ASSERT

Angelman Syndrome Support Education & Research Trust

ISSUE THIRTY FIVE MARCH 2003 **REPORT**

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REGISTERED CHARITY NO. 1021882

Letter from the Chairperson

I usually write the letter from all the trustees but this time I thought I would be a little more personal. As I write there's lots going on at home and abroad – as we all know we are poised on the brink of a war with Iraq and I guess we all feel powerless and apprehensive.

Life to some extent is like that for us every day as we care for our children and young people. So often we are made to feel as though we have to put up with what is being given, and not cause a fuss and be grateful for what we have. However, we hope you do feel empowered and informed when you read the newsletter and also stories from other families. I have just battled with services to get a wheelchair provided for Matthew by the Royal National Orthopaedic Hospital in Stanmore. We attend there since Matthew has been diagnosed with Scoliosis. We secured funding from our local authority to pay for this service and the chair, but the reality is that Stanmore cannot physically take on any more clients from out of the area – we cannot access their expertise, even though we want to.

Good news – Matthew's curve has reduced by one third since wearing his brace and our respite has been increased – wonderful! I realise that some of you may not have as much as us, and some much more so won't depress you with the amount – suffice to say it's not that great but better than we had.

ASSERT is doing well. We have booked the conference for September 3/4/5 2004 at Loughborough University again. Put that date in your diaries now! And start planning your finances...we expect the cost to be roughly the same and all AS persons and one carer to attend free. However, if we can access grant money for parents to attend or to pay for the conference itself, we may well be able to reduce costs – so watch this space!

Jim will be standing down from ASSERT at the end of the year as planned. We are pleased to inform you that Margaret Kinnear has joined her husband David as a new trustee. Michelle and Alistair Turner have also expressed an interest in becoming trustees so we look forward to welcoming them on board. Hopefully we will be able to disseminate Jim's role amongst us all and thus keep things going much the same. However, I have noticed that I am becoming increasingly jaded when it comes to the newsletter and could do with a break from it – is there anyone out there who can give me a break? If not then I may well reduce the number to two or three a year depending on the time I have.

Sorry, but this is voluntary as you know and life in our household is as hectic as yours!

I am pleased to be travelling with Finn Emmerson and Pam Robertson to Denmark to speak to the Danish Association at a meeting on 29 March 2003; the meeting is based around Communication for people with Angelman Syndrome. This is wonderful, since it's great to work together as organisations and having met with Kristian and Vibeke Nicolaisen in Ottawa in 1998, we have kept up our relationship, Vibeke and Kristian came to our conference back in September 2002.

The World Conference as you know is scheduled for July 1 – 5 2003 in Washington DC as a joint venture with the ASF. Details can be found on the website www.angelman.org for the time being it will still go ahead but depending on the Iraq situation, decisions may have to be made as to the viability. That will be up to the organising committee.

Regional Meetings – these are planned throughout the year and if you are in a region, which is hosting a meeting you will be informed. To date there are meetings planned for the Eastern Region/ South West Region/ Scotland/ Midlands and possibly the South East. If anyone can help with a meeting in the north of England, that would mean we will have virtually covered the country this year. If you can help, please contact the ASSERT helpline number 01268 415940.

Videos – we have four videos from the conference. They have not been edited, as this is a huge task and will involve extra cost and time to ASSERT. However, families are welcome to borrow them as they are, to fast-forward through and see what we did. The topics included Jill Clayton Smith, Behaviour, Seizures, Communication and Sleep. As I say, they are in a 'raw' form but may be of interest. If you would like to go on the list we could set up a rotation so that the videos can go from one person to the next instead of coming backwards and forwards to us. If you would like to see them please contact me, Sally Walburn by April 30 and I will draw up a list of viewers.

For now that is everything, please think of Ken and all our runners as they tread the streets of London on April 13 and if you can sponsor us so much the better!

Sally Walburn

The following information has been prepared by Finn Emmerson (our affiliated Speech and Language Therapist) and hopefully will be of interest to those who are thinking about or who have communication aids for their children.

British Education Communication and Technology Agency – (BECTa) Communication Aids Project

The communication aids project will run over two years; from 1st April 2002 until 31st March 2004.

The funding is intended to support LEA and school funding by providing equipment and technology for students with significant communication difficulties. It is not meant to relieve LEAs and schools of their obligation to meet their students' educational needs and consequential incurred financial support.

For children who are pre-school aged, referrals need to state current funding arrangements, so creating a starting point. This may change as the child starts school and their statement should clearly state the equipment needed and the funding source.

To qualify for Becta funding, the following criteria must be met....

- The student must have an identified communication difficulty.
- The student must be receiving education in a maintained school / non-maintained special school / otherwise educated.
- The LEA or school must show they have taken appropriate action in meeting the student's needs, prior to applying for funding.

Becta funding provides;

- Assessment
- Equipment
- Training

LEAs, schools and other agencies should fund;

- Technical support
- Maintenance of equipment
- Insurance

Becta has identified six CAP centres through which the project has been organised;

- ACE Centre North
- ACE Centre Advisory Trust
- Cenmac and The Wolfson Centre
- Abilitynet
- SCOPE
- DCCAP (Batod and Deafax)

Each CAP centre is responsible for identifying satellite centres / teams in their region, these are known as CAP teams.

A student is referred to Becta by using the application system on-line found at the following website address; www.becta.org.uk/cap_referral_forms

Applications are then sent to the appropriate CAP centre for the student's region. The CAP centre will identify the appropriate team or individuals who will assess the student. Those teams or individuals will advise Becta of the student's needs and apply for funding equipment.

