Newsletter 47 Conference Edition Autumn 2010

Assert

Angelman Syndrome Support Education & Research Trust

Useful Stuff

Freepost Address
ASSERT
PO Box 4962
Nuneaton
CV11 9FD

Website
www.angelmanuk.org

Email
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. http://458768.spreadshirt.co.uk/

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 Trustees

Every so often, and if I am honest it is becoming increasingly often, I ask myself when things will settle down. I suppose like most, albeit naively, when Claire and I got married all those years ago, we dreamed of a nice hassle free life where everything ran smoothly. They, whoever they are, never told us that things rarely run smoothly. But that was before Angelman Syndrome was added to the equation, and now I am sure that it is Ruth's intention to ensure that we have as busy a life as possible with never a dull moment as we move from crisis to crisis. That is why for us, once again at the conference up at Loughborough, it was yet again another great time meeting people whose lives resembled ours, if not in exact detail, but at least in the fact that things with an AS individual can never be described as boring. I hope that those of you who attended enjoyed it as much as we did, and found it as beneficial.

So now the conference seems an age away, although thankfully as trustees we haven't quite started planning the next one just yet! However the trustees work is never done, and we now have a number of projects that we must embark on. Probably one of our most important is to sort out our internet presence, both the web site and our email. The latter of these has, if I am honest, recently been a disaster, not least of which compounded by a total lack of support from our Internet Service Provider. We are in the process of changing, so hopefully in the next month or so when you send us an email it will no longer disappear into a black hole but will be answered in a timely manner. And for the trustees, we will no longer have to spend hours searching in vain for legitimate emails amongst all the spam.

Ruth, now 13, is just on the edge of transition, bringing us into a new realm of excitement and discovery. We rather hit it with a bump when the first form we received asked questions such as "Do you want your child to continue in full time education", and expected a Yes/No answer – where is the "Don't know" option, or "What aspirations do you have for your child in the future?". I just managed to restrain myself from putting down astronaut.

For us, October half term is always family holiday time, so we are off to Newquay again to visit our friends the Way's at Hendra Holiday Park. For those who have never been to Hendra, I would thoroughly recommend it, not least because of the welcome any ASSERT members get when they visit. It has a great family atmosphere, and a pool which all of us love, evening entertainment which the kids love and we love a little less although a couple of beers helps to numb the pain! We are also, during our visit, going to the Eden Project which is nearby. Each year, around this time of year, they have an ice rink set up which we have a very amateur attempt on. Just to make things more interesting, or is it because we are mad, we are taking Ruth on the ice this year in her wheelchair! Well we saw someone do it last time and thought why not. I am sure by the time we have finished we will have found lots of reasons why not, like all the complaints from other people as Ruth decides she wants to say hello to them by grabbing hold of them. Let's just hope her smile melts their anger. Watch this space for the results.

Hopefully in the new year we will be embarking on a number of regional meetings around the country. We will update you with the details in the near future, hopefully on our new, all singing, all dancing web site!

Jeremy Webb

Registered Charity 1021882

Useful Websites

- Challenging Behaviour Foundation
  www.thecbf.org.uk
- KidsOut
  www.kidsout.org.uk
- Orthotic Products
  www.gilbert-melish.co.uk
- Special Educational Needs
  www.senmagazine.co.uk
- MumsNet Special Needs
  www.mumsnet.com/Talk/special_needs
- Talking Point (speech & language)
  www.talkingpoint.org.uk
- Harnesses
  www.childharness.ca/index.html
- Safespace Beds
  www.safespaces.co.uk
- Intensive Interaction (communication)
  www.intensiveinteraction.co.uk/
- Baby Monitors
  www.babymonitorsdirect.co.uk
- Makaton
  www.makaton.org
- Family Fund
  www.familyfund.org.uk
- Contact a Family
  www.caffamily.org.uk
- Toys for Disabled Children
  www.tfh.com

every click

www.everyclick.com
is a search engine that allows you to raise funds for charities for free through searches, online shopping and donations.

Your Assert Trustees

Jeremy Webb
Chair and East Region

Rich Williams
Fundraising, Merchandise and Welsh Region

Sue Williams
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Lisa Court
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South East Region

Rachel Martin
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Chris Martin
Welsh Region

Neil Buchan
Scotland

Christyan Fox
South East Region

Russell Andrews
Central Region

Rosemary Teggin
(non-trustee)
N Ireland & Eire Representative

Kate Pickering has stepped down as a trustee of ASSERT. We would like to thank Kate for her dedication and hard work during her time as a trustee of ASSERT.

London Marathon 2011

Following the brilliant performances by all of our runners in the 2010 London Marathon, we can now confirm that we have places available for next year.

If you'd like to run in the 2011 London Marathon on Sunday April 17th and can commit to raising at least £1000, we need to know by November 30th 2010

We will then draw the lucky runners and notify them by December 7th 2010

Please complete the form below and return it to:

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

Assert

Angelman Syndrome Support Education & Research Trust

I am completely nuts and would love to put my body through 26 miles of torture in order to raise at least £1000 for ASSERT
Please enter me in the draw for one of those lucky places.

Name _____

Address _____

_____

_____

Postcode _____

Email _____@_____

Phone _____

Jumping For Joy!

A quick note about my sponsored sky dive which was held at Glenrothes in Scotland in October last year with the twin aims of fundraising for ASSERT and helping to raise money to take my son Kieran swimming with the dolphins in Florida.

While skydiving was the most thrilling experience I've ever had in my life it was also one of the scariest!

