Assert

Newsletter 54 Spring 2014

Angelman Syndrome
Support Education & Research Trust

Latest Research Overview

FAST Chicago 2013 report

Meet ASSERT's new Patron... and GODZILLA!

Sleep Studies latest news

Boy Bites Horse gets a haircut

Regional Meeting round-up

Latest Fundraising

Registered Charity 1021882

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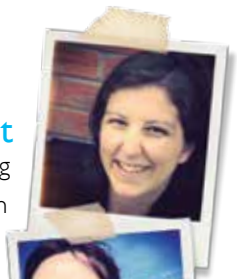

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Wear your charity with pride! T-shirts, polo shirts, bags, hats and lots more from Spreadshirt, our on-line retailer:
www.angelmanuk.org/spreadshirt

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

Conference year is upon us again. The trustee team are busy working on delivering the best weekend yet - and after 2012, the bench mark is very high. If you have not yet secured your place in Coventry, then please do so as soon as possible. Whilst we have never turned anyone away, the size of the group now does now make that eventuality likely at some point in the future. You will have received a letter with details recently. If its got lost in the post or shredded by AS fingers (as if!), then please get in touch and we will get another to you. We really don't want anyone to miss out.

The programme this year is shaping nicely and it will reflect the changes that both the understanding of Angelman Syndrome and ASSERT itself have undergone in the last two years. There has been massive interest in research across the whole scientific world- not just in one country, and our recent decision to join with other European groups to further the EU research is testament to this.

ASSERT, in the past, has been unfairly criticised for its lack of research emphasis. Whilst it is true that research is not, unlike other campaign groups, our raison d'etre, it is an important part of it. Relying totally upon volunteer time and fundraising, our resources are precious. It was felt that until the right opportunity presented itself, we would prioritize our support function. And that we have done- including the now established clinics in conjunction with the NHS. With scientific trustee Katie Cunnea joining the team last year and our recent European group move, active research is again back high on our agenda. And this will be reflected at conference.

Also new to the team is Catrina Fraser, our trustee for Scotland. Having a proven history of fundraising for ASSERT, we're sure she will be able to bring many skills to the team, along with much enthusiasm.

And we now have our new patron- Gareth Edwards. (See the article for more details). Not only is he a rising star in a global industry, unlike many people in such posts, he has seen the work we do first hand and the support that ASSERT is able to offer our families. Its a personal connection and one which we hope will benefit us both.

Looking forward to October 2015, we are planning a special adults only event in Liverpool to commemorate the 50th anniversary of the first diagnosis of Angelman Syndrome. For those that don't know, Dr Angelman was based in St Helens and Liverpool. An internationally driven scientific day followed by a black tie event will provide a contrast to our plans for Coventry. Further details will be circulated once we have them.

Hopefully by the time this is read, the winter storms will be a distant memory and we will all again be able to leave the house without risk. If not, it won't be long. And before you know it, we'll be basking in the summer sun and counting the days to conference.

Rachel & Rich

New Trustee!

I'm Catrina I live in Clydebank just outside Glasgow with my husband Nicholas and our three wonderful boys Andrew 11 - who has Angelman Syndrome deletion - Darren 8 and Jack 2, I would like to introduce myself as the new Scottish Trustee for ASSERT. I look forward to meeting those I haven't as yet and also catching up with those I have; keep an eye out for events, parties, days out and meet ups in our Scottish region. I hope I can be useful to you all. Any questions or need any help or advice you can email me: catrina.fraser@angelmanuk.org.

Siblings Just for Us!

We asked for you to send in your pictures, photos and anything you wanted to tell us, and we had some lovely responses....

Connor Dobbs, age 4 (& 3/4)

Nicole is disabled, she is my sister.
I love her lots but sometimes we fight a bit!
Nicole's favourite thing is balls.
Nicole and me love Tiger our cat, but Nicole sometimes pulls her tail.
It's fun to go places with Nicole - she likes cuddles and she's happy most of the time!
I love being Nicoles brother!

Finlay & Charlie - Siouxi's cousins

Siouxi always brings a smile to my face. Every time I see her she is always smiling. She is smiling and this makes me smile too. No matter what Siouxi smiles all the time. She is always happy. And this is a very good thing. I am going to tell you a story about Siouxi.

Once my family all came round to my house, Siouxi was there (smiling as always) we were eating a delicious Sunday lunch everyone was full. We had had desert and we were just leaving the table.

Then, suddenly Siouxi fell over. Everyone wondered whether she was all right. For a moment, she had a look of shock on her face. She had banged her head on the floor pretty hard. We all thought she was going to cry, but instead of crying, she burst out laughing. We could not believe it. She was laughing after hitting her head really hard. It was amazing and I was always remember it.

This is why I know Sioux's always happy. No matter what, she always brightens the mood.

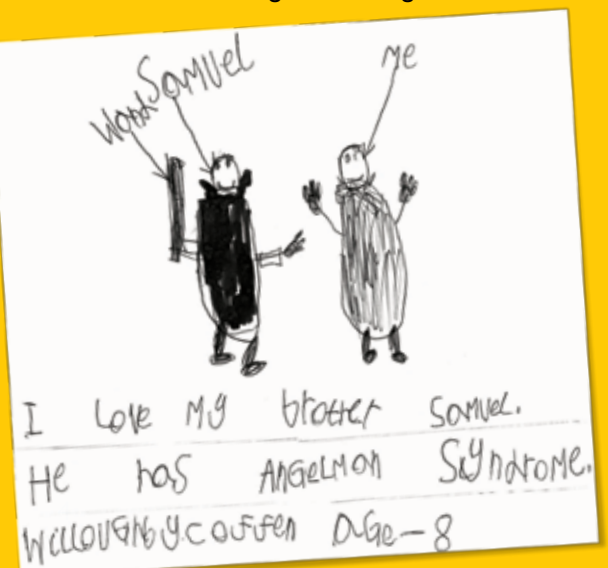
She is the best cousin in the world and every time I see her she is smiling. Siouxi is the best.

Siblings Calendar

As many of you know our 2013 calendar was a massive success! We have decided to make another one for 2015 which will include pictures of our beautiful AS children (young and older) and their siblings – howver, the calendar is open for all, not just those with siblings. The calendar will be available to buy at the conference in August. If you would like your photo in the calendar please send a photo to: catrina.fraser@angelmanuk.org

Willoughby Coffen

Here is Willoughby's picture of himself & Samuel. Samuel is holding one of his glitter wands.



Sophie My Sister

When I had a baby sister, a couple of years later we had some news because they said Sophie had a disability, we were devastated. Sometimes she is a bit of a monkey but she is funny as well but she can bite a bit and pinch but you get used to it LOL!

With my sister being an angel I have got to say she is kind of an angel, a good one. At school sometimes I feel a bit upset because Sophie goes to hospital because she has epilepsy. But I know she gets good care.

Spencer Dunne (9)

Assert Sibling Group

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.

Meet our new Patron

ASSERT are very pleased to announce that we have our very first patron.

Gareth Edwards, the award winning British director and special effects artist, has agreed to take up the post ahead of a very busy few months in the world wide media spotlight; we're sure that there are many more to come.

Gareth, from Nuneaton, has first hand experience of Angelman Syndrome as his niece has the condition. He has also seen the work that ASSERT does first hand having attended both central regional meetings and our national conference with his family in the past.

His most notable release to date is the independent science fiction film *Monsters* (2010)- a film which won him great critical acclaim and widespread media interest.

Having attracted Hollywood attention, he has spent the last couple of years working on the big-budget reworking of the classic *Godzilla* in both Vancouver and Hawaii. The film is set for release in May this year and already the global attention is building. As the premier gets closer, this will only continue to grow.

Gareth has yet to confirm whether the scenes of post apocalyptic destruction seen in the trailer currently being seen around the world are based on his real life experiences. But we are sure that they must have played a small part. How could they not?

We would like to thank him for helping us raise the profile of both ASSERT and Angelman Syndrome; and wish him the best of luck for the next few months.