Further information can be found on the Becta website www.becta.org.uk

Finn Emmerson
Speech and Language Therapist

Angelman's syndrome and IVF – is there a link?

Dear Parents,

We are researchers with experience in studying children conceived by IVF. We have contacted many of you through ASSERT to ask you to help us with a survey to find the frequency of IVF children who have Angelman Syndrome. The frequency of Angelman syndrome has been studied in the general population, but not specifically in IVF children. It is not known whether IVF children are at greater risk, but it is the purpose of this study to find out.

Thank you to all the families that have participated in the survey and have sent the questionnaires back to us. We are not yet able to draw any conclusions from the results so far because we need a greater number of respondents to the questionnaire. If any family have received a questionnaire, but have not replied, please could we encourage you to do so. We are interested in replies from all families with children with Angelman Syndrome, regardless of whether they were naturally conceived, or conceived with some help from fertility treatments.

We will report our findings back to you and hope that you will support us in this research. If you have lost the questionnaire and would like another copy, you can either contact us directly (see below) or request a copy through ASSERT. The information that you send to us will remain strictly confidential.

If you require further information about the study, please do not hesitate to contact us,

Yours faithfully,

Dr Alastair Sutcliffe
Senior Lecturer in Child Health (Honorary Consultant)
Dr Catherine Peters
Paediatric Research Fellow

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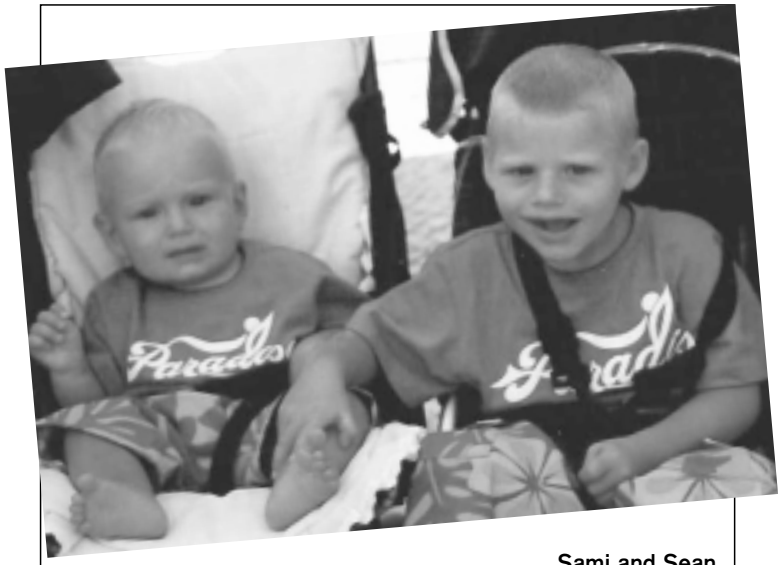
Family Stories

Here's a sunny report to brighten up the bleak winter days, from one of our family members who live in the (for now) British enclave of Gibraltar – they are the only Angelman family there and really find support and relate to us here in the UK – however, I don't think our weather would be sufficient for them to come and live here!!



Sami and Sean

Our Angel in the Sun

Our son Sean was born on 24th December 1998, weighing 6lb 8oz. Sean spent the first six weeks crying non-stop, as he didn't take very well to breast or bottle-feeding. Eventually he settled down and as soon as he started on solids there was no going back to bottles. He loved his food and must take after his father as he still does.

We realised that he wasn't reaching his milestones but as first time parents everyone told us that all babies developed at different rates. By his 1st birthday he was sitting up and blowing he even tried to blow out his candle, but no sign at all of standing or making any attempt to walk.

In February 2000 at 14 months we noticed that he was having head drops but we thought that he was just tired. Nothing prepared us for what the next 6-8 weeks were going to be like. As we live in Gibraltar, we were sent to a Paediatric Hospital in Malaga (2 hours drive away from Gib) so that they could run some tests as our doctor here told us that the EEG showed some irregular readings.

Anyway a Neurologist immediately saw him the next day and they performed their own EEG, MRI etc. They told us that the head drops were seizures and put him on medication. This type of medication made him worse with twitches all over his body that he couldn't control and having 30-50 head drops during one day. Eventually they changed his meds to Zarontin (Ethosuximide) and Epilim (Depakote) and thankfully he has been quite stable on these since.

The doctors explained that he could have something called Angelman's. As on top of the seizures he had a happy disposition, the love of water, the blonde hair and blue eyes. A Fish test was carried out, which showed that Sean was AS Deletion +. We both felt numb, this couldn't happen to us, twenty billion questions raced through our minds and we were devastated. We grieved the son that would never have a career, have his own opinion do things that we all dream our kids would achieve in life.

Well that now seems like a dream, we decided that Sean being the only child ever diagnosed with AS in Gib that we were going to have to educate everyone around us, family, friends, the medical staff and school staff. We then went to Great Ormond Street to get the diagnostic in English (as Spanish is our second language) and it would be easier for everyone in Gib to understand too. The results confirmed that it was AS. When we were in the UK we met two AS families, Sally Walburn and Tina Lockett, and although we in touch their friendship has been very important. For us it was the best thing seeing other AS children, but we said that we would take each day as it comes and never set goals for when he is going to do things.