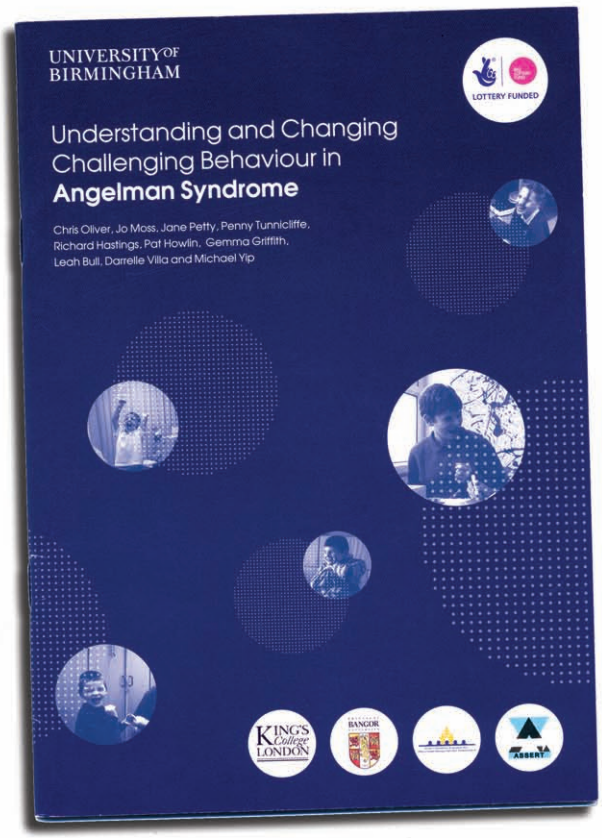
We raised over £1000 for Assert and were able to make the trip to Water Planet at Panama Beach City in Orlando. Kieran was treated to 4 hours with the dolphins in the morning then an afternoon session with the excellent therapy team.

The whole experience was amazing for Kieran and us, with noticeable changes in his behaviour.

Neil Buchan

Understanding and Changing Challenging Behaviour in Angelman Syndrome

ASSERT are now in possession of Chris Oliver's long awaited research into Challenging Behaviour in AS. This is an information pack and a DVD. All families were given a copy FOC at our recent conference. If anyone else would like a copy, please can you send a cheque for £2 to cover postage and packaging, made payable to ASSERT at our freepost address. Please put in a note to say that you would like a copy of the pack / DVD, and don't forget to provide your name and address!



Kidz up North, South and in the Middle

http://www.kidzupnorth.co.uk/

Disabled Living organises the largest UK exhibitions totally dedicated to disabled children, their families and the health and social care professionals who work with them. The events specifically focus on equipment, products, and services to improve independence and quality of life.

These FREE events were established 8 years ago with an annual exhibition at the Reebok Stadium in Bolton. It became apparent that there was an overwhelming need for these events with people traveling from all corners of the UK, Dublin, Jersey, Cornwall and the Orkneys.

In 2007 Disabled Living took the Kidz South exhibition to the Rivermead Leisure Complex in Reading, to provide people in the south of England with their own dedicated event.

Disabled Living is aware of the need to make the Kidz Events more accessible. We hope that Kidz in the Middle at the Ricoh Arena, Coventry will offer people, especially those in the Midlands, Nottingham, Leicestershire, Mid Wales and East Anglia an opportunity to attend the Kidz Events without the inconvenience of traveling too far.

The events allow both public and professionals to try the latest equipment and products solutions and talk to the specialist who supply them. There is always a full and varied programme of talks and discussions covering topics such as legal and education issues, new therapies, communication and ICT. Many voluntary organisations are also on hand with information on support, funding, holidays, publications and so on.

Disabled Living is the oldest disability charity in Manchester having worked with disabled people of all ages for over 110 years. It also has one of the largest Disabled Living Centres in the country, which amongst other services, specialises in advice, information and training on equipment for easier and independent living.

Dates for your diary:

Kidz Up North – Thursday, 25th November 2010 – Reebok Stadium, Bolton.

Kidz in the Middle – 10th March 2011 – Ricoh Arena, Coventry.

Kidz South – Thursday, 9th June 2011 – Rivermead Leisure Complex, Reading.

For free tickets or more information on any of our Kidz events please contact the organisers:

Disabled Living
Redbank House, 4 St Chad's Street, Cheetham, Manchester, M8 8QA.
Tel: 0161-214-5962 Fax: 0161-835-3591
Registered Charity No: 224742

SIBLINGS - JUST FOR US

By Russell Andrews

This year's ASSERT conference gave us an important opportunity to meet and talk with siblings and discuss together what more we can do to support brothers and sisters in the Siblings Forum. The day trips were particularly well attended by sibs (as I discovered when giving out the tickets at Drayton Manor!) and it was great to get to talk with several – we even tried to lose a couple on Saturday but alas they had a mobile phone with them.

One outcome of the Sibling's Forum was that Keri Darrock kindly offered to set-up a Facebook group for siblings (and others interested) so if you are on Facebook please check it out and join/leave a message at "My brother/sister has Angelman Syndrome" – we already have 15 members and some great stuff being shared. It's also worth mentioning that some of the sibs added excellent help to raffle on the Saturday evening. On Sunday at conference I gave a brief update on siblings support generally and was approached after by folk wanting to discuss the support for, and also the support of, older siblings which we will look into.

There were a number of helpful things which siblings discussed or wrote down during the conference and we'll share some of these over subsequent issues but for the time being I leave you with these from Lottie Fox-Jones and Poppy Cartwright:

My name is Lottie Fox-Jones, I am 15 years old and I help care for my brother Harvey who is 12 and has Angelman Syndrome and ADHD. This means he has the mental age of an eight-month-old baby and he cannot talk or understand even basic speech. Harvey has the loveliest personality and wants to play and hug all the time, but as he is extremely hyperactive and incredibly strong, he can easily destroy ornaments and furniture, pull hair out and bite or kick far too hard.

I also have a little brother Milo who is 5. I have to keep him out of Harvey's reach to stop him being attacked. I help entertain Harvey while mum is busy and keep him safe. We have locks on all the doors and we have to keep everything that could be destroyed out of his reach. He will eat anything including candles, dog poo, soap, books and toys.

I recently went to the ASSERT conference for the first time and I found it really interesting as I had never met anyone with AS other than my brother. I loved meeting and playing with all the other children and it was really odd because it felt like a whole room of Harvey's, with their jerky movements and huge smiling faces.

Lottie Fox-Jones

Hi, my name is Poppy Cartwright,

2010 is my 4th year at the ASSERT conference, although that only covers half the years I've spent living with my younger brother Euan. All of the trips to Loughborough have been beneficial to us as a family as we have found out more information, as well as about specialist equipment, such as the Tomcat Trike, which Euan uses regularly. Along with this there have been the days out which have varied across the years (and improved!) from Twycross Zoo to Twin Lakes, American Adventure to Drayton Manor, all of which have provided entertainment for all our family. There is, of course, the added bonus of Euan's disability which means that we can claim queue-jumping wristbands, much to the annoyance of the rest of the general public (ha, ha!)

