

# ASSERT

Angelman Syndrome Support Education & Research Trust



ISSUE THIRTY THREE JULY 2002

REPORT

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REGISTERED CHARITY NO. 1021882

## Letter from the Trustees

Hello Again!

It is time that if you have not registered for the conference you do so now!! It promises to be great weekend, not only from the wealth of information on offer but a good chance to meet many others just like yourselves who are all living with Angelman Syndrome. The details are on our new website which is easy to navigate and you can contact us this way if it is easier. [www.angelmanuk.org](http://www.angelmanuk.org). Alternatively, contact Jim Brennan on 01795 429061 for a Conference Pack.

The Website is largely due to Jeremy Webb who has worked hard to get it up and running along with Gerry Egan and John Outterson. Jeremy stepped in last year as he has the expertise working in this field already. We do receive quite a few enquiries through the site so it is a vital tool in supporting families complementing the telephone support line. We wanted it to look professional and be easy to use and we think you will find this has been done. Next time you are surfing have a look and if you have any suggestions for improvement or additional information/links you would like to see on there, please let us know. Many thanks to Jeremy, Gerry and John.

The new logo now appears on the website as well as new stationery. We hope that you will join in thanking Jackie Klimanska and Qd – Quarto design for their work on this and also Aleks Klimanski for printing our new headed paper and compliment slips free of charge, this is very generous and saves the charity a considerable amount of money.

We are pleased to welcome David Kinnear as a new trustee, David has written about himself on page 10 as a way of introduction. Nick Bartholomew has settled well into his new role as treasurer and we are very grateful to him for taking over from Elaine Sears. Elaine and Stephen Sears resigned at the end of April and as said in the previous

newsletter we thank them for all their hard work over 10 years. They were both involved with ASSERT, from the very beginning as founder members. We hope Elaine will keep us up to date with Holly as we have enjoyed and appreciated reading about her over the years, some of you would have read about her in the past and also in the Family Pack Information. So thank you to you both.

Next year Jim Brennan will be standing down and we have a year to find someone to become our secretary/administrator. If you can help with this then please let us know. It is a vital part of the work of ASSERT and we hope you will consider supporting us.

The IASO are looking for support in organising and fundraising for next year's conference which is to be held in Washington DC, in July. If you would like to be involved with AS on an international level, please contact Sally Walburn.

Many thanks to those who have sent in the Family stories; we have quite a few in this issue. As highlighted by Jackie Rose (page 8), we are getting an increasing number of calls from families with a 'confused' diagnosis, this is mainly due to all tests coming back as inconclusive. In some cases families are given a 'clinical' diagnosis based on physical symptoms of AS. We would say to you all that until such time as you are told differently you are all welcome as members of ASSERT. Attending conferences and Regional Meetings is often the best way of finding out whether your child/young person/ adult 'fits' into the family of Angelman Syndrome as the similarities are striking and cannot usually be confused. I know Jackie will be coming to the conference and we look forward to meeting with all her family.

Well there's not much more to say other than enjoy the newsletter, keep up the fundraising and we all very much look forward to meeting with you in September.

**The Trustees of ASSERT**

## ASSERT 2000 Club

*We wrote about this in the last newsletter and would remind you all again.*

We have reached the end of our first and very successful year of the 2000 club. We would like to thank all of you who have taken part and congratulate all the winners. It is now time to remind you that we need you to renew your membership and also invite any newcomers to take part. It costs £12 per year to join and 50% of the money taken is given back to ASSERT, the rest goes to the winners during 4 draws over the year – this is

just 25p a week to help ASSERT to run. If you would like to join in or participate again, please complete the enclosed form and send it together with your cheque payable to ASSERT for £12 (or £24 and so on if you would like two or more chances!). Last year we raised £667.47 (Jim said that the odd figures is because we had to change some Irish punts!) **DON'T FORGET TO INCLUDE YOUR NAME AND ADDRESS ON THE FORM AND THE CHEQUE – JUST SO WE KNOW WHERE TO SEND YOUR WINNINGS!!**

# Fundraising

## MISSION IMPOSSIBLE



**The brave/mad team**

The date was November 24th 2001, the time 10.00am. You may think just an ordinary Saturday for perhaps some shopping. No – think again.

This was our Mission Impossible – an Army Assault Course at Redford Barracks in Edinburgh. Our commanding officer was P.T.A Lance Corporal Hamish Leslie and we were his victims, an assortment of women and men of varying abilities and ages. It has to be said though that most of us were completely UNABLE. Prior to the course I had been keeping in touch with ASSERT trustee Jim Brennan (ex army) who had asked me how the training had gone -of course none of us had trained at all. Wish I'd listened to Jim!!!

Anyway, the day started well for me – I received lots of positive encouragement from my husband “You’ll NEVER do it Dawn”, but I was undeterred. When we arrived at the Barracks Hamish escorted us to the training fields and we all said, “Oh – that doesn’t look too bad!”

How wrong we were. This was the army and they were not going to let us off lightly. We were split up into 2 teams and really put through our paces and were MADE to complete every single obstacle before the final run.

There were a few injuries along the way, particularly on the ropes. One of the girls ended up with whiplash, another ended up in casualty, but it has to be said the worst injuries sustained

were by the spectators who were admitted to hospital with sore jaws from laughing. Particularly the army rugby team who watched the whole event!!!

But it has to be said we have talked of nothing else since.

Our sense of achievement and the buzz we all got was tremendous and not forgetting the amount we raised – a WHOPPING £1130 which was doubled by the Bank of Scotland as part of their charity fund matching scheme. This allowed us to present ASSERT with a cheque totalling £2260, which shall go towards the Conference funding in September. Brilliant.

My personal thanks go to all the people involved who chose ASSERT as their named charity and all those who contributed including Bank of Scotland’s Specialist Property Finance department and Capital Bank Motor Finance section. I have been absolutely amazed by people’s generosity and the most touching was a colleague of my husband, whose elderly father in a retirement home gave us a cheque for £50.00.

