

DIAGNOSIS

On May 22nd, in the morning, I found Matteo in his bed, almost dying; I didn't know why, but I was sure his life was in danger: it was his first epileptic status. And here I entered my second life, a life made of words I knew, but so far from my world and so terrible like: intensive care, leukemia, tumors, and many others which were completely unknown to me: neuropsychiatrist, epilepsy, karyotype, spinal sampling, metabolic analysis.....And then, many new emotions: fear "what's wrong with my son", anger "why me" , the sense of emptiness, the deep loneliness, despair, and then, little by little, giving up things I cared about: my job, my social life, maybe also happiness. The second life began from the deepest depth..... On March 5th 2001(twenty-o-one), the diagnosis: RING14 Syndrome.(Stefania – Italia)

Madam, you have to accept your daughter! Anna you don't accept Camilla ! First of all you have to accept it! To Accept, to accept, to accept! Okay, I accept.....do not accept it ! (Anna - Italy)

Cyrus' birth was terrible! It took nearly 2 days but it was natural. As soon as I clapped eyes on Cyrus I thought he was so beautiful, but I also had this knot in my stomach. I had this feeling from the moment I saw him that all was not right; maybe a mother's intuition. My stomach still churns when I think of that day, the day everything changed.(Sheena - Mozambico)

It was terrible to sit in that small room with these two doctors knowing that what they were saying us was no good! To make matters worse, was that we already had a two years old son with cerebral paralysis. Please try to to imagine how we felt knowing that now we would have had two kids with "special needs" in our home. (Melissa – USA)

Christina had little energy and emitted no sounds. I could not breastfeed because she could not suck. I fed her with a teaspoon to give her a bit of energy. She couldn't control her body. She was simply suspended right there. (Wanda - UK)

At around 11 months Matty took his first steps and that was when I really knew something was really wrong. He walked bent over like an old man and fell every 5 or 6 steps and walked straight into furniture and walls as if he didn't see them. It took me a good 6 months to come to terms with what would be our future as a family and even now every appointment I have at the hospital is never good news and each time I am surprised that I haven't become numb to the situation. (Lisa – UK)

The impact was very deep. Olivia's mother and I are no longer married and although there have been other problems in our divorce, I believe Olivia's diagnose has been a tough test for both of us. We have developed quite different mechanisms to cope up with it and our different approaches brought an increase to the conflict on how we shall manage the child. (Raymond – Australia)

The day Brooke turned 3 months old, was the day she started having seizures. It was very subtle at first. We rushed her to the ER, no one could figure out what was going on and couldn't stop the seizures, so she was mediflighted back to Amarillo, TX. That was the beginning of any parent's worst nightmare. (Sheila – USA)

It is "a condition not agreeable with life" is how the Genetic Counsellor described it. She told us Nathan wouldn't walk normally (he runs) , he wouldn't speak coherently (he tells stories of Rugrats and Pirates), he won't live long (he is now 7 years old). If I could find that geneticist I would throw Nathan on her lap and let him tell her the story of two people that loved him so much that he can do anything. (Sue – USA)

ANGER

I wish someone had been able to tell me different when Ashley was born, as we "spoiled" her rotten – getting her anything she wanted and trying everything to make her "normal". I wish I could start again, which is what we are going to try to do this summer when we move into a new place Ashley is very smart and understands everything, unfortunately with her mom and dad, she has "trained" us if she doesn't want to something, she acts "disabled". She is very smart little girl, who can control you if you let her. We are getting help with trying to "re-teach" her who is boss! (Roni – Canada)

How, I told myself, we are in 2001(twenty-o-one),and nobody can give me information on RING14? Nobody has ever had a child like my son? Nobody can tell me how his future will be, and ours? For me it was a totally UNACCEPTABLE situation. (Stefania - Italia)

INTELLECTUAL DISABILITY

Matty, as he is affectionately known, has somehow managed not to be first in the queue when it came to living out the correct set of chromosomes, something I can now laugh about and happily tell people when they encounter some of our strange behavior and repetitive signing whilst on our many trip so the lighting department in our local B&Q where Matty gets infinite enjoyment ou of starting at the huge numbers of lights! (Lisa – UK)

