PATIENT JOURNEY CONGENITAL MELANOCYTIC NAEVUS SYNDROME

NAEVUS GLOBAL

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INTRODUCTION

Naevus Global formalized in 2013 connects individuals and families around the world affected with rare forms of CMN. Together with Naevus International network, it provides mutual consultation between patients, scientists, clinicians, psychologists, and other stakeholders. Naevus Global stimulates cross-border collaboration in research, consensus guidelines and international registries to integrate efforts and map medical expertise, in collaboration with the ERN-SKIN.

ERN-SKIN (European Reference Network – skin) is a virtual expert centers network with the aim to improve quality, safety and access to highly specialized healthcare for children and adults with rare skin diseases throughout Europe.

PURPOSE

Patient Journeys represent the collective perspective on the burden of the disease and the needs of people with first-hand experience of living with a rare disease. The Patient Journey for Congenital Melanocytic Naevus syndrome was developed from the perspective of patients and parents, as a reference point for pathways and guidelines.

METHODOLOGY

SKIN ePAG advocates completed a mapping exercise of the needs of the CMN syndrome, across the different stages of the patient journey. These stages progress from first symptoms, diagnosis, possible treatment (surgery), to follow-up care and palliative care.

Patient needs at each stage of the journey are referenced under three levels: clinical presentation; patient needs; recommendations on ideal care.

A first version of the patient journey was presented at the Naevus International conference (2019) with 15 patients and ERN-SKIN patient advocates representing 10 countries in Europe, North & South America and Africa.

PATIENT STORIES

This book contains personal stories from patients, or their family members living in different parts (continents) of the world. They wrote about the impact they have experienced during a phase of the patient journey. It's a document for people whose life is affected by CMN, which can help parents of newborns with CMN have a better understanding of what can be expected in a life with CMN. It will help with empowerment and self-direction. Combined with the poster of the patient journey it will help healthcare providers to better understand the psychological, practical, and medical challenges people with CMN face worldwide.

1.
FIRST SYMPTOMS/ DIAGNOSIS

ANSHIKA

FILIP

LOUJAINE

ANSHIKA

INDIA

Anshika is our firstborn. She was born in a hospital on 22 April 1998 and that day there was so much whispering going on about her being different. During the first second, I was also a bit confused for seeing such a large birthmark. But immediately after that very first moment I did not care about it anymore. My husband and I were so happy and excited seeing our beautiful and healthy daughter.

However, the doctor was shocked. Firstly, Anshika's umbilical cord was entangled around her face and neck and secondly, the other parts of her body were completely black. Relatives and friends who came to see her were all shocked. They linked her birthmark to their superstitious beliefs. I have always regarded these views as useless. I do not believe in superstition.

Soon after, we started our journey of consulting doctors, but no one knew about her condition. After

turning to many medics, we came across someone in Delhi who told us that Anshika could be cured abroad through plastic surgery. This was not possible as we were not that fit financially.

As Anshika grew up, our worst fear became a reality: she was bullied by kids at school. Anyone who met her for the first time would ask her all sorts of questions. Some people asked if she was burnt. But it is a rare condition. Every other day, I had to go to Anshika's school to discuss with her teacher the lack of respect she received from other kids. In the end, she was afraid to attend classes. She feared leaving the house at all and a lack of confidence developed. We tried our best making her feel self-assured again.

From the day Anshika started to understand the presence of her birthmark, I kept telling her that her skin is a gift from God to make her special.

Having Anshika is a blessing to us. She makes our lives blessed and happy. Some years ago, we asked her if she wished to have her nevus skin removed. You see, initially I wanted this because I feared my daughter would feel sad because of her skin. We would try to do anything she wants, but she declined undergoing skin surgery. She is confident the way she is.

HI! I AM ANSHIKA

INDIA

Hi! I am Anshika, a 22-year-old girl from India. I have a rare condition called congenital melanocytic naevus (CMN). I have a giant CMN on my back, stomach and thighs and hundreds of satellite naevi on my whole body. Even though I was born with CMN, I called it a birthmark, because I did not know the name of my skin condition until I was 7-8 years old. At that age, a doctor explained everything about it to me. Later, I searched for doctors to have my naevus removed, but now I am confident with my skin as I got connected with many people who love themselves.

