



FACES Today

The Newsletter of FACES: The National Craniofacial Association

Living with Pfeiffer Syndrome My Life's Journey As Seen Through My Special Eyes

By Jessica Kastl

I was born with a rare genetic disorder called Pfeiffer Syndrome, which prevented part of my facial bones from growing. When I was only six days old, I had to undergo my first operation. I have since had twenty-seven operations over the past twenty-six years of my life, for which I spent nine endless summers in New York for facial reconstruction. No local surgeons knew how to perform these operations. However, these very physically painful, emotionally draining, and mentally stressful times have helped me to overcome several major obstacles and challenges in my life. I saw and experienced firsthand how and why social supports are so vital to patients.



Jessica Kastl is a former FACES client and currently lives in Washington.

I remember the exceptional kindness of nurses, social workers and

other hospital staff that helped me mentally and emotionally get through these tough times. These experiences made me commit myself to providing heartwarming support to individuals like me and sparked my passion for children in the healthcare setting. Children facing even the most critical illnesses can be tough, and my experiences have enabled me to assist them in focusing on the positives instead of the negatives. It is the struggles and victories in my life that have deeply affected my choice for a social work career.

When I realized social work was the field in which I could touch many lives, be committed to social justice, and support people in developing confidence to get the most out of life, I knew it was my destiny. The social work seed was planted when I was a Teen Mentor in high school. It was then that I realized how much I enjoyed working with children and making a difference. I received my Bachelor degree in Social Work at Pacific Lutheran University.

Having been born looking abnormal to everyone in the outside world, I truly have accomplished remarkable achievements. After

twenty-six years of my face appearing different and dealing with individuals who did not understand why I looked different, I have overcome all this and have been told I turned into a radiant young woman who is rising above all the limits and expectations that others have placed

on me. For eighteen years a tracheotomy tube helped me to breathe, a gastronomy tube fed me liquid diets, and on different occasions health professionals told my parents I would probably not be able to talk and need to use sign language. Now, the tracheotomy tube has been removed and I have achieved a lifelong dream of learning to swim after eighteen years of being medically unable. I rejoiced like a fish finally back in the water. After many hours

of demanding swimming practices, I made a giant leap forward. At the end of my sophomore year, I was awarded the only Athletic Achievement Award given out that year by Cottey College. I have slowly learned to eat solid foods for my primary diet, and since the age of five have been speaking like everyone else without any problems. Many professionals told my parents and me that I would never live a normal life. I have proved them all wrong and left them in disbelief. From these personal experiences, it is my desire to help others to succeed and overcome medical and emotional challenges they must face.

My struggles and accomplishments have given me an understanding of how difficult life's course can be. I know getting support and help during challenging times can make life's bumpy path much smoother. From personal experiences and knowledge, I have the unique ability for compassion to relate to those in extremely unfortunate situations.

A couple of years ago, to many people's amazement, but not surprise, I graduated from

(continued on page 2)

UPCOMING EVENTS

Velo-Cardio-Facial Syndrome

The 11th Annual Meeting of the Velo-Cardio-Facial Syndrome Educational Foundation will be held on July 29-31, 2005 at the Syracuse Sheraton University Hotel in Syracuse, New York. For more information, call 315-464-6590 or go to www.vcfsef.org.

FACES 4th Annual Golf Tournament

Oct. 3, 2005, Willow Creek Golf Club Knoxville, TN, Call FACES for details 800-332-2373, 423-266-1632

Moebius Syndrome – July 2006

The 7th International Moebius Syndrome Conference will be held July 14-16, 2006 in Burlingame, California. The conference will be held at the Hyatt Regency San Francisco Airport Hotel in Burlingame. For more information, call 660-834-3406 or email conference@moebiusysndrome.com

Inside FACES Today:

Pfeiffer Syndrome Report.....	page 5
Pfeiffer-Parental Lessons	page 4
Wedding Bells for Ashley.....	page 3
A Parent's Thank-You	page 2
Book Review.....	page 6

Eastern Washington University with my Masters in Social Work. After graduation, I worked as a Social Services Director where I was responsible for initiating and maintaining a comprehensive social services program at a skilled nursing facility. These days as a DSHS Liaison, I split my day between a community mental health agency and the local DSHS office, where I am a consultant and resource for my colleagues on both DSHS topics and mental health clients.