Obviously the trips aren't the only things that have changed over the past eight years, as Euan's behaviour and skills have developed dramatically, particularly once he joined me in being a troublesome teenager. There are the skills that are expected of most children, such as getting undressed, drinking from a cup, or 'simply' walking and talking. Although Euan can't talk, his communication has greatly improved over recent years, and he has learnt to express what he does and doesn't want. Aside from this, there is, of course, his behaviour. He still occasionally pulls my hair, or hits and scratches me, it's generally because he's fed up, or I'm just being an annoying older sister, and he has no other way to express himself. However, not all of his behaviour is bad, and like any teenage boy, he has a cheeky side to him, and he is almost always giggling and laughing, and just generally sharing the love!

Coming to the ASSERT conference allows us to find out about other families dealing with the problems that Angelman's Syndrome have as well as the things that Angels have in common (thrill seeking or playing with water, anyone?!) It also means that I, as a sibling, get the chance to talk to other siblings about their experiences. Euan can also be himself at the conference, in that he can be left to wander around alone and we don't need to worry about him, and none of the girls or women mind giving him hugs.

When the next conference is held in 2012, I will no longer live at home, and I'm sure many more things will have changed too. However Euan will still be my cheeky little brother, and somebody I can go to if ever in need of cheering up or a hug.

Poppy Cartwright

London Marathon 2010

I have always wanted to do a significant fund-raising event for ASSERT and the London Marathon Golden Tickets seemed like a great idea to generate sponsorship - and it would do my fitness some good to boot! I had tried two years earlier without success but this year I turned 40 and thought I had better apply again before I was too much over the hill. The good news arrived in January 2010 that my name had come up in the ASSERT lottery for the marathon on 25 April. It had been 10 years since I last ran a marathon (in Kosovo) and I wasn't sure what to expect on the day in terms of huge crowds.

Issy helps Jim's training

Being in the Army in a Brigade shortly to go on operations, my fitness levels were reasonably high. However, I still picked out a 3 month training programme from the London Marathon magazine and stuck to it. In fact physical training lessons at work added to the programme and by April I was feeling pretty confident. Some of my relations pledged to double their sponsorship if I came in under 4 hours, so the challenge was on.

On the day itself there was a short burst of rain just 20 minutes before the start, so that really cooled things down. I was running with 2 old college friends - also doing the 40 year old mid-life crisis thing! We kept together for 15 miles and our time at the half marathon point was 1 hour 59 minutes and 55 seconds. I realized that I would have to up the pace if ASSERT was going to receive the full donation. The second half got harder, of course, as was inevitable, but the London crowds were great - the route was lined both sides with one or two people deep at least all the way. They carried me through to the 26 mile point when I discovered that the last 385 yards needed to be done in about 2 minutes... aaarrrgghh! By that point the pins and needles that had started in my kneecaps at mile 18, had turned into total numbness from my knees to my hip bones. I couldn't feel my upper legs at all but, with that tiny bit of energy I had left, I tried to put on a spurt and beat the 4 hour threshold. That really hurt. Amazingly, I crossed the line without collapsing and it was quite a relief to see I had come in with only 13 seconds to spare!

Jim crosses the line!

Isabelle, our gloriously happy AS 5 year old, was of course oblivious to all the training and running that I had been doing. As the weather turned dryer in spring, she began to recognize my running kit and would occasionally fetch me my running shoes as I sat at the bottom of the stairs setting up my watch, MP3 player and sweat bands. When the big day arrived, my wife Amanda stayed at home with our 3 young kids. Isabelle would have freaked with the loud crowds and all the unfamiliarity and it isn't that much fun as a spectator for kids anyway. She seemed happy enough afterwards though back home and thought the numbers on my running vest were very interesting. We hope and know that the £2400 raised will go towards something positive for AS children and parents and are grateful to ASSERT for the opportunity. Would I ever do it again? ...maybe for my 50th!

Jim Bacon

Izzy+Jim after the Marathon

Marathon Crazy Quad

My Niece has Angelman Syndrome so the charity means a great deal to me. I'm Dubai-based but ran the 2009 London Marathon on my own; this year I wanted to run it again in with company so I talked my friend Sam into running with me: The Terrible Two!

In Jan 2010 ASSERT told me that 2 people had dropped out and they had spaces available. I later heard there were more spaces available so Sam's boss Lewis decided to join us - now we were The Three Amigos! Three became four when Sam's brother joined us and we became The Crazy Quad!

As training started we soon became aware of muscles we didn't know we had. Training was built up to 6 times a week. By February things were getting hectic; Sam managed to continue her training even though she was in New York and Vegas for the week whilst the 3 of us continued to train in the Dubai sandpit!

The runs were getting longer now but we were all coping really well. The boys were football training on a Wednesdays so had to fit the runs in beforehand, whereas Sam and I were still dragging ourselves out of bed in the mornings to get the runs over and done with before work.
By mid-April we were in our final week of training and our focus changed to getting the sponsorship money in: though we were well over £1000 we needed to put the pressure on as our target of £4000 was looking harder to achieve than the Marathon at this stage.

In the final month our vests arrived and looked great. Lewis, Matt and Sam bought new trainers (all a size too big) and were looking forward to getting the Marathon over and done with so we could return to normal life. The training had been hard and is so time consuming but the light was (nearly) at the end of the tunnel.
The final week of preparation: names printed on vests, toenails cut right back and we were all UK-bound on the Thursday and Friday. We all paid for our own flights but due to the Volcanic ash problem affecting air travel in the summer we didn't think any of us would actually make it! Sadly, Matt was the unlucky one and his flight was cancelled. My UK friend Antony was joining us for the run but he had a nice easy journey from Essex: our trip was 3500 miles each way while his was only10 stops on the underground!

25 April 2010: MARATHON DAY! No turning back now - 5 months of training - 26.2 miles to go and, despite the rain, the weather was meant to be in the 20's. It was going to be an amazing day. Our sponsorship target had reached halfway and the last week had brought in a great amount.

At about 3 miles Lewis went ahead while Sam and I continued at our normal pace - a quick pop into McDonalds for a Toilet stop we were doing ok. But at 4 Miles Sam's foot started to hurt and by 10 miles she was in pain! The next 9 miles were a struggle but we were finding amusing things to talk about. We'd rejoined Lewis by now and were making frequent stops at the St John's ambulances for Sam to get ice packs for her foot and so that Lewis could have a leg massage.