I was a troubled child because I was bullied by the kids in my school. This was so disturbing to me that I was not able to concentrate on my studies. I would make my mother come to school every other day to protect me from them. I never liked school because of some children, but as they moved to another school my

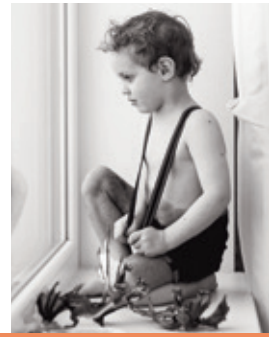
life became easier. As I grew up the bullying became less, and I gradually chose to be who I am and remain unaffected by the bullies.

I was also affected by people who constantly asked me disturbing questions about my birthmark. Thus, I resisted wearing half clothes.

My parents helped me in gaining confidence and they loved me so much that I got admission for MMBS (Bachelor of Medicine, Bachelor of Surgery). Iam dedicated to helping as many people as possible with CMN. In India there are hardly any medical professionals who have specific expertise with this condition. My medical degree could prove to be valuable in guiding people with Congenital Melanocytic Naevus in my own country.

I created the Instagram account dr_with_cmn. On social media I write short stories and organize interviews worldwide.

FILIP



CZECH REPUBLIC

Filip, our real Braveheart, was born on the predicted day of birth, after a relatively flawless pregnancy during which I developed a mild diabetes and higher blood pressure in the third trimester. All pregnancy tests turned out well, so nobody had any doubts about a smooth childbirth.

The delivery went well until I noticed the doctor's face turning completely pale. He checked with the nurses whether the amniotic fluid was green or not. I realized immediately that something was horribly wrong. Filip was not crying, and he did not breathe as well. His skin, apart from the black nevus parts, was dark blue. At that moment, I was scared that he was not alive. Fortunately, the doctors helped him with starting to breathe and soon after we could finally hear him crying. That was such a relief.

I noticed that parts of his skin did not seem to be alright. But like any other mother in the same situation,

I did not focus on his skin or appearance for too long. My prevailing thoughts were: he is alive, and he is taking his first gasps of breath. Nurses handed him to me in his small blanket and I hugged him for a moment. I was holding my little boy, who was just a few minutes between us, alive. His eyes were so restless that I wondered what he was thinking. Perhaps: "How did I get here and what am I doing here?" After that first intimate moment, Filip was taken away from me and put in the maternity ward.

The next day, it all began. Doctors were not sure of their diagnosis. None of them was able to tell me anything specific. All I remembered were a bunch of certain words and predictions for the future. After a couple of days, pressure sores occurred and Filip's skin partly teared apart. The two of us were moved from the maternity ward to the neonatal intensive care unit (NICU). A challenging treatment followed. It demanded cautious care in all thinkable areas. I had to permanently rub him, move him, and keep him in the right positions.

Then it got even worse with an infection causing neonatal jaundice. He was treated with phototherapy. ICU nurses took care of him, but since I breastfed him every three hours, we were together almost every moment. Breastfeeding was quite difficult, because wires and cannulas were attached to Filip's body and cannulas to his head. Two weeks later, neonatal jaundice had dis-

appeared, his skin slowly started to become stronger and he was discharged from Intensive Care.

After a skin lesion excision and subsequent sample analysis, it was confirmed Filip has a Congenital Melanocytic Nevus affecting 65% of his body. That was so heartbreaking for us. At last there was a diagnosis, but nobody could provide me with any specific details, so I started searching the internet. The more I read, the more upset I got.

After my initial struggle with all the information I came across, thanks to foreign nevus support groups I got hold of relevant facts. Unfortunately, these were not available in my native language. That is why I established a patient association for the Czech Republic and Slovakia. Kongenévus CZ-SK provides general support and detailed information on CMN. I hope that, because of the website nevus.cz, other parents will not have to go through the same difficult stages during early parenthood as I have gone through.

Finally, I would like to share with you a short, but important conversation I had with my son. While driving my car, I heard my son´s tearful voice from the rear seat. "Mom, I have been looking for you." "When?" I asked. "Before I was born, I had been hanging around in the mountains and the forests and I was looking for you." I assured him that everything is alright now. We have found each other, and this is how it should be.

