

THE MIRROR

The Newsletter of IDEAS - IsoDcentric 15 Exchange, Advocacy and Support

Summer /
Fall
2000

Special Family Portraits Issue!



One of the most popular features of The Mirror is called Family Portraits, in which parents tell their "stories" and describe their experiences raising children with isodicentric 15. This issue of The Mirror features several new portraits which have already appeared on the idic15 website (www.idic15.org). The stories are reprinted here for the benefit of readers who don't have access to the Internet. This issue will serve to bring everyone up to date, whether or not they have a computer. Future issues of The Mirror will go online and in print form simultaneously. Also in this issue, plans are underway as well for the first international idic15 conference in June, 2001 to be held in Philadelphia. We'll bring you more details as plans are finalized. It's going to be an exciting year for IDEAS and our idic15 community!

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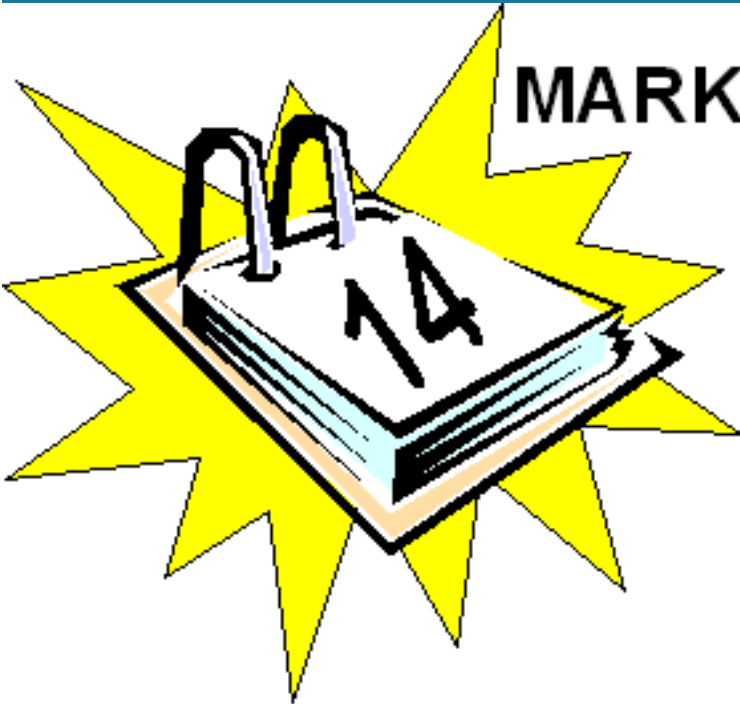


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MARK YOUR CALENDARS!!

**The First International
Conference
on Isodiscentric 15
June 14 - 16, 2001
PHILADELPHIA, PENNSYLVANIA, USA**

Conference Agenda Will Address:

- ◆ **Pre- and postnatal diagnosis of idic15**
- ◆ **Physical findings & medical management**
- ◆ **Educational strategies**
- ◆ **Behavioral interventions**
- ◆ **Medication issues**
- ◆ **Genetic aspects**
- ◆ **Research updates**

- ◆ **Research updates**
- ◆ **Sibling issues**
- ◆ **Support for moms, dads, and siblings**
- ◆ **And more.....**

**LOOK FOR MORE INFORMATION AND
REGISTRATION MATERIALS COMING TO
YOUR MAILBOX THIS WINTER!**

got news?



The Mirror welcomes the stories, insights, and ideas of all parents and professionals interested in idic (15). We'd also like to hear your suggestions for future articles. Send correspondence to Donna Bennett, c/o I.D.E.A.S., 416 Big Mount Road, Thomasville, PA 17364, Or if possible, email your newsletter items to Brenda Finucane <brenda@craftech.com>

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Megan's Story

by Paul Rivard



Where do I begin? Dec. 22nd 1995, Dawn was two weeks overdue and the doctor planned for an induction at 6 A.M. that morning. She was induced, and approximately four hours later we had a beautiful baby girl. This was our first child about to be born. Dawn's pregnancy was very normal, she was the envy of many women. Her Apgar scores were 9 / 9, everything was fine. Dawn had decided to breastfeed. They brought Megan in that evening to try for the first feeding. Megan had a very difficult time sucking and never took to the breastfeeding. Later that evening into the early morning, Dawn was awakened only to find out that Megan had been taken to NICU (Neo-natal Intensive Care Unit). Megan's temperature had dropped, and her lack of sucking ability plus the doctors' concern for infection gave her the ticket she needed to get placed in the NICU. As a precaution, Megan was in the NICU for 5 days. Born at 7lbs-7ozs she looked like a monster next to the many 1 and 2 pound premature babies that were already there. We were highly stressed, it was our first child, no one really knew what was going on, and we were just scared. Not exactly the way you want to be initiated into parenthood.

Finally, after five long days Megan was released, but with an Apnea monitor. They felt her breathing was slightly irregular. To have a baby home for the first time and to have the added stress of wrapping this monitor belt around her ribs before going to sleep. It never really went off, except for the one or two false alarms when she moved and the connection was broken. It's a lovely sound to wake up to in the middle of the night; it truly kept us sleeping on pins and needles. The monitor finally left after a few weeks, and there has never been any issue with her breathing since then.