A note from Lisa Court

I am especially thrilled with the announcement of our new patron – as Gareth is my brother!

ASSERT have been waiting a long time for the right person to come along who can represent the charity and make a difference and I truly believe this is the now case. Over the coming months and years Gareth will be able to use his time in the spotlight to our advantage. Or at the very least send over some good *Godzilla* memorabilia for the conference raffle!

Gareth is busy in LA putting the finishing touches to the film. He has sent the following statement:

"I used to think that being a filmmaker would be the hardest and most rewarding experience anyone could ever have, but it pales in comparison to my sister, who's daughter Ella was diagnosed with Angelman Syndrome some 10 years ago. As Ella's Uncle, I've been fortunate to witness firsthand how this unique condition affects both the families involved and their children, many of whom will never be able to speak or care for themselves.

Yet the unconditional love that they show to those around them is as inspiring as it is rewarding, and it is for this reason that I was very honoured to be asked to be the patron of ASSERT, the UK charity for Angelman syndrome and help bring awareness of this rare condition to the wider public."

This is an old photo of Gareth and Ella but it is one of my favourites. Gareth loves playing with Ella!

Regional Meeting Roundup

ASSERT have continued to arrange a number of regional meetings around the country. This will probably reduce through the year for 2014 because of the focus that is required for the conference – but it will pick up again in 2015. In the mean time, if you would like to arrange a meeting locally for families in your area, please contact ASSERT who can send letters to local families, assist with booking the venue etc. Please contact Linda Holmes for more information.

So in no particular order, here are some details on the recent meetings:

Folly Farm – South Wales – 22nd September

Families in South Wales and neighbouring areas were invited to the inaugural get together in southern Wales in September at Folly Farm in Pembrokeshire. The small get together, organised by trustees Sue and Rich Williams saw the attendees enjoy a subsidised day out at the popular family fun park and zoo. Offering good disabled facilities and a family friendly entry policy, the park welcomed our families and all enjoyed the day. The most popular attraction seemed to be the go-cart circuit which saw quite a few head to head races between families. We even managed to stay dry !

Rich Williams

Hop Farm – Kent – 21st September

We went to hop farm in Kent in the summer. We met up with Claire and Paul and Lucas and Brad Aston and his family and Kitty and her gran. We had a great time meeting old friends and new ones. The angels had great fun going on the rides and meeting each other.

Helen Neilson

Chester Zoo - North West
27th August

On the 27th October several ASSERT families met up for a day at Chester Zoo for our NW regional meeting. Thankfully, despite it being a very wet week the weather held for us and it was the only dry day that week! All the families met up for coffee and for lunch and everyone had a fantastic time. For some families this was their first opportunity to meet up with other families and their children with AS. Despite the 'first time nerves' all said it was a positive experience and were very pleased that they had plucked up the courage to attend. It was lovely to see the siblings and younger ones get together and form friendships that should hopefully last a lifetime. We saw the new baby elephants Hari and Bala, everyone had fun watching the penguins and the giraffes were a big hit, especially with Linda!

Rachel Martin

Christmas Party - Scotland
7th December

Our Scottish Christmas party was held on Saturday 7th December at Bathgate Church Hall. We had a few families come along with their angels and siblings, they enjoyed music, dancing and entertainment from the wonderful Jo Jingles, we danced and sang Christmas songs and played some Christmas games, we were lucky enough to have a visit from Santa Clause and Mrs Clause who brought gifts for everyone. A great day was had by all. I look forward to see you all and many more at our future Scottish meet ups.

Catrina Fraser

Fabulous Fundraisers

Great South Run

I had a fantastic time running for ASSERT on the 2013 Great South Run. I've always enjoyed running and after my brilliant experience I am looking forward to attempting more marathons/half marathons in the future. I chose ASSERT as a friend of mine I have known since school has a young son with the condition. He and his partner have managed superbly and Alistair is one of the happiest children I have ever met. However I know help from family and friends is always welcome and he has spoken highly of the support ASSERT has given his family over the years. I knew then that it would be an excellent cause for me to champion.

The day itself was fantastic, it was very windy (the last 2 miles were like running on the spot!) but the sun was out and the atmosphere was incredible, everybody cheering really helped me in those last few hundred metres power for the finish. I completed the 10 miles in 1 hour 15, which I'm pretty happy with for a first try and am aiming to get down to 1'10 next time I do one. All in all a hugely enjoyable day and has certainly motivated me to attempt more and support the incredible work ASSERT continues to do.

Calum Sanders

Thames Path Challenge

Having just come through another long, dark, wet and windy winter, it's hard to imagine that just six months ago, three friends and I were thankful that the weather wasn't too hot!

It was a beautiful Saturday in August and we were walking across the South Downs in a bid to raise money for ASSERT. My daughter Kitty had been diagnosed with Angelman's syndrome just six months previously in early 2013, and immediately what could have been a very dark period of our lives had been made so much brighter by being welcomed into the Angelman Community, thanks in no small part to ASSERT and the people we met, both online and at one of the regional events.

I immediately knew that I wanted to do something to give something back to ASSERT and do a bit of fundraising. What that should be, I wasn't so sure. I'm not the most fantastic baker, artist or craftsman, so I couldn't make cakes or things to sell. I'm terrible at organising parties and events, so that was out too. But the one thing I love to do, and enjoy doing with Kitty, is going for long walks.

So I phoned my sister and a few friends, and invited them all on a Long Walk for Angelman.

Preparation was easy: I went to the Walk4Life website (www.walk4life.info) to plan our route. They have a handy tool which allows you to plot a route in quite close detail over Ordnance Survey maps, and it will tot it all up to tell you how long it is. Seeking a distance that was a challenge but manageable, I settled on about 20 miles. After a bit of tinkering to find the right footpaths for us (for which I employed a bit of prior local knowledge), I created a nice circular walk around my home town that took us right up onto the downs, and even went past a pub that's famous for its lunches.

I also set up a JustGiving page and promoted it in person and on Facebook so that people could donate.

Before the walk we made sure that we had the right socks, boots that don't cause blisters, and a light waterproof just in case. On the day we also took snacks and a lot of water, and our cameras! I did leave Kitty behind with her grandmother though, as she was only 18 months old at the time.

The day itself was absolutely beautiful – not too hot, and not too cold. No rain, and just a light breeze. Perfect for taking in all the natural beauty that the downs have to offer. We decided to set a steady pace rather than rushing as the steep hills can really take it out of you, so were able to chat, eat ice cream (lots of ice-cream – cornettos became a bit of a theme!), and just enjoy just getting away from day to day life for a change.

In total we took about 11 hours to complete the walk, but we did stop off for a very long pub lunch, and had a few sit downs to enjoy the view along the way. Because the walk was circular we were able to look across the flood plain between the hills and gauge how far we'd come. It turns out that 20 miles is much further than I'd originally thought! Huge telegraph towers that we'd passed under that morning became mere dots on the horizon by late afternoon, whilst hills that had taken ages to climb up barely looked high at all under the huge sky. In total we climbed the equivalent height of Mount Snowden, as our path took us up onto the hilltops and back down into the valley four times.

By the end we were pretty weary, but we all agreed that it had been one of the most enjoyable days we'd had in a long time. Partly because of the sense of achievement, and partly because we'd all had the chance to simply enjoy each other's company without any of the usual distractions of modern life to get in the way. We definitely want to do another walk next year, and I'd encourage anyone who is looking for a fundraising idea to do a walk of this kind. But maybe not as far as 20 miles though – it turns out that that's a really long way!

Donna Edmunds


Calum Sanders


Donna Edmunds


Berenberg Conkers!


Jonathan Wild

Conker Championship

'Will you sponsor me for a run?' I hate getting those emails, it seems to be all the fad these days. But this one was from Jonathan Wild, who is a top lad. And it was for an unusual charity. And it was personal to him. So I thought: 'I can do better than the usual fifty quid sponsorship'. Enter the BBC (not as widely known as its media counterpart, the - first - Berenberg Bonkers for Conkers championship).

