

apologize and have a discussion if there has been an issue. Our boy has grown up! To all the parents of little ones our advice would be this: – your child will reach wherever they are meant to be in the end but may need to take the scenic route instead of the motorway. Take your time to enjoy their childhood; don't always want them to reach the next stage, as all too soon they will be grown up.

## 16p11.2 duplication

**Adam Barkworth**

Jan Barkworth



This is my 18 year old son Adam. We have had quite a heart breaking journey with Adam over the years. He was a mainstream child with learning difficulties and a diagnosis of ADHD up to year 6. I started to suspect autism around year 6, but was told it was all in my mind. He started year 7 and within 3 weeks the SENCO rang me with concerns and agreed she felt autism to. Sadly we had to move house and change his school and the new high school wasn't as supportive and that halted Adam's assessments. By year 9, Adam was suffering terrible anxiety attacks, motor tics and struggling to communicate. I had to fight to get his Ados done, by then he had had genetic testing which showed he has 16p11.2 duplication. He scored 21 in his Ados and was

diagnosed Autism/Tourette's/ social anxiety; sadly he has regressed so much he pretty much missed years 10 and 11

of high school. He started a specialised 6th Form with full time one to one support (very rare for them to allow). They

worked so hard to reduce his anxiety and help him communicate in various ways.

Adam started at another specialised unit today with full time support, still non verbal socially – unless he knows you enough to feel safe around you, he's now classed as learning disabled, yet he's self taught himself graphics design and got his own Facebook page <https://m.facebook.com/AdamsGraphicsDesign/> where he will do logos, etc for people with the help and support of myself. Two days ago we produced a price list for his work and within 2 days he's had 3 orders. Today he's become registered self-employed!

My boy, who on paperwork is classed as complex and will always need some level of support, now owns his own business doing what he loves, working from the comfort of his own bedroom with the added bonus of no verbal communication with clients, to heighten his anxiety. Instead he can do it all via online with my support. Don't ever give up hope for your child; they can achieve with the correct support and environment. I'm such a proud mum today I never thought he would be classed as owning his own business at any age, let alone at age of 18!

## Uncovering a New Genetic Disorder

Cracking that contagious grin and crinkling his bespectacled eyes, Cole dumps a bin of toy cars onto the floor. He sifts through, keeping an eye peeled for his favorites, and exclaiming over a particularly exciting find. Cole welcomes anyone who wants to join in with his play, scooting over to make room and providing helpful instruction. This scene is typical of the behavior of any number of 6 year old boys in living rooms across the country. Yet, for those who know Cole and his family, this mundane activity is the culmination of hours, days, weeks, and even years of practicing the component parts. For his peers, the ability to plan and engage in sustained, self-directed play, to grasp and manipulate toys appropriately, and to communicate thoughts verbally emerged without incident. This is not the case for Cole.

Cole has not developed at the same rate or with the same ease as other children. The clues emerged throughout infancy, toddlerhood, and early childhood, leaving a breadcrumb trail that led his parents, Haley and Eric Oyler, to several rounds of genetic testing and visits with various specialists. The long sought answer, however, led to more questions. Cole has a deletion in the SET binding protein 1 (SETBP1) gene, which impacts his neurological system. The gene is located on the long arm of chromosome 18 at position 12.3, also written as 18q12.3. The genetic mutation is de novo which means that neither parent has the same genetic difference. Unfortunately, beyond the scope of its impact and its location, we know little else about the function and purpose of the gene or the protein it produces. One barrier to progress in understanding this rare neurodevelopmental single gene disorder, is its scarcity; there are fewer than 40 known cases world wide.

Although the number of cases worldwide is very small, the impact of SETBP1 disorder is not. Haley with the help of friends and family established the SETBP1 Society to create a community for the families affected by SETBP1 disorder, to encourage awareness, and to promote targeted research. Most urgent, though, is the need for research into the function of SETBP1, as well as, effective interventions and treatments for those individuals with SETBP1 disorder.

To learn more about SETBP1 disorder, visit [www.setbp1.org](http://www.setbp1.org).

Written by Dr Trina Geye

*Director of Academic Support and Assistant Professor at Tarleton State University, and Board Member of SETBP1 Society*



## 18p deletion

**Christian, aged 20 months**

Shel Fuchs

I am writing this to tell the story of our son. We first found out about his medical problems around the age of 2 months old, but just recently found out about his genetic disorder. At 2 months old we found out he was silently aspirating on all consistencies and refluxing so bad that he was aspirating on that as well. We were immediately admitted to the hospital, where they placed a feeding tube. We've been through multiple tubes and are now just currently an AMT Mini-One Tube. After the