



Newsletter 51 Autumn 2012

Angelman Syndrome Support  
Education & Research Trust

ASSERT  
Conference  
2012 review

Latest research  
from the UK  
& US

London  
Marathon 2012

Fabulous  
Fundraising

Siblings club

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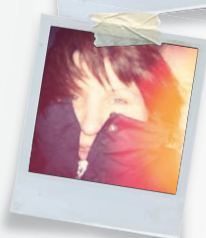
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## Useful Stuff

### OUR FREEPOST ADDRESS

**ASSERT**  
Freepost  
PO Box 4962  
Nuneaton  
CV11 9FD

### OUR WEBSITE

[www.angelmanuk.org](http://www.angelmanuk.org)

### EMAIL ASSERT

[assert@angelmanuk.org](mailto:assert@angelmanuk.org)

### HELPLINE

**0300 999 0102**

Assert Families - New Diagnosis Advice  
Contacts - Support

### ASSERT SHOP

Wear your charity with pride! T-shirts,  
polo shirts, bags, hats and lots more from  
Spreadshirt, our on-line retailer:

[www.angelmanuk.org/spreadshirt](http://www.angelmanuk.org/spreadshirt)

### FRONT COVER

Photograph courtesy of Robert  
Chadwick. We'd like to use one of  
the great photos taken at our 2012  
conference on each of our future  
covers. If your AS family member was  
one of those photographed and you do  
not want their images used on Assert  
material please let us know at the above  
address as soon as possible.

### CONTENTS OF THE NEWSLETTER

You will notice that the pieces in this  
newsletter are drawn from a wide  
variety of sources. While we are keen  
to promote discussion and to pass on  
any views and experiences, it is also  
important to appreciate that the opinions  
and views expressed by contributors  
to this newsletter are personal and not  
necessarily those of Assert.



## Letter from the chair

Another conference comes and goes, I'm sure that they pass quicker every time. It was fantastic to see so many of you at the conference, and from all the feedback we have received it seems that the vast majority really enjoyed it and found it greatly beneficial. Certainly it was great to enjoy the normality of AS, not having to explain or apologise for the action of our AS kids or adults. Our family decided to take the luxury of staying an extra night at the hotel which after you all left resembled a ghost town. Ruth certainly wasn't impressed at the lack of response she was getting from the new guests, having been able to relish so much attention previously. It isn't too late to complete our conference questionnaire, which you can find at [angelmanuk.org/survey](http://angelmanuk.org/survey). Also do send us your conference stories or photos to [conference@angelmanuk.org](mailto:conference@angelmanuk.org).

I would like to say, for a final time, a massive thanks to all those who helped make the conference what it was. To my fellow trustees whose tireless efforts were amazing and often beyond the call of duty. To the many speakers who gave up their time, the majority at their own expense, to come and give us the benefit of their vast knowledge and experience. To the hotel staff, who on the whole were fantastic, amazingly helpful, often going the extra mile to help us all out. And lastly, but not least, to all of you who came to the conference, without you it would have been very dull and quiet to say the least. It was great to see old friends and make new ones, and to be able to share quality time with people who understand the world we live in and who can offer support and comfort. Thank You!

As for the trustees, we are now taking an all expenses paid trip to the Maldives to recover. If only it were true, instead it is back to the strains of "normal" life, the joy of battling with professionals, of having to have 6 pairs of eyes to watch what our kids are getting up to and having to say sorry for what our kids get up to. We are also now busy working on newsletters, databases, trifold leaflets, fundraising, keeping up with the latest research, to name just a few of the activities that occupy us. And just to remind you, in case you were conducting your own sleep research during my keynote speech at the conference, if you would like to help ASSERT in anyway, please do contact us. Whether it be to fundraise, to help organise a regional meeting, to write something for the newsletter, or even become a trustee; we would love to hear from you. As the ASSERT family is about you and it is for you. As trustees there is no way we can manage to do everything, especially as the organisation grows, so please do get involved.

Hot off the press news - ASSERT are applying for a number of places in various runs around the country, including the Great North Run to give you a chance to fundraise for us. If this is something that is of interest to you, or your friends or colleagues, please do get in touch with us. Watch this space and our website and Facebook page, more details coming soon

**Jeremy Webb**





# Assert Conference 2012 Review



When conference 2012 was planned, little did we know that the move to the Hilton Hotel in Coventry would prove to be such a hit with everyone that attended and the hotel staff too! We filled the whole hotel- with some overspill into the adjacent Holiday Inn- representing a 50% increase on previous numbers at Loughborough University. It really was an experience to see so many families arriving.

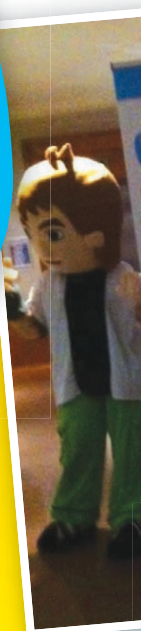
The new venue offered a more relaxed atmosphere to all of the proceedings- and the smiles (some mixed with a little trepidation) were obvious to all in reception. A welcoming atmosphere, an adjacent car park complete with luggage trolleys, no more stairs, no more single beds and great food soon dispelled any nervousness.

Friday evening saw the usual mix of catching up with old friends and making new ones. Of overcoming that first conference fear of the unknown. Of people relaxing in understanding company and being able to laugh at life again.



As ever with conference, the mornings can be chaos. Too many the night before, mixed with excitable families heading out en masse, breakfast and a tight timetable of presentations, workshops and seminars. But after the children and carers had departed to Drayton Manor in their six coaches, Saturday started positively and only slightly late. Keynote presentations were from Dr Stephen Calculator (Enhanced Natural Gestures), Angie Mabb (Huang-Philpott-Zylka Research) and Professor Gregory Stores (Sleep problems and management). A large lunch was quickly followed by the usual variety of seminars and workshops, including some professional 1-2-1s. Topics included, epilepsy, genetics, physiotherapy, benefits, communication, leaving home, eating and drinking, grants and funding, wills and trusts and general medical issues.

The arrival of the coaches signalled the start of the hands on sessions- where suppliers bravely allow their products to be put through a full AS workout and a siblings trip to the local bowling alley for some much needed time out.







Dinner followed and then the evenings entertainment started. Disco, karaoke and the infamous ASSERT raffle- which this year raised over £600. Thank you for all of the donated prizes.

Sunday morning involved packing (sadly) before the coaches headed off for a shorter day at Twin Lakes. The keynote speeches were given by Bernard Dan (general research update) and Professor Chris Oliver (behaviour research update). Workshops followed on topics including sibling issues for parents, research Q&A, Makaton, behaviour, adult services/ future care and communication.



After the return, there was a great opportunity to say goodbye to everyone over some delicious cakes as the weekend drew to a close- and then it was over.

**Is it really two years until we can do this again?**

**Thank you to everyone who came, contributed and enjoyed conference 2012.**



A special mention has to go to the staff of the Hilton who were simply brilliant. Not only did they cope with our 'interesting' group with good humour and fantastic service- but the comments from them about how inspirational they had found the weekend is testament to us all.

**We've left being asked to return again soon. When was the last time that happened?**



# Siblings

## Just for Us!

**Q: WHERE DOES A BABY APE SLEEP?**  
**A: IN AN APRICOT.**

**Q: HOW DOES AN ELEPHANT GET DOWN OUT OF A TREE?**  
**A: IT SITS ON A LEAF AND WAITS UNTIL WINTER.**



These are the sort of weighty matters (geddit?) that some sibs were turning their attention to as the 2012 conference came to an end. It struck me afterwards that just about every AS sibling I've met has a well developed sense of humour and so I invite siblings of every age to send in their best joke and we'll arrange a prize for the best ones.