To be honest we are so proud of him, its amazing to see the world through his eyes, everyone is funny and perfect and although he is extremely hyperactive and very strong, we have a very good routine and he eats and he sleeps well however, at present we're going through a bad patch. His favourite food is Spaghetti Carbonara with extra Parmesan cheese on top!! We are also trying to teach him to sit at the table, with us and he's been really good so far, when he's hungry, if not, forget it.

He eats everything we do, and now we are trying to teach him to eat with a spoon, he manages to do it when it's a pudding, as he prefers them!! He's does walk un-aided about 6-7 steps but he has to be very motivated in getting whatever we are showing him at the other end of the room. He can walk by holding someone's hand but he knows that he'll get there quicker if he crawls! Sean tends to sleep well when he gets worn out during the day, and although we are tired too, if he's had a good day its all that matters. We try to take him everywhere, what's getting more difficult is when we go away he never sleeps too well as his routine is disrupted, but that will only happen for a couple of nights then he'll get back on track.

Like any family with an AS child we have had to make our house Angel proof, his room is padded with gym mats on the floor, so that he feels more confident that if he falls he won't hurt himself, and he has his own TV & video up on the wall and he loves it. I think everyone loves it, its so light and loads of pictures its our happy room!!

We have the advantage here that its sunny & hot from May – September so he does a lot of swimming in the sea and pool, and we try to get him to walk a lot in the sand. Although we did go through a phase of eating the sand and drinking the seawater!!!! He now stays in his rubber ring and swims around unaided.

We have found that trying to keep to his routine is the best thing, if we stray even by the smallest margin it can cause him to get ratty, nervous, annoyed and inevitably disturb his sleep pattern.

Now Sean has a baby brother Sami, he was born on the 2nd December 2001. We were a bit concerned as how Sean was

going to react, but Sami is completely mesmerized by his brother and finds him really funny, and Sean shows off even more! Sean also loves giving Sami cuddles but Sami is not allowed to have any toys, Sean takes everything off him, even if he has one the same. But we never thought that they would be able to sit beside each other, as Sean is very strong, but I think he accepts that Sami is a baby and his brother, and is quite gentle with him.

Sean goes to a Child Development Centre three times a week and his concentration and interacting with other children has improved a great deal. We are also getting him to use a jelly bean switch and play very simple games on the computer, ones that you have to press the switch for the picture/shape to change and when you complete it the music come on as a reward, and he loves it.

We have noticed that with Sean if he gets a throat infection or high temperature he can fit or become edgy. We had a bad spell in July, and he had the longest seizure for us (lasted 15 minutes) and we had to give him the rectal Valium. Although we have come to accept the disability you feel so helpless when they are so poorly and there is nothing you can do, but wait for it to pass, and hopefully pick up where you left off.

Life here is pretty hectic with two little boys but every day is a challenge and a blessing, and when he smiles and gives us a extra big hug and a wet kiss, we know that its all worth it!!

Jane & Wayne Tunbridge
Sunny & Hot Gibraltar

Update on Cara

I know that many of the older ASSERT members will remember my grand-daughter Cara and the really bad times we went through in Cara's early years battling epilepsy and asthma, with Cara spending many months in hospital. We began to think that we would lose Cara, but we underestimated Cara's strong will power and determination and now five years on Cara is going from strength to strength. Her walking ability is getting better, and wherever she goes Cara makes new friends, especially when she goes abroad. Cara has a mind of her own and makes it quite clear when she wants something. The photo is of Cara paragliding in Spain. Cara is also going to Florida in November for two weeks and we know she will enjoy every moment.

Many parents and grandparents may think there's no light at the end of the tunnel but I will always remember meeting Pauline and Derek Webber who I met many years ago with their three AS girls and I know that these children can achieve many things in their lives whatever and however long it takes. Cara is happy and loving and I can say with all honesty our lives would be very empty without her smiles and hugs

June Pearson (Cara's grandmother)

Celebrating 23 years of Jonathan

Today we are celebrating 23 years of unconditional love . We are celebrating 23 years of giggles, laughter, slobbery kisses, and Teddy bear hugs.

At 23, we are celebrating because there were birthdays when we didn't feel much like celebrating. In 23 years there have been birthdays where the syndrome couldn't be separated from the child. There have been birthdays filled with endless nights, seizures, parental depression and countless questions about the future.

At 23, we celebrate survival! We celebrate knowing from birthdays past that we can celebrate the future. We have relaxed. We celebrate knowing what we did or didn't do ...was survivable. We celebrate learning to separate the syndrome from the child. We celebrate what we have learned from each other. We no longer look at what was lost but what was gained. We celebrate special friendships that have formed. We celebrate what is real not pretentious. We celebrate the purity we see in our son's eyes. We celebrate the reflection of love and honesty. We celebrate an appreciation for the small things that everyone else is sweating. We celebrate 23 years of learning; of seeing joy in mud-puddles; of chasing butterflies; of still enjoying what others long ago started to take for granted.

Today we celebrate 23 years of Jonathan.

Brenda Dixon

Scoliosis

We read this on the Angelman listserve and feel it is a good explanation of the condition. So often we blame ourselves for allowing conditions to happen for whatever reason, but often it's out of our hands and nothing we have done.

Scoliosis is muscle related. There is a muscle in the back called the psoas muscle and for unknown reasons in certain people, the muscle will begin to tighten and become shorter on one side...and since it is a muscle that is attached to the spine, it causes pulling on the spine, thus causing the spine to curve. Calcium deficiencies would only cause the spine to weaken, it would not make it turn or curve, unless this muscle was already pulling. Scoliosis also seems to be hereditary, both my mum and sister have it, although my sister's case is considered pretty severe and my mum's is slight.