LOUJAINE

FRANCE

Hello, my name is Hanane. My husband Mohammed and I live in France. I am of Moroccan nationality and he is Egyptian. We have two charming girls: Loujaine (12 years old) and Jana (7 years old). Loujaine was born with a giant CMN covering 90% of her body.

August 15th, 2009 was the big day when I prepared for a normal childbirth. I had lost water a few days before. The delivery kept waiting, and finally on Monday evening she was born with forceps. They hurt her head, I heard the nurses saying that her heart would stop, doctors arrived in the room, I saw the fear in their eyes, I remember the anesthesia they gave, my husband gave me a kiss on my forehead telling me that everything was going to be fine.

Loujaine was born on August 18th. Her first name, Loujaine, means 'river to paradise'. I wanted a unique first name, which has a meaning of rarity, a first name that we are not used to hearing.

When it comes to the diagnosis, I heard phrases from a doctor who knew nothing about psychology, or how to give messages to new parents. He talked about risks of skin cancer and said things he should not have said. Certainly, there is still work to be done on the medical side for the care of new parents. The words were said, but I had not yet seen my baby. She finally came to my room. An incredibly beautiful girl! I saw the moles that gave her a charm. Beautiful black hair.

We stayed in the hospital for 15 days to do the tests and for the wound on her head to heal, before going home. It was a great relief to be at home.

Two days later we traveled to two countries: Morocco and Egypt for the baptism and to enjoy the warmth of our big family.

Three months later, we returned to France. A heavy program of operations started. Every six months another procedure. In total sixteen operations ...

When Loujaine was little I covered her from head to toe, to protect her against the weird looks she would get. Growing up she asked me if she could wear dresses like little girls. It hurt me because I deprived my daughter from the minimum of things without paying attention to her desires. I protected her from my view, but I forgot that she is a princess. She wanted to wear shorts, skirts, sleeveless shirts, half-sleeve T-shirts, swimsuits. But the heart of a mother does everything for her child, be safe.

One day, there was a children's party in the park in our neighborhood. Loujaine was two and a half years old. Children were dancing. There was music. Loujaine approached and gave the children a hand, nobody gave her a hand. I approached and tried not to dramatize the situation. Once again, the children took their hands off and turned away while dancing, leaving Loujaine apart. I burst into tears. Loujaine came to see me and said: "It's because of me that you are crying, mum." I went to the parents and explained CMN to them. It is something I do all the time. But there are parents who do not understand. They took away their children from me, saying that I am not telling the truth. Behind my back, they even said it is contagious.

Loujaine taught me a lesson for me to understand why people screwed up our believes. We did not ask them anything.

Before the start of the nursery school, I asked Dr Béatrice de Reviers, to write a document to explain Loujane's condition to the head of school. This document is valuable to me. I present it to the teachers and parents at the start of every school year. It takes me 5 minutes to explain Loujane's condition. I anticipate in order not having to deal with surprises.

In elementary school, with Béatrice and Sonia from association ANNA, I raised awareness about CMN. Since then, the principal has asked me to repeat my presentation every year to help other children.

CMN was never a taboo subject at home. We always communicate with Loujaine. Only sometimes I feel it is too much.

Since she was little, I taught her before going out to arm herself with self-confidence, to trust us, that we are still here, to be strong. Basically, that is the strategy * EN PISTE * by Béatrice from the association ANNA, which we use all the time.

When she comes home from school, I do not give her time to isolate herself. If there are stories to be told, she is not on her own. You need to communicate to explain or solve problems.

The years go by, Loujaine is getting older, but she is always accompanied by us. We parents became experts, psychologists, nurses, doctors, friends…. we learn and we adapt to situations.

Loujaine taught me a lot of things, I became an expert mom thanks to her. Her strength to face all that she has gone through wakes me up with every depression or feeling of weakness.

I have lost some friends since she was born. I felt bad at first but realized that they were not real friends.

Some people avoid inviting you because they are scared or whatever. I ended up understanding that these people never change, moreover we too will not change for them.

Finally, I would like to tell you that we are all different on this earth, by the color of the hair, eyes, skin, nationality, religions… Surround yourself with love, trust and communication between you and your child. Do not let your children isolate themselves.

I wish you good luck and god bless you.