Speaking of the conference a group of 18 siblings went off to the nearby bowling alley on the Saturday after Drayton Manor and despite us managing to break the tv scoring system we all had a good time. It was great to be able to get away for an hour or so and do something a little different together and if at all possible we will extend the events for sibs even further at the next conference. There was opportunity to talk about individual situations and to let off some steam - leading to some titanic scores (you know who you are!).

It was an honour too to lead a couple of workshops for parents on sibling issues and share thoughts and experiences together. I was deeply impressed by how keen folk are to ensure no-one in their family gets left out. I was able to share some research into siblings along with my own experiences and hopefully we will be able to make some of the slides available through the website.

I'm going to leave it there but thanks to all the siblings for making conference a great experience and remember to send in your jokes. We are also still publishing any drawings that you want to send in.

**Russell**

**DON'T FORGET**

our Facebook page:



**Assert sibling group**

[www.angelmanuk.org/siblings](http://www.angelmanuk.org/siblings)

It's growing all the time. So if you're old enough to be on Facebook, join us and let's hear from you.



## New Young Sibs Club!

Everyone gets  
a T-shirt!

Well, not everyone gets a T-shirt... only special people. If you are under 13 we want to encourage you to send in a piece of artwork or writing (a story, poem, that sort of thing) that we can publish in the newsletter in the Siblings section – it can be about what it's like to be a brother or sister of someone with AS or it can be about something that matters to YOU.

And everyone that has something printed can choose between an *I'm no Angel!* T-shirt or one of the new Smiley Angel designs (below).

So what have you got to lose?

Send your entries to Russell Andrews either via email to:

[Russell.Andrews@angelmanuk.org](mailto:Russell.Andrews@angelmanuk.org)

...or by post to:

ASSERT  
Freepost  
PO Box 4962  
Nuneaton  
CV11 9FD

**...and don't forget to tell us  
your T-shirt size!**



Picture drawn by Alex Eland whose brother Elliott has AS and is aged 2.



Picture drawn by Katie Munro, aged 8  
whose sister Sophie has AS and is aged 12.

# Stress in parents of children and adults with Angelman syndrome

**Mary Heald, Prof. Chris Oliver, Dr Anna Phillips, Dr Debbie Allen, Darrelle Villa, Ana Vitlic, Dr Lucy Wilde, and Carmel Bennett. Cerebra Centre for Neurodevelopmental Disorders ([www.birmingham.ac.uk/cnodd](http://www.birmingham.ac.uk/cnodd))**

**At the 2010 ASSERT conference 38 parents took part in a study conducted by the Cerebra Centre for Neurodevelopmental Disorders looking at stress in parents of children and adults with Angelman syndrome (AS). Families completed a range of questionnaires and gave blood samples. This article is a summary of the initial findings.**

## BACKGROUND

Previous research looking at the effect of caring for a child or adult with an intellectual disability has found that parents/carers may have higher levels of perceived stress compared to parents of typically developing children. Parents who care for an individual with an intellectual disability have also been found to have poorer antibody responses to different types of vaccines, indicating that they may be more susceptible to viral/bacterial infections.

To date, there is limited research addressing parental stress in AS. In our previous research we have found that mothers of children with AS are more likely to experience high levels of stress, anxiety and depression than mothers of typically developing children and children with Cri du Chat and Cornelia de Lange syndromes (see our website for more information).

The aim of the current study was to further investigate the levels of stress, anxiety and depression in parents of children and adults with AS, and to assess whether certain factors influence the levels of stress which parents experience.

## CURRENT STUDY

At the 2010 Assert conference at Loughborough University we met 38 parents and asked them to give a blood sample and fill in a range of questionnaires. The questionnaires included measures of stress, anxiety and depression, sleeping habits, support and child behaviours.

The parents who took part were aged between 24 and 62 years. The children of those parents who took part in this study had all been diagnosed with AS and were aged between 2 and 40 years of age.

## RESULTS

### Stress

As suggested by previous research, parental stress levels were high. Parental stress scores were significantly higher than the scores of parents of children without an intellectual disability.

### Anxiety and depression

42.1 % of parents who took part in our study had scores for anxiety in the normal range, 15.8% had high scores and 42.1% had very high scores. Similarly, 44.6% of parents had scores for depression in the normal range, 26.4% had high scores and 29% had very high scores.

57.9% of parents who took part in our study scored above the cut-off for anxiety and 42.2% for depression. This is significantly higher than those for the general population, which are 12.6% and 3.6% for anxiety and depression respectively.

### Factors influencing depression/anxiety and stress

After looking at the levels of anxiety, depression and stress, we carried out a range of statistical analyses to explore whether specific factors influenced the levels of parental stress. Four factors were found to be associated with the scores on the stress questionnaire:

#### 1) Self-injurious behaviour:

The presence of self-injurious behaviour led to an increase in parents' stress levels.

#### 2) Conduct problems:

In general, parents whose children showed more general behavioural problems, experienced higher levels of stress.

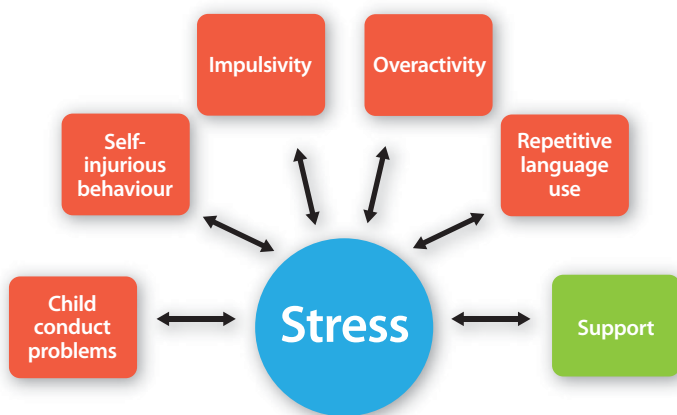


### 3) Impulsive and repetitive behaviour:

More impulsive (not being able to wait for things) and overactive child behaviours and more frequent use of repetitive language (this was only the case for a small number of children and referred primarily to repeated signing) were related to higher parental stress levels.

### 4) Support

We found that parents who reported that they had more support felt less stressed. Many parents reported low levels of support, suggesting that services need to be improved and need to be made more accessible.



## BLOOD ANALYSIS

The blood sample which was taken from parents was analysed at the Sport and Exercise department at the University of Birmingham. These results were compared to non-caregiving parents. The concentration of cortisol and DHEAS, were measured in the serum. Cortisol is a steroid hormone released in response to stress and is mainly considered to be anti-inflammatory with the function of suppressing the immune system. DHEA, another steroid hormone, is considered to have an enhancing effect on the immune system. DHEAS counteracts the effect of cortisol.

Researchers looked at the ratio of cortisol:DHEAS, as previous research has suggested that prolonged stress leads to a rise in the cortisol:DHEAS ratio. Contradictory to what was expected, caregiving parents had a lower cortisol:DHEAS ratio in comparison to non-caregiving parents. This difference was mainly driven by the difference in cortisol levels between the groups. The results suggest that chronic stress may eventually lead to down-regulation of cortisol as opposed to its up-regulation seen in response to short-term stress. In other words, the body may adapt to long-term exposure to stress by releasing less cortisol.

## WHAT DOES THIS MEAN?

Our results provide further support for previous research which has suggested that parents of children with AS experience high levels of stress, anxiety and depression. Factors that were found to influence these levels of stress included children's levels of self-injurious behaviour, conduct problems, and impulsive and repetitive behaviour. This suggests that more needs to be done to help support parents who care for a child or adult who displays challenging behaviour. In addition to child behaviours, the level of support received also influenced parents' levels of stress. This suggests that more support should be provided for parents.