We are a close group on the back shift at Bank of Scotland Phoneline service and they have all been a brilliant support to me since Lauren was diagnosed – I couldn’t have got through it without them! Thanks also go to Lance corporal Hamish Leslie and all at Redford Barracks.

The video is available on demand (blackmail only).

**Dawn Fraczek**



**A well-earned cheque**

### Specialised Clothing

I’m sure this has been an issue for other parents and it has certainly been one of the major behavioural problems for us, the smearing during daytime/night time. Night time was a particular problem for us and we tried everything to prevent it.

Eventually we managed to get a contact called KC Sleep suits that manufacture vests and sleep suits etc for special needs children/adults. We ordered 2 sleep suits and a vest which have been a brilliant success, but quite expensive –

the sleep suits being almost £30 and the vests £18.00. I have since been given information from a fantastic support group called PAMIS about a charity manufacturer who supply a wide range of clothing (including bibs) at very reasonable prices. They are called “Friends of Fashion Services” and their telephone no. is 01274 597487. Address: Greenfield Centre, Green Lane, Baildon, Shipley BD17 5JS.

**Dawn Fraczek**

### **Another Bank of Scotland Phoneline Charity Event**

I would also like to write to you all about another charity event held by some of my friends on the back shift at BOS Phoneline.

They organised especially for ASSERT a theme night for Halloween based around the Wizard of Oz. All of them got dressed up and had decorated their working area in a Halloween theme packed full of homemade baking and goodies. They also made a yellow brick road, which ran all the way through the office. A competition had also been set for our kids to draw the best Halloween picture and these were judged by our Call Centre Director. What a night!

It was a brilliant success and they managed to raise around £700. Again, I have been totally amazed by the generosity and support and it was decided that the money raised would go to the Conference.

We will never forget our time at the Conference where we met some fantastic families and children and hope to see you all again in September. It has been a difficult couple of years, since the original diagnosis, but without the help and support of our friends family and ASSERT I really don't know how we would have managed. I have never forgotten that first phone call to ASSERT and my very long discussion with Gerry Egan. We then knew that there were other families who felt the same as us and of course life would be difficult and hold many challenges, but we know that Lauren will also make us very happy.

**Dawn Fraczek**

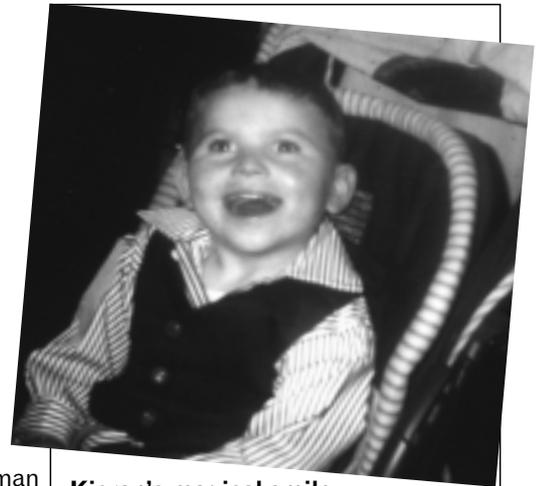
*Many thanks from all of us at ASSERT for such sterling efforts – the contribution from both fundraising events is a wonderful boost for our conference funds. We look forward to meeting with Dawn and family there – well done!!*

## **Ideas for Fundraising**

*Thanks to Linda Buchan for her fundraising efforts, she wrote the following letter to us to let us know what she had raised for ASSERT and how she did it.*

Dear ASSERT,

I enclose two cheques, one for £107.45 which I raised through Webb Ivory Fundraising Catalogue and I will continue to do this throughout the year. The other cheque for £110.00 was raised from having a jewellery party and the 10% commission going to Angelman Syndrome. My son Kieran, who I enclose a photo of, gave everyone a big smile and I'm sure that helped everyone spend a bit more! Kieran is now three years old and is a happy smiley little boy who is up to mischief all the time. Kieran was diagnosed with Angelman Syndrome a year ago. He has progressed so much over the last few months and happily the future looks bright for him as he is almost walking which I thought I would never see after the tough time when Kieran developed epilepsy.



**Kieran's magical smile**

**Linda and Neil Buchan**

## **Communication Aids**

I have received some information, which, I think may be, of interest to you it is about a Communication Aids Project. Full details can be found on [www.becta.org.uk/cap](http://www.becta.org.uk/cap) We have taken both George and Chloe for an assessment for their need for communication aids and computer equipment and were given lots of information and the opportunity to try out different keyboards, mouse etc and switches which would be particularly suitable for both of them.

There is also another address [www.inclusive.co.uk](http://www.inclusive.co.uk) and [www.semerc.com](http://www.semerc.com) which is a magazine showing all different kind of software and equipment. I believe the aim of this project is to help pupils who have communication difficulties by providing technology to:

- Help them access the curriculum and interact with others.
- Support their transition to post-school provision.

Parents can also purchase items themselves but it obviously costs money. I hope you will find the above of interest.

**Jennifer Darlow**

*Read all about George and Chloe on page 5.*

## **Conferences**

As you will all know we do like to keep you informed about events going on elsewhere in the AS world. Hopefully we will be able to bring you reports of other conferences taking place over this year.

### **Canadian Angelman Syndrome Society (CASS)**

Angels by the Water, 10 – 14 July 2002 Alliston, Ontario  
More information [www.angelmancanada.org](http://www.angelmancanada.org)

### **Angelman Syndrome Foundation (ASF)/ IASO**

Washington DC, 02 – 05 July 2003  
More information [www.angelman.org](http://www.angelman.org)

## Easter Concert Organised by Fran Childerhouse

Our son Joshua (dob 27.6.97) was diagnosed with Angelman Syndrome at the age of two. Prior to diagnosis we were reassured that it was unlikely that he would have Angelman Syndrome as clinically he showed few of the signs. A diagnosis was therefore a shock and he was re-tested. The results of this showed he had UPD and very little could be told us about this 'type' of Angelman Syndrome. "Joshua will probably just do his own thing" was the reflection of the Geneticist.