Hanane Douibi

2.
TREATMENT

LUCAS

TYLER

LUCAS

COLOMBIA

The pregnancy went perfect. All the tests showed great results and there was nothing to be afraid of. Lucas would come in good shape and health, so we thought. When Lucas was born, his skin condition was a surprise for everyone in the room. Unfortunately, in the days that followed no-one could provide answers. We as new parents looked at him with delight, but in the background, there was this fear of not knowing what happened to our son.

I started searching on the internet, especially on social media. I came across several cases and treatments, but it got to a point where I only found the worst cases, of kids that died from cancer (melanoma) and disorders related to congenital melanocytic nevus. My father-in-law had melanoma the year before Lucas was born. I consulted other people and after digesting many opinions and considering many options, we decided to start a treat-

ment by Doctor Bruce Bauer in the USA. He is regarded as one of the best plastic surgeons in the world, especially when it comes to removing nevus skin.

Dr. Bauer and his team presented a series of 12 surgeries and he recommended that most of these had to be done before Lucas would turn two years old. I had just lost my job because of the oil crisis that affected the world at that time, so financially it was a hard decision to take. At the same time, being without a paid job was the best thing that could happen to me, because I focused all my energy on collecting the money for the surgeries. At the same time, I managed to help others with the same condition as my son.

When 9 months old, Lucas underwent his first surgery, having two skin expanders implanted: one in his head and the other one in his shoulder. Our role changed into being his personal nurses, as we regularly filled the expanders with saline fluid. I learned how to make a puncture in my son and how to inject a liquid solution. For 12 weeks, we took care of him every day until the next surgery came about.

Two weeks before this second surgery, another kid suffered from complications during her treatment, resulting in the amputation of her leg on which the nevus was located. This scared me and I felt terrible because Lucas experiences a skin tear that week. I called the surgeon and he ensured, "everything is fine; we are going through this together".

At that point it was impossible to reverse our decision. We had faith in God and trusted in what doctor Bauer was able to do. We could only wait for everything to go well. And indeed, the surgery was a success. I cried like a baby when my son came out of the surgery room.

After these first steps followed 6 more surgeries in 5 years. We had to overcome many obstacles, and today we can say as a family that we took the best decision for Lucas. The main nevus (the most extensive one) has been totally removed. He still has small satellites, but we are not going to remove them unless this is necessary.

Now Lucas must conquer new challenges with his scars, his skin allergies, and his hearing loss in his left ear, but he does not remember having had several surgeries in his life.

By Pedro Lopez, father of Lucas Lopez, our little nevus owner.
From Colombia, South America

TYLER

USA

My name is Tyler Kudej. I am ten years old and in the fifth grade. I live in the United States with my parents and two older brothers.

When people first meet me, they usually notice right away that I look different. The reason my face looks different is because I was born with a large Congenital Melanocytic Nevus (CMN) on the right side of my face. My family has never made a big deal out of my nevus, it is just a birthmark that I was born with. When I am asked, what is that on your face? That is what I tell them.

When I meet new people, for the most part they are curious, ask questions and want to learn more. Occasionally, there are some people who are not so nice. I have learned that having a facial difference is not something that people are used to seeing, so I do not mind being asked questions and I do not get upset about rude comments. I think it is important to teach people that it is

okay to be different and not to judge someone because of how they look, whether it is a nevus or a scar.

My parents made the decision to get my nevus removed when I was young. Due to the size and location of the nevus it could not be removed in one procedure and required several surgeries and a combination of surgical techniques.

The surgeries began when I was four years old and my nevus is almost completely gone now, except for a small line of pigmentation that runs along my bottom lip. I have been in the care of three surgeons who specialize in craniofacial surgeries and together they have used excisions and tissue expanders to remove the nevus. Although we consider my nevus removed, I still have a few surgeries left to correct the shape of my mouth and lip.

Removing my nevus has been a long process and this had some challenges. My parents had to find the right surgeons, decide on treatment, and explain the tissue expansion process to people who had never seen or heard of it before.

People were really inquisitive about the bubble growing out of my face, especially my classmates at school. To help with those questions my mom put together a letter explaining my nevus and what would happen over the expansion process. It was helpful for all the questions and I discovered that as my expander grew, it would glow when light went through it. It was a neat trick!

People have said that I am brave because of all the surgeries, but I do not think it is a big deal. I get nervous whenever I go into the hospital even though I really like my doctors and know a lot of the nurses.