Megan began to bottle feed, but very little at one sitting and very slowly. One week later we brought her into her first pediatric visit. Megan had been slightly cranky, or had what we thought might be colic. (It lasted for six months after that.) The doctor did a routine reflex test and observed that her left arm had no reflex. She would pick it up and it would fall limply to her side. We nervously went through many tests and found nothing. Over the next several weeks it did get better and finally back to normal. So far this has only been Megan's first week of life. In retrospect, these could have been beginning signs, but to first-time parents and unsuspecting doctors we just moved on.

Dawn and I carefully followed her milestones. We would read what her progress was supposed to be and then watched to see when it would happen. I remember each new month reading "what your baby should be able to do". Megan rarely accomplished anything early or on time, but somehow managed to come through with the milestone just under the wire. It seemed like forever before she even made her first smile. I think she was three and half months before she actually cracked a smile. Even with that first smile, the smiles were very far and few in-between. People would often comment on how serious she always looked. If we look back on pictures, we now notice her serious look and her floppiness. Her head was commonly off to one side or the other, and she was looking very mellow.

By six months old she was able to sit up, but even that was not very solid. Megan

continued to go for her regular doctor visits and the doctor would always ask questions about her milestones. I think back on it now and realize that there were some questions that we answered in a positive manner that we probably should have answered "no" to. We began to notice around nine months old that Megan would sit in one spot and just stay there. Even though she had just learned to crawl, she was very content to stay idle. At times she was so content that she would ignore any attempt from us to get her attention. She would just stare right through us. As we approached the one year old mark, our concerns started to build. We had her hearing tested, only to find out that it was perfectly normal. Tubes were placed in her ears due to the chronic ear infections that she had in the previous six months. The next step was to see a neurologist. At twelve months, Megan was not walking and showed no real signs of walking. She had very spaghetti-like legs. This is probably the milestone that bothered us the most and caused us to seek more answers with a little more aggressiveness.

The neurologist saw no immediate need for concern. He said everything was fine and that she was just delayed. He did suggest early intervention through Easter Seals which we did get Megan started on, but that did not satisfy our void. In the meantime we sought the opinion of another doctor (who is still Megan's neurologist today). He was unsure, so he suggested three tests for Megan – an EEG, an MRI, and a standard chromosome test. One by one the test results started to trickle in...EEG came in normal, the MRI was also normal, and then we got the call for the chromosome test results. I can still remember that day as if it was just yesterday. We had just gotten home from work and had a message to call Megan's doctor (of course when you get messages like that they are never good). We quickly called back. The doctor was brought to the phone and started to relay the message that they had found something in the test results that was extra genetic material. They were continuing to do tests but wanted to talk to us at Boston University where the tests were taking place. I can still see my wife's face watching mine as I was taking all this information over the phone. After hanging up and explaining what I just heard, it was like the world just stopped. It was kind of a numbing feeling. We had always wanted to know what was going on, and now we had something, but now I didn't want to have the answer. It was a bittersweet time.

We made the appointment with the geneticist and brought Megan down to Boston University to meet with him. He explained what he had found preliminarily; the tests were not complete so he could only speculate on the actual diagnosis. He left us that day with only thoughts of Megan having 'something'. They were not sure what, but based on the extra material on the 15th chromosome, maybe it was Prader-Willi syndrome, and that was going to be tested further to see what the real results were. So we went home armed with that information, only to go and research the disorder and have fear driven into our hearts. It was a very tough disorder to handle, and yet we still didn't know what was going on. After what seemed to be an eternity, we got the call to meet with the geneticist once more to discuss the final result. As you all know, it was not Prader-Willi, but indeed Inverted Duplication of the 15th chromosome. So now he had very little to share with us, except for

the gloom and doom of mental retardation, no speech, and the expectation of a very depressing quality of life for Megan.

Next began our journey with Megan. We tried to put his words aside and start doing some research. Megan was now qualified for more services with her diagnosis, so we continued to do what we could to expand her learning, although progress was very slow. Megan was approximately 20 months old by the time we got the actual diagnosis. She was still not walking. Slowly she made progress and by 22 months she was starting to walk. This was the start but she was very clumsy still - at least it was progress. Her speech was still close to none. She could say "ma-ma", "da-da" "all done" and "more".

Ages 2 to 3 seemed to be tough times with Megan. Her progress was very slow. Her processing of ideas seemed to be minimal, if any at all. Her speech remained the same and her walking was still clumsy at best. She could climb up and down stairs but only with assistance. Self-help skills were also lacking. She could not dress herself and feeding was also minimal, although that was starting to get better and she was beginning to use a spoon on her own, but not with much finesse. We continued to learn more and more about Megan and her ways. She definitely had an aversion to loud noises and certain touches. She definitely had sensory issues that were surfacing.

On a positive note, Megan was a good sleeper at night and was a relatively mellow child. Her behavior was not yet a problem, although anxiety in crowds or in public areas was starting to increase. Some of the bigger changes started to happen around 3 years of age. Megan, all of a sudden, began to be a parrot. She would repeat almost anything that she heard. Her articulation was not the best, but it was progress and it was nice to hear her voice. Megan's latest milestone has been to actually put two and sometimes three select words together. There hasn't been much of that, but it has been nice to hear some. Her walking has become slightly stronger and not so clumsy. Her eating skills with a spoon have become more and more refined, and she has started to show some thought process in everyday activities. Another thing that we noticed was her ability to anticipate sequential events that she was used to. For example, if she was watching one of her favorite videos (Barney), she would start to say the word "duck" five seconds before the duck would actually appear or get mentioned on the screen. She could also anticipate other sequential things in her daily routine, such as when we would get her down from the table after lunch, she would start to say "nap", "nap". This was all very welcomed progress.