The special friendships and bonds I have made with the amazing and unique people I met during my journey through life are my most cherished memories in life. Each and every one of these relationships has shaped a part of who I have become. My family and friends have always been there for me, every day encouraging and giving me hope, even when I could not push myself forward. No matter how tough and unbearable things seemed to be, my family has constantly been at my side cheering me on.

Despite the ordeals I have faced, I have had these loving and caring people by my side at all times. I have also had the generosity of health professionals who went out of their way to ease the physical pain, as well as the emotional and mental anguish. Through all the supportive people in my life has come my passion to help others in dire need like the ones with which I was dearly blessed.

Throughout my journey, I have viewed my life as something on which to continually work. Every day, every action, each

person I meet, my education, my personal experiences, as well as social work professional experiences, have shaped who I am today and will continue to form who I become until my last day on earth. In all my life's adventures, the communities of people with their beliefs, cultural diversity, and values, have influenced my approach to life.

As mentioned earlier, family is a very important value to me. From the day I was born, my mother has deeply affected how I view life and what I do with what life delivers to me. My mom taught me to always look at the positive, no matter what — it is what I do have, not what I do not have. Because of having been born differently, there were many negative medical issues that, at times, have made me want to give up, but my mom's emphasis on the positive instead of on the negative has pushed me through many extremely tough tests and trials. Viewing life in this dimension has made me realize what is truly important in life: family, friends, and helping people that are less fortunate.

As the saying goes, "Life is a journey, not a destination." My life is continually being written. The journey of my life with each experience I have encountered, people I have met, and values I cherish, have shaped who I am. For all the people that have been a part of my life in assisting me during my journey, I plan to make a difference in others' lives by aiding them to help themselves.

You can email Jessica at racoon21@hotmail.com or call the **FACES** office for more information.

A Parent's Thank-You Letter to FACES



Cameron Royes lives in Mansfield, Texas, and has been a FACES client since May 2004. He and his family travel to see Dr. Burt Brent in Woodside, California for ear reconstructive surgery.

Dear FACES,

I am writing to thank you for the financial assistance you granted to my son, Cameron, and me, which allowed us to travel to Mountain View, California for treatment of Cameron's microtia.

Cameron was born seven years ago without a right ear. I learned of Dr. Brent through my mother-in-law when Cameron was only a few weeks old. I've always felt a maternal obligation to seek the best treatment available for my son. I was relieved to hear there was a surgical treatment for microtia that was proven to be very successful.

Earlier this spring, as his surgery date approached, Cameron began to have reservations about the procedure. I searched for advice and support groups to find other families coping with the same situation. I found your organization and was very quickly connected with a family whose son is Cameron's age and also awaiting the surgery. The connection with this wonderful family proved very helpful, helping to ease my fears and reservations, as well as Cameron's — knowing that another boy was having the same treatment helped Cameron to feel differently towards surgery.

During my introduction to FACES, I also discovered the availability of financial assistance. While I was committed to the best treatment for Cameron, the financial burden of fulfilling this commitment made it difficult to achieve. Simply stated, your organization's financial assistance allowed us an otherwise distant opportunity. The resources of your organization and the gifts of your donors bring families together and help them seek treatment for their children.

Cameron has successfully undergone phase I of the four part external ear reconstruction surgery. While in California for surgery, we were able to meet other children who also traveled to California for treatment of microtia. Cameron felt a connection with each one of the children as I did with each of their parents. He is looking forward to completing all surgical phases and having the gift of a realistic looking ear. His smile says it all!