The hardest part of having had so many surgeries was that I was unable to play sports because I either had a tissue expander in or was recovering from surgery and the doctors did not want me to get hurt or risk open any incisions. If there was anything that I could change, it would be that I would not have had to miss out on playing sports.

3.
FOLLOW-UP

VIOLA

VIOLA

ITALY

My name is Viola, I am 23 years old and I live in Florence, Italy. I was born with a CMN on my right arm, neck, head – but I have too much hair there for other people to see it – right side of my breast and back. I also have got many satellites on both legs.

I underwent 16 surgeries in total and most of them consisted of skin grafts and laser therapy. At the age of 18, after a bad result with a skin expander, I decided to put an end to plastic surgery, because I was tired of it.

I felt I only had been a patient and a girl that never would be normal. To be honest, that is why I choose the last surgery, the one with the expander. I was trying to be like the others, and I was fighting to become a normal person, and I was having these surgeries because I wanted a normal skin. But after every surgery new scars appeared on my body and being a normal person seemed further away than before.

Then I asked myself: "What does 'normal' mean?" At a certain point I understood that I would never be normal, because I was born with a rare skin condition. I was born different, not only with a different skin but also with a rare and different heart. Without my skin, I would never be the person that I am today. I would never have learned all the important things that matter in life. I would never have gained the confidence that I am enjoying now. I also never would have loved my skin the way I am doing now.

Maybe I will regret having had most of my nevi removed. At the same time, I know that I reached this level of confidence because I had to deal with all the surgeries, and I had to face all the pain and every scary moment as well to become the girl that I am today.

Nowadays, when I look in the mirror, I understand this is who I am: a girl with a difference who would never have loved herself without it. This is me. I tried to change myself, but every time I was disappointed because all the changes diverted me from my inner soul and my well being. So, I just stopped putting myself down, and I stopped hiding myself in turtlenecks or dresses that cover all the scars, and I stopped regarding myself a non-sensual girl. In a way, I felt reborn.

Now I have a profile on Instagram where I talk about skin diversity, self acceptance and what it means to live with a rare skin condition. I want to raise awareness

about this condition in Italy. I also want to show how beautiful a different skin can be. By doing so, I challenge all the social standards that make people believe otherwise.

We are all beautiful in our uniqueness and we must fight to obtain the right to be different and to be proud of it.

4.
ADDITIONAL SYMPTOMS

ALDRIN

SALLY

EREZ

ALDRIN

INDONESIA

Aldrin was born on the 19th of October 2012 with a cape nevus.

At the age of six months, we did an MRI scan on his brain and spine as per Nevus Outreach's recommendation and we found out that there are deposits on his amygdala. Tests were also carried out to find pigment deposits on his eyes and his ears.

Aldrin remained asymptomatic until he reached the age of 5 years and 2 months. During that age, Aldrin developed seizures. His early signs were very subtle: he felt nauseated. This early symptom lasted a few months until the real seizures (jerking) came about. Since then, Aldrin has always been accompanied at school and at home. We were worried because the seizures could come at anytime. Aldrin is currently taking epileptic medication but still experiences seizures, despite having tried multiple anti epileptic drugs. It has been three years

since Aldrin's first epileptic seizure. Now Aldrin's seizures only occur during sleep and mostly in the morning towards waking up.

Aldrin has no other disabilities but does have a problem learning and concentrating, which we believe is ADHD. The psychologist confirmed that by the age of nine Aldrin would be "back on track", which means he should have an easier time learning and concentrating and would hopefully grow up normally.

He often daydreams during lessons and his impulsiveness is a big problem during schooltime. Since March 2020 Aldrin has been home schooled.

Aldrin is currently wearing glasses with a remarkably high astigmatism of 2,5 in one eye and 0 in the other eye. We are not sure whether it is neurologically related. Aldrin is very sociable, and he can carry conversations with adults but not with children of his age. Intelligence-wise he is generally above average. He is also capable of remembering people that he came across a while back.

SALLY

FRANCE

My name is Sally, I am 22 years old, and I am a carrier of a Congenital Giant Nevus with neurological complications. After a hydrocephalus at 10 months and epileptic seizures when I was 8 (treated), the most interesting thing began in the year I turned 15: every day, at the slightest flick, I fell over. I had one leg that could not get up enough and was stumbling.