Overall, this article should make parents of individuals with AS aware that they may be experiencing high levels of stress. Many parents may also be experiencing high levels of anxiety and/or depression. For any parents who are experiencing some or all of these feelings it is really important to contact your GP, as this will ensure that you get the right kind of support and advice you need. Below is a list of resources aimed at providing parents with information on stress management and pointing you to groups and courses.

## RESOURCES

### Information for parents of children with an intellectual disability:

- [www.learningdisabilities.about.com/od/parentsandfamilyissues/tp/Stress\\_Management\\_Tips.htm](http://www.learningdisabilities.about.com/od/parentsandfamilyissues/tp/Stress_Management_Tips.htm)
- [specialchildren.about.com](http://specialchildren.about.com)
- [familylives.org.uk](http://familylives.org.uk)

### To download or request Dr Ann Edworthy's book *Managing Stress for Carers*:

- [www.cerebra.org.uk/parent\\_support/support/stress](http://www.cerebra.org.uk/parent_support/support/stress)

### For more information on stress, anxiety and depression:

- [www.nhs.uk/Pages/HomePage.aspx](http://www.nhs.uk/Pages/HomePage.aspx)



# London Marathon 2012

## **This year we had 7 runners in the London Marathon**

– 5 Golden Bond runners and two runners who very kindly ran on their ballot place to raise funds for ASSERT. Between the 7 runners they raised the staggering amount of just over **£15,000**. We are extremely grateful to all of our runners for their fantastic efforts.

If anyone would like to put their name forward for the chance of a Golden Bond place for the **London Marathon 2013**, please email **Lisa.Court@angelmanuk.org** by **7th December 2012**. The minimum sponsorship is **£1,250**.

## **A massive thank you to our 7 runners:**

Ed Winter, Duncan Dale, Nigel Flook, Haidee Moldram, Chris Stratford, Chris Rons and Laura Fraser.

## Ed Winter

### **Taking part in the 2012 London marathon has been something I will never forget for many reasons.**

In 2011 I decided that it would be a great idea to take part in the next London marathon, even though I was getting married the month after.

What drove me to run for Assert was my soon to be brother in law (Matthew Walburn, 21 years old) who has Angelman syndrome. I met Matthew in 2005, soon after this he was undergoing a back operation. During this time Matthew had some complications which meant he would either be paralysed or at worst die. It was a very difficult time for the family and for me to see a close family go through something like this.

Thankfully Matthew was a tough lad and managed to get through this along with the support of his family.

What I've seen and been told by the Walburn family for a number of years was what Assert was doing to help and how they help other families in the same situation. The advice and support they give to anyone with this unknown syndrome, is second to none. This for me gave me the drive to decide to run the 2012 London Marathon for Assert.

I had a great trainer as Ken Walburn (Matthew's father) who in the past has run 5 London marathons for Assert, he pushed me and help me to train.

I never knew how hard the training would be but soon it hit me that this was not going to be any easy task. I continued to push forward but seeing Matthew most days and the family advising me as to how

the money would be spent gave me more ambition to run further and faster. This was not the only hard task I found, it was raising the money for Assert because my friends and colleagues I asked to sponsor me had never heard of such charity or syndrome, it showed me how unknown Angelman syndrome is to the public that haven't been affected by it.

As soon I explained more about the syndrome, my own personal link and how Assert offer help to many children and families around the world, it soon opened their eyes and wallets and I'm proud to say I raised over 3,500 pounds.

With all of this in my mind, it helped me complete the London Marathon in 5 hours and 5 minutes, but not without some troubles along the gruelling 26.6 mile course. I had been running at a good pace for the best part of 19 miles (aiming for 4 hours 30 mins) but then I hit the wall (now I know what people were talking about), feeling the an intense pain for at least 3 miles. I had to stop and stretch a few times and at one point didn't feel like I could carry on.

Lucky enough I found the Walburn family along the way and they soon explained why I was doing this and that giving up was no option (thank you Ken), this did push me on and after a mile I found the strength to carry on running suddenly be overwhelmed with an emotion of relief and happiness that I had completed this mountain but also, raised money for an amazing charity which will always be close to my heart.

As I'm writing this, it still makes me feel proud knowing that the little amount I raised could help families understand life doesn't stop but starts for children with Angelman Syndrome with the help of Assert.

Hours after the marathon I said to myself never ever again but, guess what....I have applied again through Assert as that feeling of crossing the finish line is amazing and (apart from getting married) I may not feel this again.





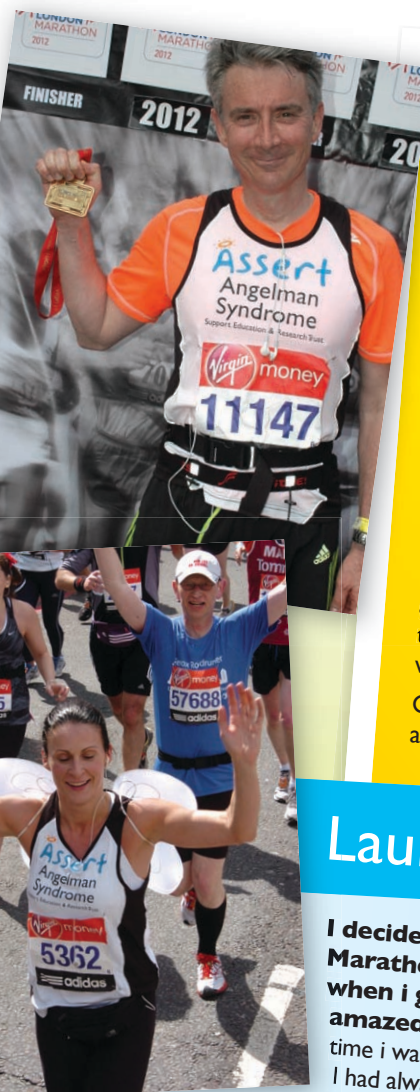
## Duncan Dale

**This was my first and only Marathon.** I did it because a friend at work had done it the year before and was beaten by a man carrying a fridge. Three of us joshed each other along to taking part in 2012 and it became too late to pull out. I thought it would at least be a good opportunity to raise money for ASSERT as my 8 year old daughter, Juliana is an "angel". She is a total delight – always happy and generous with her cuddles.

What I had not realised is that it takes so much time to train. I guess I did 400 miles in all. This took a lot of support from my wife who was looking after our three kids whilst I was off "enjoying myself", jogging around the country side. On a typical run I might see one or two people in the odd car or passing a pub. I caught up on a lot of music listening too. I think the strangest time was running in the dark listening to loud rock music along the country lanes – I had to make sure I stuck on the light grey bits and not the dark black ones. I hope no one heard me singing.

On the race day itself there were 36,000 runners and millions cheering. Everyone was running for something they believed in. There were bands playing and the crowd slapping hands with the runners as they passed by. It was a truly life re-affirming experience. Many people were in fancy dress or in some cases – almost no clothes at all. I was determined to beat a stegosaurus that always seemed to be just ahead of me and kept it up for 15 miles. The last three miles were hell though and it was bloody mindedness that kept me going. I came in at 4 hours and 5 minutes which I was delighted with.

Completing the marathon made me feel a bit of a hero. The real heroes though are the mums and dads who have a little Angel to look after and make it work day after day.



## Laura Fraser

**I decided to run the London Marathon again so I applied and when I got that letter to do it I was amazed.** I had ran it once before but this time I wanted to run for another charity and I had always thought if I get a place I would run for ASSERT, and the reason for this was because my best friend Shelley's son has Angelman Syndrome and I know what a great help the charity has been to Shelley and especially to Louis her little angel.

I started running twice a week and upped it to three times a week. I used to put up face book status to keep me motivated and the cold weather can really make you want to curl up on the sofa but the thought of how hard Shelley has to work every day helping Louis and although I know she must love spending time with Louis there is no doubt its a very hard job and thankfully every second you spend with Louis is amazing as he is a very happy and content boy and he has the most infectious laugh and smile even when he is pulling your hair out your head.