This brings us to the present. Megan just turned 4 years old in Dec. of 1999. As you all know, in parallel to Megan's progress in the last year, we found IDEAS, met some friends, and then eventually it spun off into the "onelist", our email community. The onelist has proven to be the best thing that has ever happened for us as parents and students of this anomaly. We look forward to continuing the book of Megan as she continues to progress. Each day is a challenge still, but it is all worth it when you see her smile back at you, or when she just leans over and plants a wet kiss on your cheek.

[Click here if you would like to send a message to Megan's parents, Paul and Dawn](#)

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Naomi

by Kat Amundsen



Naomi is a beautiful brunette angel with a way with animals and brings out the best qualities in people. I had a normal pregnancy with Naomi, she took her sweet time coming and on December 7th, my labor was induced. I was full term and she was born after 13 hours of labor. I was pretty relaxed during labor and didn't really get going until late that afternoon; the hard part was the last 4 hours. I scared my family that was there for the birth. I had a sheet covering me and so when I got too hot and kicked it off, my family saw that the bed was soaked in blood. The nurses were not expecting my labor, which had been pretty slow, to speed up as fast as it did that last 20 minutes. Everyone said, don't push yet, the doctor is not here yet, hold on. I was pretty tired when I finally pushed. She was a good size baby, 7lbs 8 oz.

Naomi was not born with any rooting reflex and I had to teach her how to nurse. We went home on the third day. Naomi was losing weight, 1.5 oz each day. I was pretty concerned that

she has not had a bowel movement yet. After 14 days of trying to keep her awake with ice-cold wet washcloths, she was readmitted to the NICU (Neonatal Intensive Care Unit) for “failure to thrive”. The nurses wanted to put her on formula and I came unglued. I didn’t want to have to fight with nipple confusion and breast milk refusal. The nurses were surprised, and tried to explain to me in condescending tones that “the baby needs milk” as if I was not producing any! “Ha,” I said, “I will give you the breast milk, you can feed that”, and they gave me a very tiny 4oz cup with very doubtful looks. I parked myself in the rocking chair in the NICU unit and proceeded to hand express 4 oz in about 2 minutes. Supply was not the problem. While they fed her the milk in a bottle, I sat in the reading room off from the Nursery and read the La Leche League pamphlets. What I found was that some premature babies are born before the rooting reflex develops. Some babies fold their tongue to the back of their throat and suck on that rather than latch on to the breast properly. I learned in that room that night that I had to force her jaw down to drop her tongue from the pallet of her mouth and get the nipple up over the tongue so that she could get the milk into her tummy so that she could start growing. I had to leave my little angel in the hospital that night just a few days before Christmas 1990, and go home to sleep without her. That night I realized that she might not be here on earth more than a year. After just one night in the hospital she got enough breast milk in her that she was able to have a full blown bowel movement and clear all the meconium out of her body. She got to go home and we started the long difficult process of teaching her to eat on a schedule, waking her up every 2 – 3 hours and feeding her so that she was getting regular feedings. Teaching her to nurse ‘properly’ was just the first thing that we found that made her unique. She was not born with any instincts. Diapers became scheduled, feedings were scheduled.

At 6 weeks, she was thriving but not smiling, not rolling over. She was not meeting the developmental milestones at 4 months so when we went to the NICU follow up clinic, the clincher was the visual tracking. She would not track visually or turn to unexpected sounds consistently. She would not turn to noises so she was referred to an audiologist. That test came back normal. She was referred to an ophthalmologist for her vision and that test came back normal. She was still pretty tiny and the pediatrician ordered bone development x-rays. They were conducted at 4 months and they showed that her bones were those of a 3 month old. Exhibiting a physical growth delay of 4 weeks at 16 weeks of age. She was orally and generally tactile defensive. Her muscles were floppy and hypotonic, she was referred to a neurologist and that test came back for additional testing at Shodair Genetics Hospital in Helena Montana.

Those blood tests were sent to Houston, Texas in December 1991 for analysis. The initial diagnosis hinted at Prader-Willi or Angelman Disorder. I was busy researching Prader-Willi and Angelman disorder and grew very concerned. Severe eating disorder was very scary. I was terrified. But rather than a deletion in genetic material, Naomi had additional material. Not just duplication, but a triplication of a segment of the chromosome. What did it mean, no one knew. She was one of 4 or 5 children known to have any abnormality in this region. There was no information in January 1992 when we got the diagnosis.