By paying more attention, I noticed flaws in sensitivity (hot / cold / pain), and by talking to my parents, I also learned that having constant pain somewhere was not normal. An MRI revealed cysts (always present) that are compressing my spinal cord. I had operations two years in a row to empty them, missing one semester of class each time. It did not help me making friends, but I never gave up.

Today, I have a wheelchair all the time to free up space in my mind, because walking is still an effort.

As for the sensitivity, from my chest to the tips of my toes, either I do not feel anything, or I sense things in an intensified manner. There is no rule, I take it as it comes: I can hit myself hard without hurting, or brush against something and be in great pain. I never leave anything on my knees, because in 100% of cases, I get up and it falls because I did not feel it and forgot about it.

Then the year of my baccalaureate arrived where I missed half of the year and the second half, I missed part-time. Severe fatigue appeared. It had nothing to do with depression, I still wanted to do a lot of things but once sitting or standing, it seemed impossible to use my mind properly. My brain was completely taken by trying to process information from my body.

After a detour to England to find out what was happening to me, the verdict came: the communication between my body and my brain is scrambled. Instead of receiving the right information at the right time, my brain is constantly receiving a tsunami of loose information because of a mutation in certain cells.

The result is I am in a constant state of exhaustion, even lying down doing nothing, and I must be focused on everything I do (talking, digesting, breathing ...), to make my body work properly. Any gesture or action is an effort. The more I remain standing or sitting, the more information accumulates (too hot / too cold / pain in 1, then 2, then 3 places / sweat ...).

But I still managed to get my baccalaureate which turned out to be the achievement of my life!

Today, between physiotherapist, Chinese medicine to better digest and sleep, housekeepers to help me with my domestic duties in my apartment, and my social duties, I am already constantly in survival mode, so I had to give up having a job. Instead, the time I have left is spent lying in front of my TV and developing my mind.

I analyze what my life is like, I imagine what I do not have in live, I now even manage to live just by imagining it. I have never been saddened by my situation. On the contrary, I immediately saw the life my situation offered me, because I always wanted to live an extraordinary life, living and experimenting with all that is possible to do, and that is what my life offers me today.

EREZ

ISRAEL

1. How did CMN affect your life?
2. What was the impact on your family's life for parents, siblings, grandparents, etc.
3. What was the impact on your social life?
4. When you visited the hospital, what were your feelings?
5. What could be done to make life with CMN easier? This could be your advice for researchers, doctors, patient support groups, insurance companies, policymakers, or even your employer!
6. What is your dream? Describe your ideal situation.

I am not sure if Erez gets medical treatments. In the attachment, I add an infographic of what we used for the patient journey.

Hi, my name is Yoav Gaon, father to Erez Gaon, a 12-year-old beautiful and smiling boy, suffering from NCM at its extreme, including giant back, shoulder and scalp nevi, multiple satellites, internal tumors, a very severe scoliosis, and seizures.

Erez was born with his rare condition, with little hope for his future from the medical staff as they knew little about this rare disease or syndrome. The impact on our family was quite hard, from the happiness of bringing a new baby boy to join our family to the anguish, sorrow, and constant worries about the hope of him and us living with a complex and unknown disability.

The impact on our social life was immediate, a cocooning way of digesting the unknown combined with shock, shame, and depression on why and what has happened to us.

The looks and remarks of people not knowing what he had, ignorance combined with "fear eyes" to get infected were there, while we were trying to stay strong and build a life support line, so maybe we can make it through somehow.

And so, while Erez was showing surprising signs of cognition and understanding, incredible tolerance and resilience to hard medical procedures, he always laughed back to us and to the doctors. Teaching us a lesson of humbleness, that we know what we know and there is

still a lot to learn about the human spirit and human body.

Erez today, following numerous of medical procedures and paramedical training, is a special needs boy in an amazing happy world who learns every day in school. Erez has friends and through his personal showcased story on national television, his mom's inspiration book (Quilt Blanket, soon to be translated into English) on how to deal with uncertainty, my start up www.yoocanfind.com which is the global collaborative community for people with disabilities and our licecombot.com special needs comb inspired by Erez, have made this journey well worthwhile. Without him, as individuals and as a family we would never have evolved like this.