And Joshua has!! As he's got older he has developed clearer signs of Angelman's (e.g. he started having seizures at aged 4). However, we are fortunate to have a fully mobile child who is making great progress in his school with communication aids, toileting, self-help skills etc. Joshua surprises us and his teachers with what he seems to grasp and be able to achieve. He is also, like all 'angels' a generally happy child who loves life and people.

Having passed through the initial pain and coming to terms with his diagnosis and prognosis, we felt that we would like to do something to support ASSERT (who's telephone line was invaluable in those early months) and to raise awareness of this little heard of Syndrome. Consequently we have organised and participated in 3 events where we were able to address approx 400 people and tell them about Josh, the Syndrome and the work of ASSERT. The first was a village cabaret where all the raffle proceeds (£500) were given to ASSERT. The second was a classical Easter Concert where Fran and others performed to a sell out audience and raised £1453.58. An additional donation of £238.00 was also received after the event. And finally a choir concert in Milton Keynes raised £212.60 at the end of April.

Many who attended these events said that they learnt something about disability and Angelman's Syndrome and others said that they would like to do some fund raising for ASSERT in the future. Great News!!

So thank you to ASSERT and all those who work so hard to support families like ours.

If any readers would like to contact us our address is in the directory. We would especially like to hear from parents of children with UPD.

Finally we have various 'musical' contacts that may be willing to perform at any other events that members may like to organise in the Midlands area (including Fran).

**Andy, Fran, Joshua and Oliver Childerhouse**

*Many thanks to the Childerhouse Family – they emailed me (Sally Walburn) all the bits and pieces about the concert and I could clearly see that it took a large amount of preparation and organisation to 'stage' the event, so well done and thank you for all your hard work, what an achievement!*

### Contents of the Newsletter

You will notice that the pieces in this newsletter are drawn from a wide source of places. This is tremendous, and we hope you will agree that it makes interesting reading. However, while we are keen to promote discussion and to pass on many views and experiences, it is also important to appreciate that the opinions and views expressed by contributors to this newsletter are personal ones and not necessarily those of the trustees of ASSERT.

**Please note that the  
deadline for the  
next edition of the  
ASSERT Report  
is 15 September 2002.**



### New Website!

Have a look at Paul Chick's new website – this will be of interest to all of you regarding problems with your child being able to sleep in a normal bed.  
[www.chickies.co.uk](http://www.chickies.co.uk)

### Error in the last Report

I apologise for the error on page 6 of the last Report about the Personal Passport, the last sentence was missing! For those eagle eyes that spotted this mistake and want to know what the last bit was, here you are!

"It's worth either encouraging your school to adopt a similar idea or do one yourself – believe me it can save the heartache when you are confronted by tactless people and questions that you don't want to answer."

**Sally Walburn**

**HELPLINE**  
**ASSERT Families/New**  
**Diagnosis/Support Line**

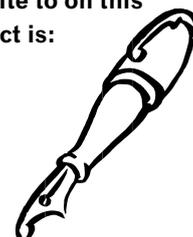
**01268 415940**



**If you enjoy these articles  
please think about writing one  
about your family, we really find  
this the most interesting part of  
the report.**

**The address to write to on this  
or any other subject is:**

**ASSERT  
P0 Box 505  
Sittingbourne  
Kent ME10 1NE**



# Families Stories

## George and Chloe Darlow



George, Chloe and friend

Are there any parents that have an Angelman Child who has a brother or sister with special needs too? We have two children George and Chloe. Chloe who is almost 3 who has Angelman Syndrome and George who has a left sided hemiplegia. We would be very interested to know if there is any link between our children or is it just coincidence.

Chloe is a very happy contented little girl with quite a sense of humour. By this I mean, deliberately turning her bottle upside down and shaking it so the contents go all over her and her chair and the rest of the kitchen. When you say to Chloe "drink it nicely" you are greeted with a huge grin and chuckle and Chloe continues to tip her bottle. Is Chloe doing this on purpose I wonder as she obviously gets a reaction every time?

Actually talking about bottles, Chloe finds feeder cups that are on the market impossible to use and has now started to make very large holes in the teats, which are not Chloe proof. Or maybe this is because we need new teats. Anyway, we would be really interested if this is a common problem and would recommend some helpful tips. Chloe's sucking reflexes are quite poor.

Chloe loves her balloons and makes us cringe at what the balloon has to be put through by Chloe especially by being squashed, squeezed, sucked, and dribbled on and bit. I think when the balloon has finally had enough it pops.

Chloe will be starting Nursery after Easter for 2 mornings, which is very exciting. We have been a couple of times and taken Chloe with us and she was off straightaway into the water trough and sandpit, albeit eating the sand.

Chloe is now able to stand at the furniture and move along, but the best way of moving is at lightning speed by crawling. Is there an Olympic event for crawling?

I think one of Chloe's favourite past time is getting to the toilet when George has not closed the door and Chloe seems to think the water in the bottom of the toilet is great. I must say

this does not happen often. I can imagine you all reading this and thinking surely mum is not letting her child play in the toilet. Honestly, I don't. I think another job is a toilet lock but then I can see it now. George struggling to open the toilet seat and just managing to pull his trouser half way down, which is a huge achievement for George and having an accident as we struggle to get the lock off.

Sorry I must stop nattering I could talk for England.

Let me just tell you a little about our George. George had a stroke when he was 21 months old and developed Epilepsy although this is well controlled at present. It is now becoming very obvious that he has other difficulties i.e. Dyspraxia, Behaviour difficulties, speech and language to name a few. Although with careful planning each day and routine George copes much better.

George loves to be outside in the garden and helping dad do DIY and going out on his trike.

I would like to tell you about our sleep that never happened in our house until very recently. We continually got up at least 7/8 times a night for George and Chloe came a close second 6/7 times and obviously all at different times. My husband Adrian and I would bed hop and often ending up sleeping with George to get some sleep. This has only been the last 5 years with George and 2½ of those years for Chloe.