Jill Malchiodi

Please note that the deadline for the next edition of the ASSERT Report is 15 May 2003.

News From New Zealand

Before I start I would like to reply to Eileen's update on Lyndsey in the last edition of ASSERT. I am in New Zealand and my Kirstie is the Angelman that Eileen wrote about. I could not believe how these two – Lyndsey and Kirstie – got on together. It was out of this world and I had a few tears when I read the article, as did Eileen and I at the time!!

Now I would like to bring your readers up to date with my Kirstie's life and the wonderful positive attitude we have now. It has been some time since I have written an article for the ASSERT – like two or three years I guess.

In New Zealand we have what is called the IHC (Intellectual Handicapped Corporation) which some time ago they (being the Society) attempted to bring the name up to more modern standards but had no success. I think they have been known as the IHC for so many years that to change it became a futile exercise.

Kirstie went to live under the IHC roof about 8/9 years ago. They have been good over the years. I was also so terrified about what would happen to Kirstie when I leave this world, and my mind was now eased by how she was looked after. Kirstie would come home each weekend and on taking her back to her home (which consisted of 2 caregivers and 3 clients – who were all less able than Kirstie) we would have world war three with hitting out etc. I then got the IHC to bring over their mini bus to pick her up which did ease the problem a bit.

I would always phone about 10 minutes after her going back to the house (which was called the Dales and was on farm land – really nice outlook) and she seemed to settle down quickly and all was well. Naturally there were a few problems, which mostly involved her behaviour – hitting, rowdy, bed wetting, banging on walls and just being a right pain in the neck. Even when she came home for weekends, although I haven't had wet beds for about 20 years, her behaviour still left a lot to be desired – some would say a typical "Angelman"!!!!!

Kirstie is now 28 – going on 29 in February.

At the beginning of this year Kirstie was taken from the Dales and put into a house in town (Wanganui has about 40,000 folk). I will admit 99 percent of me didn't want this to happen, as I just didn't know how she would react. She was to have complete new staff except one (who only stayed about 3 weeks) and two other clients both considerably better than Kirstie – with speaking etc.

The big day came and she shifted. The staff from the new home (Sarjeant St – they call it) had meetings nearly every day for the first fortnight, then weekly and now monthly to which I am invited and attend. They have worked so hard and the result has been so wonderful. Kirstie is almost a different person. They have gone for a month without a wet bed, she is not ripping the sheets at night (which became a very expensive exercise), is still ripping her t shirts which she wears as pyjama tops but not nearly as much and her general behaviour is much quieter. We went out to a Halloween evening and I kept on thinking something was missing but couldn't think what!! It wasn't until the end of the night and we were on our way out that I realized Kirstie hadn't said an "ello" all night. When the staff take her out they now can't get over how gentler and quieter she is.

I can now take her back from the weekends – which I have altered to every second weekend giving me my first spare weekends since Kirstie was born I guess and it sure is neat to be able to do something for myself at last (not that I don't love her to bits) – and do not have one hassle. I can go into her home for coffee in the evening and she doesn't want to come back with me. It is so NEAT!! She is so happy there. In actual fact she now pushes me down the corridor and out the front door (which some part of me says – fancy doing this to her Mum who she has relied on for all her life, took some getting used to!!!!) although on a Saturday she is excited to see me and come home.

If someone had told me a year ago that this could have happened, there is no way I would believe them. It is like a miracle come true.

But of course she still is Kirstie – an Angelman – and we will always have the behaviour associated with this – but SO much less of it. I guess I had better stop – but please take heed folks, our Angelman people can, given the right circumstances, alter as they get older. So take heart guys!!!!!

Anne Fulcher
Email – anne.fulcher@clear.net.nz

Question Time

This was a question from the Conference back in September. The reply came from Lucy Bennett who spoke about Behaviour.

"My son bangs his head repeatedly against anything behind him. Why does he feel the need to do this and should we try to prevent him? He cannot stand or walk"

This could be caused by lots of things for example – pain or sensation in head, self stimulation, excessive/enthusiatic rocking. It is important to try and check out if there are any physical problems and deal with them. It would be best to try and protect the head from harm by placing an absorbent cushion in area where head is likely to strike. Holding the person to stopthem doing it is likely to be difficult to do and lead to confrontation in the relationship.

More on behaviour in the next newsletter – which I am working on already – Sally Walburn

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.

Development and behaviour in Angelman Syndrome:

- A study of development in 15q11q13 maternal microdeletions and behaviour across genotypes.

The above study was conducted by Angela Reason, Clinical Psychologist, as part of her Doctorate in Clinical Psychology.

The study takes the form of two parts, one section focusing on the development of people with Angelman Syndrome by deletion aged between 3 and 14 years. The second section focuses on behaviour across all ages and all genotypes. For clarity, the sections will be presented separately. The first part on development, will appear in this issue, and behaviour in a subsequent issue.

Part one: Development in Angelman Syndrome

Background to the study:

To my knowledge no comprehensive study exists to date specifically exploring the cognitive (mental) development of individuals with Angelman Syndrome (AS). I felt this was a significant omission and attempted to address this gap in the present study. Both in services and in research individuals with AS are referred to as having a significant learning disability, but with no real sense of the extent or nature of this impairment. Looking at cognitive ability aims to provide parents, caregivers, those in the educational system, and other professionals with a sense of where best to pitch interventions so that understanding can be maximised. In addition, determining specific strengths and weaker areas will allow for greater focus to be placed on areas of skill, tailoring learning through the most effective methods.