Miles 19 - 21 weren't too bad but by 22 we could see the pain Sam was in. We ran/walked our way through 23 - 25 and Sam was definitely at her happiest when we turned the final corner and the finish was in sight. Lewis crossed the line after 6 hours 10 minutes, followed by Sam and me at 6 hours 11 minutes.
The amount of people that turned out was unreal and the experience of running with 2 of my closest friends is a memory that will live with me forever: Rhino's, Mr Men, Telephones, Army Soldiers, War hero's, Lion's and thousands of other brilliant fancy dress costumes – we can all be proud to say that we were part of the first Virgin London Marathon 2010!

Now all we have to do is hit our target so if you haven't already then please sponsor us, every little helps and after what we all went through today I think we all deserve your pledges! I would like to thank Sam and Lewis from the bottom of my heart for all their hard work for ASSERT. Running with you guys was so good and made the day 1000 times better than last year!

Would we do it again? 100%. Anyone reading this who has ever thought about doing the Marathon, my advice is DO IT! And running for a charity makes you do the training. It is the most fantastic day and a achievement that will stay with you forever. From all of us that ran for ASSERT I would like to say thank you for the opportunity, and there are several people I know that want to run it next year!

Jo Parker

Angels without Voices have created a children's book called *Just Like You,* written, illustrated and printed all free of charge to help in the fundraising for ASSERT

Look!

There is also a CD of Music from the Movies which features *Sam's Song*, a very moving song written for ASSERT and all the Angels in the UK.

Both the book and the CD are available to purchase online at www.angelswithoutvoice.co.uk at a cost of £5 each + P&P

If you are a Blue Badge holder you can now get the Blue Badge map sent directly to your mobile phone. It covers the whole of the UK and shows parking bays, accessible toilets, petrol stations and local council parking rules.

You need a phone that can connect to the internet.
Text Blue to 83377 (standard rates apply)

Conference Photographs

During the Loughborough conference in September ASSERT were lucky enough to have photographer Rob Chadwick donate his time and expertise in taking some great photographs of our AS family members. ASSERT wants to hold a database of photographs to use in future literature for the charity.

Shown here are just some of the wonderful images that were taken on the day, but don't worry if you can't see your loved one, the full range of photographs can currently be seen on Rob's ASSERT web page:
www.robertchadwick.co.uk/assert
And while you're there why not check out Rob's other great images on his main site to see what good company you're keeping:
www.robertchadwick.co.uk

If you were one of the families involved in the photographs and would like copies please write in or call ASSERT with your details. We can't make prints for you, but for a minimum donation of £5 to the charity we will gladly send you a disc of your images to keep and print as many copies as you like for yourself, family and friends.

Please note that ASSERT can only supply images of your own family member. Make sure you clearly identify your images from the website as they're currently only identified by number.

So whether you just want copies for yourself or the ability to make some great Christmas gifts, write in as soon as possible.












Assert Conference Report

Loughborough 3rd-5thSeptember 2010

Friday 3rd September saw the ASSERT family again return to Loughborough University for the 2010 conference. As always it was a friendly reunion for some, a catch up for others and for the new members of this strange family, it was a brave first step.

As is the tradition with the conferences, every year throws up a problem for the Trustees, which we have not encountered before (despite this being our sixth visit to the site). Although there are months of planning beforehand, staff changes and system changes mean it sometimes feels like this is our first visit! This year, we had a room layout change that had not been brought to our attention until the day before our arrival. Hopefully, the vast majority did not notice the frantic changes being made to the accommodation lists. Some however, were affected. To those families that offered room moves and room shares to accommodate our diverse group we can only say thank you. A handful of very accommodating families meant that everyone could concentrate on what the weekend was really about.

Friendship is, as ever, a central theme to the weekend. Through late night conversations over a drink or three, sharing experiences at regional break out sessions, chatting over lunch to riding the Shockwave at Drayton Manor; the weekend is about making new friends, rekindling old friendships and generally relaxing in the company of others who know what a world touched by Angelman Syndrome is like. New faces were quickly befriended, and by the time Sunday came around, the time it takes to say goodbye each year just gets longer and longer.

Families are at the heart of everything we do. This year, this was no exception. We even arranged the weather this year to minimise the risk of man flu once the families had returned home. A new direction over the last few months has been a determination to ensure siblings are catered for in their own right, rather than as an afterthought, and this became real during a small session which has since led to a facebook group being established.

With so much information available, how do you fit sufficient detail into the very tight timetable so that everybody gets everything that they need ? The simple answer is you can't. But we do try the impossible at each conference, and 2010 was no different. Key note speeches and workshops from old friends like Bernard Dan (neurology and medication), Chris Oliver (behaviour) and Jill Clayton-Smith (genetics) and newer ones like Dan Hindley (neurology), Gina Davis (attention), Nigel

Pugh (the law), Nic Bayliss (rebound therapy), along with many other speakers, presenters, exhibitions and one to one sessions saw a packed timetable struggling to fit into two days.

And lastly fun. The conference is a weekend in which normal routines and restrictions are put on hold for a couple of days. A time when nearly everybody wins a raffle prize (although experience tells us that letting angels deciding on the prize from the table is not always the best idea). A time when the families head out en masse to theme parks and our needs are taken seriously, without fighting too hard to get it. A time when the party entertainers really do earn their money, playing to the crowd and then leaving still with all of their equipment accounted for, and a time when as organisers, we really do remember why we put months of voluntary work into making it happen – even though by Friday we had no idea where we were going to find the stamina to make it through the weekend!

To those that helped make the weekend a success behind the scenes, the kitchen staff, the bar staff, the reception team, the raffle prize donors, the coach company and of course our experts, thank you. To those that came in 2010 we owe you the biggest thanks – you and your families all helped make the weekend the success it is, and we hope to see you in 2012.

Sue Williams

Brand New!