Each run got a little easier and it really helped me feel amazing health wise and also knowing more people would start to know more about ASSERT, as each person I spoke asked all about the charity and awareness for all charities is amazing but for ASSERT I was so happy as I had never seen anyone run for ASSERT before. I'm sure they have but being a small charity its not a wide known as some other charities.

So I ran 3 to 4 times a week for about 3 months and really did get much fitter and loved every second of my training and got to be honest best bit of the marathon training is the 2 weeks of carb loading you have to do which is basically eating lots of potatoes, pasta and rice meals before the race - spaghetti bolognaise, shepherds pie and chicken curry, need I go on mmmmmmm !!!!!

On the day I was really nervous as even if you train really hard

you always worry if you will be ok, and the many many facebook messages I got helped me feel happy and confident about my run. I had my breakfast and made my way to the train station then to the starting line. The atmosphere was amazing every where on the train, at the park everyone was so happy and excited about the day. I chatted to so many people that day all of who wished you good luck, so I'm now 10mins before we are about to start and I need a nervous wee - well the queue was sooo long but I had to go as really did want to hold it in for 26.2 miles, so I waited and then made my way to the start line and put my wings on. I was now ready to go oh now I'm really nervous. We all start to count down 10, 9, 8, 7, 6, 5, 4, 3, 2, go so I'm off.

I kept in contact with friends all the way round. Me and Shelley had agreed we would meet at 25 miles and I just ran and ran I was in good spirits and chatted to so many people on the way round. What a great day it was. I had a really bad stitch about 8 miles in and I had to stop off and get some painkillers which the guy in the shop gave me for free which was so lovely. I continued on and saw a friend at 9 miles in, then missed my friend at 18 miles which was gutting, then missed another friend at 22 miles and by this time even with all the great support you get on the day I really was beginning to hit the wall and need to see a familiar face. I text Shelley to say I was nearly at 25 miles and as I got there I looked and looked and I just couldn't see her. I was so upset and finding it so hard to finish then about 5 mins later I looked up and heard Shelley and saw her and her mum. I ran over and me and Shelley were just crying and she was saying thank you and I was just so happy and relieved to have seen her after missing everyone and especially as I was running for Louis her son. Now I had enough energy to run with my head held high although I did cry the remaining of the marathon but oh how amazing once you see that FINISH line I just gave it my all thinking of Louis and every other person that has AS.

I enjoyed every second of my training and the day but best of all I'm very thankful to having Shelley, Jay and Louis in my life.



## Nigel Flook

**'The London Marathon is an all-consuming event** which has sucked me in and spat me out in 2011, but I was not deterred and thought I won't let it beat me, so entered again! In 2012 I approached it with respect, but also with a view to beating it. It turns out that 2012 sucked me in, but this time I came out smiling having had probably the best 3 hours 43 minutes and 22 seconds of my life! I took the attitude that I would smile and look around this year and really try to enjoy it....and it worked. So my advice for future/potential marathon runners is run the race twice as the first time you will probably be over-awed like me and not enjoy it, but if you come back again and prepare properly then just perhaps it will turn out like my second effort smelling of roses!

To make it even sweeter the raising of the money for ASSERT proved to be relatively easy and successful despite the economy – just ask anyone who came to our Man V Curry Night, a great success, but maybe the curry was just a little too hot!



## Chris Stratford

**I hadn't planned to run the marathon this year** having ran it in 2011 raising money for ASSERT, I remembered how much it hurt!

However, I got "the call" in February from ASSERT's Lisa saying they had a place and it took me exactly 5 seconds to decide and say Yes! I was therefore very much behind with my training and the fitness level of my fellow runners but got on with it.

Once again it was a fantastic day and it was particularly nice doing it in the same year as London hosting the Olympics. I had forgotten how loud the crowds were and took a far more laid back approach to the run than last year to enjoy it as much as possible (hence the slower time). I hadn't forgotten the pain though of the last few miles. I would encourage anybody who has the chance to run the London Marathon to do so, it is one of the best things I have ever done, and if this can mean raising funds for ASSERT then all the better.



## Haidee Kerr

**London 2012 was my 2nd marathon in support of ASSERT.**

The reason I fundraise on behalf of ASSERT is because I've seen first hand the support you provide to those living with AS. I work with Tim, whose little boy William has AS. I remember when he was born, after some months all the tests, the uncertainty of what was wrong and then the life changing diagnosis.

This is where ASSERT came in, somewhere to turn for help and advice, a network of people who could really understand what you are going through.

So this is the 'why', as for the day - it was painful! I went in with an injury and it really hurt after about 10 miles however I managed to finish in my target time of sub 5 hours (just) - I completed it in 4.59.45!

As I came over the finish line, I grabbed a rail to catch my breath and try to avoid collapsing, a guy ran past me, also having just finished. He looked at my running vest in surprise and told me his son has AS... it was an amazing moment. This my friends made every painful mile all worth it, out of the 40,000 people running, I bumped into someone who had hopefully been supported by ASSERT and possibly a little by the money I raised.





# Assert Conference 2012

## official photos

ASSERT are very pleased to announce that the official portrait photos taken at the conference are now available. They are fabulous, so many lovely smiles and happy faces! If anyone would like a copy of their child's photos on a CD then all we ask is for a donation to ASSERT to cover the costs of creating and sending the CD (and a little more if you are able to). Please send a cheque made payable to ASSERT with a note of your name and address to our freepost address. If anyone has any queries, please contact  
**Lisa.Court@angelmanuk.org.**



Participation

needs YOUR

Participation!

I am looking for parents, guardians and/or carers to complete my online questionnaire for my research project titled 'Factors associated with participation of children with Angelman Syndrome, between 6 and 21 years of age'.

This research project aims to explore aspects of children's participation in everyday life (outside of school), and the factors that facilitate or hinder in social participation. My older sister, Amy, has Angelman Syndrome so I have some insight into the issues around 'participation' so wanted to use research to follow up on this.

This project is one of my final year units of the 4 year BSc Speech Sciences degree, which provides a direct pathway into Speech and Language Therapy. Being an AS sibling was a contributing factor in choosing the degree course, and my career as a Speech and Language Therapist.

## Useful Websites & interesting blogs

### For ipad/apps

- [www.autismpluggedin.com](http://www.autismpluggedin.com)
- [atmac.org](http://atmac.org)
- [blog.friendshipcircle.org/2011/02/02/the-special-needs-ipad-app-series](http://blog.friendshipcircle.org/2011/02/02/the-special-needs-ipad-app-series)
- [www.lilliespad.com/special-needs-ipad-blog/tag/special-needs-apps](http://www.lilliespad.com/special-needs-ipad-blog/tag/special-needs-apps)
- [www.techlearning.com/Blogs/37722](http://www.techlearning.com/Blogs/37722)
- [momswithapps.com/apps-for-special-needs/](http://momswithapps.com/apps-for-special-needs/)
- [www.gadgetsdna.com/10-revolutionary-ipad-apps-to-help-autistic-children/5522](http://www.gadgetsdna.com/10-revolutionary-ipad-apps-to-help-autistic-children/5522)
- [www.oneplaceforspecialneeds.com/main/library\\_pick\\_great\\_apps.html](http://www.oneplaceforspecialneeds.com/main/library_pick_great_apps.html)
- [www.ikidapps.com/2010/10/apps-for-children-with-special-needs.html](http://www.ikidapps.com/2010/10/apps-for-children-with-special-needs.html)

### Pre-loved equipment

- [www.disabreg.pwp.blueyonder.co.uk](http://www.disabreg.pwp.blueyonder.co.uk)
- [www.askdes.org.uk/index.html](http://www.askdes.org.uk/index.html)
- [www.disabledliving.co.uk/Home](http://www.disabledliving.co.uk/Home)

### Clothing and swim wear

- [www.incywincy.net](http://www.incywincy.net)
- [www.disabled-clothing.co.uk](http://www.disabled-clothing.co.uk)
- [www.togs4specialsprogs.com](http://www.togs4specialsprogs.com)

### Legal

- [www.ipsea.org.uk](http://www.ipsea.org.uk)
- [www.scope.org.uk](http://www.scope.org.uk)
- [www.mencap.org.uk](http://www.mencap.org.uk)
- [www.challengingbehaviour.org.uk](http://www.challengingbehaviour.org.uk)

This website offers very useful tips and advice, and is run by an AS mum:

- [www.netbuddy.org.uk/newsletter/netbuddy-tools](http://www.netbuddy.org.uk/newsletter/netbuddy-tools)

Your participation in the study just involves completing the online questionnaire or if you would rather complete a printed version please contact us and we will get one sent to you. The study is based solely on the information you provide and it will not be possible to identify you or your child as no names or addresses are required.