Aims of the study:

Therefore the aims of this study were to:

1. Determine an approximate level of overall development
2. Explore different aspects of development
3. Determine a profile of strengths and weaker skill areas
4. Obtain parent/guardian perspectives on their child's development

Selection of participants:

Recent literature has indicated several differences between the different genotypes of AS. The scale of this study could not however look at all groups, so the largest group 'deletion' was selected. Additionally, only children were selected. Recruitment took place via ASSERT.

In all, 14 children took part (11 boys and 3 girls) ranging between 3 and 14 years with a mean age of 6.4 years. All suffered with epilepsy.

Procedure:

The assessments chosen and reasons for this are as follows:

- **The Bayley Scales of Infant Development – II**

This was chosen as it is a measure of pre-verbal intelligence. A pre-verbal measure means speech is not heavily required to pass items. Such a measure was needed due to speech difficulties in AS; if I had chosen a measure which relied too much on speech to pass test items it would have meant individuals reached their limits early on. This is an infant development scale and was selected in the absence of non-language reliant tests for children with learning disability. It provides an overall developmental level as well as level of ability in the areas of mental skills, language, social skills, and physical abilities.

- **The Vineland Adaptive Behaviour Scales**

This is an interview which is conducted with the main caregiver(s) to allow for their perspective on their son/daughter's development. It covers a range of aspects, namely communication, daily living skills, social skills, and physical abilities.

All participants were visited at home and assessment took place in the presence of a parent/guardian.

Issues affecting the results:

There are several issues which affect how the results are interpreted, which are important to discuss.

- Firstly, conducting a one-off assessment means I was not able to be certain that their performance on that day was representative. For example, they may have been more tired than usual and therefore did not perform to the best of their abilities. The accompanying caregiver interview went some way to try and address this, however.
- Secondly, I noticed that all of the individuals in this study began to cease passing items at a similar level. Whilst this is likely to reflect an approximate level of their ability, two other factors are involved. Firstly, despite the Bayley's being predominately a pre-verbal measure, as the scale progresses in difficulty more language items are included. The point at which most individuals stopped coincides with the point where language items increase. Secondly, the sorts of materials used also begin to change at this point and these were observationally of less interest to individuals (e.g. shape boards) meaning they were less motivated to attempt the task which thus may have affected their score.
- Thirdly, there are some problems interpreting the results of the test on a population for which the test was not designed (it was designed for infants). This means that the overall development scores can only be recorded as a range (e.g. 9-11 months).

Results:

1. **Overall developmental level:** Taking into account the issues highlighted above, this study can only conclude that estimates of developmental level across this sample of children with Angelman syndrome by deletion, are on average between 9-11 months. The range across the group spanned 7-14 months (meaning in this group the lowest developmental age score recorded was 7 months and the highest was 14 months).

2. **Specific areas of development:** Furthermore, a profile of skills across different areas of development, namely mental (cognitive) ability, language, social, motor (physical) skills, was elicited from the assessment. This aimed to provide a more comprehensive understanding of ability. Results indicated that language and social skills for the group averaged 6 months, motor skills were 9 months, and cognitive skills were the highest reaching an average of 13 months. This shows that in some cases individuals had 'splinter skills' – this means their cognitive abilities were above that of their general developmental level.

3. **Strengths and weaker areas:** In particular all individuals showed strengths in 'object permanence'. This is the ability to understand objects still exist when they disappear out of sight. The group also showed skills in the following tasks: making a bell ring, cooperating in a game, retrieving items despite a barrier, removing a pellet from a bottle, securing a ring when out of reach, using gestures to make wants known. Areas where performance was weaker included completion of shape boards and peg board, putting items in containers (taking out was no problem!), imitation, and using a pencil.

4. **Caregiver perspectives of development:** An overall score of 'adaptive behaviour' was on average 13 months. Once again profiles of skills in different areas of development were established. Communication skills emerged at 10 months on average, social skills at 12 months, daily living skills (e.g. washing, dressing, feeding) at 13 months, and motor (physical) skills at 16 months.

Conclusions:

The children with AS by deletion that participated in this study showed an average level of development of 9-11 months. Their 'adaptive behaviour' was on average 13 months. They displayed strengths in several areas, some clearly higher than their general ability, such as tasks of 'object permanence'.

These results show that estimates of developmental level were slightly lower on the clinical assessment than that of parent/caregiver reports. However, the two assessments measure slightly different things and should therefore not be used in direct comparison. It does show that any assessment of development should be comprehensive, not relying on just one measure, and should take into account the perspectives of people who observe the individual on a day to day basis.

Implications:

Aiming teaching and interventions at approximately the 9-11 month range will be likely to promote empowerment and a sense of achievement. Tasks aimed too high will produce feelings of frustration if they repeatedly fail. Of course, individuals will benefit from being 'stretched' beyond their limits at times to enhance learning, but this should be within reason and not at a level which is unachievable.

Popularity of assessment materials is likely to play a key role in the individuals' willingness to attempt tasks. Popular items were those that were noisy or made of plastic. Use of such items in education and teaching of skills will be likely to facilitate their learning, and keep their attention.