Assert

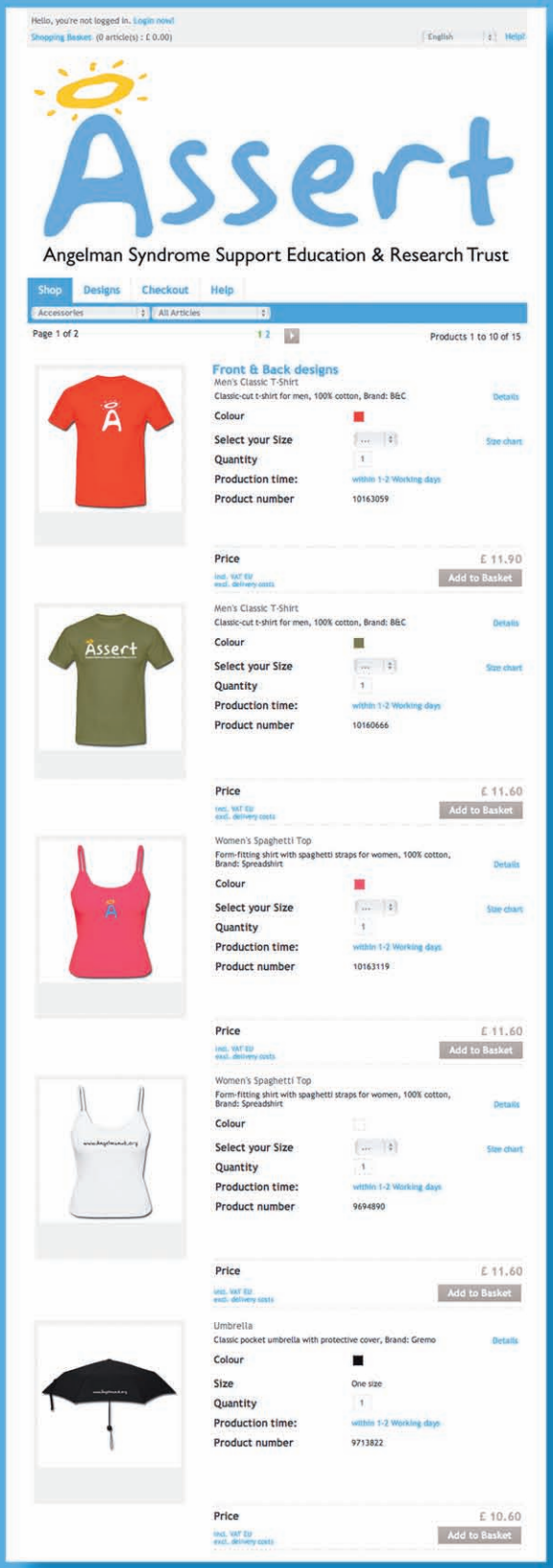
Merchandising

In association with Spreadshirt, our on-line merchandising shop is now open. Whether you just want to support your charity or looking for that perfect gift for friends and family, you'll find something here to please everyone.

This link:
http://458768.spreadshirt.co.uk/
will take you straight to ASSERT's merchandising page - or you can reach it via the ASSERT website. Select from mens, ladies and childrens clothes, along with some great accessories - you name it we've probably got it, or there's a good chance it's coming soon.

And taking note of your conference feedback we've added some looser-fit shirts along with other great new products.

Shown here are just a few samples of what's on offer. Make sure you bookmark the Spreadshirt site and check back regularly for more great designs soon.



http://458768.spreadshirt.co.uk/

"Too often we underestimate the power of a touch, a smile, a kind word, a listening ear, an honest compliment, or the smallest act of caring, all of which have the potential to turn a life around."

Leo Buscaglia

Angelman Syndrome

by Bernard Dan

This comprehensive guide to the clinical management and basic science of Angelman Syndrome will be of value to clinicians and researchers as well as parents and relatives.

Usually £71.25 but available for the special price of **only £37.50** to ASSERT members. Contact us for for a copy of the publisher's pdf file for mailing details.

Paperback: ISBN 978-1-898683-55-1

Mine's a Half...

I completed the Humber Bridge Half Marathon on 27th of June 2010 in aid of ASSERT. I raised £712. The run was enjoyable but extremely tough, it was the hottest day of the year with temperatures of 34 degrees. I managed to complete it in 2 hours 28mins (not bad considering the heat). We ended the day with an after party in the garden to say thanks to everyone for donating... the first drink I had had for a month!

Adam Smith

Climbing Angel Mountain

How do I start to tell you about life with my Angel? Well, first I decided to confirm the definition:
ANGEL: *A person of great beauty, kindness, or virtue.*

So, what do you think? Is this right? Of course I hear you all say, they are wonderful children and life is better when you have an angel in your family. Now be honest with yourself and everyone else. Life is difficult, tiring, a struggle, frightening, a constant battle of wills and a never ending world of what ifs and what will life hold for our Angels and the rest of our family in the future.

Going back 16 years to the happiest day when I held my newborn daughter in my arms and she cried and cried for hours at a time, I remember the nurses saying she will wear herself out and settle down soon for a long sleep. What did they know? I am still waiting for that long sleep. At 14 months, I waited 3 hours to see a Health Visitor who wouldn't listened to my concerns that Emma wasn't developing the way I expected and then I was informed I was a neurotic, attention seeking mother who needs to stop looking for things that were not there and then proceeded to write in Emma's notes that she was walking and talking to the development level expected at that age. Shame she never actually looked at Emma at the time as it would have saved her a lot of embarrassment a few months on. At 19 months Emma had the Hand, Foot and Mouth virus and when I visited a new GP about it, she commented that Emma wasn't really developing properly. Finally, someone was thinking the same as myself. Emma was again admitted to hospital where blood tests with various other assessments were carried out. At 22 months, Emma started taking drop fits and on admission to hospital after her 49th fit in 14 hours she was given EEG's and nothing much was said. At 23 months, Emma was given a possible diagnosis of Angelman's Syndrome and I was so relieved (but a definite diagnosis of a mutation wasn't received until she was 9 years old). I know this is a strange thing to say and yes, I was upset but after spending nearly 2 years of my daughter's life trying to be believed that something was wrong, it really was a relief that I had been right all along.

Things progressed, regressed, progressed, regressed and so on. I was given the number of ASSERT from a distant relative and after a number of times picking up the phone, dialling then putting the phone down again, I actually dialled one night and before I could put the phone down someone answered. We talked and talked and I was given honest answers to my frightening questions. After a long conversation with a total stranger I felt much better and decided I was doing all the things I could do at that time so was reassured I was being a good mum to my Angel. My most vivid memory of my calls to ASSERT was my second call, I was feeling positive (or so I thought) and when the voice at the other end of the phone asked how I was that day, the tears overwhelmed me and I cried and cried for almost an hour before I could even answer the poor woman. Funny thing was, she cried with me, she knew how I was feeling, understood what I was going through and the fear of what I was facing in the future. I will never forget that phone call or Sally Walburn who was on the other end of the "conversation". From that moment on I knew I wasn't alone and never would be.

