Understanding chromosome disorders
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+44 (0)1883 723356

Duplication 15q11.1q13.1

A Dad's View of 15q – An insight to the first 2 years of a "Chromo Parent"

by Ian Hill

Chromosome disorders and their diagnosis on your child are a lonely and isolating event, as a Dad you must stay strong, keep it together, maintain that stiff upper lip and keep the ship steady, or so we are told by society. Reality is somewhat different, your children and your family are your world. Never fear shedding a tear for them, be it pride or pain they are worth every last one and never be too proud to show it.

Shortly after receiving diagnosis of duplication 15q for our son Matthew I registered with Unique, the loneliness decreased a little and I had valuable information at my disposal. The information was great, the welcome so warm, but there seemed to be something missing from the groups, something so familiar, something relatable, something so simple, other Dads. So, the following is a Dad's view of 15q, a recollection of events of Matthew's life so far, emotions, thoughts, and musings from the heart, typed at times through tears of pain and pride at the little boy that is my son, Matthew Hill – Dup 15q11.1q13.1.

22nd July 2013, the day had arrived of the planned C-section, my wife Debbie and I would find out if I would be truly hen pecked and Father to another beautiful daughter or get the boy I so longed for that would complete our family; either way the love would be instant and I would burst with pride. As we turned on the car and the radio switched on,

news filtered through that the Duchess of Cambridge was in labour, our special day would be extra special and our new child would share a birthday with a future Monarch. The birth was relaxed and straightforward, the surgeon as promised did not reveal "the flavour" as my father in law would say, and it was left to me to find out. A tear hit my eye and as I looked to Debbie my voice went hoarse, "a boy, we got a boy, thank you Debbie" I choked out.

All appeared normal following the birth, Matthew seemed just like any other baby, a little difficult to settle into a feeding pattern but with determination from Debbie that passed. At around just 4 weeks we had our first taste of the children's ward at Warrington Hospital, Matthew stopped breathing while feeding, described as a "blue moment" by Doctors. Following two days of tests and observation, results proved inconclusive and it was attributed to reflux, whether it was linked to what was to follow we are not sure. Time passed by and at his 6 week check the first concerns were raised as he did not smile as expected, initially passed off as "nothing to worry about, we will monitor it" immediately the mind was set racing and the inevitable worries. In the following weeks I found myself constantly imploring him to smile but to no avail, when he finally did the feeling immense, yet the smile was fleeting and not full and gummy like our daughters' were when they were the same age. "Why?", "Is it just a boy thing?", "Just ignore it, there is nothing wrong with my boy!" I told myself, deep down and secretly I feared there was something not quite right, but denial was in full swing.

As summer passed to autumn and in turn winter his development lagged in many areas, milestones were missed but child development specialists told us to just wait and see, deep down I still held a fear something was wrong, but denial stood firm. As a new year dawned we were full of hope and expectation as we prepared to move house to accommodate our new larger family, unfortunately a week prior to exchange and completion our onward house purchase was delayed, our buyer gave us 2 weeks for us to sign and move out or he pulled out. Reluctantly we agreed to his demands, completed and moved in with Debbie's Mum for the 6 weeks it was anticipated that it would take to resolve the onward issues, hopeful for a quick resolution we shoehorned all 5 of us into one bedroom of a 2 bed terrace.

The 6 weeks approached and we looked forward to being able to get space again in our new house when our attention was diverted back to Matthew and something that made our development fears come back to the fore. The smiles he had for us had gone, he wouldn't eat, he slept most of the day and it was more of a question of how long is he awake rather than how much does he sleep, our beautiful boy held a vacant stare, he had no interest in toys and had almost become a zombie before our eyes and we had no idea why. Were we being paranoid, neurotic parents? This was our third child; we knew what was normal for a child his age and what was not. "Something is wrong" I suggested to Debbie. "IT IS NOT, leave my boy alone!" she replied, I knew, and I could see that she did

too, but the denial had kicked in again, the acceptance was just too hard to take. A few days later Debbie phoned me at work to say Matthew was doing something funny with his arms, "It will just be wind", I dismissed as I had a busy day at the office, "But phone the Doctor and see". The local GP was mostly unconcerned but made an onward referral just to be sure. That night he did it again, I thought it was strange, "Probably wind". I told myself unconvincingly, but I knew it was far more serious and it terrified me.

I began to withdraw from Matthew, I hated myself for it, but I couldn't help it, the unknown was scaring the hell out of me. If he was waking I found I would avoid him as I knew one of the episodes was inevitable and seeing it upset me too much. This was my Son and I was scared to go near him, I had to fight back to protect him and be strong.