I want now to tell you something about Alessandro (Alle), about joys and satisfactions for achievements, as luckily there's not only anxiety and fears even if they will join us forever in our life. Alle has difficulties in speaking, he says only a few words correctly, but he found his own way to be understood: he issues syllables looking at the face of his interlocutor trying to reach a reply of consent, he's often able to be understood using his gaze and his hands. (Daniela – Italy)

Matteo never wants to be alone and we can't leave him alone as he isn't self-sufficient and he could get himself in danger. We are trying to teach him some independences such as dress, wash, some house works and garden works and we are trying to improve his language. Our life is a very routine, we shortly get out home because of seizures, respiratory infections and many other different problems that prevent us from making plans but it's okay for us. If we wake up in the morning without crisis is already a good achievement and it will be a good day. (Stefania – Italy)

We try to treat Elizabeth as a regular kid, but now that she is a teenager, we give her a little more freedom to pick out her own cloche, etc. She knows how to use the microwave and loves TV, videos and music, mostly Disney music. (Rachel – USA)

Kim's education and like skills are slowly improving. Everything must be repeated and done thousands of times before she can understand. She does very well on a rigid routine with people she is familiar with. This does limit our ability to travel and do some things but we have come up with very creative ways to stay engaged with friends and family. (Karen – USA)

Ethan needs support to do EVERYTHING; for example, to dress, to put on his socks and shoes, to wash and to take care of himself. Ethan continuously regress because of his uncontrolled epilepsy. For Ethan is very difficult to stay focused for more than two minutes, we have to actively encourage him to play and to learn.. Ethan seems to take three steps forward and two steps back. We have to be very patient with him and each time we have to teach him everything again (Lisa – UK)

At the end, Samy is a little, very smiling boy, he doesn't walk, he doesn't speak, he doesn't eat by himself, he has to stay lie down 24 hours a day, he's completely dependent on me. I think he doesn't understand anything, he isn't aware of anything. A ten years old boy's body with an infant living inside of him.....I call him "zippatore" because it's very difficult to keep his attention for more than two minutes on an object.... (Lydia – France)

Today, Cami is 13 years old and has no way to communicate. She does laugh and she cries if hurt or upon hearing loud angry voices. She loves textures and to run, though she must always have someone close at hand or she'll run into traffic or out an open door. She is very loving and sweet but does not interact very much with other people. Her receptive language skills are very minimal and her expressive language skills non-existent. She is mostly silent, but occasionally will vocalize for no apparent reason. (Cary – USA)

LOVE

Dear Camilla if I had conceived you in 2002 instead of 1972 surely you wouldn't be born, knowing how I was before making your acquaintance! Certainly I wouldn't choose you. Is it possible that you have chosen me? If so I don't know if I have to thank you or if I have to be angry! Couldn't you leave me peaceful? I wanted a male and I wanted him to be absolutely normal and perfect! I felt a wide range of emotions and feelings about you that you have no idea! First of all a big angry that covers great pains, than despair, feeling of guilt, shame and powerlessness. But you are tough, very tough, my darling! You managed to get you off, you don't have given up, in spite of everything, you wanted me at all costs. Sometimes you wanted me in a very difficult way, difficult to interpret. Compliments my darling you did it, I succumbed. I love you! (Anna – Italy)

I have wanted to share my story about my beautiful little girl Shelby for quite some time now, but I have been overwhelmed with emotion, heartache, despair and boundless amounts of love and joy also. I am very blessed to have such a wonderful family; not that I am a religious person by any means. I believe God gave me a wonderful gift who's not perfect but whose love is so unconditional, she truly does complete us. We refused our situation for two years, no more. (Tanya Australia)

Ari is a very social little boy and he loves to drive his little sister crazy. We have come to realize, Ari knows a lot more than he lets on and uses it to his advantage. Cognitively.... We think he is a genius! (one can dream!) (Silvia – USA)

Whenever I am asked to describe Cyrus I think of the qualities he possess with abundance. He is happy, he is calm, affectionate, loving, gentle, content and kind. It humbles me to think that these are the qualities to which we all aspire. Looking after Cyrus is a huge challenge but also a blessing. (Sheena Mozambico)