Your organization and your donors have been a beacon of hope for my family. For this, I am thankful and want to express my heartfelt gratitude.

Thank you,
Sheri Royes

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Congratulations Ashley and Patrick!

Ashley Allen, our FACES Client Services Coordinator, recently celebrated her marriage to Patrick Rhodes. Although we rejoiced with them on their happy day, we also had to say "goodbye" to Ashley, as she moved to Virginia where Patrick is in sales with Mohawk Industries. Our FACES staff and clients will miss her dynamic



personality and her faithful service; however, we wish her nothing but happiness as she starts this new chapter in her life.



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 Donate Now! Call 1-866-332-1778 or follow the links on FACES' website: www.faces-cranio.org

FACES Today

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Parental Lessons

By Laurel and Mark Sanborn

The end of one journey is the beginning of the next. This we learned on December 23, 2002. Mid-term pregnancy blood work showing a high risk for Down's Syndrome was dismissed by an amniocentesis which said our soon to be born baby girl was chromosomally "normal". So with that, the rest of the pregnancy had been a journey of anticipation and excitement as we prepared for the arrival of our second daughter. The planned c-section went well, and we were soon holding our 9 lb 14 ounce daughter named Olivia. One of the first things we noticed was that her toes were a little large, but later these thoughts were discarded as we were reminded by family that everyone has different looking feet. We also thought something looked a little peculiar about Olivia's face, but again, the nurses reassured us that it was because she had been quite bruised during the birth process. Then there was the difficulty she seemed to be having breathing. Of course, the nurses must be right when they counseled that it was because all babies rattle a little getting rid of the excess fluid from their lungs after birth. So, things went as planned for about 24 hours.



When the pediatrician visited our room in the morning, we told him about our concerns. He "checked" Olivia out. He was somewhat concerned about her small head size. At that time, the journey we have been on for the last two and a half years began. Within minutes, the Geneticist had come and looked Olivia over and given us her preliminary diagnosis of Pfeiffer Syndrome. One of the residents did some internet research and gave us a few printouts about Pfeiffer Syndrome. We couldn't help but think that somehow an awful mistake had been made. The amniocentesis said "normal". This couldn't be our baby they were talking about.

Over the days and weeks that passed, we had an "official" diagnosis of **Pfeiffer Syndrome Type I**. Since those early days, we've learned some important parental lessons that I would like to share:

- Don't feel like you have to explain everything to everyone. The birth of a child is time of great joy, and as we look back, we wish we had celebrated more. We were so caught up in the "diagnosis" that we didn't take the time to enjoy our precious daughter.
- Don't take too much stock in the "official" diagnosis your child has been given. While Olivia's diagnosis is Pfeiffer Syndrome Type I, she has some of the characteristics of other types.
- Take the "research" and "scientific" information you find with a grain of salt. To this day, we are still terrified when we search for Pfeiffer Syndrome on the internet. Honestly, there are rough days, but it's not like it sounds in a Medical Journal.
- Find a support system. It's sometimes difficult to find this support even from family and close friends

(Continued on back page)

Pfeiffer Syndrome (continued from page 5)

The overall care of a child with Pfeiffer is too complex to adequately review in this article; however, here are some of the basic treatment issues:

- Craniofacial teams will typically first evaluate the infant's breathing, to ensure that there is a safe airway. This may require a bronchoscopy (a procedure in which a lighted scope is passed down the windpipe to look for narrowing) and evaluation with a pediatric pulmonologist. In some rare cases a tracheotomy may be needed.
- Next, it is important to ensure that brain growth has not been significantly limited by the fused sutures of the skull (craniosynostosis). There are many ways to follow this: head circumferences, eye exams, overall development, and MRI scans (looking for cerebellar tonsillar herniation – a condition in which a portion of the brain is squeezed down into the spinal canal that may commonly occur with Pfeiffer syndrome, and hydrocephalus – a build-up of fluid within the brain). With thorough frequent assessments, it may be possible to delay the initial operation to enlarge the front of the skull, which if not done too early, may reduce the total number of operations that the child will need. Severely affected individuals, and children with a cloverleaf skull, will routinely require surgery before 6-months of age; many other children may be delayed to 9 to 18 months of age. Children with Pfeiffer may need anywhere between 2 and 4 skull operations before they turn 18 years old. MRI scans (some doctors prefer CT scans) are done almost yearly to monitor the brain, until growth slows down.
- It is important to carefully evaluate the ears, as ear infections are common early in life, and repeated infections may cause hearing loss.
- As children grow older and approach school age, sometimes sleep apnea (not getting enough oxygen at night) can get worse; therefore, children are often followed with yearly sleep studies.
- Advancement of the mid-face with a LeFort III does more to normalize the appearance of a child than any other single operation. Most children will undergo 2 or 3 of these procedures before age 18. The use of distraction with the LeFort III appears to give much better results, and may reduce the total number of operations required. There are two basic LeFort III distraction techniques: one uses buried devices (first done in San Francisco) and the other technique (developed in Dallas) uses an external halo device called the RED system.

PFEIFFER SYNDROME

Jeffrey A. Fearon, MD

Director, The Craniofacial Center, Dallas, Texas

www.thecraniofacialcenter.org

Pfeiffer syndrome was not recognized as a syndrome until 1964 when R.A. Pfeiffer first described eight individuals with this condition, all in the same family, spanning three generations. Pfeiffer syndrome is not common, and it appears to be even more rare than its "cousins" Apert and Crouzon syndromes.

Pfeiffer Syndrome is a condition resulting from premature fusion of the sutures of the skull and deformity of the skull.

Children born with Pfeiffer syndrome share some similarities to those born with Apert, or Crouzon in that the skull and facial sutures are either fused at birth, or fuse early in childhood. Most parents will notice that when their child is born, his or her eyes look larger than other children, and the eyelid openings may slant downwards. These changes are the result of the bones of the skull and face not being able to grow forward in a normal way. The mid-face, or area from the top of the nose to the teeth, in addition to not growing forward at a normal pace, will also be short and wider. Geneticists usually rely, in part, on their examinations of the hands and feet to tell the differences between Apert, Crouzon and Pfeiffer syndromes. With Apert syndrome, most all the fingers and toes are fused together from birth (called "syndactyly"). With Pfeiffer syndrome, there may be a couple of fusions (or more commonly, none at all), but usually the thumbs and big toes are slightly wider than normal. With Crouzon syndrome, the hands and feet usually appear normal.

Children with Pfeiffer syndrome may have problems with breathing, and may be noisy sleepers at night. This is most commonly because of the poor growth of the mid-face, which results in the nasal passages being smaller (recall how difficult it seems to breathe when you have a cold and your nostrils are all plugged up). In addition to having a narrowed nasal airway, sometimes children with Pfeiffer syndrome will have a section of their trachea (the "windpipe") be abnormally narrowed, as well. If this narrowing is severe enough, it may require surgical correction at a specialized center.

There have been two different genes linked to Pfeiffer syndrome. Most children born with Pfeiffer syndrome are the result of a "new mutation," which means that neither parent carried the gene for Pfeiffer syndrome, but in the process of bringing together both parent's genes, this new gene occurred. There is nothing a mother can do to prevent having a child with Pfeiffer syndrome; that is, Pfeiffer syndrome is not the result of the mother doing anything wrong. This gene is inherited in an "autosomal dominant" fashion, which means that when a child grows up and chooses to have children, there is a 50:50 chance that the Pfeiffer trait will be passed on for each birth. For parents and older children interested in learning more about the genetics of Pfeiffer syndrome, meeting with a geneticist can provide the opportunity for an interactive discussion.