Erez's story has touched millions globally and taught us all the lesson that we should always look at a person from an equality point, humble to accept diversity, be inclusive and optimistic of the "big plan" the universe has.

Photographer Rick Guidotti created the "Positive Exposure Exhibition" showing the personalities of children with disabilities through his creative lens and photography. I have embraced his exhibition phrase as a life moto: "Change how you see, see how you change". I think that when looking at each one's abilities and soul you are already a better person contributing to a better world.

My recommendations to doctors and medical staff are to stay humble, always look at the parents and patient's eyes, hear the parents that better know their child and have the patience and be open minded, to ideas, treatments, and co-doctors.

To researchers I can say, you have already made some big discoveries and changes, understanding the NCM mechanism and ways to treat NCM, but the way for prevention or an ongoing cure is still ahead.

Be bold and learn from everyone, families, parents, patients, and co-researchers. Never give up. If there is a will, there is a way. If mankind has sent people to the moon and now soon also to Mars, everything is possible!

My dream for Erez is to live a long healthy and happy life, in a place with friends and family that love him endlessly and see him grow to his full potential as the true amazing person he is.

My message to all: parents, patients, families, medical staff is to always calculate the quality-of-life factor vs the treatment potential of success or improvement. The best thing we decided doing with Erez was to let him live and flourish. Better to let him be him than to try and fix him!

I am happy to be of assistance to anyone in need.

Sincerely yours
Yoav Gaon

Our patient journey

- Born with giant CMN
- Identified also as having NCM
- Seizures and internal tumors and scoliosis
- Serial excisions for suspected cancerous skin lesions (not full grafting as this is impossible)
- C4-C5 tumor excision
- Long treatment as being constrained to wheelchair
- Ongoing treatments and checkups due to the complexity of his situation

HTTPS://
YOOCANFIND.COM

5.
PALLIATIVE CARE

SULLIVAN

SULLIVAN

USA

Sullivan Rye Tomblin was born on January 5th, 2017 at a military hospital on Fort Hood. He was born from a perfectly normal pregnancy and came into the world with 3 other siblings. Sullivan was born with very "few, small" nevus. We had no idea what it was, and his medical team knew very little as well. They called in for resources from outside hospitals and the verdict was that his nevus did not fit the description of anything serious and he was very low risk to develop any other complications. At this time, we had no idea what those complications could even be, but we left the hospital thinking he would develop perfectly normal, and he was just extra special with his "spots".

Fast forward to one week old, I notice that Sullivan is making jerking movement with his eyes. They would shake and go "sunset". Family just kept stating he was a baby and learning to look around and see, but I knew it

was something more. At two weeks old I noticed his head was beginning to swell at his soft spot, something was not right. The pediatrician immediately scheduled a CT scan and it showed a serious amount of fluid on his brain. He was then transferred to the nearest children's hospital, a small hospital in Temple, Texas. It was determined that he had hydrocephalus and a shunt was placed, and we were told that "due to the anatomy of his brain, the shunt had to be placed in a different location than he would have normally place it". I could never get clarification on this but was later told that Sullivan's brain structure was abnormal.

At this point it was decided that an MRI needed done of both his brain and spine. This would be done with and without contrast. It showed that his entire spine was "lighting up" but it was not clear why. His neurosurgeon was always very honest with us and stated that he believed at this point Sullivan had symptomatic NCM and that there was malignancy in his spine. This is something I would not accept, after all how could this be? We were told he was low risk; he only had a few small nevi.

This is when I went on the search for a new medical team, our first stop was Houston Children's Hospital. Turns out that was a quick dead end, as they knew less about Sullivan's diagnoses than his small hospital. We made the decision to move to Kansas with Sullivan

when he was about 6 months old. There was a doctor there who was running a cutaneous clinic and stated that he had experience in this area. Upon meeting Sullivan, he was thoroughly impressed with his development, he was a completely normal little boy who even was ahead of all the milestones! He made the claim that despite Sullivan having hydro, he only had a few moles, and he was not symptomatic because he was developing perfectly normal. This was wonderful news! Finally, a doctor with some sense telling us exactly what we wanted to hear… until it wasn't. We did 6-month scans, and it all looked pretty stable compared to prior, even better due to fluid coming off the brain. Now though, a chiari malformation was noticed. Still though we were told not to worry, it was a common find and plenty of people have those.

