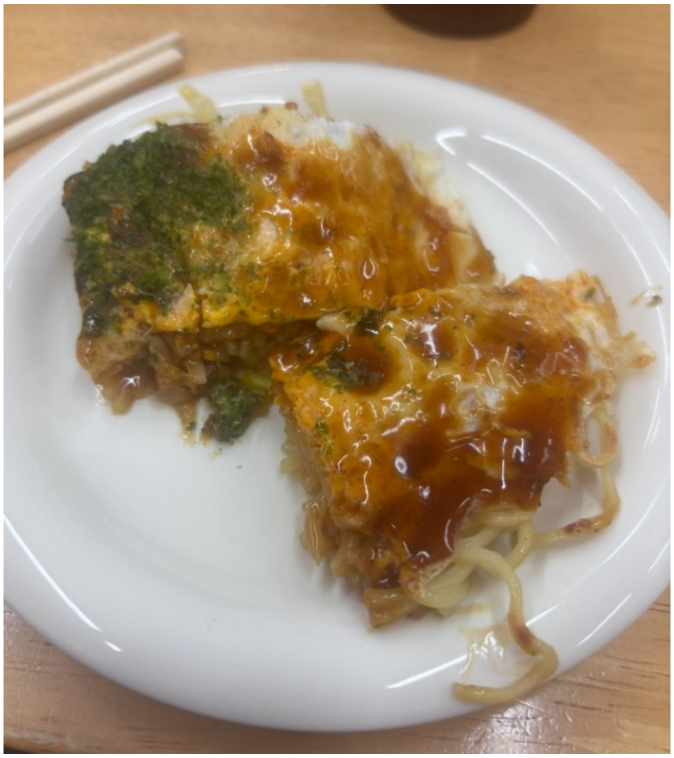
Japan in a few bites …

These great pictures are courtesy of one of our registrants who recently returned from a trip to Japan! Our registrant is a true foodie and he works in a restaurant which enhanced his interest in sharing these photos. The pictures are an example of some of the delicious food that was sampled during their trip.









A Moving Experience

Raymond Cheng
Director of Peer Groups and Newsletter Editor

It’s been more than a month since I’ve been living in my new place in Yonkers, and I have to say, it’s been an awesome experience so far. I grew up in a close suburb of New York City in Westchester County and then lived in three different boroughs of New York after graduating from college. For more than ten years, I bounced around from Astoria in Queens to the Upper West Side of Manhattan, and then to Brooklyn. Yonkers is in Westchester County and is the third largest city in New York State with a population of approximately 212,000 people which is larger than the capitals of 31 state capitals. In New York State, only New York City and Buffalo have more people. I grew up in a much smaller town in Westchester. Moving away from the familiar surroundings of New York City by myself with my dog was a terrifying concept to me. I knew that life would be different. On the positive side, I grew up nearby in Scarsdale. The house I grew up in is only a ten-minute drive, and it is easy for my parents to come visit me and help take care of my dog if needed. I knew it would be different. Even as a sizeable city, Yonkers doesn’t have the hustle and bustle of the “The City.” By comparison, it is peaceful and quiet. The air is fresher. There are no rats running around the streets, and very little worry about dangerous people in the streets.

My apartment is near a shopping complex called Ridge Hill, and within that area there is a Whole Foods right across the street, a well-maintained dog park directly next to it, a TJ Maxx, Starbucks, Shake Shack and many more stores and activities. The drive or train ride into New York City is only about 30 minutes from Manhattan, and only 5 minutes or so from the northern most part of the city, the Bronx which borders Yonkers. By comparison, it is so quiet outside. No sirens, no drama or fighting heard in the middle of the night. My dog has already made a lot of friends in the dog park (although sometimes he still gets scared of the bigger dogs). And I love that my parents are so close. It was extremely difficult for them (or anyone) to visit me in Brookly which was at least an hour away. The amenities are abundant (there is a swimming pool here!), and the apartment itself is so spacious, you wouldn’t think it’s a studio.

The downsides of the apartment, I’ve discovered, is that the walls are paper-thin. The neighbor directly next to me can hear me playing music if loud enough, and she has a baby that I can hear crying through the walls (luckily, she is friendly, and we’ve exchanged numbers). There is also construction going on in the back (they are installing more amenities for the building), and sometimes the tractors and jackhammers wake me up early in the morning. Because this is the suburbs, there are more bugs that are spotted throughout the apartment. Since living here, I’ve killed multiple spiders and flies that have entered. That is pretty much the only downside of living in this place and I can’t complain about it much because the pros outweigh it by far. Nowhere and nothing is absolutely perfect.

It's a scary thought to move somewhere completely new and different, something that I know can be difficult for people with VCFS. I will admit that packing was hard, including dealing with the moving company, installing the Wi-Fi and electricity in the new place, the finances that went along with it (moving is expensive!), and everything else in between. It was stressful. Organization was key. The last few months I've struggled financially micromanaging finances to make sure I don't go under. Hopefully by living here, the next few months I can save up some more and get my life back on track.

It was terrifying to move to a completely new location, but it was also exciting. Sometimes, change is good. A fresh look in a new life is sometimes necessary, and it's what I realized I would need. As much as I wanted to keep living in the city, I realized that life would be much better here for me and my dog, both spiritually and financially. I can see myself living here for the next few years at least. Hopefully I can make that happen. I can't wait to see what the rest of the year has in store for me.

To our readers:

We are always looking for new material to present in our Newsletter. If you have suggestions, curiosity about a particular subject, or questions you would like answered, please submit them to us by emailing our Editor, Raymond Cheng, at raymond.cheng@vcfscenter.org, or to our Director of Development and Community Relations, Jodi Bloom at jodi.bloom@vcfscenter.org. You can also call our clinical coordinator, Christine Phillips at +1 (315) 884-7676 or email her at christine.phillips@vcfscenter.org.

The Virtual Center for VCFS