Chloe would need a bottle to settle as this appeared the only way to get her back to sleep but we then had to stay awake until the bottle hit the floor. Then Chloe would be wet and her cot so it was a complete change of bedding etc. Chloe must of thought it was the beginning of the day. George was physically sick if we tried to leave the room and would climb over his stair gate and come up and down stairs all night. We thought things would never change and thought Chloe was following the normal pattern of not much sleep and thought George was unable to sleep because of his difficulties. It was a viscous circle until after finally trying medication prescribed by our paediatrician to help George sleep over a 2 week period which still did not make George sleep and if he did it would be only for a brief time and we would be back to square one.

We had tried so many things with George and at Christmas we decided that we were not happy about given any medication that was unnecessary but we were desperate. We then spoke with our health visitor and she said we could try a sleep program, which was initially for George to try and find out why he could not sleep well. So at the beginning of January 2002 we were very focused and raring to go and decided to do Chloe at the same time. Were we mad! No.

We stopped Chloe's bottle completely and this made Chloe very upset as she had learnt her bottle was her comfort to sleep. So we had a job to break this comfort and replaced the bottle with one special cuddly, which is kept just for bedtime. After about four weeks and lots of tears from us all Chloe was able to sleep with less fuss as each night progressed until now we have a little girl that goes to bed at 7pm until about 6am which is fantastic. We have had odd relapses when Chloe has been unwell but she has soon got back into the routine.

Well as for George it took a lot longer as we needed to do a

# Families Stories

very gradual approach and it took us 4 weeks to get to his bedroom door and another 3 weeks to sit outside his room and then 2 weeks at the bottom of the stairs and now we are in the lounge straight away after a lovely story and hug and a goodnight, mummy will see you in the morning.

Both Adrian and I took it in turns to have a different child each night, which was great for us to stay focused, and Chloe and George are happy that turns are taken and again keep them in a routine.

I just want to let any parents out there know that with a lot of dedication and hard work our sleep program really worked. It was hard but the rewards are amazing both Adrian and I can make a cup of tea, talk, read the paper, which was impossibility before. One really important message is that we have a good

routine after our evening meal when the house is quieter and we have no music dancing etc just quiet time and it almost appears to prepare George and Chloe that sleep is the next stage. I hope this may give other parents inspiration and would be happy to discuss in more detail if you would like me to.

I will write again soon to keep you all posted on our progress.

One final message, although George and Chloe are very demanding they are also a joy to be with especially George and his funny personality, I am sure he will be on the stage one day and Chloe just gives you that gorgeous smile and it makes everything worth while.

Bye for now

**Jennifer Darlow**

## More new members!

*The next piece is from Diane Box and introduces her son Scott Cooke.*

I have just joined ASSERT as I have just found you. I did ask doctors and health workers if they knew of a support group but always got the reply "we don't know as Angelman's is very rare". They did give me a group a few years ago but sadly it was no use to me, they did not appear to be very helpful unlike ASSERT.

Scott is 10 years old and is a twin. He was not diagnosed until he was six years old. I was told I would probably never get a "label", their words, as many parents didn't but I just kept pushing much to the annoyance of everyone around me at home and in the medical sense although Scott's consultant often seems to be amused by me!

Scott's diagnosis came from a change in the gene causing Angelman Syndrome. He does not have epilepsy much to my relief as I have it and know how it feels.

Scott has a twin brother Nathan who does not have AS or anything else, he is just like any other 10-year-old boy. Scott also has another brother who is 19 called Stuart – he is only home during holidays as he is at university. There's me, Diane, and his dad, Andrew. He goes to a wonderful special school where the headmistress is fantastic. He likes school and I must admit I dread the school holidays (Editor's note: who doesn't?!!) and when his dad comes home from work I am shattered. He's very hyperactive; I can't keep up with him. He is very pleasant most of the time and has a smile that can

light up a room, and a very infectious laugh. He can twist anyone around his finger – mainly his older brother and me.

However, he can have a temper, and he can be very stubborn and very strong, he will do what he wants to do, whether I like it or not – things like breaking the fridge lock off the door (we have had 3 different types up until now) because he wanted another jelly and nothing was going to stop him. He has worked out every cupboard lock I have bought mainly if there are biscuits or chocolate inside them! He learnt to climb over the safety gate at the bottom of the stairs, we raised it so he went under it and got stuck. His dad had to take the gate off the wall to get him out. He loves water and once in the middle of the night got in the bathroom, put the plug in and turned on all the taps. I woke up to the sound of running water, luckily the bath wasn't filling (plug wasn't quite in properly) but the sink was overflowing on the floor and Scott though this was very funny.

Looking after him is hard work but there is never a dull moment. Like all other Angelman parents I have laughed, and cried, shouted and got very frustrated at the medical profession when having to fight for Scott's rights like getting regular speech and physio therapy (which he is not getting). I am always tired, Scott is with us 7 night a week, he won't sleep alone, but the laughter and love we get from him more than enough compensates for it all. It is nice to know there are parents like ourselves and finally I believe that we belong somewhere, it's a little like joining a family.

**Diane Box**

**HELPLINE**  
**ASSERT Families/New Diagnosis/Support Line**

**01268 415940**



# Families Stories

## The Most Dynamic Guest at Our Party!

*ASSERT always tries to keep up with our other AS families in other parts of the world. Jackie Golden has agreed that we can print the following article about her son Joshua who is 20 years old. Jackie and her husband set up a scheme whereby Joshua lives in his own home but close by with his own carers. This is what she wrote to the Angelman Listserve back in February.*

Had to share this. You know things are going right and that you have the right supports for your child with AS when you have a night like I had Saturday night! Yesterday, my daughter threw my husband and I a surprise 25th wedding anniversary party. It was truly a surprise, and she got this over on us by telling us that it was a surprise birthday party for my mother. Mike (my husband) and I had the job of keeping my mother out until exactly 6:30PM, then when we walked my mom into the party room at her apartment, we were floored when everyone said "Happy Anniversary, Mike and Jackie!"