By this time she was 1 year old and after weekly physical therapy she was starting to crawl. She was weaned and drinking from a cup and eating finger food. All in all, she has a 50% delay in all areas. She was still on a schedule, she was not able to vocalize to get her needs met so I continued to get aid from physical therapist, speech therapists, occupational therapists, even

enlisting swimming lessons to get her into the water. The water helped in the ability to tolerate different textures. She could only tolerate 100% cotton, diapers, clothes, blankets, everything. But in the water, she would touch rubber duckies, and foam turtles and plastic cups and lots of textures. It was wonderful, slowly she was making progress. One day when she was 18 months, we were in the front yard, she was on a cotton blanket watching me weeding the flowerbed, I had the water hose going, the yard was pretty much flooded, the grass was ankle deep in water. She crawled off the blanket and touched the grass - major accomplishment for her. She crawled through the water and grass to get to me. It was a wonderful transition for her. She took her first tentative steps at 22 months and was walking solidly by her second birthday, December 1992. Potty training was a nice idea but a totally unrealistic goal. We worked on the word "more" for food and drink needs. By the time she was 4 years old, she could say "more", I was thrilled. She had been in preschool for years, and the therapy was paying off.

We moved to Texas in 1996 and she was in a classroom of 6 students and 3 adults. She had learned to identify the Texas flag and the state shape. We were making tremendous progress on self-help skills. Four years ago she was ordered by the courts in Montana to go for visitation with her biological father from Texas to Montana for the summer. So now she is not allowed to leave the county and I have to have all visitations in Montana and her sisters have to miss school to see her. Visitation is 3 hrs a day, 4 days a week, which makes me sick to my stomach. But I refuse to give up and go up as often as I can. She is growing and developing and I find that each day she is making progress. Or perhaps I am making progress in learning about her world.

Naomi has been visiting our world for some time now. And when stress gets to be too much, she would drift behind a veil and shut us out so that she could get some peace. I feel that she is an angel and she would simply slip off and commune with the other angels and then come back to us when she was ready. I felt lonesome when she would leave. I felt sad when she refused to make eye contact with me and put me in visual "time out". But I am patient and I am ready to love and hug her when she "returns". The more I am drawn into her world, the more I am aware that she retains everything. She takes in all the information and stores it like a computer. She has a very slow processor, somewhat like my old 8086 with a 10 GiG hard drive. She has shown me how much more complex that she is that I am now alternating between sheer awe and sheer terror at the magnitude of the situation.

For instance, she lives far away and can only talk to me on the phone most of the time. I had a baby 10 months ago. She has seen him for 3 months off and on in the last 10 months. She becomes very upset when we are talking about him. At first I thought that it was sibling rivalry, but I spoke with her last night and she told me that she misses him. She is worried about him when he cries. It hurts her heart that he is sad. She wants him to move his bed in her room so that he can be with her. She wants to come home to my house (sigh). I told her that baby Erik says that she is HIS Naomi, but I tell him that she is MY Naomi and he says HIS Naomi. She laughs hysterically with sure pure joy that we would fight over her.

Naomi has a tremendous understanding. She has poor communication skills, but the thoughts are going on in that head and heart of hers. My advice is never assume that the "processor" in our little angels is ever broken. The information is converted and translated several times in those brains, and the time that it takes is beyond most parents' attention span,

so we miss the important answers / revelations. This for me has been such a phenomenally important revelation, not just for myself but for all parents of these displaced angels. I don't know how many parents realize how lucky we are to be chosen to care for these totally awesome little creatures! It is so hard for grandparents / outside family members to understand what is going on. The reason that THEY were not chosen is simply because they just do not have the right qualifications of PURE unconditional love that is so critical for the care of these angels. I can see into Naomi's world. I have been drawing her into my world for nearly 10 years, but lately she has been sharing her world with me. She has made an effort to come into mine, but she is opening hers up to me and that is so awe inspiring. I just want to share this information with you. These wonderful kids are "in there" and they are some of the most beautiful people that I have ever known.

About Naomi's family:

My name is Katherine (call me Kat). I am mother to Jasmine, Krystal, Naomi, and Erik. I am wife to Sverre (pronounced Svada). Sverre is a computer systems engineer at Compaq. I am a WAHM, I own my own business, making websites on the internet (<http://www.ausome.com>). Sverre and I met on the Internet in 1996. He lived in Norway and I lived in the US. We moved here and were married a year later. We have no pets, I travel too much and Sverre has no interest in caring for them. We live in Houston. In my former life, I received my bachelor's degree in Forestry and I was a publication's Editor. I am from Montana and spend 3 months total a year there. I watch movies incessantly, surf the net uncontrollably, and shop profusely. I am mouthy and opinionated but I hope that I am a good friend.

Erik is the sweetest baby that sleeps through the night and is so pretty with his golden auburn blonde curls. When Erik was born, he was kept in the nursery for 5 hours rather than with me because he could not warm up and his blood sugar was too low. From three days to six weeks he had pretty bad jaundice but no bili-light treatment. His face, and occasionally his arms turned blue, frequently. He has gastroesophageal reflux: the acid from his tummy was coming back up into his throat and irritating his heart and lungs. He had low oxygen levels in the blood, so they put him on an apnea monitor. They x-rayed his chest and found that his heart was enlarged due to irritations from the acid reflux. The whole problem was from lactose intolerance. I stopped consuming all dairy products and he is never sick. If I consume even one slice of cheese, he gets reflux all over again. We live off of Rice Dream Milk.