If anyone would like to discuss issues relating to this study, Angela can be contacted on: 020 8921 3506 (direct line/voicemail) or email: angela.reason@oxleas.nhs.uk

Donations

Thank you, to everyone who has donated to ASSERT since the last Report back in July, 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:

Anderson
In memory of Margaret Bates
Linda Buchan
Mrs J E Capen
Mrs J E Childerhouse
Sue Donnarumma
18th Edmonton Scout Group
Fiona Gibbins
In memory of Gwen Goad
A S McGinley
Mr & Mrs Munro
Mr A K Nunn
Ranelagh Masonic Lodge
R T Simpson
Tomcat trikes
U Levers
B & J Willis (Patriot Scooter Club)
Mr Wyatt

2000 Club Draw

Three draws were made at the last trustees meeting in January – Winner receives £40, runner up £30

Winners

June – Mr L H Capen
Sep – Mr W S Hynes
Jan – Wayne Tunbridge

Runners Up

June – Joyce Egan
Sep – Mrs L Hynes
Jan – Pauline Herbert

April 2003 will be the last draw to be drawn at the next trustees meeting on April 13th 2003. Many thanks to all who have taken part and to the Tunbridge's who gave back their winnings to ASSERT as a donation – thank you!

Baylor College – Research (USA)

I will be glad to give you some information about the ongoing study here at Baylor College of Medicine.

We still do not have results but we have preliminary data. In Houston we already enrolled 20 patients. 10 are under the age of three and 10 are four and above. The study is designed to ascertain if administration of folate and Betaine provide any dramatic therapeutic benefit in patients with Angelman syndrome. Each participant with Angelman syndrome has a 50/50 chance to be randomised to the placebo or drug group. We have patients with deletion, UPD, Imprinting, UBE3A. 11 patients already completed the study.

After completion of 12 months in the study, a number of families have asked if their child could be placed on folate and Betaine regardless of whether they were on treatment or placebo during the double-blind portion of the trial. We were able to offer this opportunity to the families who asked for it and only if they have completed twelve months on the double blind trial. During the study each patient will be studied as an inpatient in Texas Children's Hospital General Clinical Research Centre during 1-day admission, three times during the year of study. The child will have height, weight, body circumference, routine physical and neurological examinations to assess motor function. A total of 20cc of blood will be drawn to assess complete blood count, kidney function, and levels of Folate, Betaine and Amino Acids. The Child will also undergo a developmental evaluation test and a one-hour EEG test.

Three months after the study starts the child will have 22cc of blood drawn. We also call the families once in two months and ask questions concerning any problems with the medication and questions concerning whether the child has shown improvement in behaviour, sleep, walking, drooling, breathing, alertness, communication, seizures, motor skills, attention span.

Lina Shinawhi

Siblings

Parental Strategies

1. Equal does not mean the same
2. Be open and honest in communication
3. Schedule special time with the siblings
4. Limit care giving responsibilities
5. Accept individuality
6. Use respite care and other supportive services
7. Provide opportunities for a normal family life and normal family activities
8. Parents do not forget to look after yourselves

Appeal from Abroad

Now that ASSERT is part of the World Wide Web we do get enquiries from ex pat families who sometimes are thinking of relocating to the UK to get better services for their children. One of the appeals, apart from wanting to know about benefits and respite, involves schooling and the provision across the country. If you know of a particular school or residential provision which is good with Angelman persons then it would be very useful to let us know about it, in order that we can pass on the information these people need. We also get requests from time to time from UK families and professionals about residential schooling and colleges and whether there are any good ones out there. If you have any information, which would be useful to begin a database, then please do get in touch – it seems to be a fact of life that we live in a postcode lottery and some families have moved around deliberately to receive a better service. Thanks for your help.

Can you Help?

Research Project

I am a parent of Matthew he was diagnosed with Angelman Syndrome in 1999 at the age of 2. This year I am writing a project on Angelman Syndrome for a course I am studying. I am looking for information from other families around the UK and overseas regarding what information was made available, the quality of therapy received (if any), general awareness of this syndrome as well as, most importantly, your experiences as parents/families of AS individuals. Although this article will not be published and will remain the property of the college I hope to raise some awareness to this little known syndrome.

You can contact me by e-mail or in writing. I look forward to hearing all your views and experiences.

Sincerely

Lois Windle

6 Kearsley Road, Sheffield,

South Yorkshire, S2 4TE

or e-mail: markandlois@yahoo.co.uk

More Research Information from Kate Horsler

Thank you to all those parents and carers who took time to talk to Chris and myself at the conference. We both learnt a lot about children with Angelman Syndrome and the information we gathered will inform our research. Over the next couple of months we will be contacting people who live within the West Midlands area via ASSERT and asking if they would like to participate in the study. Alternatively, if you live within this area and you would like to take part in the study please get in touch with either Chris or myself by email or phone.

Kate Horsler,

Clinical Psychology Doctoral trainee

email: KJH191@bham.ac.uk

Chris Oliver, Professor of Clinical Psychology, School of Psychology, the University of Birmingham.

Telephone: 0121 414 4909

email: C. Oliver@bham.ac.uk

Useful Workshops,Conferences, and Publications

Developing Effective Advocacy – for those with severe and multiple learning disabilities
8th April 2003 – London. Organised on behalf of the Learning Disability Forum by Kith and Kids and supported by BILD Professionals £80 Parents, Advocates and Self Advocates – £20 Contact Kith and Kids 020 8885 1517

National Parent Partnership Network (NPPN) can also provide information on parent partnership services. Visit their website at www.parentpartnership.org.uk for more information.