At around 9 months Sullivan started to walk, this was fantastic! But he was not sleeping at all, he would arch back at night as if he were in pain and uncomfortable. Scans were done and more was found on his spine, a syrinx and multiple cysts. Finally, it was decided that a decompression surgery would be in his best interest. Sullivan got much better at night and we had a few good months. Still his doctors continually insisted that he was not symptomatic, and these were all separate finds aside from the diagnosis of NCM.

At 17 months Sullivan started walking with a tilt, I emailed his doctors on numerous occasions with worry.

I even sent them pictures that you could see a visible curve in his spine. They stated that his routine MRI was coming up in a few weeks and that I was just "over-reacting with worry". That Sullivan was walking, and he was just fine, there was no reason to move up his MRI. So, when it was finally time for his MRI, it showed a large mass in the bottom of his spine. At this point his neurologist is telling me he does not know what it is and that it could be completely unrelated to NCM. He insisted that his neurosurgeon go in and take a sample of it to send to the lab so that we could properly treat it. His neurosurgeon said this was not a good idea and that he was worried because of the location that he could do serious damage to Sullivan. His neurosurgeon in Kansas, was much like his in Texas, brutally honest. He always believed that Sullivan was suffering from disease progression, despite what the neurologist said.

We made the decision to do the biopsy, and this is when the nightmare really began. After the surgery Sullivan did not wake up, for days. Once he finally woke up, he was never himself again. It was like he was in this shell that could communicate with me as normal, but his face drooped and he suffered from palsy in his eyes. He never walked again. It was confirmed that Sullivan did have a malignant tumor and he did indeed suffer from disease progression and it was progressing quickly. He was quickly put with a new oncologist as I requested

to never see his neurologist again. He should have known better, I knew better.

This is when we began to reach out to whoever we could, doctors from MD Anderson, Memorial Sloan Kettering and many more. He even took a trip to St. Judes. It seemed that the overall recommendation was immunotherapy. The combination of ipilimumab and nivolumab. The second recommendation was an MEK inhibitor. This was a difficult decision but due to the medical state Sullivan was in and his weakness, a simple pill he would take seemed like the best decision. It brought us three months of beautiful moments together, he became stronger. He could crawl again and stand with support. It brought back his appetite and we saw minimal side effects. The only negative side effects he experienced were a small rash that was quickly healed with lotion, and he developed one ingrown fingernail. It was a wonder drug, until it stopped working.

When MEK stopped working we had to act quickly. He had another scan done, and two more areas of worry appeared in his cerebellum and brain stem, even though his spine mass remained stable. At this point he was healthy enough that I thought we should try immunotherapy, because we had to do something, we had to try everything. For his first infusion we stayed in the hospital overnight. He handled it well and we went home the next day. It was decided that a port would be placed

because of the amount of blood draws and infusions he would need. Because of the odd placement of his shunt the port had to be placed on his left side vs his right. For Sullivan, the placement of the port was horrible, he was uncomfortable for days and he absolutely hated having it accessed, it was torture.

Fast forward to his second dose of immunotherapy, he still handled this well maybe more exhausted but in my honest opinion it was not stopping the disease progression and his symptoms from that were getting worse. He quickly declined in health and unfortunately new scans showed those areas in the brain still getting bigger, almost tripled in size. His doctors recommended that we stop treatment and make him comfortable in hospice.

While Sullivan was one of the lucky ones who did not have severe symptoms from immunotherapy, the infusions were still torture for him to sit through and it wreaked havoc on his body. He was already suffering so much, there were so many blood draws and pokes to make sure the level of toxicity from the drugs was not too high. He was not even two, so it was difficult to say how his nausea was. He stopped eating again though, when the MEK inhibitor stopped working, he only breastfed and his appetite never came back with immunotherapy. As the disease progressed Sullivan lost all mobility completely and a short month later, he passed away.

COLOPHON

Patient Journey Congenital Melanocytic Naevus Syndrome is based on the first version of the patient journey which was presented at the Naevus International conference (2019) with 15 patients and ERN-SKIN patient advocates representing 10 countries in Europe, North & South America and Africa. Ten strories are published in this booklet.

CONCEPT

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