Jasmine and Krystal are identical twins - extremely gorgeous and too pretty for their own good. At 14 they are already 5 ft 7 and 140 lbs with long blonde curly hair. There was a complication and the second twin was born breach. Jasmine is ADHD and Bipolar. The Bipolar makes our life very interesting. She is like having a Pentium III 800 mHz computer with 1 Megabyte of RAM. She is a rapid cyler so she can go from Happy Happy Tigger to Super Super Sad Eeyore. Each day we medicate to find the Pooh within and help her have a good day.

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Robert

by Marion and Steve Mitchell
West Sussex, England



Robert was born by elective caesarean on 28 April 1994, two weeks prematurely because he was in breech presentation. He weighed 6lb 13oz. He was born with a small hole at the base of his spine and was taken straight to x-ray. They could see to the bottom of the hole and therefore called it a dimple and said it was nothing to worry about. Three days after Robert was born, three paediatricians were examining him and found that he had a heart murmur; so when he was six days old we were taken to Royal Brompton Hospital in London where he had all the chambers of his heart examined and it was then that they discovered a small VSD. The specialist thought this was just due to immaturity and he was right because after 6 months the hole had closed. We left that hospital and on returning to Crawley Hospital were discharged with what I thought was a perfectly "normal" little boy. Robert slept a lot and most of the time I had to wake him up to feed him. He was, however, a very colicky baby for about four months.

On attending his eight month check at the local health clinic, he wasn't able to do any of the tasks set before him and I was asked to bring him back one month later to see if there was any change. There wasn't. After seeing our family G.P., we were referred to a paediatrician at Crawley Hospital where Robert spent a few hours undergoing tests, mainly for fragile X. It was some months before we got the results back. With Robert at the age of 12.5 months, we were given the news that he had this very rare chromosome disorder, in medical terms written as Inverse Duplication of Chromosome 15, which gives him a 48 chromosome karyotype (which means that instead of one pair of chromosome 15, he has two extra inverted duplicated 15's). At that time, Robert could only lay on the floor; he wasn't able to roll over or sit up. We were referred to all sorts of different professionals: Physiotherapy, Occupational Therapy, Speech Therapy; suddenly our lives became an endless round of appointments.

By the time Robert was 18 months old, the help that we had been given by these professionals was starting to improve Robert's life. He started to crawl. He spent a long time trying to pull himself up the furniture and eventually succeeded. He started to walk at the age of three, although he couldn't understand how to get back up from the floor to the standing position, and this took him about another four months to work out. At the age of six, he doesn't walk that well as he has an abnormal gait where his feet turn inwards; he also has subluxating hips (they dislocate). He is also hypotonic (has low muscle tone) and has severe dorsiflexion (he can bend all his joints backwards). He is very wobbly and falls over a lot. He has been to A&E several times with a head injury because of this. He doesn't understand the concept of inclines and declines or kerbs or different surfaces (although he is getting better with the kerbs), and he doesn't know the difference between a road and the pavement. He has severe learning difficulties and absolutely no fear!

It can be very difficult to take him out, and it is virtually impossible to use public transport because I have to carry him on board as well as folding his special buggy which cannot be done with less than two hands, as well as carrying the bag with all his essentials. As he gets older and more strong-willed, his behaviour becomes more difficult especially as he has no communication - he doesn't talk or sign. It can be very difficult to understand his

needs, and the worst thing about that is not knowing when he is in pain. Except that he screams very loudly! He cannot do anything for himself such as washing and dressing and feeding himself with cutlery. - he will pick up things with his fingers regardless of what is on the menu and on occasions has eaten cat food, £1 coins, sweets with their wrappers on, and used teabags. He is doubly incontinent and he dribbles constantly so I have to change his clothes more than once a day.

Robert attends a school for children with severe learning difficulties - the staff there have been very good with him and he has been learning social skills. Before he went to school full-time he used to have Portage - which is a home learning programme for children with special needs. This was very beneficial to us and I cannot recommend it highly enough. He also used to attend the child development centre where he took part in an Occupational Therapy class for a very small group of children. This was also very beneficial.

As well as the everyday problems, Robert also has severe eczema. This can involve lots of applications of ointments several times a day and also wet wraps (bandages) when it is very severe. This can be traumatic as Rob screams and the treatment takes up a lot of time. Robert doesn't sleep all night either - I can be up once, twice and sometimes three times in the night with him, changing him, or redoing his bandages if he has them on etc. or just cuddling him. He seems to need constant reassurance that he isn't on his own. Robert also has problems with his ears, and since having grommets fitted in July 1998, he is still unable to go in the hydrotherapy pool at school because he is still getting ear infections.

I'd say that despite all of these trials, on the whole Robert is reasonably content most of the time, but is getting more and more frustrated as he gets older. He loves being outside in the garden. He likes adults who give him lots of attention; lights and the computer at school fascinate him. He is also a climber - he climbs on anything and everything. He bangs on the windows, switches lights on and off constantly, opens and shuts the microwave door, plays with the cooker, tries to get out of the doors and windows, and gets very frustrated when he wants something quickly and I'm not quick enough. He loves water, and bath-time is a favourite, but you have to watch him constantly around any sort of water. Taking him to the beach is a nightmare because he just wants to go into the water all of the time. If he sits on the beach he will eat pebbles (or anything else he sees that takes his fancy). But he is a lovely little boy who likes a cuddle and a kiss, and he loves children's music and stories.