### angelmanuk.org/participation

I currently have 23 completed questionnaires but I would be SO grateful and it would be of great benefit to the study if I was to have more!! A big THANK YOU to those that did hand one in over the weekend and thanks in advance to those who complete my questionnaire online. **Keri**

Here's something we think a lot of you might be interested in. They're expensive, but if they're just what you're looking for you might be able to get funding from a local authority or charity to help with the cost. Let Assert know how you get on if you try them.

# Tomcat

Special Kit for Special Kids



USUALLY  
RETAIL AT  
**£99**

## How does it work?

1. Pull the wheel and the heel counter drops



2. The foot (with or without a splint or AFO) can be inserted.



3. Press and twist the wheel and the shoe gathers together again in seconds for a perfect fit on each foot.



## Tomcat Twisters™

Here at Tomcat, we meet lots of families who find it difficult to find suitable footwear for their child, especially if they use splints or AFOs. Some have to buy two separate sizes, or have to struggle every morning to get shoes or boots over splints, which can be painful for everyone involved! Tomcat discovered these special shoes whilst in Italy. We were there showing people our custom built tricycles at the time (which you may know us better for!). When we saw what they could do, we couldn't wait to show our customers and hopefully ease a lot of the footwear problems faced by parents.

Tomcat Twisters™ are specifically designed for splint or AFO wearers and those with arthritis or fine motor problems. Styled as designer sports shoes, Twisters have an innovative lacing wheel behind the heel instead of conventional laces. The lace wire is guaranteed for the life of the shoe.

Tomcat Twisters™ take just seconds to fit and are always snug and supportive whether the foot is in splints or not, because Twisters automatically adjust themselves to conform to the shape of the foot from heel to metatarsal. Because of this unique feature, specially sized shoes are unnecessary.

Orthotists can reshape the Twisters' sole if the foot is pronated or supinated by heating then peeling back the VIBRAM rubber sole and reshaping the EVA midsole by adding or removing material. Orthotic insoles can also be used.

Winter Twisters have all leather uppers in two standard colours, and summer Twisters have leather ventilated and composite panels, also in two standard colours. All shoes are leather lined.

Tomcat Twisters™ are beautifully made by one of Italy's leading orthotic footwear manufacturers and exclusively distributed throughout the UK by Tomcat SNI Ltd. Sizes are available from child sizes 7-13, and adult sizes 1-5, including half sizes and each shoe will self-adjust over four foot widths.

If you have any questions, please do get in touch!

Telephone **01452 616 900** Email: [info@tomcatuk.org](mailto:info@tomcatuk.org)

Or see [www.tomcatuk.org/Tomcat-Twisters](http://www.tomcatuk.org/Tomcat-Twisters) for more information, photographs and an order form.



**We are in the fortunate position of being able to offer 1 pair of these fantastic shoes to the highest bidder!!**

All proceeds of the bidding war will go to ASSERT. Please email ASSERT at [shoes@angelmanuk.org](mailto:shoes@angelmanuk.org) with your name, contact details and your highest bid by 31st December 2012.

The lucky winner will be contacted by email for style and size preference and will be given details of how to transfer your bid to ASSERT's bank account.



## DON'T FORGET

You can also support Assert by buying from our great range of T-shirts and other gifts. You can find our on-line shop through the link on our new website or by going directly to:

[www.angelmanuk.org/spreadshirt](http://www.angelmanuk.org/spreadshirt)



# SOCIAL AND SENSORY REINFORCEMENT IN ANGELMAN SYNDROME

**Researchers at the University of Birmingham are inviting parents/carers of children with Angelman syndrome aged between 2 and 15 years inclusive to take part in study investigating the use of sensory and social reinforcement.**

## WHY?

Previous research has suggested that some children with Angelman syndrome may have a preference for certain sensory and social experiences. For example, some children with Angelman syndrome really enjoy playing with water. The aim of the current study is to assess the use of these preferred experiences as rewards to teach new behaviours.

## WHAT WILL HAPPEN IF YOU/THE PERSON YOU CARE FOR DECIDE TO PARTICIPATE?

Participation in the project will involve the following:

- You will be asked to complete a questionnaire pack and take part in two interviews in order to provide us with background information about your child/person you care for and their behavior.
- We will conduct a day of activities/assessments with your child at their school or your home depending on which is your preferred location.

## HOW DO I TAKE PART?

To be sent an information pack about the study, contact Mary Heald at: [mxo988@bham.ac.uk](mailto:mxo988@bham.ac.uk) or on 0121 414 2855, or Prof Chris Oliver at: [c.oliver@bham.ac.uk](mailto:c.oliver@bham.ac.uk). Alternatively, write to Mary Heald, School of Psychology, University of Birmingham, Edgbaston, Birmingham, B15 2TT.

This study has full ethical approval from the Science, Technology, Engineering & Mathematics Ethical Review Committee at the University of Birmingham.



UNIVERSITY OF  
BIRMINGHAM

# Financial Matters for your Child - Jargon Busting

Dealing with financial matters for your child can be difficult, as you seek to navigate the different rules that apply to bank accounts, benefits and other financial entitlements which may arise from time to time e.g inheritances etc. **Donna Holmes from Anthony Collins Solicitors LLP seeks to assist your understanding by explaining some of the confusing terms used (sometimes interchangeably) in relation to your child's financial affairs.**

## APPOINTEE/APPOINTEESHIP

An Appointee is a person approved by the Department for Work and Pensions to receive state benefits, such as Disability Living Allowance, Income Support etc on behalf of a person who is unable to manage their finances for themselves. An Appointeeship is a relatively straightforward arrangement to put in place. If your child's only income is benefits based and they do not have any savings or other capital assets (e.g. bank accounts, property) in their own name, an Appointeeship should be all that is needed to manage your child's finances.

To apply for Appointeeship, form BF56 will need to be completed and the Appointee may need to attend an interview. The form and further information about the role and responsibilities of an appointee can be found at **[www.direct.gov.uk](http://www.direct.gov.uk)**

## LASTING POWERS OF ATTORNEY (LPAS)

LPAs are documents which authorise a person (an Attorney) to act on behalf another person known as the Donor. These documents can only be made if your child (the Donor) is over 18 years old and has the mental capacity to:

- understand what a power of attorney is and what power would be delegated to the Attorney,
- understand their finances sufficiently to recognise that they may need help; and
- be able to make a decision for themselves that they would like to appoint their chosen Attorney.

An independent and suitable person has to confirm that the Donor has the necessary capacity and the document has to be registered with a Government body called the Office of the Public Guardian before it can be used. This registration process takes approximately 12-14 weeks.

For many children with Angelman's Syndrome, an LPA may not be appropriate but LPAs may be useful documents for you and other members of your family to have in place to enable your chosen Attorney(s) to assist or take over your financial management in the event that you were ill, had an accident or otherwise lost your capacity.