That weekend we took the kids to Knowsley Safari Park, we stopped for lunch and Matthew waking from a slumber started doing it again, spasm like movements, this time more pronounced, all at once arms moving together, eyes wide, legs up! "What is he doing?" Debbie whispered to me close to tears but not wanting to raise concern with the girls. He continued, arms, eyes, legs, and again and again for several minutes, all the time unresponsive, then it stopped and he cooed. "Probably wind", I thought, but I knew now it wasn't, we both did. Later that day the phone rang, it was the vendor on the onward house purchase; our house purchase had fallen through, though it barely registered at the time through the concern for Matthew.

That night I foolishly turned to Google, typing in the symptoms it arrived at a diagnosis, one that was ultimately correct but reading it made my heart pound and my head spin, I closed the laptop and tried to pretend I hadn't just seen what I had read, denial was taking over again. Early the next week and manning the office of the family business alone as my Father was away on holiday I was fraught with fear, the Google "diagnosis" still fresh in my mind, the solitude made thoughts run through my head, I cried, emotions of anger, fear, upset and more denial! I dreaded the office phone ringing and having to deal with customers, my head and heart were both elsewhere. "My boy, my boy, not my boy, what is wrong with my boy?" I asked over and over again. The phone rang, it was Debbie, and he was doing it again and had done so a few times that morning. "Just take him to hospital", I said choking on tears, we were both crying, we now knew there was something seriously wrong.

Having already googled the symptoms I had an idea what the diagnosis would be but still denial kicked in, if I didn't admit it, it may not be true. I grabbed the phone, called my Father to interrupt the final day of his holiday and tell him I was abandoning all at the office, the business was important to me but right at that moment nothing else mattered but Matthew. I raced back down the M62 to Warrington to be with Debbie and Matthew at hospital, he was already on the ward and I was taken through to see them. Multiple doctors came and went, questions asked over and over, tests done, bloods, Matthew prodded, poked and pulled in all directions, an MRI, a trip to Alder Hey for an EEG, fear, diagnosis, numbness! The Doctor sat us down and told us, "The EEG, shows hypsarrhythmia consistent with West Syndrome/Infantile Spasms", my throat burned, I wanted to be sick, tears filled my eyes, the prognosis and its effects still etched in my mind from Google, "Our little baby boy has a rare form of epilepsy, what now?" I thought.

Tests of all kinds were done to determine the cause of the epilepsy, the medication failed to take hold and the seizures came again and again, we became scared to wake him as that was when they would usually happen. Emotions ran high, sleepless nights followed, all the while cramped in a house with no space to let it out. Another long seizure episode of over 20 minutes, curtains twitched from neighbours as an ambulance arrived for the 6th consecutive night to take Matthew away to hospital, lights flashing, the girls woke confused to find us gone again. "When will this stop?" we asked, "Help our boy!" we pleaded. Easter weekend was spent at the children's ward with our daughters packed off elsewhere without us, oblivious to events but our eldest was acutely aware that something was not right.

The Vigabatrin was not working on its own so steroids were introduced for a short sharp burst, Matthew bloated, a week or so passed and still the seizures came it had been nearly a month since the medication started, then on the night of 7th May it happened, he woke and he smiled! I looked, Arms? Still! Legs? Still! Eyes, normal! "Had they stopped?" we asked ourselves cautiously. They had! Seizures under control we waited, more tests were done and seeing Matthew whimper and sob bent double in a Doctor's arms as they performed a lumbar puncture broke our hearts again, no child so young should be put through this and no parent should have to see them suffer like that but we declined the option to wait outside, there was no way we could leave him alone, still we waited for the cause.

Months passed and summer came, our life seemed to be passing us by and we felt guilt as unintentionally we neglected the girls at the expense of Matthew and the atmosphere at home grew more and more tense. Slowly though Matthew began to smile again, he became more alert and started to show an interest in things around him, the first signs our little boy was coming back. As Matthew's first birthday approached we received the call we wanted, the call we dreaded! They had an answer, a diagnosis, a cause. Tensions at our temporary home were at breaking point we knew the news was going to be hard to take; again we had nowhere to hide. Meeting the consultant on that hot Friday in July my heart raced, I held Debbie's hand one way and Matthew's the other, the consultant paediatrician spoke explaining what they had found and it hit us both like a kick to stomach.