ROBERT'S STORY

My mum says that sometimes I can be a real pain. I don't mean to be, but I find it difficult to understand things a lot of the time. I don't know how to talk yet, but I can shout very loudly! I get a lot of pain in my ears, which is very difficult to cope with. I have this condition called eczema, which I inherited from my mum (thanks mum!). It drives me mad and all I can do is just scratch, scratch, scratch all the time.



I like climbing anything and I like hanging upside down. I also like chasing my cat, Trudy, but she gets cross with me and sometimes scratches me. My mum tells me off! I like water too, but my mum won't let me go swimming because the doctor says it isn't good for my ears. I am hoping that one day I will be able to go and not have any pain in my ears afterwards. The last time I went swimming my ear drum perforated. I go to Catherington School, and I have lots of friends and a very nice teacher called Pauline.

Lots of Love from Robert xxxxxx

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Kevin

by Nancy Chamberlin



Kevin is a very sweet and special 4-year old boy. He has a large trisomy 15 (isodicentric) diagnosis. Kevin was born on December 30, 1995. He was a week overdue and came into the world weighing 8 lbs 8 oz. My husband Mark and I took Kevin home unaware of any problems. Kevin has a sister, Natalie, and a brother Matthew, who both were very excited about their new brother. At about 2 months of age, Kevin did not seem able to see and his eyes were looking upward a lot. The doctors began tests to check for tumors or hydrocephalus. Kevin was bottle fed and eating fine, but he was not developing fast. His head was weak and we thought he was blind. Kevin had a startle reflex that could not be pinpointed as seizures, so we continued to watch and wait.

When Kevin was 10 months old he woke up with a scream one night. He was seizing with constant myoclonic-clustered seizures. We raced him to Children's Hospital where they treated him with four different drugs. By the second day, Kevin was overdosed and stopped breathing. He was put in ICU and on a respirator. It was so awful, and we cried and learned a lot. The doctors couldn't diagnose him but did look for many diseases. After about a week, Kevin came home. He was put on ACTH shots that we had to give in his thighs. This treatment did not work so we began other drug treatments. At age one, we tried the Ketogenic diet and this did not help his seizures either. Kevin's seizures increased and by age two he had many myoclonics and ten minute long tonic-clonic seizures. We use diazapam rectal gel to stop these long seizures. Our lives continued and our children's schedules kept us busy. We tried to be as "normal" as possible and thanks to family, friends, and God, we have survived.

The chromosome test was overlooked at first, but then done at about age 2 ½. The results were 48 chromosomes, one large inverted dup 15 and one small unknown chromosome. Kevin is four now and has myoclonic seizures all day; he has tonic-clonic seizures a couple of times a day. The big seizures last up to 2 or 3 minutes. He is low-toned and cannot sit up alone. He attends pre-school with a mix of special needs kids and mainstream kids. He eats well but cannot chew well, and he now weighs about 35 lbs. Kevin kisses a lot, he can hug and love us and he seems mostly happy. Kevin loves music and to look at books. He is a big, exhausting boy, but it has been a very rewarding life that we share.

This past April we met another family, the Edwards, with a beautiful daughter, Brianna. She has the same diagnosis and is very similar to Kevin. It was a special and touching moment to meet with them and to share our children's stories. Angie, Wayne, Brianna, and their 8-year old son Aaron became our instant family. Kevin is a gift from God and we are lucky to know him. The newsletter, IDEAS, and new web-page are helpful. Kevin seems more severe than the other kids, but it is still interesting for me to read about the children's achievements.

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Olivia

by Pamela Taylor



Hello, my name is Pamela Taylor. I am married to Bob, and we have three wonderful children: Rebekah 11, William 10 and Olivia 5. Olivia is the reason that we are with IDEAS.

Olivia was born a week later than her due date. I was induced, and had a pretty easy delivery (as easy as deliveries can be!), which was very quick. She appeared to be a healthy, normal baby weighing in at 7lb 11oz. She slept almost constantly, needing to be awakened for a feed. We did, however, notice that she seemed to stop breathing every now and then. When I called for the nurses to come and see, by the time they managed to get to us, she had started to breathe normally again. We went home after four days.

At 14 months, Olivia had her MMR vaccination. She was such a brave little thing and didn't utter a sound. One week and a day later, she took her first febrile convulsion, she was hospitalised for observation and allowed home the following day. Two months later, the same happened again, and up to now we have been convulsion-free.

Up until the age of 18 months, when Olivia was becoming increasingly late reaching all her milestones, we put that fact down to her being content and being the third child. Deep down, I had an idea that something wasn't right. She couldn't sit up unaided without folding totally in half. She didn't crawl until she was 19 months, didn't talk or babble, seemed to have a fixed stare, and had recurrent ear infections.

Eventually, when Olivia was 18 months, she had a development check with her health visitor. It was very clear to her that something was not right, so we were then referred on to a paediatrician. He ordered a battery of test to be done. Liver functions, full blood count, thyroid function (I have thyroid problems), and, as he said to me, "I will do a chromosome test too as we are doing everything else! I don't think that we will find anything there, but we will do a process of elimination."