We count ourselves very lucky to be blessed with such a unique bundle of fun and happiness. Underneath that cheeky face we have a little girl who has so much love

to give not only to us but to all those whom come into contact with her. Laura certainly leaves her mark wherever she goes. (Carol – UK)

Kim was born in Seoul South Korea in November 2001 and was born in our hearts in September 2002. We adopted Kim when she was 16 months old with a known diagnosis of Ring14, however no one really knew at that time what all the implications were for Ring14 children. (Karen – USA)

Eli is an angel. He's happy. He enjoys his life. Like any mom, I want the best for my son. I'm doing whatever I can to provide the best care for Eli, help others by sharing my experience, and support research to find a treatment or cure for Ring 14. There are advances in medicine every day. That's why Eli is alive. You have to look to other parents, and you have to be there for each other. (Terry – USA)

In a few words, next to his three years old he is our imp and we love him the way he is. When we learnt he had a rare chromosomal abnormality his father named him "my rare birdie" and I thought it was nice. Every day we do our best so that in the future he may fly with his own wings like each of our other babies! (Florence – France)

We were confronted with serious, sometimes life-threatening decisions. However, we never lost sight that our little girl was who we loved more anything in this world and we would do whatever was in our power to help her. We live our life one day at time and enjoy every one of those days watching our little girl grow. (Graciela – UK)

Nathan has taught us many things since his arrival. Most of all the feeling of an unconditional love that we as parents of special needs children can know. He has taught us patience.....and how. He has taught me to see people for WHO they are, not WHAT they are. Nathan is and always be his Mommy's and Daddy's Little Hero. He is Brave and Courageous. He is the love of our lives. (Suzanne – USA)

Ruby is now a happy, contented 13 month old, who now even enjoys her food! She loves music and being outdoors in the garden. Ruby has a cheeky character and a smile that would melt your heart. Greg and I are still coming to terms with the fact that Ruby is different to other children, we do not know if she will walk, talk or to what degree she will understand us in the future, but she is our special little girl and whatever happens we will do our very best to help her. The name Ruby means "precious jewel" and to us that is what she is. (Debbie –USA)

Celine has a lot of love for everyone around her. She hugs and kisses everyone. She's our sun. She can lighten up a whole room. Celine may not be like everyone else and

*that's no goal anyway – she's unique and that's what's making her so special.
(Christina – UK)*

Cristina has epilepsy seizures, cerebral palsy, tracheotomy, a tube for feeding and hearing aids. She can't talk, she doesn't make gestures, she doesn't walk, she doesn't crawl and she doesn't grab her toys. However she has a beating heart, she has tears and feelings. She feels hungry, she has fun when she bathe, at the park, at the zoo. She loves country music and she loves when we make her dance or when we sing for her. (Hanna – USA)

For smiles you give...for that joy that invades my heart when you dearly call me MOM....for that constant fear of being without you... for that anxiety I feel when you are sick...when you throw tantrums and I don't go along with them... for the patient person I became... for all the good that I feel about you and as I always say " I never wanted a child different from you " (Daniela – Italy)

*Brooke is a very happy child and she has so much unconditional love to give. She has taught me to have strength I never knew I had and because of that, I have made it through some really tough times, but have become a better person because of her.
(Sheila – USA)*

PAIN

Write made me cry. Tears have a little dissolved frost and pain. Perhaps they returned you a smile, my sister walk. Come on Camy, let's dance with life (Anna – Italy)

I'm full depressed!!!! Aside from that, everything is all right!!!!!! (Lilya – France)

During Christmas of 2000 and continuing through to the New Year period, Paul was ill. He was suffering from recurring chest infections and became very poorly indeed. The chest infections progressed to fluid being present within his lungs and the fluid led to the eventual collapse of his lungs. Paul was very ill and hospitalised. Staff at the local hospital found that treating Paul was more complicated than with most people. Paul can not explain which parts of him hurt, when they hurt, which treatments ease the condition or which treatments aggravate his condition. It was a puzzle for doctors to work out what was causing Paul's breathing difficulties and it was equally as difficult to determine how he could be helped. (Kaith – UK)