This ticked all boxes, in my little mind. I had tons of conkers in the garden so I picked (the easy bit) and personally drilled 200 of them, put them in and on the Aga (on is better than in, I discovered, for the purists out there), bagged them up and took them into work. We got immediate go-ahead from Berenberg management (being a 420 year old private bank, they still like to enjoy life). After we did some initial arm twisting and cajoling, it was like a snowball. We ended up with 50+ competitors. Judging by the 'observers' (really, what is this weird English game?!), next year we will have many more competitors (oh yes, it was very competitive).

So we charged each competitor a tenner, threw in a gopping trophy (the winner has to keep it on her/his desk for a year) and proceeded to have six knock-out rounds over the day (rather conveniently, it culminated in one person from research vs one from sales - research 1-0). Berenberg were also brilliant in donating wine left over from a wine tasting which allowed us to have a silent auction as well as a raffle.

All told, we made about £500 from the BBC (sic), £500 on the raffle and £1700 from the silent auction. So we had great fun, socialised at work, made an awful mess (sorry, cleaners) and raised a fair wad of cash for my chum's charity. Result, as the youngsters say. Injuries were minor, fortunate as a few punters had to duck flying conkers as the string gave way.

Roll on BBC 2014. A big thanks to Hannah and Natalie who were brilliant at making it happen.

Lawson Steele

Great South Run

When I was at school, and dinosaurs roamed Earth, 200 metres or less was my kind of distance. I then started jogging in my late 40's in an effort to find a free and easy way to get fitter and to see more of nature. As time went on I even started to enjoy it. In early 2013 I hadn't thought of entering a race of any kind but then my wife, Sarah, volunteered me for the 10 mile Great South Run to raise money for ASSERT!

We've attended two ASSERT conferences and really appreciate the chance to swap ideas with other parents and keep up to date with the AS issues tackled in the seminars. I also remember ASSERT as being a lifeline when we were floundering around in the early days of getting to grips with what AS could mean for our eldest, Oliver (10) and for us as a family. The idea that any AS parents could make / spare enough personal volunteer 'bandwidth' to keep ASSERT prospering remains pretty mind boggling to me. I hope that the money that the run raised goes a little way to helping ASSERT remain such a valuable resource for all those trying to make the right decisions both for their Angels and for their broader family.

With the help of the Nike+ app (highly recommended!), a head torch and some sturdy shoes I put the miles in around the beautiful Hankley Common area in Surrey (where they filmed the house blowing up in *Skyfall*!) which looks a lot like some sandy Scottish hills.

I didn't know how quickly I could run the 10 miles in Portsmouth, I had no experience of running this distance or as part of a huge crowd so I guessed at 2 hours... a time which put me in the last wave off together with people dressed as Guide Dogs, teams carrying Rocking Horses etc. In all 30,000 took part. There was loads of generous verbal support from onlookers and many of the running tops carried very touching messages. I thought about Oliver a lot as I ran and wanted to do my best for him.

I'd hoped that training on sandy hills and racing on a flat road course would get some speed benefit relative to my training runs. A combination of the extra bounce the road gives and maybe the occasion, speeded me up by more than 10% on the day. I found it a lot easier than I had feared and got around in 1 hour 25 minutes despite having to zigzag through the field a bit.

I had some very generous support for the run from friends and family and we had an unexpected bonus after the run when a friend set up a conker competition and wine auction at his workplace which raised a really healthy additional sum.
To all at ASSERT thank you for your great efforts.

Jonathan Wild

Continued....

More Fabulous Fundraising...

CHESTER MARATHON

Stephanie Thandi participated in her first ever marathon at Chester in October 2013. She clocked a magnificent 3 hours 46 mins and raised a fantastic **£1,039** towards her chosen charity ASSERT. Steph is a lovely friend to the Turner family in Derbyshire. Harry (AS age 19) was there to greet at the finishing line with a great big hug and smile.

Deb Turner

SPONSORED EVENING

A great evening held in Sutton Coldfield raising funds for ASSERT with standup bingo, an auction followed by a disco. Thank you to all who donated prizes and a special thanks to Jan and Paul Mcgrail who hosted and organised the event. A great total of **£1,720** was raised.

Deb Turner

THREE PEAKS CHALLENGE

Five friends of Kerry Houghton took part in the Three Peaks Challenge and raised the fabulous sum of **£939.** The participants were Brendan Brogan, Gareth Best, Lisa Fawcett, Jim Macmillan and Adam Walklate.

GREAT BIRMINGHAM RUN

I am Sukhninderjit ('Pukka') Braitch, my wife is Parmjeet (Pam), our AS son Manvir, daughters Serena, Bethany & Jessica. Manny is now 15 but at 18 months the news he had Angelman Syndrome was devastating. We cried and couldn't believe how this had happened - nobody within our friends or family circle had a disability. ASSERT's Sally Walburn was amazing; also having an AS child she related to our story, comforting and encouraging us with just the right words at the right time. Manny began to crawl at 18 months and walked soon after. His understanding is amazing, he speaks a few words and loves going to church - as a Christian family the prayers of our church and friends helped us - he carries his children's Bible everywhere, along with his iPad!

Manny loves the entertainment of Haven holidays; other favourites are playdough, McDonalds, fish and chips and watching football - going with me to watch Wolves. Our aim from the outset was to give Manny a normal, full and enjoyable life - we've found the 'social' education when we take him out has really helped, he loves to interact, especially with the young women.

Manny has opened so many doors to so many wonderful people and places. Some days can be very challenging but when he smiles it melts away your problems and we feel so blessed.

This year I had the opportunity to run my first ever half marathon in Birmingham on behalf of ASSERT which I loved and it was good to raise awareness and give something back.

Sukhninderjit Braitch

Sleep Interviews

At the Chicago FAST Conference 2013

Two members of our team, Mary Heald and Jayne Trickett, were invited to interview families about sleep at the Foundation for Angelman Syndrome Therapeutics (FAST) conference in Chicago in December 2013. With the help of research students from the Universities in Chicago, Mary and Jayne spoke to over 50 families. Parents were asked questions about their child's sleep, with a focus on the primary issues that families may experience, current strategies used to improve sleep and priorities for the future. We are now in the process of looking at the information collected from parents, and hope that it will guide the next stage of research in sleep interventions for children with Angelman syndrome.

Mary and Jayne took time out from conducting interviews to attend Dr Keith Allen's presentation on the results of a behavioural intervention to reduce sleep problems in five children with Angelman syndrome. Here is a summary of the work that was presented.

Evaluation of a behavioral treatment package to reduce sleep problems in children with Angelman Syndrome

Dr Keith Allen and colleagues from the Nebraska medical centre have trialled a behavioural intervention to reduce sleep problems in children with Angelman syndrome.

This intervention was conducted with five children aged 5-11 years from across the United States experiencing sleep problems, such as unwillingness to go to bed or lying calmly in bed without parental presence, difficulties falling asleep and waking up during the night. Parents completed a sleep diary, challenging behaviour and sleep questionnaires, and children wore watch like devices on their ankles to measure their movement for between two to six weeks prior to the start of the intervention to identify stable scores on these measures prior to the intervention.

The intervention had three components:

1. Improving bedroom environment by ensuring that there was no light or a very low level of light (e.g. by using a nightlight), by removing stimulation such as turning off televisions and music devices, and ensuring the room was a comfortable temperature.

2. Adjusting the children's bedtime to ensure that they were tired when they went to bed. Children's bedtimes were calculated based upon the time at which they usually fell asleep and 30 minutes was added.

3. Helping parents to ignore children's inappropriate bedtime behaviour such as leaving their bedroom, crying and calling out. Parents would enter their child's room when they were calm and quiet, or if the parents were concerned over their child's safety, they were permitted to check on their child after a predetermined interval such as 10 minutes.

Parents were given telephone support by a researcher on a weekly basis.