Well, here we are, Emma is now 16 years old, a happy, loving, demanding, funny, kind, beautiful, hyperactive, babbling, challenging, wonderful, hormonal determined young woman who is the biggest, brightest star of our lives. She loves swimming, horses, candle making, felt making, art, shoes, clothes, hide and seek, playdough, playing practical jokes on people and partying like there is no tomorrow (well, there is no tomorrow when you don't sleep from one day to the next, there is only one very long day that never begins or ends). I have not had much contact in the past few years with other Angel parents for various reasons, but thankfully through facebook, I have come back to Assert and I am now hopefully helping other parents of Angels some of whom are in Canada, New Zealand and also getting help from parents of older Angels who have passed the stages we have reached in our family with Emma and are giving me advice and hope for the years ahead.

Like all children EVERY Angel is different but they all have a few things in common. They are special, wonderful, loving and happy children who bring blessings to our lives as both members of our families and also as part of the local communities. A large part of our lives with our Angels are laughter, fun and seizures. Seizures come in many different forms for our Angels but every seizure filled day is a frightening experience for all the family. Medications are wonderful when they work but finding the correct dosage and cocktail of anticonvulsants can be a long, hard, distressing time and you feel like there will be no tomorrow.

To sum things up, I couldn't ever imagine my family without Emma. She is an inspiration, a reason for living, a blessing from a higher place and to quote the definition a person of great beauty and kindness. She has made us all stronger as a family and better people for having her in our lives. If there's one song that would sum up the lives of being the parent of an Angel it would be *The Climb* by Miley Cyrus - take a look at the lyrics if you have time.

So there it is, I am climbing the Angel Mountain and I know there are many other friends and families on the same expedition. To everyone out there, ASSERT yourselves, keep on climbing and I'll see you at the TOP.

Linsay Dinnis

Pizza and Pasta Party

Held in March at the Elmbank Drama Studios, Ayr, to raise funds for ASSERT. I'd like to thank everyone who attended the party and especially the Angelman families, some of whom travelled for up to 5 hours to be there and I really appreciated the effort they made.

A great night with £300 worth of various large pizzas donated by Domino's Pizza, drinks donated by ASDA and the Post Office/General Store in Dalrymple, FIB Musical Theatre entertained with a wonderful presentation of song and dance while Jacquii McGivern donated the premises and organised the presentation.

This party was also the launch of the ASAC (Angelman Syndrome Awareness Clothing) which has so far raised a fantastic total of £769 for ASSERT.

Prices start at £10 for children's t-shirt and £15 for adults. Sizes go up to XXL with XXXXL available in some items. ALL PRICES INCLUDE POSTAGE IN THE UK! If enough people are interested, the company are willing to sell them internationally on Ebay and all profits will go to ASSERT.

Solve your Christmas present problems and raise money for ASSERT at the same time.
Just email me on linni.22@hotmail.com. Or contact me at 0796 972 2959.

Safari Party

On 1st August 2010, a group of families met up at the Blair Drummond Safari Park for a day of fun, laughter and getting to know each other.

The day began when we all met up and had a drive through the animal enclosure. Several parents commented that our angels would have been just as happy in a car show room, as they seemed to enjoy watching the cars more than the animals but hey, they were happy!

We then had a lovely packed lunch which everyone contributed to. This was destroyed by the hungry families during a noisy and fun packed time – kids getting to know each other and parents trying to talk and establish their friendships that we have on facebook. No-one will ever tell me that facebook friends are not real friends.

Every angel was presented with a specially made hat which were worn with pride.
Next was a walk about followed by a wonderful show in the sealion enclosure. There were lots of squeals and clapping and lots of laughing and smiling faces. Our angels faces spoke volumes and wow did they enjoy themselves.

All in all we had a great day and cant wait to do it again.

An update on Philip

by Rosemary Teggin, Dublin, Ireland

In September Philip Teggin celebrated his 21st birthday in style, with a party in his residential home with gorgeous finger food and huge birthday cake, and a musician playing for 2 hours. He got lots of presents from family and friends, it is difficult knowing what to buy a 21 year old AS, so he has lots of new clothes and toileteries and DVDs... though his favourite is still El Dorado and Aladdin.

Philip has been in residential since November 2008, which is 7 miles/15mins drive from our home. It was also a time of transition between school and adult services (both within the same grounds), which is one mile from our home. He adapted instantly, we found it more difficult. They have a bus which takes the residents who attend the day service in from 10 – 3.30. We bring him home on Sundays.

We haven't taken Philip on holidays for a long time, but this year we took him for a week's holiday (with volcanic ash, and other flight delays we felt it would be better to stay within Ireland). We went to a lovely self catering lodge which has a leisure centre in the main hotel and lots of walks around a forest area. Irish families might like to check out www.shannonoaks.ie which is in Portumna. The lodges sleep 10, with 2 bedrooms and shower room on the ground floor and 3 bedrooms, large bathroom and one ensuite shower room upstairs. We preferred to use the shower in the leisure centre as it was for special needs and more spacious.

He didn't sleep at night, (different environment, he sleeps well normally) and I sat with him every night on the couch watching Aladdin, and he was up and about as soon as the titles came up at the end. Nigel took him out for walks, on the second day for 3 walks a total of 5 hours, and Philip decided that it was too much, and dug the heels in at the gate and refused to go again, much to Nigel's disappointment. (Philip no longer uses a wheelchair, but Nigel wishes we had brought one as the walks had a good surface). He enjoyed the indoor swimming pool which had easy access steps.

A few weeks before his birthday we had another celebration in the family, when Philip's oldest brother Andrew got married. It was a lovely day, and Philip attended with his keyworker and he was very well behaved throughout the service and he loved the music in the evening, but he didnt want to get up to dance, but Philip loves to sit and watch other people and sway to the music.

We also had an informal Angelman Syndrome family get together in Dublin, during lunch from 12.30 – 3.30, the last Sunday in August - the week before the ASSERT conference in UK. In the restaurant we were allocated a spacious corner section, where 8 families, some only diagnosed a short time and meeting other families for the first time. Some travelled from the north, south and west of Ireland. We chatted and shared experiences and information.