"A duplication what?" I enquired. "Duplication 15q, it is a rare chromosome disorder", the Consultant said slowly. People were speaking but little registered, tears filled Debbie's eyes, I tried to hold mine back with little success, the voices continued but the room felt silent behind a whirling high pitched noise in my head. After regaining some degree of composure we were told calmly and with an ironic combination of compassion and directness that our little boy may not be the boy we hoped and dreamed for, that the hopes and dreams may not be fulfilled, we had to grieve, we had to let go and we had to adjust. Our precious little boy, our very own Prince had a rare chromosome disorder and it would change his and our life for ever. The Consultant pointed us in the direction of Unique and that night reading information given to us about 15q disorders the reality began to dawn, thoughts went back to what the Consultant had said, those hopes and dreams began to slowly fall apart and I feared the worst. Like with many Dads I had unrealistic

hopes and dreams, Matthew was going to play football for my team, Manchester United! I already had him down as their future centre forward, dream shattered! Football in the park? Shattered! University? Shattered! Matthew's future independence? Shattered! All those little things you take for granted, how many would we experience with him? Numb, I went to bed, I felt helpless, useless, devastated, full of self-loathing and blame and asking the questions, "Why Matthew? Why us?". "What happens when we are old and gone, who will be there for him then?". At that point there seemed to be no light in the tunnel and I didn't know where to turn.

The next day I met with a group of friends I have known since school to bid farewell to one who was going abroad to work, I had said I would go and I felt that I couldn't bail and let him down. As I arrived the first question asked was inevitable, as was my response "How's Matthew?" one enquired. Instantly I broke down in tears, 34 years of age, in front of the lads on an afternoon beer session, the ultimate shame? Not one bit! They were great, by their own admission they didn't attempt to understand what was going on in my head, but they were supportive, they had my back, that's what friends of near 25 years are for and they knew that this was serious.

Matthew's 1st birthday was a subdued affair, the diagnosis just days before made it all so hard, but we had to maintain a brave face for the girls, they wanted cake and balloons for their baby brother, we did our best to celebrate despite the burning pain inside but it just seemed so forced and difficult. Slowly the haze that had shrouded me for the past 5 months began to clear, we had an answer on diagnosis and now I had to be strong! I was Matthew's Dad, his advocate, his rock! I also needed to be there for Debbie and the girls, my attitude began to change and new found strength followed, I realised that if I crumbled it would affect the business, my wife and all of my kids and all were far too important to me.

The house move finally happened in late August, 6 long months of waiting over, within weeks of moving in Matthew began to thrive, his smile bigger, giggles, laughs, playfulness, hugs and cuddles all we ever wanted was back. My strength grew again and I began to reach out to contact other families via Unique, they became an invaluable resource and I realised we were not as alone as we first imagined, there were many more families out there who had been through the same emotions, pain, confusion and come out the other side, the other side of that tunnel that weeks before was totally dark which I was now beginning to emerge from. It helped me to focus on the now and cherish what Matthew was doing today, I realised that his condition I couldn't change, but if I could make the most of what he did today and any progress he made then I would be sure I didn't miss anything, the small things became big and the joy at each one immeasurable and since reducing and then stopping the Vigabatrin we saw a new boy emerge with every step down of the weaning process. We remain thankful and feel blessed that thus far the seizures have not returned, we accept that may not always be the case but that reinforces the need to embrace and cherish every part of today.With the positivity we showed, Matthew responded and although today he is still behind his milestones for his age he is making progress and with a smile that could melt a thousand hearts. No longer is he the boy of shattered dreams, he is our brave little boy who is fighting his condition and with each day surprising us over and over again. Seeing comparisons to his peers is still hard, seeing Prince George in articles or TV appearances brings the stark reality of difference in development to the fore, at times it hurts as I consider where he could be right now, but above all I value the massive steps he has taken from that baby without a smile to who he is today. He is a cheeky little guy, very attached to his immediate family and wary of those he does not know and even some who he does, he has developed an incredible dynamic with his eldest sister who at eight years old shows a maturity with him way beyond her years, their closeness and bond is a real joy to watch and both our girls have and will learn so much from him that we as parents and the education system cannot teach.

Approaching two years old he is attempting to take his first steps and bring us a whole new dimension of fun and trouble. Like with many chromo-kids, feeding is a slow and unpredictable process and sleep is erratic, he has very limited, if any recognition and understanding of instruction or language and as yet he is still to speak his first word. We know there is a real possibility he may never, but hearing a babbling "DADADADAD" is enough for me right now and hearing a hummed response to "Love You" that sounds like and matches it is a gift. He may not get to play football in the park, if he got my footballing genes he was never going to play for The Dog and Duck reserves never mind United, he may not go to University or do any of the other things we hoped for but one thing he has that you can see in his eyes is a bravery and determination that will go a long way! The future we cannot predict and we know the condition will bring challenges and struggles but with the help of family, friends and all those associated with Unique we know we have great support whatever happens. I am Matthew's Dad, just an ordinary bloke with amazing kids and an incredible wife who is stronger than she will ever admit, who in a short space of time has been elated, deflated, devastated, touched by the kindness of friends and family and become wiser and stronger and all from someone so young. He is Matthew, he has duplication 15q but duplication 15q does not have him!

As I said at the start, your children and your family are your world. Never fear shedding a tear for them, be it pride or pain, they are worth every last one and never be too proud to show it.