Contact A Family has produced an information pack for parents of disabled children explaining the new disability legislation in education, which came into effect from September 2002. The Special Educational Needs and Disability Act 2001 makes it unlawful to discriminate against disabled children's participation in all aspects of schooling. Contact: 0808 808 3555 Mon – Fri 10am – 4pm for details. (Community Care 29 Aug-4 Sep 02)

Cerebra – The Foundation for the Brain Injured Child has moved to: 13 Guildhall Square, Carmarthen SA31 IPR Phone: 01267 244 203

The Disability Rights Commission (DRC) was set up by parliament to stop discrimination against disabled people. They offer a helpline on 08457 622 633 Monday – Friday 8am – 8pm

Older Family Carers Initiative is a three year project aimed at supporting Learning Disabilities Partnership Boards across England to identify and meet the needs of older family carers of people with learning disabilities. This will involve a partnership of organisations and individuals with expertise in this area. The project will start by completing a detailed audit of current work with older family carers around the country.

Contact: Dalia Magril at the Foundation for People with Learning Disabilities on 0114 221 8655 or 020 7802 0329 or e-mail: dmagrill@fpld.org.uk for more details.

Daily Care Ltd
They provide services to individuals who require advice and assistance with activities of daily living; they can also offer occupational therapy and physiotherapy advice and treatment. Unit 4 & 5 The Coach House, Phoenix Business Centre, Low Mill Road, Ripon HG4 1NQ. Tel: 08700 345200 www.dailycare.demon.co.uk

Fledglings
Toys, equipment, ideas, aids for special needs children – enquiries to Fledglings, 6 Southfield, Ickleton, Saffron Walden, CB10 1TE enquiries@fleglings.org.uk
Tel: 0845 458 1124 Fax: 0845 458 1125

Publications
The Easy Guide to Physical Interventions – This guide, published by BILD, encourages people with learning disabilities to know their rights about physical interventions. It uses easy language and clear pictures to explain what physical intervention is and why it may be used and gives good practice guidelines for carers.
ISBN: 1 9025 1997 3 £4.50
Contact: BILD publications on 01752 202301 for details.

Special Educational Needs (SEN) – a guide for parents and carers – is available from the DfES Publications Centre. This publication also has a list of all the parent partnership services available plus a good list of contact details for other voluntary agencies. Contact: 0845 602 2260

Parents have needs too! This booklet aims to increase understanding of the emotional aspects of having a child with special needs and the help that counselling might offer. It is hoped that parents of children with special needs will find helpful the thoughts and feelings expressed by parents in the booklet and feel less isolated with their experience. For a free copy, please phone: 3Cs 07813 095 383 or write to: 3Cs project, Ealing Mencap, 44a The Broadway, Greenford UB6 9PT

Transport for London
Congestion Charge – as you all must know by now all vehicles entering Central London will have to pay £5 per day. If you have a disabled badge you will be exempt from the charges, however, you will need to register and this will cost £10. If your vehicle is exempt from road tax, you are fully exempt from the charge and you do not have to do anything to be eligible. For more information check out the Transport for London Website at www.cclondon.com

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

HELPLINE
ASSERT Families/New Diagnosis/Support Line
01268 415940

Marathon Man

Jason and his nephew, Brandan

Last year, I decided to run the 2002 London Marathon with the main goal of completing a life's ambition whilst raising awareness of the Angelman Syndrome condition and raising as much money as possible for ASSERT & the White Lodge Centre.

My nephew, Brandan Jones (aged 2½) was born in 2000 and was diagnosed at eight weeks old with two rare conditions called Angelman Syndrome & Colaboma (Eye development disorder). Brandan has been in and out of hospital since birth, but with all his troubles Brandan always manages to shine a ray of light with his beaming smiles for his family.

I would like to thank everybody who helped me raise £4,000, which was equally shared between ASSERT & White Lodge Centre based in Chertsey, Surrey, which supports disabled children and adults. The day itself was a fantastic and very emotional I was shocked at the amount of support, encouragement and energy I received from the crowd all the way round. After all the highs and lows of the gruelling the 26.2 miles, the 8 months of training and the effort in raising all that money the first thing that came into my mind once I crossed the line, was. 'I can't wait until next year'.

I was happy just to have completed my first marathon in 4 hr 43 mins. This was special to me as I was nearly going to pull out due to injury, which left me out of action for the whole of March, which left me only 2 weeks to do the best I could to get fit.

This year my two friends, Phil and Justin are going to run with me and help raise money for both ASSERT and White Lodge Centre. If anybody would like to sponsor me either send a cheque into ASSERT or go online at the following URL: www.justgiving.com/jasonthornton

Jason Thornton

Ken, Sally and Brad with the Patriots Scooter Club

Patriots Scooter Club

Back in December Ken and Sally Walburn went along the Patriot Scooter club meeting to be presented with a cheque for £200. Jan and Brad Willis are members and together with fellow members had organised a live band evening as a fundraiser for ASSERT and Essex Air Ambulance. We had our pictures in the local paper and ASSERT and Angelman Syndrome got a mention. Many thanks to everyone who helped with the fundraiser and to the club for thinking of ASSERT.