When Cyrus was 4 I remember experiencing the pain of people's prejudice towards him. The parents at his nursery had all signed a petition demanding his removal from the school. They threatened to stop sending their children if he continued there. He stopped going and was stuck at home bored and miserable. There are no nurseries for special needs children in Mozambique and so it took me many months of searching, but finally I found a nursery that was happy to take him. (Sheena Mozambico)

Frantically I searched for something to help my daughter, as she was slipping away and the doctors did not have any answers. (Cary – USA)

Sometimes I'd like to know why some children suffer so much. My Kacperek in November 2012 will turn six years. Everything was okay after his birth, but he didn't sit down in the right time but he didn't have problems with feeding. When he was 8 months old he had epilepsy seizures and since that time our fight began, from a hospital to another and nobody knew what was going on.(Dorota – Poland)

Prognosis was HORRIBLE (dead by 1 year, or if she lived, she would be a "vegetable" to put her in a home or just take her home and love her while we could. We spent the next 12 years coddling her, babying her, loving her, trying everything to make her progress. The first 7 years were spent trying to get her to do anything. (Roni – Canada)

I'm very confused, angry, sad and discouraged about information, I'm praying that Madelyn will not have that characteristics but it will be probably like that and I have to learn to deal with it. I know that every child is different from others but I continue to ask why this has happened to my baby and my family. (Bethany – USA)

EPILEPTIC SEIZURES

At 5 we noticed that Cyrus had stopped doing some of the things that he had done a year earlier. We hoped it was just a bad phase and kept an eye on him. As the months went by, he was doing less and less. His language disappeared completely, he could no longer feed himself, he could no longer hum to songs and he lost interest in everything around him. His walking also deteriorated. His left leg developed a limp and his foot was twisting outwards. He was a lot less steady when walking. A recent EEG showed that he is still suffering from numerous sub-clinical fits. His left leg has also worsened. His whole leg is set in the hip joint twisted round slightly, his leg is growing outwards from the knee down, he has slightly lower muscle tone in this leg

and his foot is also twisted outwards. He has just had a split fitted which has kept him walking, but we have recently been told that his right leg has also started to twist outwards. (Sheena Mozambico)

In February of that year (I think it was 1993) Cami's seizures escalated quickly until she was having many seizures a day. She was put on new meds, but nothing helped. The happy, communicative little girl was gone. She was either having a seizure, post-ictal or recovering from the heavy doses of medication used in a vain attempt to stop the seizures. She was clearly experiencing brain damage from the seizures and/or meds. From age 4 to 5 Cami seized almost everyday and cried constantly. She would wander aimlessly around the house, moaning, unable to focus on anything. Sleep became elusive - it seemed that her brain was unable to shut down and she continued to decline cognitively. At this point, she lost the use of her epiglottis and was unable to safely swallow, so a Gastric tube was placed surgically. After 2 weeks she was stable and was released but was unable to walk. She no longer signed or used any kind of communication. (Cary – USA)

The doctors were puzzled as something was causing these seizures but they didn't know what, as babies are at least 6 months old before they have seizures where Tyler started at just 2 weeks, at this point he was 5 weeks old. (Jaimee – UK)

The seizures have been steady and have seen no improvement, then, suddenly, when Brooke turned 8 years old, she only had one seizure the entire year and her progress soared. She learned to talk better and even put a few short sentences together. We had finally mastered using silverware to eat and using a napkin to wipe our face. It was amazing! Then just as suddenly as the seizures had ended, they began again. (Sheila – USA)

FAIR

I love my daughter with all my heart and soul, but I fear for her, she is so dependant on us, she is an only child (tanks to the doctors telling me not to have anymore children). We have had a very trying 12 years, some good, some bad, some ugly! We have almost lost Ashley 3 times, once during a surgery that went bad, once during an allergic reaction, and once in the very beginning when she was lack of will to thrive and not able to hold down anything. My life is her, I don't know what I would do without her. But no, I realize that I have to let her live HER LIFE, she is so used to us treating her like a baby, that is what she has become.... It is so hard to differenziate

*between what is her level and what isn't, as she is anywhere from a 2 – 7 year old.
(Roni – Canada)*

I'm scared for his future as i don't know what will happen, and neither does anyone else. I am prepared for some things as I have been reading other peoples stories about their children with Ring14. I understand that he will have language problems when he grows up, and other symptoms. But for now, we are taking it day by day and I'm enjoying every minute with him. We will worry about the future when it comes.(Jaimee – UK)