Variability in Angelman Syndrome

What Does This Mean, and Why Does This Matter?

An important question that may arise with the diagnosis of Angelman Syndrome, and that may persist is "what will my child be like?". This is a very natural question following a diagnosis as families and caregivers seek to understand the implications of the diagnosis.

This article will address some basic scientific concepts upon which variability in genetic syndromes is grounded, as well as discuss the effects that the ideas and realities of genetic variability actually have for both the child and the family living with Angelman Syndrome (often referred to as "AS").

What Will My Child Be Like?

You may have noticed that your child is not achieving expected milestones and sought help with a medical professional. Due to the fact that it is a rare disorder, it is common for doctors to not at first suspect or test for Angelman Syndrome. As such, a diagnosis is sometimes not forthcoming. However, some diagnoses of Angelman Syndrome are made before one year of age.

When a parent is told the diagnosis is Angelman syndrome and they are made familiar with the characteristics of this disorder, such as seizures, sleep problems, non-verbal communication, severe cognitive impairment and more, it is quite natural to wonder what the future may hold.

Early diagnosis can be beneficial. First, one has more time to come to terms with what it means to raise a child with special needs. This can be intimidating for there are many factors to consider including the following: special education; doctors and specialists; daily living; and long term living options. An early diagnosis also means an early start of therapies and access to services, which has been shown to be of enormous benefit to the child. A late diagnosis is equally important. While the parents/caregivers may already suspect that their child has special needs, getting a genetic diagnosis of AS opens the door to a wide base of knowledge and strategies that are effective in AS, as well as providing access to a community of others with shared experiences.

What will your child be like? It is a natural question to wonder if they will be able to sit up or walk. You may ask, "Will my child talk or use sign language? How will they play with other children? Will my child go to school? Will he/she be able to ride a bike? How will my child's development progress and how does that affect plans for the future?"

While no one can predict milestone attainment, we do know that not all children with AS experience the same degree and severity of symptoms. For example, while most children with AS will experience some form of seizure activity, some children with AS never have seizures.

In order to understand the manner in which these differences occur, we need to explore additional aspects affecting the genetics of Angelman Syndrome. (Please visit the Genetics page on the FAST website for an in-depth review of the basic genetics of AS). Two key concepts that can help explain variation in Angelman Syndrome are genetic penetrance and genetic expression.

Genetic Penetrance

Penetrance is a term used in genetics to explain the likelihood that a gene (genotype) will express an associated trait or appearance (phenotype). Populations expressing a genetic variation (like the loss of UBE3A function seen in Angelman Syndrome) are described by associating that genotype with certain characteristics. In Angelman Syndrome, the characteristics define the syndrome. For example, 70% of deletion children will have a sleep disorder, or 1% of all AS individuals will have mosaicism.

Penetrance can be described as:

- Complete penetrance - all individuals having the gene variation have clinical symptoms associated with the variation.
- Highly penetrant- the trait caused by the gene variation is almost always apparent.
- Reduced penetrance - some individuals may not express the trait, although they carry the gene variation.
- Low penetrance - a trait will rarely be apparent even when the gene variation exists. In this case it is often difficult to differentiate between genetic and environmental factors as a 'cause' of a trait.

The current body of research suggests that Angelman Syndrome falls into the realm of complete penetrance. That is, anyone with a mutation that affects maternal UBE3A expression will present with characteristics of Angelman Syndrome. However, there are different expressions of the phenotype among individuals. If we take into account the other 99.9% of inherited genes, as well as differentiated environmental influences, it is quite normal to find differences in abilities and development among those diagnosed with Angelman Syndrome.

Genetic Expression

Angelman Syndrome is just that....a syndrome. If developmental milestones and abilities are plotted, a normal bell shaped curve of distribution occurs. Some individuals will have minimal self-help abilities and need maximum care, while others may have more developed self-help skills and need less assistance.

While there is a recognized relationship between genotype and the severity of phenotype in Angelman Syndrome, the correlation is not definite. For example, there may be two children with Angelman Syndrome that share the same class of deletion size, yet there are manifest differences in milestone attainment and expression of abilities.

The following concepts can help explain this:

- In Angelman Syndrome, there is a functional loss of the maternal UBE3A gene. When the maternal copy of the gene is nonfunctional, (or non expressive), Angelman Syndrome results. Without that functional copy of the gene, a myriad of problems occur, which among others results in a brain that cannot process experiences correctly, or lay down memories in typical fashion. This translates into cognitive delay, non verbal behavior, seizures, and more. The paternal UBE3A gene is still present, but remains 'silent', which is the normal state of the paternal gene.
- Generally, children that are born with a deletion of the UBE3A region (encompassing more genes than just the UBE3A gene), tend to have greater challenges in terms of motor development and seizure control. The reason that deletion positive children are generally more severely affected is likely due to the loss of expression of many other genes in the region. Other genotypes of AS (UPD, ICD, UBE3A mutation) show a tendency to be more mildly impacted than deletion positive types, again likely due to the fact that people with these genotypes have not lost additional genes in the Angelman region of chromosome 15.
- Environmental influences also play a large part in determining the developmental progress of children born with AS. Getting timely medical care and follow-up, experiencing physical, occupational and speech therapy and having a firm plan for schooling that supports the particular needs of the child can account for some variations in development, all other factors aside.

The message here is that your child is both the product of their ~29,000 other normally functioning genes, as well as their life experiences. A child who is diagnosed with Angelman Syndrome due to a genetic deletion may also look very different in terms of characteristics than those diagnosed with a non- deletion error (such as uniparental disomy), or an UBE3A mutation, or some type of mosaicism.

However, the genotype is not alone in determining the phenotype. While intense therapies such as those provided by Early Intervention Programs are normally directed toward children from birth to three, obtaining resources and providing therapies shown to be effective in Angelman Syndrome are important even if a child is diagnosed later in life. Effective therapies, and positive environmental influences, have been shown to increase developmental abilities, as well as enhance the quality of life for many individuals with AS. Medical professionals and local human services agencies can provide guidance toward resources that result in meaningful developmental enhancement.