Findings

- All five children showed an increase to 7 nights a week in the number of nights that they were falling asleep independently without parents in their room.
- The time taken for the children to fall asleep reduced by 5 minutes after the intervention.
- The children were sleep for an average of 30 minutes longer after the intervention.
- Parents also reported a reduction in the frequency disruptive behaviour around bedtime and during the night after the intervention compared with before the intervention.

Conclusion

The study suggests that children with Angelman syndrome may respond to a bedtime behavioural intervention, with the intervention showing particular success in eliminating the children's need for parental presence to fall asleep. It is important to note that the study used a very small number of children and no direct effects were shown for decreased periods of wakefulness at night, which remained constant. Parents indicated that they would recommend the intervention to other families, intended to keep using it.

Reference: Allen, K. D., Kuhn, B. R., DeHaai, K. A., & Wallace, D. P. (2013). Evaluation of a behavioral treatment package to reduce sleep problems in children with Angelman Syndrome. Research in developmental disabilities, 34(1), 676-686.

Angelman Syndrome Support Education and Research Trust

Research Report

On 11th October 2013 I attended the Italian AS international conference (Or.S.A) in Rome. I have written some notes from most of the scientific talks. This is certainly not an exhaustive description of everything that was said. There are also other scientists working on AS who didn't attend the conference, but I learnt a lot, and I have done my best to explain this as simply as possible. Over the next year I hope to gradually gather more and more information from these, and other scientists, so that we can all stay up to date with what is happening in terms of scientific research into Angelman Syndrome.

Katie Cunnea (ASSERT Science & Research Trustee)

UBE3A/E6AP: Not Just an ubiquitin-protein ligase?

Martin Scheffner
http://cms.uni-konstanz.de/scheffner/lab-members/scheffner-lab/prof-dr-martin-scheffner/

Martin is a biochemist based at Universitat Konstanz in Germany. His work focuses on proteins involved in a process called the ubiquitin-conjugation system. The gene that people with Angelman Syndrome are missing normally enables a protein called Ube3A to be made, which we now know is involved in the ubiquitin-conjugation system. This is why Martin's laboratory is interested in Angelman Syndrome.


Martin Scheffner

Ubiquitin is an incredibly important protein and plays a very important role in the body. Most proteins in our body are modified by ubiquitin. A major job of ubiquitin is to signal which proteins should be broken down and removed from the body. It does this by binding (sticking) to them. However, it can have other roles, including changing the activity of the protein, or affecting the proteins ability to interact with other molecules.

There are many proteins involved in the ubiquitin system. In fact there are about 1000 genes involved (4-5%) of human genome. So deregulation of this system results in many different diseases, including some neurological disorders like Angelman Syndrome.

Ube3A

Scientists do not actually know very much about the Ube3A protein. We do know that it is a ubiquitin ligase. This means that it is a protein that recognises other proteins and earmarks these for removal from the body by attaching ubiquitin.

Ube3A is just one ubiquitin ligase, there are many others too. They will all recognise different proteins for alteration/removal from the body. One of the main things we don't know is which proteins Ube3A specifically recognises.

Martin's research group is interested in finding which proteins Ube3A works on.

Interestingly it has been discovered that one molecule that Ube3A recognises is a Human Papilloma Virus molecule, E6. Certain types of human papilloma viruses cause cervical cancer, and it is these viruses that we vaccinate teenage girls against. While this doesn't directly relate to Angelman Syndrome it does mean that in recent years there are more scientists interested in finding out how Ube3A works. So indirectly this could benefit Angelman Syndrome because more scientists have heard of Ube3A and are looking at how it works.

Ube3A and AS

Scientists know that loss of Ube3A results in Angelman syndrome. They also know that too much Ube3A results in some autism spectrum disorders. So it is very important that the amount of Ube3A in the body is very carefully controlled. How the body controls the amount of Ube3A, is one thing that Martin's group is interested in discovering. Alongside this they need to work out the best ways to identify which cells Ube3A is working in.

He is also trying to identify which molecules Ube3A recognises and targets for modification.

Another area of interest is that Ube3A might not be active on its own. It might be inhibited from working or activated to work by other molecules. A protein called HERC2 is one such molecule that has been identified.

The gene for HERC2 is found on the same chromosome,

close to the gene for Ube3A. HERC2 is like Ube3A because it is a ubiquitin ligase. HERC2 is deleted in 70% of patients with AS. It is not known whether this additional deletion of HERC2 has an additional clinical effect on an individual with Angelman Syndrome.

Martin's group looked at whether HERC2 destroys Ube3A or the other way around. They found that neither molecule destroys the other. Instead they believe that when HERC2 interacts with Ube3A it activates Ube3A. (Kühnle et al. J Biol Chem. 2011 Jun 3;286(22):19410-6. doi: 10.1074/jbc.M110.205211).

Evidence for this theory has been provided in a collaborative effort with the group of Andrew Crosby (a human geneticist), London, and published by Harlalka et al, 2013. http://jmg.bmj.com/content/50/2/65.abstract

This scientific paper describes the clinical features of people who lack HERC2 but still have Ube3A. They found that patients have similar characteristics to people with Angelman Syndrome, but in a much milder form. These observations make sense if HERC2 is an activator of Ube3A, because Ube3A is still present in the body, but it is not as active.

Martin stressed that it is important to realise that Ube3A interacts with other proteins. There is likely to be a complex chain of events that result in the different clinical features of AS. If we can understand these processes and all the molecules that are involved it will give us a better insight into how we might correct processes that do not happen properly in people with AS. We might find that it is difficult to replace Ube3A, but easier to correct processes that occur in a later chain of events.

Some molecules that have been proposed to interact with Ube3A include Ring1b and ARC. It is unclear what the role of Ring1b in AS is. ARC is an important molecule for synaptic plasticity. So this could cause problems in synaptic transmission.

Martin has tried to create a set of rules to define how we identify which molecules Ube3A targets for removal from the body's cells.

Martin is not sure that Ube3A interacts directly with ARC and that Ube3A removes ARC by attaching ubiquitin to it. Instead he thinks that Ube3A affects nuclear hormone receptors, and these in turn affect ARC levels. So Ube3A indirectly regulates ARC by regulating the ARC gene.

Summary:

- Ube3A does more than target proteins for degradation.
- There are other factors involved in Ube3A's regulation.

Investigating the impact of rare CNVs on the clinical heterogeneity of Angelman patients carrying deletion.

Silvia Russo

Silvia is a geneticist who works with patients. She is based in the Molecular genetics laboratori, in the Istituto Auzologico Italiano, Milano Italy.

Silvia has studied whether larger deletions in the region of the chromosome that causes Angelman Syndrome, result in patients with more severe disabilities.

She identified two places on DNA where a deletion might begin and called these class I and class II. She then looked at the patients clinical features compared to the amount of DNA deleted.

Silvia noted that a similar thing was described in a paper, Valente 2013. However, she felt that this study was done on too few patients, and so could potentially be misleading.

She looked at 47 patients who were deletion positive. She did not find any significant difference between patients with the larger deletion verses the smaller deletion. However, she did note that patients with a larger deletion did appear to have worse epilepsy, were slower to learn to walk, and more likely to be wheelchair bound.

Silvia also tried to identify any additional genes in the regions that are deleted. One gene that she felt was important was CHRNA7. This is a candidate for causing severe epilepsy in AS (according to their study).

Lessons learned from induced pluripotent stem cell models of Angelman syndrome.

Stormy J. Chamberlain

http://facultydirectory.uchc.edu/profile?profiled =Chamberlain-Stormy

Stormy is in charge of a laboratory at the University of Connecticut.

Her specialty is in the creation of human stem cells. Stem cells are the cells that are first created when a sperm fertilizes an egg. These stem cells then go on to become all sorts of different types of cells. There are a couple of ways you can make human stem cells artificially in the laboratory.

...Continued

One is to take discarded human embryos (from IVF treatment). Stormy does not feel this is something she would want to do from an ethical point of view, and so uses an alternative method.