DEATH

There was a very critical situation at the end of November last year, he had a flu and after three days we found him very pale and unconscious in his bed. We went with the ambulance to MC in Amsterdam and Elmer stayed at the intensive care for almost 3 weeks. The doctors told us there was a small chance for Elmer to survive this. Wonderingly he did. We are very glad Elmer is still here with us and hope we can enjoy his smiling face for a long time. (Antoniette –Olanda)

My son Peter is affected by 14 Chromosome Ring syndrome. Doctors told us that he would survive a year only and in 1978 (his birth's year) we knew nothing about this chromosomopathy. Peter lived 14 difficult but good years. He was the angel of my life. He was inside and outside wonderful. We spent a lot of days and nights until the last few four years of his life but he could spend most of the time at home or at school. At home we had all necessary medical equipment for his disease, when feeling well he went to school. He enriched our lives, he was my heart and my soul. I miss him every day even if seven years have passed, he always lives in our hearts. I envy the parents who have their special children next., I miss those days. Please love your special Kid, enjoy him and remember how much you special are and how much enriched you life is from your angel's touch. Try not to be too much worried about their future, live day by day and thank God for every day you 've got them. They are special gift from God for special parents. (Ami – UK)

Obviously we were devastated. We were unprepared to dedicate our lives to our son's care for 24 hours a day. Our living became an intensive care unit. What this meant for his two years younger brother? What our normal family life have became? Nobody told us how many human resources, equipments, concerns we would endure.. We saved his life a lot of times! We weren't prepared to breast and to stop his apnoea, if we were, we would have avoid so many trips to the emergency room. I could take care of children, to nurse them but I was inexperienced in situations like

this one. "Is Eli breathing?" Even if we never said it out loud, it was our fixed thought. (Terry – USA)

When Peter was 13 years old an epilepsy seizure almost killed him. (Nancy – USA)

FUTURE

Our daughter life is a bit uncertain and we only fix short-term objectives. We go ahead day by day, working and trying to solve daily troubles. What do we wish for Sara? We hope she becomes autonomous and independent, but first of all we hope she's happy, very happy! (Eva – Spain)

To-date we are calmer than a few years ago, because we know we are not the only ones who think about Giada's future. For this reason, I hope that many other people will trust and cooperate for the future of our children. (Lucia – Italia)

The future scares us a little, but at the same time it commits us to do more and more, to become those "angels" who shall help Bruno in his path of growth. (Vincenzo – Brasile)

Usually the parents wish the happiness and fulfillment of their son. This is what I would like for Olivia. Evidently, she likes her environment of home-family and her life is a whole of learning, physical activity and amusement. Given her current health status, I would not be surprised if she approached a normal life expectancy, although I believe it is unusual for the people affected by the chromosome 14 syndromes to reach 40 years of age. (Raymond – Australia)

My expectations for the future are pretty fuzzy. Although I have had the pleasure of meeting many of the older children having ring14, it is very hard to envision Marie at an older age. She lost her first tooth this past week and it really came as a shock to me! Although I know she is growing and getting older, there is still a sense in which because growth and progress happen so slowly with Marie that the normal rules of time don't seem to apply to her. This makes it difficult for me to form concrete expectations for the future. I expect for us to have a happy and fulfilling life with Marie - outside of that, I just have a lot of hopes. I hope that her language skills continue to develop, so that she can more easily interact and enjoy other people. I hope that she learns how to walk and becomes less physically dependent upon us, so that she can enjoy a certain amount of independence (as well as us!). I really, really

hope that her seizures just go away!!! And I hope that Marie can continue to inspire us and teach us about what is really important about living life and loving others.
(Yssa – USA)

I hope the association may continue to offer help and support to the families; I hope our choices meet their real needs; for my son, as for all of the children of RING14. I don't want to delude myself with the hope of finding a cure to the syndrome, but it would be much to be able and locate effective treatments and therapies to heal the heavier symptoms such as epilepsy, the respiratory infections, the mental delay and the autistic symptoms. Am I asking too much maybe...? (Stefania – Italia)