While there is no present 'cure' for the genetic defect in Angelman Syndrome, it has been shown that physical, occupational, and speech therapies are beneficial for children with developmental delay, regardless of genotype or present developmental attainment. (For more information on these types of interventions, visit the Get Informed section of the FAST website.)

How Genetic Variation in Angelman Syndrome affects Your Child

As discussed above, there are variations in the presentation of Angelman Syndrome from individual to individual due to the combination of genetics and environment. As a parent or caregiver of an individual with AS, it is important to remember that nothing is set in stone. It is a mistake to presume that all of the characteristics associated with this syndrome will present in each individual to the degree described by clinicians for the syndrome in general.

In assessing developmental achievement or progress in such areas as "when did the child start to sit up, walk, potty train, play games, etc...", the variance is great, even among AS children of similar genotype. There are a few key factors to keep in mind that impact developmental attainment as well as "phenotype", or appearance:

- Genetic Penetrance and Expression: While Angelman Syndrome may fall into the realm of complete penetrance, the expression of the other genes in the genome play strong roles in your child's abilities and personality.
- Epigenetic Regulation: Outside influences (environment, diet, medications, etc...) may influence the level of skills a person will have through enhancing inherent potentials and managing developmentally disabling medical conditions with medical intervention.
- Seizure Disorder: This can have a significant impact on the development of an individual and should be closely monitored by a licensed medical professional. Medications and dietary modifications have been shown to alleviate epilepsy and should be discussed with the child's licensed medical professional.
- Therapy and Services: There is ample scientific evidence that points to the success of Early Intervention in terms of physical, occupational, and speech therapy for persons with disabilities. Therapies for older individuals can also be effective in enhancing skills and the quality of life.

- Diet and Nutrition: Like any individual, a person with Angelman Syndrome needs a nutritious diet. Given the feeding and reflux issues many individuals with AS deal with, it is beneficial to seek advice from dieticians and GI specialists If a child is unable to take foods orally, parents should consult with a medical professional about a G-tube, which delivers nutrients through a surgically manufactured opening in the stomach.

Establish a 'circle of support' for the individual with Angelman Syndrome as well as for the parents/caregivers. This can include caregivers, friends, caseworkers, teachers, medical professionals, therapists and family members. The idea is to build a support network for the individual and parents/caregivers, so that as the individual grows, there are multiple resources to draw upon when needed. For example, it could mean something as simple as needing good respite care while the primary caregiver does household errands, or something as complex as arranging for special care when traveling. Also, having people around the individual that care about his/her welfare and growth, will go a long way toward allowing the individual to achieve his/her fullest potential through early childhood, the teenage years, and into adulthood.

A final thought to impart, is that while a person may be born with a loss of function of the one gene, UBE3A (the "Angelman" gene), she or he is, however, born with thousands of other fully functional genes.

This, more than anything else, will determine "what your child will look like". Yes, an Angelman Syndrome diagnosis may be life-changing, but many other factors will influence development and determine the overall quality of life experience. Foremost are the variety of enriched environment experiences that allow for learning, growth and achievement.

The best thing any parent can do, is to get informed. Find a great doctor. Seek out other parents and join an Angelman Syndrome organization that best meets your values. Become an advocate and aim high. Small miracles happen every day. It makes good sense, to leave the door open to experience them.

(All of the individuals pictured in this article have Angelman Syndrome and are deletion positive.)

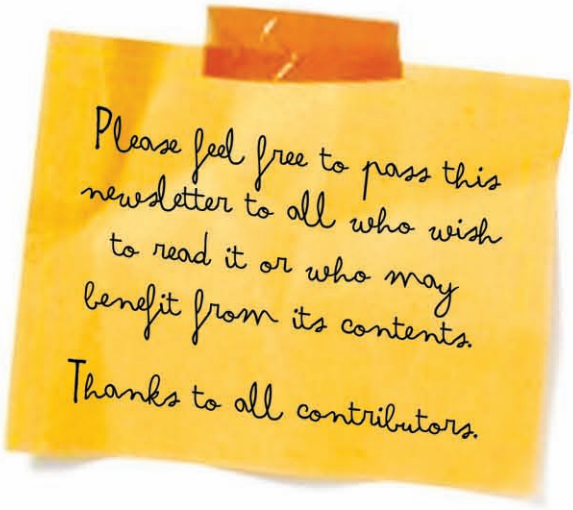
This article was authored by Elke Sprow

Reviewed for accuracy by Lynn M. Bird MD Rady's Children's Hospital, San Diego, CA and John R. Waterson MD PhD FACMG Oakland Children's Hospital, Oakland, CA

ASSERT Bank Account

Please be aware that we now only have our current account. All other accounts are closed. There have been some receipts paid into the other accounts which at the moment the bank are automatically transferring to our current account but I am sure there will come a time when they will not do this. So please do not pay any donations, collection tins etc into these other accounts. If you would like our current account details, please contact the support line.

Lisa Court, treasurer



REGIONAL MEETINGS

Could you host or help to organise a regional meeting in your area? It could be a coffee morning or a full on fun day. Recent meetings have been held in Special Schools and have proved very successful. Most schools are well equipped for our children with changing facilities, play areas and separate dining areas. Other ideas include local Soft play centres i.e. Wacky Warehouse or community rooms. If you feel that you would like to help families in your area meet up and build relationships then please contact ASSERT and we will help in whatever way we can. Moderate expenses can be covered if necessary.

CLASSIFIEDS

If you would like to advertise to the ASSERT community send your adverts to us and we'll endeavour to publish them in the next newsletter.

Britax Traveller Plus car seats

We have 2 Britax Traveller Plus car seats for sale. The exact specification of the car seat can be found on the Britax website: http://www.britax.co.uk/car-seats/traveller-plus

In summary it is a seat that is specifically designed for children that need additional support / security when in the car. It has a 5 point harness and is for children between 15-36 kgs.

We have used one car seat in each car. They have proved invaluable in transporting Ella. She needs the 5 point harness to stop her slipping down, and to keep her sat in a safe and comfortable position.

The only reason for selling these is because Ella has outgrown them. They retail at between £400 - £500. One car seat is in a better condition than the other, so one is for sale at £100 and one is for sale at £50.

If you would like further information, please contact me at: lisa.court@ciber.com or phone on 07870 234947.

DONATIONS

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:

General Donations:

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(Humber Bridge Half Marathon)
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Graham O'Neill
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