Instead she takes skin or blood samples from patients and reprograms them to become stem cells. She then uses another technique to turn these stem cells into neurons (this takes about 10 weeks). She thinks the resulting neurons are the equivalent of neurons in a foetus of about 14 weeks. These cells are not designed to be put into humans. They are created so that we can model what happens in human brain cells. This means we have a way of testing chemicals, including potential drugs, on human brain cells.

Angelman Syndrome

Stormy described how the chromosome we inherit from our father is switched off in Angelman Syndrome compared to the one we inherit from our mother.

She has found that the Ube3a antisense RNA associated with AS in humans is a bit different to the equivalent RNA in mice. In humans there is a much longer stretch of Ube3a antisense RNA that stops in a different place than the same RNA in mouse. This stretch of RNA becomes even longer in neurons, and prevents the Ube3A gene from being switched on from the fathers chromosome.

She has grown cells that only contain chromosome 15 from a father, and cells that only contain chromosome 15 from a mother.

Stormy has taken cells from individuals with Angelman Syndrome and using the technique described above, made and grown brain cells in a plastic dish in the laboratory.

The next step is to check that these neurons (brain cells) are working and that they have Angelman Syndrome. This is obviously important to check so that she knows her results are valid.

When she checked the activity of the neurons to make sure they are functional, she found it takes 10 weeks to grow functional brain cells.

Stormy then took the neurons (brain cells) with Angelman Syndrome and tried to work out how the gene from our fathers is switched off.

Comparing DNA from patients with Prada Willi Syndrome and Angelman Syndrome provided some clues.

Her results indicated that the chromosome from our fathers is folded up into a shape that acts as a physical barrier to stop the Ube3a antisense RNA in non-neurons. This Ube3a antisense RNA prevents Ube3a from being produced from the fathers' chromosome in neurons.

Stormy also looked at whether non-brain cells express Ube3A in the same way as brain cells. She found that normally they don't, but if you remove some elements that regulate the gene, you can make non-brain cells silence the father's copy of Ube3A.

Mouse and humans regulate Ube3a slightly differently, so it is important to understand how humans do it so we can understand how well the mouse models Angelman syndrome.

Preventing and reversing synaptic and memory deficits in Angelman syndrome

Eric Klann
http://www.cns.nyu.edu/corefaculty/Klann.php

Eric Klann leads a research group at New York University. He is a molecular neuro-scientist, interested in brain disorders.

He recently returned from a year abroad where he was working in association with a pharmaceutical company with an interest in AS.

To look at the brain and how it is affected by AS, their group worked out how to breed mice with AS. They then wanted to identify molecules that caused the problems in the brain of mice with AS.

They looked at multiple molecules and their potential role in AS. This is quite complex but to summarise some findings:

- They found a drug called PD158780, that can reverse the memory deficits in AS mice. He tried to work out the way that this worked.
- They found some structural differences in the brain cells of mice with AS compared to those that did not have AS.
- They found the amount of certain molecules were altered between the two brains.
- They tried to breed problems out of the AS mouse to prove their hypothesis.
- They think that the synapses are there but not working properly ... so if we can find factors that improve the way synapses work we might be able to help them to work better.

Understanding the molecular mechanisms underlying the neurological deficits of AS

Ype Elgersma
http://beta.neuro.nl/people/y.elgersma.html
http://beta.neuro.nl/research/elgersma/

Neuroscience Institute, Erasmus University, Rotterdam, The Netherlands.

Ype is interested in looking at how we might switch on the Ube3A gene that we inherit from our fathers to correct symptoms of AS.

He also wants to understand the role of Ube3A in the synapse and indentify targets for developing drugs. He is looking at indirect therapeutics. So it might not be easy to replace Ube3A, but this molecule is likely to trigger a chain of events, and we might find drugs that act on molecules later in this chain.

They have discovered some novel proteins that interact with Ube3A. They then looked at whether they were targets for Ube3A? A substrate for ubiquitination?

They did some molecular modelling and saw similar amino acids in two molecules that interact with Ube3A.

Summary

1. His group wants to know if AS is reversible in adults. Is there a critical age, above which we can no longer correct AS?

2. His results have not found anything positive so far, however they are not conclusive. Much more work needs to be done.

3. Activating the Ube3A gene in adult mice does not rescue the selected behavioural deficits that they looked at in mice.

4. Activation of the Ube3A gene at 3 weeks (but this could be equivalent to an older child/teenager in humans) rescues the rotarod deficits. More research is needed to see which behaviours can be rescued at which age.

Pharmacogenetic insights into AS

Ben Philpot
https://www.med.unc.edu/physiolo/faculty/philpot

Ben is a professor at University of North Carolina School of Medicine. He leads a research group which is interested in Angelman Syndrome; Autism Spectrum Disorders; Neurodevelopmental Disorders; Experience-Dependent Synaptic Plasticity; Learning and Memory; and Restoration of Plasticity in Neurological Disorders.

A new angle on AS therapeutics

Deletions or mutations of the maternal UBE3A allele are the main cause of Angelman Syndrome. It is also known that increased genes dosage of UBE3A can be found in common forms of autism. It is therefore clear that it is very important to accurately control UBE3A protein levels. This is obviously important for scientists to consider if they are trying to work out how we might go about replacing missing UBE3A protein in people with AS.

Ben has looked at how the paternal Ube3A allele is switched off, and how this switch can be turned back on. He works with mice that lack maternal Ube3a, and thus model AS, to carry out experiments.

We know that UBE3A protein is expressed throughout the brain and that a deletion on the chromosome we inherit from our mothers stops this happening.

Ben is trying to discover therapeutics for AS. To help achieve this he has formed a collaboration with Mark Zylka and Bryan Roth, both at the University of North Carolina. Mark Zylka is renowned for neurogenetic and molecular biology research, while Bryan Roth is world renowned in the field of drug discovery, as he leads the NIMH Psychoactive Drug Screening Program. This is a very exciting collaboration for both Ben's team and the AS community.

Ben's research team is studying how the paternal Ube3a allele can be switched on.

To test whether the gene is on or off he has taken advantage of a fluorescent reporter mice developed by Scott Dindot while he was in Art Beaudet's lab. This mouse has a florescent tag to see if the Ube3Agene is active. He took neurons that have the fluorescent reporter on the paternal Ube3a allele, and then added drugs to see if they could switch on the gene as indicated by an increase in fluorescence.

His research found one compound that increased Ube3A levels in cells, called irinotecan. This is a drug called a topoisomerase inhibitor, currently used to treat cancer.

So they looked at other similar drugs to see if they could find a drug that would increase levels of protein made from the paternal copy of Ube3A gene. They found that a drug called Topotecan did.

So next they needed to see if it would work in a living animal – e.g. a mouse. So first they injected the drug into the spinal column of mice. They used this method of getting the drug into the body because this is the way that the drug is already administered to children to treat leukaemia. This has been done for many months with few side effects.

He needed to see if Ube3A is produced in the same way in the spinal cord, as it is in the brain.

Then they injected mice with topotecan. When they did this they saw an increase in Ube3A. However, Ben stressed that there are limitations to this research. For example they found smaller amounts of Ube3A than you would expect in a normal cell.

They wanted to find out if the increase in Ube3A was long lasting, or whether you would have to keep giving injections of the drug all the time to keep producing Ube3A? So they injected a group of mice for two weeks, and then compared the levels of Ube3A, immediately, after 4, 12 etc weeks up to 1 year, and compared the mice. They found that even a year after treatment Ube3A was still being produced.

However, they then did the same experiment in cultured cortical neurons and found the production of Ube3A could be initiated but it was lost within about 48 hours. So very different to spinal cord in vivo. The differences in these two findings could be because of the type of cell, but it could also be because one study looked at what was happening inside mice, while the other was on cells grown in a dish outside the body. There are other possible reasons for these differences, but it is possible it is simply different in the spinal column verses brain cells.

They have asked the question...Will topoisomerase inhibitors switch on Ube3A on in human neurons with AS?