The tests came back. Everything was normal ...except of course the chromosome test. Her karyotype was 47 XX +psu dic (15) (pter-q13::q13-pter) Ish psu dic (15) (q11-q13). We weren't actually given Olivia's karyotype until November 1999. I suppose the paediatrician didn't think that it was important to us. He had given us a verbal diagnosis for her: "de novo maternal isoduplication of chromosome 15 in the Prader-Willi/Angelmans syndrome critical region". Well, as you can imagine, this left us totally baffled. Most of the specialists seemed to latch on to the Prader-Willi part, even though Olivia didn't actually have that!

After her diagnosis, we were referred on to The Children's Centre, which houses many specialists. She was put into an early intervention programme, which was fantastic. There she got the help that she needed. She had speech therapy, occupational therapy, and physiotherapy; she attended a small group, and was being observed and assessed by clinical and educational psychologists. As well as being at the Children's Centre, we tried a joint placement at a mainstream playschool and nursery. Although they tried their best to accommodate Olivia and her needs, it was proving to be far too traumatic for her. It was at this point that the autism was becoming more obvious.

Olivia had a battery of tests, done from evoked response tests under sedation for her apparent selective hearing, to EEG's for her absences. Both were negative, and her EEG showed no signs of epilepsy but an immaturity of the brain. Again I now believe this to have been the early signs of the autistic disorder. Olivia eventually walked just after she was two. For the next year, her walking looked like she had just that minute taken her first steps. She still has an unusual gait, and walks with her left foot turned in slightly, lifting it high to take a step. She will charge around just avoiding a collision, and her spatial awareness seems to be a little confused at times.

Olivia's speech was very slow to develop, and she was an expert at making animal noises, before she could say a word. Her speech is very good at the moment. She can say anything, and can sing even more. Her understanding of the spoken word is very delayed though, being that of a child of 2 years and 10 months. This doesn't seem to have improved over the last six months since her last assessment with her speech therapist. She will be assessed again soon, as her speech therapist feels that the gap between expressive and receptive language is widening.

Olivia uses stock phrases when she becomes anxious and will repeat them over and over again. She parrots a lot of things, and even copies mannerisms of other people.

Toileting is another one of Olivia's delays. She shows no signs of wanting to use the toilet. She will sit on the toilet and use it appropriately (sometimes), but only if we initiate it. She will soil 7, sometimes up to 11, nappies a day, denying it if she if we ask. She is on co-phenotrope medication for this, as her transit time is around 3 hours, and the medication slows every thing down.

Olivia has no sense of danger or their consequences and is an expert absconder. Consequently, doors need to be kept locked and higher fencing has been put up in the garden. She also has a very high pain threshold, and unless there are physical signs of an injury, we are unaware that she has hurt herself.

Over the last year Olivia has become increasingly difficult, showing very challenging behaviour. When we are out in public, she becomes very distressed and usually begins to show this by saying, "the crocodile bite me". This is the start of her ritual. She then follows with crying, screaming, spitting, lashing out with her arms and legs, trying to take her clothes off, and calling everyone, regardless of their gender, a "stupid lady". She has very erratic mood swings from aggressive to almost angelic. We can never anticipate what will trigger her off, so trying to avoid a potential tantrum is very difficult.

Olivia is now in full time school. She attends a fantastic special school locally. She travels to school on a bus, which she really seems to enjoy. The only problems we encounter there is if there is any change to the regular surroundings whilst we are waiting for the bus to arrive (e.g. roadwork, loud noises, someone different standing at the bus stop with us, or if anyone tries to talk to her without an invitation from her first). Her class is very small with only 8 or 9 children, depending on which day of the week it is, as most of the children are only there 2 or 3 days. While

she is there, she has speech therapy twice a week, physiotherapy once a week incorporated into a P.E lesson, and an individual learning programme.

Olivia is a very affectionate little girl, and if she chooses you, will smother you with hugs and kisses. She tends to favour older children and adults, especially if she is getting that golden one-to-one attention. She is also a real character and has us all laughing with some of the strange little things that she says. Rebekah and William adore her, and although she quite often spoils their fun and is aggressive with them, they love her to bits and are very protective of her. Olivia is quite small for her age and has small hands and feet. She is a really beautiful child with long golden hair and gorgeous brown eyes. Sometimes her eyes seem so deep that you could just fall into them. Although she has made our lives very different, she has shown us not to take for granted what you have. It has also made us appreciate even the tiniest of steps. This is our story so far.

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Julia's Story

by Kathy Wise



There was nothing that would ease the pain of having a miscarriage except for getting pregnant again. When, 3 months after the miscarriage, I discovered I was pregnant both my husband John and I were elated. We had been married for 5 years and were both ready to start a family. My pregnancy went well. I was put on modified bed rest in my 8th month, though, because I had contractions. Julia was breech, so she was delivered by Caesarian section on September 12, 1996. She weighed only 5 lbs 10 ozs.

We had music playing in the operating room as she was being delivered. It was Mom playing classical music on the piano. The lights in the O.R. were dimmed. She was placed in a warm water bath immediately after she was born. It was sort of like a LeBoyer birth – supposedly less violent for the newborn. The doctors and nurses were touched by her birth. A few had tears.

I can remember later in the afternoon on the day that Julia was born. I was lying in bed with her in my arms sobbing with joy. My brother and sister-in-law walked in the room. My sister-in-law asked “Having a tender moment?”. She understood the joy of motherhood that I felt.