Anyway, one of our guests truly stood out! It was the young man wearing the khaki pants, the grey button down collar shirt and a catchy cool sweater, docker type boots and a caesar hair cut. This guest had a beaming smile and just jumped for joy when we walked in. This guest was none other than our handsome young son, Joshua!

Like all of the guests Joshua was included in the fun. Joshua came to the party with three support staff, all who are viewed by us as an extension of our family. All three of Joshua's friends made sure that he looked 20, was sitting in the middle of all the excitement and had a place at the celebration. Here is what was really amazing! Joshua, although he followed his DAD (Joshua is definitely a Dad type of guy) around the large party room with those large brown eyes of his. Joshua sat on the sofa with plenty of tempting decorations, glasses full of drinks, plates full of food and he did just that – SAT there! I looked over at him several times during the evening and saw him being served a huge plate of food, several drinks and even witnessed Joshua holding out his cup for more drink! There was a live band at the party and Joshua got up, with support, walked over to the band and just beamed at the loud sounds coming from the group. He was in his glory!

My daughter had made sure that Joshua was fully included in the events, had lots of his friends surrounding him and made sure that Joshua's Mum and Dad, did nothing in the way of Joshua's care!

After Joshua had his fill of food and drink, had made the rounds of seeing the others guests, listened to some great music, I noticed that Joshua looked a little tired and his support network noticed as well, they said their "good nights" took and dressed Joshua in his coat for the ride home. Joshua came over to say good night to the guests of honour, Mum and Dad, and off they went. Off to Joshua's house to get ready for bed and get a good night sleep.

What made me write this today to all of you is that the evening we had with Joshua came about only by making the right connections for supports. It came about not by accident,

but by making sure we worked hard to get the right things in place for Joshua's future. This doesn't mean that throughout Joshua's life it has always been this way, but I want to make sure everyone on this list knows that it is possible for our children to grow up, to live in their own home, to have people in their lives that care about them as much as we do as parents. That our young adults do have lives to look forward to, and we have lives as well.

We talk about "placement" for our children, when what we really should be talking about is planning and contributing to our children's future. Like for instance our family home. This is the home that we purchased because of Joshua. Close to schools, close to his doctors, close to stores, bus routes, good neighbours, friends and family. So when it came time to truly solidify the future plans for Joshua, it made sense for Joshua to remain in this home and for us to move on. So we gave the home to Joshua. We are not rich, by no means; we are a simple middle class family with a mortgage still on the home. Yet we found that by planning we could make a home for Joshua and for ourselves. We rent the home to Joshua and his roommate, which is enough to pay the mortgage. We receive the Home and Community Based Waiver services to pay for the support staff. Yet, we partner with this system of ours giving Joshua to best of all worlds, we oversee his supports through a microboard. This microboard includes his sister, his parents; one of his friends that has disabilities and several other friends that believe Joshua has a full right to an enriched life. Oh and yes, Joshua is also a part of the microboard.

This microboard (self-directed support corporation) is the support network that every parent dreams of for the child with special needs. Custom designed for Joshua, supports custom designed for Joshua, staff custom selected for Joshua. The SDSC is designed to continue even in the event of Mike and my death. Joshua's life will go on and be enriched.

So folks, when I watched that dynamic young man at my party, beaming from ear to ear, looking like any other guest there, taking in a bite to eat and a drink of sprite, it dawned on me that others need to know that there is a way to have a life and for our children to grow up and have a life as well.

I will tell you that before I opened that party door, I wasn't even thinking about Joshua. I wasn't worried if he was having a good time, or if he had his medications or evening if he was out for the evening. I knew before I opened that party door that Joshua was well cared for, he had plans, and he has a life. I opened that party door and there was Joshua living his life and he happened to share that beautiful evening with his parents! We are included in his full and enriched life. He truly was the most dynamic guest at our party!

Take care all,

**Jackie (Joshua 20)  
Baltimore, Maryland**

# Families Stories

## Cheeky Monkey Syndrome?

Introducing Leo Trigg D.O.B. 15.05.00



Leo, a cheeky monkey?

Leo's dad and I have been impressed and moved by the family stories in the ASSERT newsletters and decided it was about time we wrote to introduce Leo and I guess, tell his story so far.

Leo was diagnosed with Angelman syndrome in Haringey, London when he was a

year old. This followed our health visitor's referral to the Child Development Centre when Leo was 7 months old. We also suspected something but as first time parents we were constantly being told that children develop at different rates and that he would 'catch up', and we wanted to give him the benefit of the doubt. At Leo's Assessment the consultant said it was a unanimous decision that Leo had Angelman's, but blood tests have since ruled out deletion, UFD and imprinting defect, and his EEG showed abnormal spike waves not typical of Angelman's.

His diagnosis was a huge shock, which all newly diagnosed parents must go through, we simply had no idea that this gorgeous little boy would have problems as far reaching as they were saying. As the shock wore off and I found ASSERT's help line number gradually things started to fall into place especially with the help of Sally Walburn who I am indebted to for her practical advice in those early weeks.

We gathered lots of information, were particularly impressed by the ASSERT leaflet so much more positive and upbeat, than the, CaF directory photocopy we had been given at Leo's assessment. We quickly discovered that a whole other world existed and that we had simply opened the door a crack, (still have only partially stepped through) In October 2001 we finally moved to Leeds and since then I am glad to say provision and support for Leo has been excellent.