So they are now working on patient derived cell lines from Stormy Chamberlain and they have found that you can, indeed, turn on the paternal copy in human genes.

What is the mechanism of turning on the Ube3A gene we inherited from our fathers?

One thing they have realised is that the DNA strand needed to make the Ube3A-antisense, which silences the paternal Ube3a allele, is very long.

Topotecan inhibits expression of long genes, and interestingly many long genes are associated with Autism related genes.

It is a concern for the group, that if they tried to treat AS very early, would they accidently induce autism? So they feel it will be very important to determine when the critical period for treating someone is?

They found that the production of Ube3A induced by Topotecan doesn't happen throughout the brain. So the group is now trying to identify additional toposisomerase inhibitor-like compounds which can penetrate into the brain better.

They are also aware they need to:

- Determine critical periods for treatment of AS.
- Identify the best way to deliver the drugs into the body to increase therapeutic benefits while minimizing potential adverse consequences.
- Determine the mechanism of action of these drugs to guide other attempts to turn the cell on to produce Ube3A.
- Carry out additional unbiased drug screens.
- Work out the right amount of topotecan needed for it to function in an optimal way.

Mark Cushman and Yves Pommier are creating topoisomerases as drugs for treating cancer. So they are looking at topoisomerases that cross the blood-brain barrier better, which of course could be good news for AS too.

Ben told us that he has just signed a contract with a pharmaceutical company to test a new drug. He will be looking at its ability to treat AS, to see if it works better than the topoisomerases he is currently investigating. He is unable to give any specific details because he is bound by a confidentiality agreement. But said he is very excited by the collaboration.

Ben finished by saying that, increased basic science discoveries, increased Ube3A research, a number of therapeutics in the pipeline and engagement of Pharmaceutical companies hold promise for future treatments.

2014 Conference and other new T-shirts available now!

You can now order ASSERT's new 2014 Conference T-shirt via our partner merchandising site, Spreadshirt. Due to popular demand we're introducing a whole range of *'I'm No Angel'* items to tie in with the conference. The cost and final design for the conference shirt was still to be confirmed at the time of writing but you can check it out along with all the sizes and colours - and all of our other fantastic designs - by going directly to the Spreadshirt site:

www.angelmanuk.org/spreadshirt

If you just can't wait (and who could blame you?) and want to get yours in time for Summer you can order directly from Spreadshirt. Or if you'd like to make a saving on postage and collect your T-shirt from the conference in August you can order through ASSERT by taking a look at all the order details on Spreadshirt's site, choosing your T-shirt(s) and sending a cheque (made payable to ASSERT) to our freepost address. Please make sure that you write on the back your conference booking name, address and order. The deadline for ordering through us is 31st July.

Assert

Don't Forget!

You can support ASSERT by buying from our great new range of 'I'm No Angel' T-shirts and other gifts, alongside our traditional designs.

We also sell shirts without the conference lettering

You can find our online shop through the link on our website or by going directly to:

www.angelmanuk.org/spreadshirt

Useful Websites & Interesting Blogs

Support

- www.angelman.ie
- www.disabledliving.co.uk/Kidz/North
- www.cafamily.org.uk
- www.mencap.org.uk
- www.autism.org.uk
- (Princess Royal Trust for Carers) www.carers.org
- (Transition) www.transitioninfonetwork.org.uk
- www.challengingbehaviour.org.uk
- www.learningdisabilities.org.uk

For iPad/apps

- www.autismpluggedin.com
- www.atmac.org
- www.blog.friendshipcircle.org/2011/02/02/the-special-needs-ipad-app-series
- www.lilliespad.com/special-needs-ipad-blog/tag/special-needs-apps
- www.techlearning.com/Blogs/37722
- www.momswithapps.com/apps-for-special-needs
- www.gadgetsdna.com/10-revolutionary-ipad-apps-to-help-autistic-children/5522
- www.oneplaceforspecialneeds.com/main/library_pick_great_apps.html
- www.ikidapps.com/2010/10/apps-for-children-with-special-needs.html

Blogs

- www.boybiteshorse.co.uk

Pre-loved equipment

- www.disabreg.pwp.blueyonder.co.uk
- www.askdes.org.uk
- www.disabledliving.co.uk

Clothing and other equipment

- www.fledglings.org.uk
- www.clothingsolutions.org.uk
- www.incywincy.net
- www.disabled-clothing.co.uk
- www.togs4specialsprogs.com

Legal

- www.ipsea.org.uk
- www.scope.org.uk
- www.mencap.org.uk
- 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

Struggling to get the equipment a disabled child needs?

Almost one hundred items of specialist equipment are now ready and waiting to be delivered to children in need, across the UK.

Newlife's pilot project called 'Newlifeable' launches on Monday 20th January 2014 offering a large selection of new and clinically refurbished and certified equipment, all available through a grant for 'priority delivery' at no cost.

Equipment includes specialist seating, manual wheelchairs, buggies, walking frames and assisted living aids like height adjustable tables.

To view the current range of equipment and download the FAQ's on the project, please visit www.newlifeable.co.uk. If you cannot access the internet to view the range and know the name or type of equipment you are seeking, please call us.

We welcome contact direct from families. We also want to hear from professionals if they know a child in need of an item (that they would probably not be able to get through the local statutory services).

Newlife can deliver anywhere in the UK, including Scotland, Wales and NI. We also have a small budget attached to each item that can be used to make an adaptation to the item, to make it exactly right for it to fully meet the child's needs.

Newlife is committed to ensuring that children and young adults with disabilities, life limiting conditions and terminal illness are able to get 'the right equipment at the right time'.

Stephen Morgan, Newlife's Head of Operations says "In addition to giving our year round grants and loans of vital equipment, through our Nurse led services, this pool of equipment is ready and waiting to be dispatched. There are no long forms, we only need to know some basic information and have the support of a professional (who can agree the specification of items), and we will dispatch straight away"

"We suspect these items will be very popular, so we would advise families and professionals to contact us as soon as possible to register interest in an item"

To register your interest in any of these items, just call Nicole Norris, on 01543 431465 Monday to Wednesday from 9.30 – 5pm. Please note an answer phone service will be in operation on Thursdays and Fridays.

Cerebra Centre for Neurodevelopmental Disorders

Over the past three years, researchers at the Cerebra Centre for Neurodevelopmental Disorders have been conducting research examining preference and reinforcement in Angelman syndrome. Two projects, led by Prof Chris Oliver, Dr Dawn Adams and Mary Heald, were designed to further explore the reported heightened sociability and preference for certain sensory experiences often associated with Angelman syndrome. This article is a summary of the main research findings from both studies.

Study 1: Preference and reinforcement in Angelman syndrome

Background

Previous research has suggested that some children with Angelman syndrome may find learning new behaviours difficult or challenging. It is thought that this may be related to a loss of the UBE3A gene implicated in Angelman syndrome (Jiang et al., 1998).

Often, items that children prefer or enjoy (e.g. a favourite toy) can be used as effective rewards to help teach children new behaviours and increase their speed of learning. Several behaviours frequently described in parental report and the current literature suggest that children with Angelman syndrome may find both social interaction and sensory experiences extremely enjoyable. For example:

- Children with Angelman syndrome laugh and smile more in the presence of adult social interaction (Oliver et al., 2002) and compared to children without Angelman syndrome (Oliver et al., 2007).
- Some children show frequent social approach behaviours towards both familiar and unfamiliar adults (Mount et al., 2011).
- Children are often reported to have a 'fascination' with certain sensory stimuli, including water and shiny/reflective objects (Didden et al., 2006).

Together, the literature suggests that social interaction and sensory experiences may function as highly effective reinforcers to increase children's speed of learning.