The problems started immediately. She was found to have a dislocatable left hip and was placed in a Pavlik harness on day 2 of her life. The harness was supposed to keep her hip in place so that hopefully it would develop more and stay in place. On day 7, she was hospitalized for a week for hypothermia. We never did find out why her body temperature dropped. It was during this hospitalization that I saw my husband cry for the first and only time in the 13 years that I have known him.

At age 2 months, Julia was put in traction for a week then put into a spica body cast. The Pavlik harness hadn't worked so the cast was needed. Julia was in this full body cast for her first Christmas. When, at the age of 12 months, she wasn't sitting on her own, we started Early Intervention and saw a neurologist. He did a slew of tests and didn't find anything wrong. He didn't do a chromosome test. He said she had mild cerebral palsy. Julia was able to sit on her own shortly after starting early intervention and soon was crawling. At 16 months she had a grand mal seizure. She was air lifted to Children's Hospital in Boston and spent some time on a respirator. That was the scariest time of my life. I was so numb in that helicopter. I couldn't believe what was happening to my baby. For reasons I don't want to go into, we started seeing a new neurologist after the seizure.

Julia began to say words around 20 months, and walked shortly after her 2nd birthday with the help of leg braces. In August of 1999, the month before her 3rd birthday, she had a major hip operation and was placed in another full body cast, this time for her right hip. Her right hip was subluxing and she spent 7 hours in surgery so the surgeon could put a plate and pins in her hip and reconstruct her hip joint. She was in the cast for her 3rd birthday. She spent a total of 8 weeks in the cast and had months of intense physical therapy afterwards.

Around the same time that Julia was to undergo this surgery, I was applying for Julia to receive Medicaid. I was going over a neurological office visit report and saw that the doctor had said that if a chromosome analysis hadn't been done yet, we should do one. I knew that that test hadn't been done, so I asked that we get an order to do it since she would be having her blood drawn anyway in preparation for surgery. That's how we found out about Idic15.

When the geneticist told us about Idic15, my husband and I had different emotions. My

husband was relieved to finally know why our daughter was so delayed. I was crushed. Why did this happen to us? Why did this happen to Julia? Would I be strong enough to mother a child with 'special needs'? So many questions.

The geneticist put us in touch with Donna Bennett, cofounder of IDEAS, and then we learned about the support group on the internet. This group has been my lifeline. It is truly a comfort to know that there are other people dealing with similar issues with their children. We exchange information, smile stories and most importantly, offer each other lots of support. I don't know how I would have gotten through all of this without this group. It has been this internet group and the support of my mother and father that has helped me to maintain my sanity. My mother and father have been there every step of the way for me. Through the hospitalizations, my depression shortly after Julia was born, and just helping out with Julia when I need a break. John and I have been able to get away for weekends with friends or to Family Support Conferences knowing that Julia is with people that love and care for her as much as we do.

Julia started preschool in November of 1999. We are very fortunate to live in a community that can pride itself on its special education system. I have seen many improvements in Julia since she started school. She is more cognizant of the world around her, she follows directions more readily, her eye contact has improved, her low muscle tone has improved, and she learned her colors!

Now we are approaching Julia's 4th birthday. Let me talk about this beautiful little girl's personality. She loves to give kisses. She loves to give hugs. She loves dolls. She loves books. She loves taking baths and swimming. She loves Barney and Elmo. She loves to go for walks in "the jungle" (the woods near our home). She loves music. She knows the words to a countless number of songs and nursery rhymes. She loves her Nana (my mother) and Papa (my father) and has told them so. She loves to sing. She thinks there is a song for everything in life. "Car song?" "Airplane song?" she'll ask. So we make up little songs about everything. She has an incredible memory. She's just precious beyond words. Those of us who know her well, love her with all our hearts.

Julia's muscle tone is still low. She is very delayed in both fine and gross motor skills. Her speech is mostly in phrases, and she has no conversational speech yet. She can get her wants and needs across – I am grateful for that. Julia does show some autistic signs but would not be considered autistic. She has sensory integration difficulties but they seem to be diminishing over time.

Let me talk now, about the changes that have occurred in me after having a child with special needs. Before I had Julia, I was a 'go with the flow' type of person'. I didn't create waves. Now, I have become an advocate, not only for Julia, but for myself as well. I've learned that in order to get what you want, sometimes in life, you need to speak up. The 'squeaky wheel gets the grease' as they say. This squeaky wheel will squeak as long as it feels that Julia is in need of something she is not getting. I'm not afraid or intimidated by doctors anymore. We see so many of them. They are ordinary people that have furthered their education. They have faults. They are not Gods.

What the future holds for Julia I do not know. I do know, however, that I am committed to doing whatever I can to make it as bright as possible. She is a gift from above, not "typical", but a gift nonetheless. For some reason, God had chosen me to take care of one of his 'special' children. I suppose I should feel honored. Hopefully, further along in the grieving process, I will. For now, I

am just enjoying Julia and hoping that one day I will truly come to terms with all of this.



WELCOME TO THE WEBSITE OF IDEAS, a support group for families and professionals interested in the chromosomal conditions known as isodicentric 15 (formerly inverted duplication 15) and interstitial duplication 15. Through our informative newsletters, parent networking and research contacts, our group is growing every day. If you're searching for information about isodicentric and interstitial duplications of chromosome 15, you've come to the right place!

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