## ENDURING POWERS OF ATTORNEY (EPAS)

EPAs have been replaced by LPAs. However, if an EPA was in place and signed prior to 1 October 2007, it can still be used to manage a Donor's finances. It should be registered as soon as someone loses their mental capacity to enable the Attorney to take over the Donor's financial management.

## COURT OF PROTECTION DEPUTYSHIP

A Court of Protection Deputyship is a Court appointment of someone to manage the property and financial affairs for a person who is unable to manage their affairs for themselves. Such a Court appointment would only arise in circumstances where the person does not have the capacity to make an LPA and appoint an Attorney of their own choice. A Deputy can be appointed for someone under the age of 18.

A Deputyship would be required if your child does not have capacity to manage their finances for themselves and they have capital assets e.g. property or savings and/or receive income over and above any benefits entitlement.

A medical professional has to confirm that your child lacks capacity to manage their finances and the Deputy has to make some promises (officially known as undertakings) to the Court of Protection. The Court of Protection and Office of the Public Guardian oversee the Deputy to make sure that they manage finances appropriately and for the benefit of the person they are appointed for.

**If you have any queries about managing your child's finances or if you would like to discuss whether an LPA is possible or a Deputyship is required, contact Donna on 0121 214 3671.**



## Regional Meeting Splash Landings Hotel at Alton Towers



### Would you like a family party weekend at Alton Towers Splash Landings Hotel?

If so, read on for more details ...A Caribbean themed fun packed family weekend has been arranged for **Friday 1st February 2013 - Sunday 3rd February 2013**. A 2 night bed and breakfast stay in a family room (to sleep up to 4 people) can be booked at the very reduced rate of £200, a large family room (to sleep between 5 and 6 people) can be booked at another very reduced rate of £250. (The usual rate is over £400). This includes entry to the amazing pool and sea life centre, although please note that the theme park itself is **NOT** open at this time. For anyone who has been before, they will know how family orientated this hotel is – the

lifts are like Captain Nemo's submarine, there is a huge pirate ship in the middle of the bar, there is children's entertainment every night, the hotel rooms are decorated for the kids, cartoon characters walk around the hotel ...is that enough to convince you? If you would like to attend then please contact Linda Holmes or Diane Box as you will need a reference in order to book at the discounted rate.

For more details, please contact:

**Linda Holmes**

[lindaholmes65@hotmail.co.uk](mailto:lindaholmes65@hotmail.co.uk) / 07739 363456 or

**Diane Box**

[dianebox9@hotmail.com](mailto:dianebox9@hotmail.com) / 07833 372828.

## Kidz Up North Thursday, 29th November 2012

Premier Exhibition Suite,  
Reebok Stadium, Bolton



One of 3 of the largest, **FREE**, UK exhibitions dedicated to children with disabilities and special needs, their families and the professionals who work with them.

- Over a 100 exhibitors offering advice and information on funding, mobility, seating, beds, communication, access, education, toys, transport, style, sensory, sports and leisure.
- Running alongside the event are **FREE** seminars for parents and professionals.
- New to all our Kidz events - come and visit the Car Zone.



### NEXT EVENTS

- **Kidz in the Middle** 21 March 2013  
Ricoh Arena, Coventry
- **Kidz South** 13 June 2013  
Rivermead Leisure Complex, Reading
- **Kidz Scotland** 11 September 2014  
Royal Highland Exhibition Centre, Edinburgh

For Visitors free entry tickets or more information contact organisers Disabled Living on

0161 607 8200 / [info@disabledliving.co.uk](mailto:info@disabledliving.co.uk)

[www.kidzupnorth.co.uk](http://www.kidzupnorth.co.uk)

 **Disabled Living**  
Practical solutions supporting disabled adults, children & older people

Registered Charity No: 224742

# Our fabulous fundraisers

## Battling the arctic for Angelman charities around the world

### Alistair Blackburn

Back when my son Callum (Del +) was born two great friends (Giles Derry and David Drake) kindly agreed to be his Godfathers. They probably expected a few birthday parties, a baptism and a wedding ahead and an excuse to make sure to keep in touch. Now Callum is nearly eight years old and together with Callum's sister's Godfather (Paul Day) they have also become expert fundraisers in support of Callum and Angelman Syndrome. Every couple of years they come up with some crazy idea as to how to raise money and capture potential donors' sense of adventure and humor to entice them to part with fundraising dollars – and to have fun while doing it!

**Rule 1:** The idea must have a transport "theme" to it

**Rule 2:** Most people will not have done it, or even thought about doing it

**Rule 3:** Each time the event must be more "out there" than the next

Past fundraisers began with a "simple" sponsored bike ride and had progressed to a banger car rally across Europe from France to Croatia in a car purchased for less than \$300, performing tasks along the way.

So for this year's fundraiser, why not go dog-sledding in North Sweden at the arctic circle? And why not make it a bit more hardcore by turning down the option of cabins at night and instead sleep outdoors in open air snow graves and truly experience the extreme cold conditions and winter nights?

Following the amazing success of past events, and with Callum's health more stable than past years, I was convinced to join the three others in Yxsfaftkalen in Sweden in February (I still can't pronounce it properly, but it means Axehandle Mountain). Following some basic instruction we each were shown our team of six Siberian Huskies who were desperate to get going, and off we went for our three day, 200km trip over mountains, across frozen lakes and through forests. Sledding for about three hours between breaks standing on a thin runner required some significant concentration. We each fell off a couple of times on the first morning going too fast around corners, but following that got the hang of it!

Throughout the year we have had a number of fantastic events to raise as much money as possible for ASSERT. Here is just a small selection of the amazing efforts our families and friends have gone to in order to maximise their fundraising. If you would like to arrange a fundraising event on behalf of ASSERT then please contact us to see how we can help. We really do appreciate all the efforts by everyone involved, no matter how big or small the donation.

We were very lucky that the weather was kind to us – sunny and about -8C during the day, and ONLY about -30C at night. Food was luxurious boil-in-the bag high energy food, and lots of water to stay hydrated. At night we each had to dig our snow graves (exactly how they sound – a trench about 3 feet deep in the snow). Whilst very cold, even in about 8 layers of clothes, the nights were so still and serene, and the snow not as uncomfortable as it sounds.

After our first night my sledding colleagues' first comments were about how badly they had slept and asked me how I had slept. "Much better than an average night at home" was my response, having not had to get up and attend to an Angel for a night being a luxury for me! No more complaints from them about their sleeping after that...

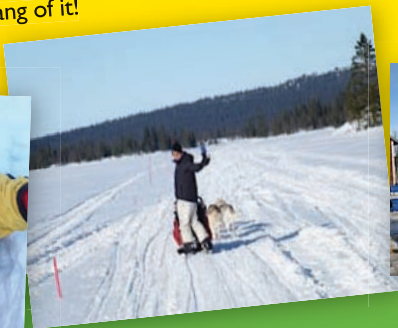
The scenery was simply stunning, we ice-fished for arctic char for dinner, saw almost no sign of human existence during our three days, and were treated to a 90 minute Northern Lights display on our last night while sitting around the campfire after dinner. Something you really can't describe in terms of its scale, beauty and colours. One of our guides has been there for five years and described it as the best display he had ever seen.

A warm shower and a change of clothes were most welcome after three days, and not having to dig for an hour to build your bed was a bonus. It was an unforgettable experience and one where I could catch up with friends and they could get a really good update on Callum and all the exciting activities going on in the Angelman world in terms of research and potential therapies. Plans for the next trip are underway – though this trip will take some beating in terms of creativity and adventure for a bunch of daytime desk-jockeys (three accountants and a lawyer). Any thoughts welcome!

The money was shared between UK, Canadian and US Angelman charities – and I am very pleased to say that ASSERT has received over £6,100 (plus gift aid).