We quickly were given an appointment to see the consultant Paediatrician because of the information already sent to him from London. We have already had another assessment, and been put in touch with all the services Leo will need health wise and many of the educational ones as well. However, since coming here all of the professionals who have seen Leo, Consultant paediatrician, geneticist, and

neurologist have all doubted very strongly the Angelman diagnosis. We now feel as if we are back in 'no man's' land but are prepared for the fact that we may never find out what exactly is wrong with Leo. The next step is a brain scan and he is on the waiting list, along with eliminating other conditions which can be tested for. The geneticist is reluctant to test for UBE3A because there are not enough Angelman indicators present, so we wait to see the results of other tests. What we do know is that Leo is a charming, cute, loveable little boy with a sense of humour all his own and that we love him beyond all comprehension. He loves water, swimming, going in the bath, and even washing his hands. He loves playing with his toys particularly musical ones and he loves listening to stories. Food is also a great love as he becomes more aware of what different packets contain. (Crinkly packets are a particular favourite as well as purple foil, and fromage frais pots!) His greatest love at the moment is exploring, no new door is left un-banged, no new plant pot left un-soil removed! He has just mastered walking and he thinks he is so clever when he crosses the room purposefully stopping two paces away and falling into your arms, with a huge cheeky monkey grin on his face !! His major problems concern his language which has not developed beyond 'Ah' (sometimes very loudly, and usually in supermarkets!!) his understanding of and making simple connections between simple things like eating food off a spoon, which he doesn't connect with loading up again. He also struggles with fine motor skills; picking up small things takes a lot of concentration. (The dog has eaten more bits of Easter egg than Leo as he has dropped so many on the floor!) He is also epileptic. His seizures happen in the early hours of the morning and cause him to fit rigidly with his eyes open, breathing very lightly. This goes on until we can get him to a hospital for anti convulsant medication stronger than Diazepam. Facially he is not typical of Angelman's, he is not hypopigmented, and he is not ataxic, his balance is good, in fact he is not typical of anything just a lovely and very special little boy. So why am I writing an article for ASSERT when we are now being told Leo does not have Angelman's? Well really because we are hugely grateful to ASSERT for helping us throughout the tough times and for accepting us into its family whether we fit there or not. We just feel glad that we can latch ourselves onto a support group such as ASSERT, because we haven't yet found one for Cheeky Monkey syndrome! I know because I have talked to Sally that there are other families who are members of ASSERT with children who are as yet undiagnosed and it would be good to hear from them especially if their children are similar to Leo. We are also looking forward to the conference in September and meeting other families. Until then, Thank you ASSERT you are doing a very valuable job.

**Jackie Rose, mum to Leo.**

# Learning Disability Helpline – 0808 808 1111

The first national Learning Disability Helpline in England – set up with a £750,000 three-year grant from the Department of Health with match-funding from Mencap – has been launched.

The helpline, provides a much-needed 'one-stop shop' information and advice service on a wide range of issues – including support, welfare benefits, health, housing, learning and employment – for people with a learning disability, parents, carers and social care professionals.

The Learning Disability Helpline will operate via Freephone 0808 808 11 11 from 10am to 4pm, Mondays to Fridays, with a

round-the-clock answering service. All calls are confidential and free. Contact can also be made 24 hours a day via e-mail, website, post and fax. From April, the telephone service will be extended to outside normal office hours.

The setting up of a new national helpline targeted at people with a learning disability and their carers in England was a key objective of 'Valuing People', the Department of Health's Learning Disability White Paper, published in April 2001 – the most important milestone for people with a learning disability in thirty years.

## British Epilepsy Association Launches Epilepsy Action

On May 1st 2002 Epilepsy Action is being introduced as the working name for British Epilepsy Association. With a new logo and a striking new colour scheme, Epilepsy Action aims to raise the profile of this often forgotten condition.

Epilepsy is the UK's most common serious neurological condition affecting 450,000 people but there are still too many myths and misunderstandings surrounding it. The vast majority of people with epilepsy (75%) have their seizures controlled and are able to work, drive and lead a fully active life.

Epilepsy Action is the largest member led epilepsy organization in the UK with 22,000 members. As well as campaigning to improve epilepsy services and raise awareness of the condition, we offer assistance to local people in a number of ways including a national network of branches, accredited volunteers, regular regional conferences and a Freephone Epilepsy Helpline

(0808 800 5050). Epilepsy Action also provides a website <http://www.epilepsy.org.uk> which is the most frequently visited epilepsy website in Europe.

Philip Lee, Chief Executive of Epilepsy Action said: "We are very excited about the new look. Epilepsy Action is a bright and modern approach which puts epilepsy at the forefront and will help us in our vital work. People tend to forget about epilepsy, but our members live with it every day."

### Questions About Epilepsy?

Visit our website: <http://www.epilepsy.org.uk>

Email the Helpline: [helpline@epilepsy.org.uk](mailto:helpline@epilepsy.org.uk)

Phone the Helpline: 0808 800 5050 (UK),

+44 113 210 8850 (International)

Published by Epilepsy Action, <http://www.epilepsy.org.uk>  
New Anstey House, Gate Way Drive, Yeadon, Leeds LS19 7XY  
UNITED KINGDOM

Epilepsy Action is the working name of British Epilepsy Association

## Obituary

*From time to time we receive donations from families who have lost a loved one and these are given in lieu of flowers to ASSERT. Occasionally we have written about the person at the request of the family. The following was received from the Taintourier family who live in France, Lauren (nee Frith) is English and her father passed away recently. Many thanks to the Frith family for the donation to ASSERT. Grandpa will be greatly missed.*

I'm English but my husband is French and we live near the Belgian border. We have three children – Jade 11, Luke 8 and Anna 5. Luke was diagnosed as having AS at 8 months. Of course we know the trials and tribulations of all parents of AS children. Luke now goes to the local special needs school which is far from satisfactory but the choice in this region is severely limited. We nip over to England at least twice a year and I'm always interested to read your newsletters.

### In memory of Sydney Frith

Grandpa of Jade, Luke (AS) and Anna. Ever inventive, he made

stools for Luke

when he was

learning to sit,

trolleys to help

him to learn to

walk and was

too big for

shop-bought

ones, special

pedals to

keep his feet

on bike

pedals and

was

generally

good at

generating

bellyache

laughs by just

sneezing or throwing

his slipper in the air.