Pfeiffer syndrome has also been broken down into three different types, Type I, Type II and Type III. The difference between these three types is not always clear. Type I is the most common, or "typical" form and children usually have normal intelligence; Type II and Type III are the more severe cases, and are more likely to have problems with the brain and development. Both are associated with a greater risk of death early in life (more aggressive management of the skull and the airways in these children may prevent these deaths). Type II can be differentiated from Type III in that the Type II children are born with a "cloverleaf" skull (also called: "Kleeblattschädel"), and Type III is used to describe the most severely affected individuals without the cloverleaf skull shape.

Craniofacial teams may approach the treatment of a child with Pfeiffer in significantly different ways, but all should closely monitor the child for the various potential problems and strive to perform the fewest number of operations. The best advice I can give parents is to be proactive about your child's care and be your son or daughter's number one advocate. Don't be afraid to ask your doctor lots of questions, feel free to discuss other techniques that you have heard about with your doctor, and ask your doctor why he or she chooses a particular technique (and see if that makes sense to you). Always feel free to get second opinions (good doctors never mind their patients getting second opinions!). Finally, get in touch with other families who have children with Pfeiffer syndrome and share your experiences; you may learn things that can help your child.

(continued on page 4)

COMING UP:

If you would like to submit information on upcoming events, please email Editor@faces-cranio.org before July 30, 2005.

The next quarterly issue of *FACES Today* will feature *Easing the Stress of Back to School*. Also, don't miss the ANNUAL FACES' Golf Tournament coming up on October 3! Call us for details.

Parental Lessons

because even though they mean well, they honestly don't have any idea what you are going through. It was important for us to find a family with an older child with Pfeiffer Syndrome who had been through most all of the major surgeries. We still keep in contact with the first family we "met", even though they are clear across the country. Their daughter is a great inspiration to us. Also, it's important to find a family whose child is similar in age with yours. This, also, has been invaluable to us. Most days, those families are the only ones that can truly understand what we are going through.

- Realize that while this all seems complicated to you, you really are the expert when it comes to your child. Pfeiffer Syndrome is so rare that unless you live in a large city, there is a good chance that you won't have a seasoned Craniofacial Team in your backyard. Your local doctors, medical professionals and care givers will rely on you for information that you learn about Pfeiffer Syndrome. Be ready to be in charge and be an advocate for

(Continued from page 5)

your child.

- Do your research and find the Craniofacial Team that is right for your child and for your family. Living in a rural area, it wasn't a matter of whether we traveled to a larger center but more like a matter of which center we traveled to. Meet with or talk to several qualified teams in person, on the phone, or via email. You will know when you find the team that is right for your family.
- Expect to have your baby enjoy and have an abundant life and grow into a happy, well adjusted individual.

Some days, it seems like our journey has already spanned 1,000 miles, and other days it seems like it's just begun. Thank goodness for the joy we've experienced and things we've learned along the way thus far. Olivia is a bright, energetic, happy 2-year old who has taught us more than we could ever imagine. She has a special love for life and is a happy toddler. She truly amazes us every day.

Olivia Sanborn lives with her sister and her parents in Richmond, Vermont. She has been a FACES client since March 2003.

BOOK REVIEW

Missing Genetic Pieces: Strategies for Living with VCFS written by Sherry Baker-Gomez.

Velo-Cardio-Facial Syndrome (Chromosome 22 Deletion) is one of the most common genetic disorders; however, with up to 187 known symptoms, it is often not diagnosed or misdiagnosed. The information packed into this 550-page book will not only educate and enlighten parents, but will also provide practical information for physicians and educators.

In addition to writing her book, Ms. Baker-Gomez has also set up an excellent website.

Visit www.vcfs-22q11.com for more information. Her book can be ordered through the website or by calling the publisher at 602-789-6416.

Return Service Requested

We would like to remind you that contributions to FACES: The National Craniofacial Association are deductible for income tax purposes under Section 501 (c) (3) of the Internal Revenue Code. You may be assured that each dollar is carefully managed to do the utmost good for those with severe craniofacial disorders.

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