You can see more photos of this adventure at Callum's website: [www.ourangelcallum.com/Site/Photos\\_from\\_Sweden.html](http://www.ourangelcallum.com/Site/Photos_from_Sweden.html)

Alistair and his wife Heidi live with their Angel Callum and his sister, Keira, in Oakville, Ontario. Giles, David and Paul live with their families in the UK - where Alistair is from.







## Firmin Transport Faye Rapley

Firmin Transport are a transport company from Kent whose latest charitable initiative is to link the success of their Pallet distribution service to donations that Firmin will make to a nominated charity. ASSERT was nominated by Faye Rapley, an employee of Firmin Transport, as her 5 year old niece Ruby Brooks has AS.

For every pallet handled by the company, a donation of 10p was contributed and between April 2012 and June 2012 5,837 pallets were handled, therefore £583.70 was donated to ASSERT.

Faye Rapley is donating the cheque to her sister Jane and her partner Jon Brooks with Ruby and her siblings Alfie (8) and Gracie (6 months). As you can see in the photo, the truck has been named after Ruby!

## Charity Cricket Match Kerry Houghton

Our little angel Hannah has captured the hearts of our local community. So when it was mentioned of doing a charity cricket match Hannah came into their heads. They politely asked if we minded and of course we said to fundraise for the charity we are connected to – ASSERT. So planning got underway and Liam (the fundraising organiser) arranged to play against the cricket team where we live - Wedgwood Cricket Club. As they all know Hannah too they accepted. The amount of work these wonderful people put in to make this happen was immense. On Saturday 16th September they held the cricket match. It was Plume of Feathers pub regulars against Wedgwood Cricket Club team. We all rallied around to get donations for a raffle and tom-bola stall. We had a BBQ which the meat was donated from the Plume of Feathers (thank you Mr West) and Wedgwood Cricket Club donated all the condiments and facilities and equipment (thank you Paul Williams). We also had an auction, for a signed Stoke shirt, and power league kids party. All in all we raised £765. There are so many people we would like to thank, but too many to name, so a big thank you to the organiser, our good friends from the Plume of Feathers, and our good friends from the Wedgwood Cricket Club, also for the people who turned out to watch and spend money. It was an amazing day and we are so proud to be a part of this day. Thank you again, Hannah, Kerry, Mike, Connor and Alistair Houghton.



Photo courtesy of Paul Bentley Jones

## Sponsored head shave Kerry Houghton

These are the before and after pictures from the Plume of Feathers pub. They are Liam Rogers, Adam Walklate, and two brothers Nick and Neil Moorhouse. They raised £425. Well done to them and thank you so much. Thank you to all the support given by the locals from the plume of feathers in Barlaston who donated kindly.





## Manchester Marathon Darren Keighley

I started running in 2007, just before my Aunt passed away from cancer. I told her that I would run a marathon for her if it made her better. She never did get better however I had vowed to one day run a marathon in memory of her. Within a few weeks of my Aunts passing I ran my first 10k, the first Jane Tomlinson run for all and from that day forward I have never looked back running a number of races including 10k, 10 miles and half marathons. In May 2011 I ran my first full marathon in Edinburgh raising money for the Jane Tomlinson appeal. The feeling when I crossed the finish line was immense, an absolute buzz! But still the inevitable words crossed my tongue – never again !!!!

In January 2009 my cousin gave birth to a beautiful little girl, however not too long after this happy occasion she was diagnosed with Angelman Syndrome. So within 6 months of vowing to never run a marathon again I found my thoughts turn back to fund raising and myself applying for the Greater Manchester marathon. I wanted to raise both funds and awareness of Angelman Syndrome for my cousin's very special little girl Holly.



Darren raised the excellent sum of £450

## Charity Football Match Christy Welsh



Hi all, we're a new family from Liverpool who wanted to give something back to ASSERT. After we got our diagnosis in September last year I turned to the ASSERT Facebook page where I met Rachael Martin and many other families. During our first few months of diagnosis this page was a great source for both support and advice.

Martin has always played football and has helped one of his friends from the cast of Hollyoaks out in other charity matches so we decided to arrange one of our own. Luckily we have a lot of great friends and family who helped find the ground, sponsors for the kit and anything else we needed. The day itself was very stressful but we had a great time meeting families from as far as Scotland. It was also nice putting real faces to names but the best part for us both was meeting all the amazing angels! We managed to raise £1000 with 10% (£100) going to the Hollyoaks charity. We would just like to say another huge thank you to ASSERT and for me particularly Rachel Martin who is always there with great advice.



# Welcome to Holland

## Part 2



**For those of you that are familiar with 'Welcome to Holland' there is now a follow up which is detailed below. For those of you that are not familiar with 'Welcome to Holland' it is something that is quite often given to new diagnosed families (of any condition) to try and help put into words what they are going through, and how they might be feeling. The original 'Welcome to Holland' by Emily Perl Kingsley can be found at: [www.our-kids.org/Archives/Holland.html](http://www.our-kids.org/Archives/Holland.html)**

I have been in Holland for over a decade now. It has become home. I have had time to catch my breath, to settle and adjust, to accept something different than I'd planned.

I reflect back on those years of past when I had first landed in Holland. I remember clearly my shock, my fear, my anger, the pain and uncertainty.

In those first few years, I tried to get back to Italy as planned, but Holland was where I was to stay.

Today, I can say how far I have come on this unexpected journey. I have learned so much more. But, this too has been a journey of time.

I worked hard. I bought new guidebooks. I learned a new language and I slowly found my way around this new land. I have met others whose plans had changed like mine, and who could share my experience. We supported one another and some have become very special friends.

Some of these fellow travellers had been in Holland longer than I and were seasoned guides, assisting me along the way.

Many have encouraged me. Many have taught me to open my eyes to the wonder and gifts to behold in this new land. I have discovered a community of caring. Holland wasn't so bad.

I think that Holland is used to wayward travellers like me and grew to become a land of hospitality, reaching out to welcome, to assist and to support newcomers like me in this new land. Over the years, I've wondered what life would have been like if I'd landed in Italy as planned. Would life have been easier? Would it have been as rewarding? Would I have learned some of the important lessons I hold today?

Sure, this journey has been more challenging and at times I would (and still do) stomp my feet and cry out in frustration and protest. And, yes, Holland is slower paced than Italy and less flashy than Italy, but this too has been an unexpected gift. I have learned to slow down in ways too and look closer at things, with a new appreciation for the remarkable beauty of Holland with its tulips, windmills and Rembrandts.

I have come to love Holland and call it Home.

I have become a world traveller and discovered that it doesn't matter where you land. What's more important is what you make of your journey and how you see and enjoy the very special, the very lovely, things that Holland, or any land, has to offer.

Yes, over a decade ago I landed in a place I hadn't planned. Yet I am thankful, for this destination has been richer than I could have imagined - Anon

# New Frontiers in Angelman Syndrome Research

Angie Mabb



**There has been a surge in Angelman syndrome (AS) research in the last decade. This surge is most likely due to an increase in the identification of AS, greater awareness of AS through the efforts of private foundations, and the scientific belief that therapeutics could be developed for this syndrome.**

Scientific intrigue has been further escalated due to the complexity of AS and the unique biology underlying the control and regulation of UBE3A, the primary gene that is disrupted in AS. Typically we inherit one gene copy from our mother and another copy from our father, and these genes are expressed equally from both parents. However, in the case for UBE3A, the copy that we inherit from our mother is only expressed and the father's copy is turned off. Most AS cases are caused by disruptions or mutations from the mother's inherited copy of the UBE3A gene. Remarkably, this process has only been observed to take place in neurons (brain cells), and other cell types express both copies of UBE3A. Thus, research efforts have attempted to understand how UBE3A is only expressed from mom's inherited UBE3A copy. Additionally, scientists have also been fascinated with how a loss of UBE3A expression in neurons can lead to some of the core changes in brain circuitry that give rise to the symptoms seen in individuals with AS.