Aims

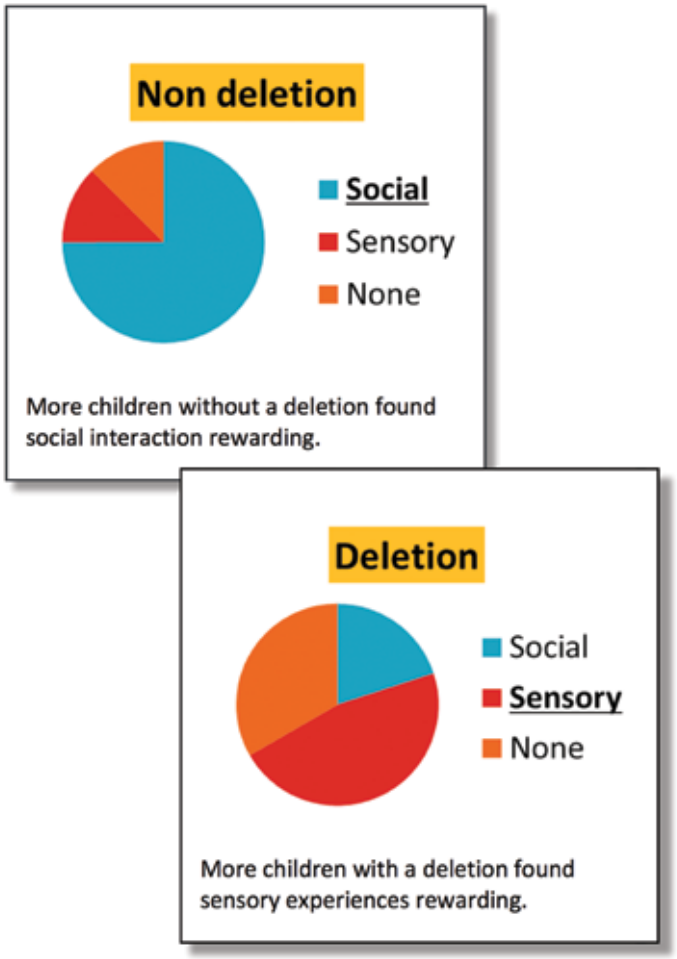
The main aim of the study was to examine the use of social interaction and sensory toys as rewards to increase children's speed of learning.

What did we do?

- We visited 22 children with Angelman syndrome at their school or home (14 children with a deletion, 7 children with Angelman syndrome caused by other genetic mechanisms).
- We asked children to complete a simple task e.g. 'can you put the block in the bucket'. If children completed the task, they were given a reward.
- We alternated the type of reward (social/sensory/no reward), to see if this would affect the number of times children completed the task.

What did we find?

- All children completed the task more often in the presence of a social reward.
- Social and sensory rewards were equally rewarding across all children.
- There were striking differences in the type of reward children found reinforcing when we compared across children with a deletion, compared to children without a deletion:



What does this mean?

- The use of social and sensory experiences as rewards with children with Angelman syndrome may increase children's speed of learning.
- The type of reward given may needs to be **targeted for each child;** some rewards resulted in almost zero levels of task completions for some children.
- Children with Angelman syndrome not cause by a deletion (UBE3A mutation, Imprinting Centre Defect, UPD) may show **increased motivation for adult social interaction.**

Update on Research

Study 2: Teaching children when to approach for attention

Background

Both parental reports and research conducted within our team suggests that some children with Angelman syndrome find adult social interaction extremely enjoyable, and will often approach both familiar and unfamiliar adults to gain access to attention.

Parents have reported that sometimes children will approach for attention even when it is not available e.g. if a parent is busy doing something else. This can be frustrating for both the parent and the child.

Aims

The main aim of the study was to teach children to discriminate between times of adult availability. We used a bright orange jacket as a signal to children that attention was not available.

What did we do?

Four children with Angelman syndrome were visited at their school by Mary. Each child was visited for three days.

In order to teach children to associate the orange jacket with times when attention was not available, Mary alternated her responses to children's social approaches:

What did we find?

After three days of training, all four children began to show lower rates of social approach behaviours when Mary wore the coat.

What does this mean?

- The results suggest that all four children were beginning to learn that if the researcher wore the coat attention was not available.
- This means that using a cue in this way might be a good way to make the environment more predictable for children.

Reference for full text

Heald, M., Allen, D., Allen, D., & Oliver, C. (2013). Discrimination training reduces high rate social approach behaviors in Angelman syndrome: Proof of principle. Research in Developmental Disabilities, 34, 1794-1803.

FAST Gala Chicago 2013

In December 2013 I represented ASSERT at the annual FAST Gala in Chicago, as we were kindly offered a ticket by Paula Evans. It really was a flying visit as I flew in on the Friday evening just as the Gala started, attended the seminars on the Saturday with the various scientists and flew home on the Sunday. Although very tiring, it was a fabulous experience meeting other AS parents and listening to the scientists explain what they had found so far and what they were hoping to achieve. I also took part in the sleep study with Mary Heald. I was very pleased to see a few familiar faces, as well as meeting some new mums from the UK that had travelled over to Chicago.

The review of the weekend has been written by Claire Lerman who I met for the first time in Chicago. Claire sums it up perfectly, so there isn't really anything else for me to say except to add that these are exciting times ahead.

Lisa Court

It was with a mixture of excitement & trepidation that I boarded a plane to Chicago on the 5th December. My daughter was only diagnosed with AS this summer when she was 16months old & the night she was diagnosed I had 'googled' a lot & during the process I stumbled across an organisation called FAST who were actively researching AS & I don't mind admitting it gave me a lot of comfort at a scary time.

I quickly realised over the following months that opinions on FAST were divided so I was anxious to go to the Gala & try to get a feel for myself as to what it was all about. Upon arrival I had an interview for a 'sleep study' & I was comforted to be greeted by 2 ladies who told me they had travelled from the UK (Birmingham). I was pleasantly surprised to learn that they worked on AS research in respect of sleep/challenging behaviours as I hadn't realised there were people actively doing research back home in the UK.

During my first evening there I met some other parents including a couple with a daughter of a similar age to mine & who looked just like her (I have since realised that Eva has many looky-likeys in the AS world!) I was stunned when I was telling them about Eva's standing frame & they had never heard of one. I quickly realised that even though the US may seem ahead of us in many ways, that parents don't actually get equipment & services the way that we do. I went to bed that night feeling sad for this little girl with her lack of equipment & also feeling very grateful for the NHS!

The following day I attended a literacy seminar by a lady called Erin Sheldon. This was unbelievably moving as there was absolutely no doubt in this speakers mind that all our Angels have the ability to be able to be able to read & write. She was very passionate about the fact that we must always presume competence with our children & I left the seminar vowing that I always would even if I am still trying to help Eva learn to read when she is 30yrs old then that's fine by me as I will never give up hope on what she might achieve.

The Gala itself was a glamorous affair & it was great to have a little chat with Colin Farrell in the flesh! He asked me lots of questions about Eva & told me a little about his son James. It was

wonderful to meet lots of parents in the same position as me & I also met lots of people with Angelman Syndrome from babies through to adults. One of the most moving moments of the entire trip for me was chatting to a mother who told me that her daughter was part of the Minocycline trial. She was passionate that she felt her daughter had progressed in all areas at a very rapid rate since taking the drug. Naturally, this excited me & like many others I am eager to hear how future trials will turn out.

The day after the Gala I attended the scientific round table seminar where different scientists gave presentations on the work they had been doing recently in the field of Angelman Syndrome. It was extremely informative/exciting to hear of the progress which has been made & the scientists were open to questions at the end of the presentations. It was admittedly quite difficult to keep up with the jargon at times but Dr Jason Yi did share the amazing news that they have found an on/off switch on the UBE3A protein itself (as opposed to in the gene) & the mechanism that turns the proteins on/off.

All in all I am really glad that I had the opportunity to go to Chicago & it is always uplifting for me to hear of the scientific advances of AS. Of course FAST are not the only organisation in the world that are working hard in this field and its wonderful for me as Eva's mum to know that there are lots of things being done by some amazingly talented people that may one day be of benefit to my gorgeous girl.