Luke with Grandpa

# The Family Fund Trust

The Family Fund Trust was set up by the Government many years ago to help families of disabled children, or where children had high care needs because of an illness, and where the families were on a low income. The help that the Trust can give is very wide, and covers things that families can't get from any other source. For instance, the help can include things like a washing machine; tumble drier, play equipment, travel costs, Holidays, and even driving lessons for a carer.

The Trust has to set a financial limit for an income, and this Year it is £20,300, but benefits such as Disability Living Allowance are excluded from the calculation. The income level goes up every year.

If a family makes an application for help, a local Family Fund Visitor comes to see the family, and together they discuss the help requested by the family, and the affect that the child's disability is having on the family. The Trust is committed to making life better for children, and even if a child's disabilities do not come within the Trust's remit, a visitor can usually provide a lot of information that is useful.

The visitor then sends a report to the York office where decisions are made. The family is told the decision by letter as soon as possible. Sometimes, in order to clarify issues around the child's disability, letters requesting further information are sent to the child's school, or to the doctors who know them best.

Once a family has received help from the Trust they can apply again after 12 months. The only exception is that if the family are in desperate need of a washing machine, they can apply again before the 12-month limit is up. Families should not see the work of the Trust as providing charity. The Trust is fully funded by Government, which has given it the responsibility of providing practical help and targeting it where it make the most difference for families.

If you would like an application form you can contact the Trust on 01904 621115, or simply write to them at Family Fund Trust, PO Box 50, York, YO1 9ZX giving full details about your child and his or her disabilities or illness.

## Research

For those with Internet access Dr. Dan Nolt has sent an email on research he is conducting into the use of anti convulsants for epilepsy. If you can participate, please do, all the information is as follows:

I'm a Pharmacist at Children's Hospital Central California working with Dr. Terry Hutchison. We put together an Internet questionnaire regarding how parents and caregivers of persons with Angelman syndrome feel about the effects of antiepileptic medications on their Angels. It can be found at: <http://www.childrenscentralcal.org/angelman.asp>

It is a very brief, confidential, questionnaire not affiliated with any pharmaceutical companies. We hope to post the results of the study on the Internet once completed.

Sincerely,

Dan H. Nolt, Pharm.D.

Paediatric Pharmacy Resident, Children's Hospital Central California, 9300 Children's Place (GW19), Madera, CA 93638 U.S.A

e-mail: [dnolt@childrenscentralcal.org](mailto:dnolt@childrenscentralcal.org)

## David Kinnear

David is our newest trustee and we are very grateful to him for agreeing to serve on the Board. As you can read he brings a wealth of experience and we look forward to working with him in the future.

Born 9th April 1938 at Aberdeen.

### Career

Entered the Army at the age of 18 and retired in 1993 in the rank of Brigadier. Now self-employed and work from home as a Law Costs Draftsman.

### Decorations

Appointed OBE in the New Years Honours List 1982.

### Family

Wife Margaret who trained as an SRN and then took additional qualifications specialising in Children's' nursing. She trained at the Birmingham Children's' Hospital. Margaret returned to nursing some years ago and retired as a Community Nursing Sister in 1995.

Daughter Lindsey. Three children Imogen (8), Alice (6) and Isabelle (3). Isabelle was diagnosed with AS at the end of 2000. Married son Alastair. No children. Bachelor son Andrew. Married daughter Morna. One child Thomas (3). Expecting second child in October.

### Interests

Was keen mountaineer/rock climber until advancing age made such pursuits a touch on the silly side. Keen runner until about 10 years ago. Have completed 4 marathons. Took up sailing when climbing faded out of the picture. We sold our boat last year when a combination of work and the residual effects of a mild stroke 3 years ago made it all too difficult. Very keen DIYer – with reasonable success! Trustee and member of the Executive Committee of a large Army Benevolent Fund. Chairman of a local committee which organises an annual activity week for some 70 – 80 children. Active in local Church affairs.

### Major current activity

Avoiding mirrors and trying desperately to lose excess weight. All suggestions gratefully received!

## Purchasing New Cars

We have been looking into purchasing a new car as our current vehicle, a Ford Galaxy is under the Motability scheme and up for renewal in September. We have found that the rules for Motability have been tightened up and some of their new strategies are quite harsh. Hence we wish to come out of the scheme. It has been an interesting debate as to what to buy and we have found that we no longer need a 'people carrier' as our other children are not in the car so often these days – teenagers – need I say more!! And Matthew is getting more difficult to lift up and into the Galaxy (he still is not walking and is almost 11). We have decided to buy a new car but something that will be easy for him to access and also room for a wheelchair.

Under the current VAT rules, if you purchase a new car with adaptations, i.e. a hoist for a wheelchair, the whole car purchase is exempt from VAT – this is well worth bearing in mind as this represents a substantial saving on the purchase of the vehicle and also may mean the difference between having the car you want rather than the one you can afford, or between a new or second hand car. **THIS ONLY APPLIES TO ADAPTED VEHICLES.**

However a word of caution – you have to follow a sequence and if you don't then you would be liable for the VAT on the car but not the adaptation.

- 1) You must register the car in the child's name before the adaptation.
- 2) The adaptation must be carried out **BEFORE** you buy the car
- 3) The dealer must pay for the adaptation (including VAT which they can recover)
- 4) The dealer must add the cost of the adaptation excluding VAT to your total invoice as a **SINGLE** transaction
- 5) You must sign a declaration form for the VAT office

If you purchase a vehicle and then have it adapted this is not acceptable, you will only get VAT exemption on the adaptation.

For confirmation of information, please contact the VAT office.

**Sally Walburn**

## Spanish Conference (April 2002)

First at all I am very grateful to Sally Walburn for giving me the opportunity to share with all Angelman families in the U.K. the summary of our first Spanish Angelman Conference.

The Conference was held over two days and we shared deep feelings with many Angelman families. It was for many, the first time we met face to face, sharing experiences about our children, learning about Angelman syndrome with our specialists.