Most recent scientific research has attempted to understand changes in the brain that lead to various AS symptoms. In AS individuals, microcephaly (smaller brain size) is quite common but all of the brain structures look normal and are present. Research from Dr. Elizabeth Wilde's lab at Vanderbilt University and Dr. Senthil

Sundaram's lab at the Children's Hospital of Michigan have suggested that AS individuals have decreased white matter integrity in the brain, which could mean that AS individuals have a misrouting of their neurons, decreased levels of neuronal connectivity, or delayed axon myelination that could lead to disruptions in synapse communication. The decrease in white matter has been hypothesized to result in the developmental delay observed in AS individuals.

Mouse models designed to mimic AS have been incredibly useful tools for scientists to understand AS and to allow for intervention therapies that could benefit humans who have AS. For instance, Dr. Larry Reiter's laboratory at the University of Tennessee Health Science Center has used an AS mouse model to discover that these mice have increased levels of serotonin and dopamine. Some of these changes may reflect the behavioral changes that are observed in individuals who have AS. In regards to seizure susceptibility observed in AS individuals, Dr. Ben Philpot's laboratory from the University of North Carolina has discovered that AS mouse models have an imbalance in excitatory and inhibitory (E/I) circuitry that progresses with age. This E/I imbalance may be the result of seizure susceptibility in AS individuals observed at different ages.



On the therapeutic forefront, scientists have attempted multiple strategies that may one day lead to beneficial treatments for AS individuals. Recent work from Dr. Ed Weeber's group at the University of South Florida has utilized viruses as a way to replace UBE3A back into some neuron populations in AS mouse models. This replacement strategy appears to rescue some of the cognitive anomalies such as learning and memory in these mice, which could equate to learning difficulties observed in AS individuals. For these studies, obtaining global delivery of UBE3A virus to all neurons in the brain is a huge challenge and the potential for using viral therapies to treat rare disorders is far off. However, this work highlights the possibility that restoration of UBE3A in the brain could lead to recovery of AS-related symptoms in humans. Dr. Weeber and Dr. Ype Elgersma's groups have identified molecular pathways, such as  $\alpha$ CaMKII signaling, a pathway that appears to be disrupted in AS mouse models. Crossing these mice with a model that has disrupted  $\alpha$ CaMKII signaling can rescue some of the behavioral changes that are seen in AS model mice. Interestingly, Dr. Orsetta Zuffardi's lab from the University of Pavia Italy found that  $\alpha$ CaMKII controls the dopamine transporter, which could partially explain the disrupted dopamine transporter function observed by Dr. Harald Sitte's group at the Medical University of Vienna.

The finding of disrupted signaling pathways in AS mouse models has led to a human AS clinical trial that will test the antibiotic minocycline, which has been suggested to result in cognitive improvements in AS model mice (although unpublished). Results from this clinical trial should be forthcoming in spring 2013. Dr. Eric Klann's group at New York University found that ErbB inhibitors were able to reverse the deficits in learning and memory

of an AS mouse model suggesting that these inhibitors may also be a potential therapeutic for cognitive anomalies observed in AS. A final approach to ameliorate some of the symptoms related to AS has been based on an attempt to restore UBE3A function by a small molecule inhibitor. A screen in mouse neurons led to the finding that FDA approved topoisomerase inhibitors could turn on dad's inherited copy of UBE3A, which is normally silenced in neurons. However, it is currently unclear if turning on dad's copy of UBE3A could alleviate some of the symptoms of individuals with AS, something that first needs to be explored in AS mouse models.

### So what do these scientific studies and breakthroughs mean for individuals with AS?

1. Work from basic scientific research has allowed us to begin to understand how UBE3A contributes to brain and synaptic physiology. Scientists now have an idea of some of UBE3A's targets and pathways that are afflicted during its absence.
2. We know that the brain circuitry is disrupted in AS and can speculate as to how this disruption leads to certain symptoms observed in individuals with AS. This will allow scientists to continue to rationale molecular targets for therapeutic intervention.
3. Novel scientific findings have allowed for continued research in the AS field and the development of new tools and resources to advance AS-related research.
4. Research results from these studies could one day be beneficial for the treatment and improved quality of life for individuals who have AS.

## HOT OFF THE PRESS

ASSERT now have 5 places for a number of **Bupa 'Great' runs 2013**... we are very pleased to announce that ASSERT have secured 5 places for each of the following runs:

**Great Manchester Run** (May 26th),  
**Great North Run** (September 15th),  
**Great South Run** (October 28th),  
**Great Birmingham Run** (October 20th),  
**Great Edinburgh** (TBC).

We have set the minimum sponsorship at £250. If you would like to apply for one of the places please email: [Lisa.Court@angelmanuk.org](mailto:Lisa.Court@angelmanuk.org) confirming which race you would like to be put forward for. You are welcome to put a second choice in case your first choice is not available, and then we will put you forward for your second choice instead. The cut off date to apply for all races is December 31st 2012.



# DONATIONS

Thank you

Thank you to everyone who has donated to ASSERT.  
Your contributions - no matter how small - all make a difference. Without you there would be no ASSERT and we know how important we are as a support to our families within the A/S community. Many thanks to:

## Assert Calendar

Have you ordered your 2013 calendar yet? Why not buy some calendars as Christmas presents for your family and friends. The calendar is full of amazing photos of our AS individuals and will bring a smile to your face each day looking at all those happy faces.

The calendars are available from Catrina Fraser at [Angelandrewfraser@hotmail.co.uk](mailto:Angelandrewfraser@hotmail.co.uk), or on Catrina's page on Facebook, or using the Ebay link below:

[www.angelmanuk.org/calendars](http://www.angelmanuk.org/calendars)

The calendars cost £10 plus £3 postage.  
The full cost of £10 goes to ASSERT.

Assert

Helpline 0300 999 0102  
[assert@angelmanuk.org](mailto:assert@angelmanuk.org)

Angelman Syndrome Support  
Education and Research Trust  
Freepost, PO Box 4962,  
Nuneaton CV11 9FD

Mrs H M Tait

The Oxtail Group

Mark Emmins

Lloyds Charities Trust

(Haidee Kerr London Marathon 2012)

Gleadless Primary School, Sheffield

Parsons Brinckerhoff

Mrs K Brundle

Mrs G Brown

Mrs Linda Philpott (in memory of Doreen Wroth)

Christine McQuoid (in memory of Doreen Wroth)

Jackie Klimanska (in memory of Dorothy Barton)

Statkraft UK (Duncan Dale London Marathon 2012)

Royal Sussex Chapter 342

Neil and Linda Buchan

In memory of Mrs Vera Hopkinson

Ken and Carole Wall

In memory of Mrs Winifred Parry

St Nicolas Church, Nuneaton

Peter Mullen

Kay Wellbelove

Eveline Knapp

Bernard Marshall

Alan Firmin Ltd

Giles Derry, Paul Day, David Drake & Alistair  
Blackburn (dogsledding in Sweden)

Christy Welsh (Charity Football match)

Darren Keighley (Manchester Marathon)

Stephanie Houghton (Sky Diving)

Luke Munt (Assault Course)

Laura Spink (Duke of Edinburgh Cycle ride)

Finn Curry (Tunbridge Wells Half Marathon)

Catrina Fraser (Glasgow Womens 10k Run)

Ben French (Brighton Marathon)

Jonathan Hooper (Bath Half Marathon)

Haidee Kerr (London Marathon 2012)

Ed Winter (London Marathon 2012)

Nigel Flook (London Marathon 2012)

Chris Stratford (London Marathon 2012)

Duncan Dale (London Marathon 2012)

Laura Fraser (London Marathon 2012)

Chris Rons (London Marathon 2012)