Claire Lerman

New study: Quality of sleep in children with Angelman syndrome and their parents

The Cerebra Centre for Neurodevelopmental Disorders
UNIVERSITY OF BIRMINGHAM

We would like to invite all parents of children with Angelman syndrome (whether your child has a sleep problem or not) to complete an online questionnaire about sleep, health and behaviour. The questionnaire will also ask you about your own sleep and functioning during the day.

This study forms part of a larger study comparing sleep quality and the nature of any sleep problems across children with different genetic syndromes.

Please visit this link for the information sheet and the consent form: http://tinyurl.com/pt2enjs
The password is **cerebra**

If the above tiny URL does not work, please enter the full link
https://lesweb2.bham.ac.uk/surveys/index.php?r=survey/index/sid/293485/lang/en

If you require any additional information, please contact:
Jayne Trickett : jxt292@bham.ac.uk or 0121 414 2855

BOY BITES HORSE

NOTES FROM THE FRONTLINE OF AN ALTERNATIVE CHILDHOOD

Some of you might already be familiar with Boy Bites Horse, *an entertaining blog that reports the life of a (so-caled) Special Needs child. For those that aren't, sit back, buckle-up and enjoy the ride.*

More at: www.boybiteshorse.co.uk

Haircut

My hair used to be a lovely light blond colour, which with my blue eyes gave me the look of a young Steve McQueen. As I grew, my hair darkened a little and people remarked that I had the look of a young Leonardo DiCaprio… presumably before he got fat. Lately I've been trying to project Daniel Craig, but people have stopped making comparisons so I can only assume that I look like me.

My hair grows thick and fast: luckily I seem to have more genes from Frowny than Smiley in this respect as his hair is getting thinner by the day and the very top part is disappearing altogether. Perhaps he has ambitions to be a monk. Curly Top – as his name suggests – also has a lovely crop of thick hair, which I often admire by yanking a tuft of it out. Sometimes there are tiny pieces of scalp still attached.

In my experience hairdressers are a pretty mixed bag. Those that have been lucky enough to work with me are often very young girls of a nervous disposition. They'll start by tying a smock around my neck which, like most new things, I like to study in very close detail. If it's of a plastic or rubber texture it's usually worth a taste and I'll follow this up with a test of the tensile strength with my teeth.

Bib abandoned – Boo! – she'll start snipping away. Now I don't think it's very polite for people to start hacking away at me unless I've had a chance to examine the tools they're using, so at this point I think it's perfectly reasonable for me to grab the blades as they're passing for a closer look. And when I say closer look I like to hold them very near to my eyes.

I ***always*** do this.

Smiley always forewarns them that I ***will*** do this.

And yet they're always surprised, sometimes traumatised when I do it.

Some girls have been known to give up at this point. One poor girl was reduced to tears – I really don't think she had the temperament for hairdressing. This has become a problem: as long as the hair is out of my eyes I don't particularly mind how I look but Flower Girl keeps telling me that it's social suicide to go about with half a haircut. Sometimes a more senior hairdresser will appear and make the best of what's been started and we make a hasty exit. Sometimes it's made plain that we aren't going to be welcomed in that establishment again and we have to find another hairdresser. I've been to a lot of hairdressers.

Aside from the need to inspect the equipment I'm afraid I also suffer with a short attention span, and the need to sit still for 10-15 minutes while someone minces around behind me with a pair of scissors is so *Booooooooorrrrrrrriiiiiing…*

I try to amuse myself with a look around the shop but Smiley and the hairdresser seem to think this is a bad idea and try to get me to look at my own reflection. I can appreciate other people's high regard for my appearance but I really don't feel I can stare at myself for that length of time without appearing vain. So I'll look down at the hair falling in my lap or on the floor, or if there's a sink I'll sit as far forward as possible to see if any water has appeared. Sometimes I'll try to slide down in my seat to the floor, or tilt my head back as far as I can and take a look up the hairdresser's nose. Mmm!

Over the years Smiley has developed a lot of techniques to entertain me. These usually involve hand-held mirrors (easily dropped), water sprays (which make me jump, with unfortunate consequences to the haircut) or toys which he'll give me, then immediately remove when he realises the hair is sticking to them. If it's a particularly dull session I'll take a handful of hair clippings and give them a quick chew, though I'll admit this is probably an acquired taste and not for everyone.

For the last couple of years we've been going to a big, smiley Northern man who tells rude jokes and waves at me every time I pass his shop. This is about as far removed from the cry-baby little girl hairdressers as it's possible to

get; although jovial and entertaining, he doesn't stand for any nonsense. He gives me all the tools to examine before we start and he lets me sit in my own chair and do my own thing: as I look one way he swoops in and cuts the back. As I look up he swoops in and snips the front. He even lets me play with the buzzy clippers that he uses on the back and the sides (though last time I tried them on my tongue so we might not be doing that again). He is always happy when he cuts my hair – though he does sometimes work up a sweat – and I've never left with half a haircut.

He's a professional, and I admire that.

Respec'.

About The Boy Who Bit The Horse

I am a fifteen year old boy with so-called special needs. Primarily those are: I need to eat (a lot), I like plenty of exercise or I get really bored, I grunt instead of talking, I'm not much of a reader, I'm not too big on personal hygiene, I don't like being too hot or too cold and I like playing with my… well… y'know… so I'm pretty much like every other fifteen year old boy.

As I look around me I see there are lots of ways in which I can help people with the benefit of my autobiographical anecdotes. On the whole I think everyone works too hard and worries too much; in my experience of putting in as little effort as possible I still find that people bring me food at regular intervals, provide all the clothes I need, take me wherever I need to go and generally go out of their way to indulge me.

So I dedicate this blog to all those of you who are trying too hard in life.

Slow down. Sit Back. Relax.

Knowledge Exchange Project Update: The build has begun!

In October 2013 we started to build online accessible resources for families and professionals. The aim of this project was to make sure that reliable and accurate research findings were available quickly and in a useful format. You or a member of your family may have taken part in a discussion group about this project where we asked families what they wanted from these resources. We have now finalised our draft design for the website and have now begun building the site. The website is coming to life not only with written information that we are uploading but through the videos, stories and photographs that families have contributed – we could not have done this without your support! Over the next six months we be getting back in touch with families to find out what they think of the information and we will be extending the site to provide assessment measures for professionals.

If you would like to get involved with the project email us at: **buildwithbham@gmail.com**

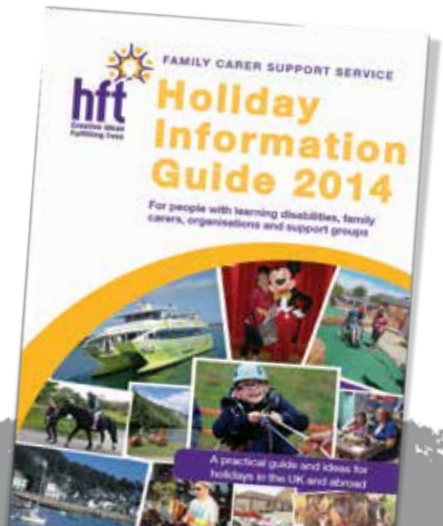
Holiday Information Guide

The 40-page Holiday Information Guide has been put together for people with learning disabilities, family carers, organisations and support groups, and includes:

- Where to go for useful advice, holiday guides and information
- Details about tour operators
- Holiday providers in the UK and abroad
- Organisations which provide benefits, help and insurance

Don't wait until the last minute - take a look now at the wide selection of activities on offer, and the wonderful places to visit.

www.hft.org.uk/holidayinformationguide



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 all know how important we are as a support to our families within the AS community.

many thanks to...

BUPA Great Runs

ASSERT have places available in a number of the Bupa 'Great' Runs for 2014. If you would like to be considered for a place, please email: lisa.court@angelmanuk.org.

The minimum sponsorship is £250 for all of the events.

The races we have places for are:

- Great North Run
- Great South Run
- Great Manchester Run
- Great Birmingham Run

Assert

Helpline 0300 999 0102
assert@angelmanuk.org

Angelman Syndrome Support Education and Research Trust
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