Our speakers were:

- Dr Elisabeth Gabau (paediatrician and geneticist) (Parc Tauli-Sabadell Hospital)
- Dr Jaume Campistol (neurologist) (Sant Joan de Déu University Hospital)
- Dr Josep Artigas (neurologist-paediatrician) (Parc Tauli-Sabadell Hospital)
- Dr Isabel Lorente Hurtado (neurologist-paediatrician) (Parc Tauli-Sabadell Hospital).
- Dr Carmen Brun (psychologist, speech pathologist) (Parc Tauli-Sabadell Hospital) and lecturer Psychology Faculty University Barcelona).
- Dr Conxita Escofet (neurologist-paediatrician) (Parc Tauli-Sabadell Hospital)
- Emili Soro-Camats (speech pathologist) (specialist in Alternative Communication)
- Pilar Ribera Villanua (Social worker – Paediatrics Services) (Parc Tauli -Sabadell Hospital)
- Dr. David Poyatos Andujar (biologist – Cell Biology Department Barcelona University).
- Cristina Camprubí (biologist – Cell Biology Department Barcelona University)
- Dr Miriam Guitart (biologist – head of department of Genetic and immunology) (Parc Tauli-Sabadell Hospital).
- Dr Jose Ignacio Lao Villadóniga (doctor and geneticist) (administrator of Molecular Medicine in Dr. Echevarne Laboratory).
- Inmaculada Garrido Jiménez (special teacher education)
- M<sup>a</sup> Teresa Lloria i Yacer (general manager of hippotherapy) (Riding for the Disabled centre)
- Laura Segovia Encimas (occupational therapist)

The Conference was closed by Jill Clayton-Smith.

The subjects included, diagnostic clinical criteria, development, neuro-motor functions, epilepsy, sleep problems, language, phenotype behaviour, associated medical problems, alternative communication, school and family, social necessities, genetics in Angelman syndrome, diagnostic strategy, genetic counselling, family presentations and finally an overview from infancy to adulthood.

I would like to emphasize the importance that every one of the speakers tried to explain their presentations in an easy format, with a comprehensible language for parents. The biologist presentations were great, because for the first time, many Spanish Angelman families were able to understand the "mechanism" of what caused their child's syndrome.

The families had lots of facilities for their children, even childminders in order for parents to attend sessions without problems. There was a great programme for children too and the last day we had a lunch with the children, childminders, some of the speakers and families all together.

At this moment we are working hard to edit all the presentations on our Spanish website, Jill Clayton-Smith's presentation can be read in the English version too.

It struck us that when a group of Angelman parents are together you feel like we are part of the same family.

I am sure all Angelman families in U.K. will enjoy like us, your next Angelman Conference in September.

Thanks again for this opportunity and good luck with your Conference.  
Love to everyone from Angelman families in Spain.

**Maria Galan – Elena's mother 18 (+)  
Madrid – Spain**

# Telephone Support Line

For the moment we have streamlined the support line into one new number, which is 01268 415940. This will also accept faxes. Please do not use the old numbers.

We are still providing the 'On-call' service, which is not limited at the moment. You can still contact ASSERT at any time. Instead of trustees on call at different times we are currently monitoring the volume of calls and you will get through to Sally Walburn. Of course if you wish to speak to a particular trustee, Sally will pass the call on and get them to phone back you as soon as they can. You will at times get the

answer machine, please be patient and leave a message. It is vitally important to leave your details clearly and also a time which is best for you to be contacted. As before we are here to share your frustrations and at times, your despair, but also the good news too! ASSERT is not able to 'fight your battles' for you but we can offer support, a listening ear and we will endeavour to point you in the direction of further assistance.

You can contact any of the trustees by letter or E-mail: [assert@angelmanuk.org](mailto:assert@angelmanuk.org).

## HELPLINE

**ASSERT Families/New Diagnosis/Support Line**

# 01268 415940

## Contact Register Updates

Every year we like to update our records. Many of you let us know when you change address and telephone number, but we like this confirmed each year in order to produce an up to date version of the Contact Register.

We will be sending out forms later this year and it would be appreciated if you could complete and return them EVEN if there's been no change.

## Donations

Thank you, to everyone who has donated to ASSERT since the last Report back in December, your contributions no matter how small all make a difference, without you, there would be no ASSERT and we know what a lifeline we are at times. We also collect foil, cans and stamps; stamps must be trimmed as close to the perforations as possible (don't trim those or the stamps will be no use). Please send stamps to Jim and contact Bernie Silver concerning the details for our account with Alcan and recycling aluminium.

Many thanks to:

Balham BA Sports & social Club

Mr & Mrs Bell

Ms Buchan

Ms Chiapinno

Mrs J Darlow

Mrs L Davies

Mr & Mrs Donnarumma

Finnigan's Pub (via D Kinnear)

Dawn Frazcek

Jenny Gray

A S McGinley

A K Nunn, Masonic Lodge

Stephen Penlington

Slimming World Sittingbourne

Mrs G Slowcroft

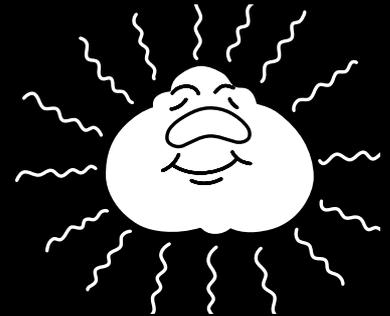
South Kilworth, village Hall Committee

South Kilworth Masonic Hall

Statonbury Chamber Choir

Stockwood Medical Centre

In memory of Dawn Surman, nanny of Ben, Luke & Ross Yates



## Holidays

It would be very helpful if you have any articles, information, or practical experience on holidays that you have loathed or enjoyed to share with our readers. It is always a challenge to find a holiday suited to all the members of the family. Please send your ideas and info to Sally Walburn who will be compiling a file on holidays which we can all use as a resource base for future reference.