London Marathon

Yes it's come around already Sunday 13th April 2003 – and too soon for Ken Walburn who is attempting the Marathon for the fourth time, after having said last time 'never again – I am too old!' Once again Ken is running to raise funds for the conference. Along with other fundraising initiatives, many of you who attended the conference benefited from those who generously donated funds to pay for all AS persons and their carers to attend free of charge. We want to offer this opportunity again in 2004, and so once again Ken is running specifically to support families in attending the conference. If you would like to sponsor Ken please complete the enclosed sponsor form. This time we are including Gift Aid forms – if you are sending in a single donation from just yourselves, providing you are a UK taxpayer, ASSERT can claim 28p for every £1.00 you donate from the Inland Revenue at NO cost to yourself. If you are sending in multiple donations and would like more forms, please contact Sally Walburn on 01268 415940. You must complete the form fully and it is most important to date the form – last time Ken raised £3000 so had we been able to claim Gift Aid then this would have resulted in an extra £840 for ASSERT – so you can see why we need to have these forms completed.

We also have some other runners taking part – Jason Thornton (see Marathon Man), Justin Drake and Phil Neate (Jason ran last year and raised £2000 for ASSERT) – if we have forgotten anyone we're sorry. We wish you health and a successful run on the day and many thanks for your support and hard work...it's not just the run, it's all those months of training as well. Go for it!!!!!!!!!

Making Plans for Joshua

We read this article on the AS Listserve and thought that the information may be of interest to you all – although Jackie and family live in the USA, you may get some inspiration from what they have managed to set up for Joshua.

First there was an article written about Joshua several years ago, which you can read on line and I think the ASF has it somewhere on the web. You can locate the article by going to www.self-determination.org then under the Common Sense News Letter, then under editorials here you will find the article entitled: Joshua's House

I have to say that Joshua's life is indeed his own and is living proof that mountains can be moved. We didn't get Joshua's official diagnosis until he was 16 years old, so we were loners out there, never meeting any child like Joshua. Boy did this change once we got the wonderful news of AS.

Joshua's seizures were always life threatening, from 15 months of age Joshua's seizures were non stop, or rarely a day without any seizure activity. Joshua spent most of his early years in and out of PICU's across the state of Maryland, PA and Washington, DC. We made just over $300.00 than the limit to receive any supports. We did have respite dollars, but no one was willing to watch Joshua with his seizures. (This is why I tell people to get people use to their children...have friends, family, students, neighbours come and help out while you are home so they do get comfortable with our children. We did have a few family friends and I trained a few neighbourhood students who were comfortable with Joshua) Anyway, by the age of 8, our resources, energy and so forth took it's toll and we looked for some long term supports. We found that the government was willing to pay total care if Joshua left our home and went into a paediatric hospital. We rejected this for a while, then out of desperation we took this option, since there wasn't anything else. (Note: we gained full control over Joshua's seizures when he was about 9 years old. We went three years without any seizure intervention, but he is now back on depekene and L-carnitine)

In this awful place, he was overdosed, beaten and neglected. Needless to say we lived in this place as much as Joshua. This experience is what people have said "Put fire in my belly!" The injustice for people with disabilities is so criminal in my eyes. We fought the system to change this. Got involved, or I should say continue to be involved to make things right.

Joshua moved out of this place, but we just didn't take Joshua, we took all 45 children out of this place and forced the system to provide supports in the community. Yet, Joshua still couldn't come home, he had to receive services through a "traditional" provider. He moved into a group home with two other children. He lived here for another three years. It was about this time that we first heard the term "Self-determination". Folks know these principles and live by them. (www.self-determination.com to get more information)

Basically how this works is that the person with the disability directly designs what services makes sense for them, with the assistance of family and friends. We did just this. We gave our family home, or I should say rented it to him (WE ARE NOT RICH) to Joshua. We rented out a room to someone else with a disability and between Joshua's SSI rent and the boarder rent, this paid the mortgage, allowing us to move and find a home. Joshua was then 16 years old.

We then looked at what made sense for Joshua. We became his case managers and assisted Joshua to hire his own staff. Yes, Joshua can fire his staff as well and has done so. (We hired one person out of the newspaper and this is the person that Joshua let go. He didn't say "You're fired" he just let us know he wasn't happy with this person, as the person never developed a true relationship with Joshua. This guy was great, but just didn't have that connection that Joshua needed. So we helped Joshua by letting the guy go.) Other people that work with Joshua are people that he has made a relationship with in the community. Example: Joshua's one on one left the school system and moved in to Joshua's house to become his live in person. (Cannot get better than this) Anyway...Joshua took back his life through self-determination, and we became the case managers, simply by being Mom and Dad. We worked through an agency, our local Arc, that was willing to do this for us.

We then pushed our system one more time. We decided that the agency was getting too much of Joshua's budget (self-determination talks about individual budgets) and every time Joshua needed to access his budget the provider had spent it on someone else. Not very individualized! We decided to become the provider through a microboard (www.inclusionresearch.org under Self-directed Support Corporations and www.microboard.org Vela www.tnmicroboard.org for more information on this)

We set up Joshua's House Incorporated and became a Medicaid provider for one person, Joshua, in the State of Maryland. This means that Joshua's microboard, the board of directors are made of people that care about Joshua, family and friends, design and assist Joshua in his supports. Truly self-determination. So Joshua's home looks like this: Joshua has his own home that he shares with his live in support person (formerly his one on one from school) Joshua has an additional staff of 5 people, all of whom know Joshua and care about him. These people are gifts from God I have to say. We found them by listening to Joshua. We also respect them in what they bring to Joshua's life. (This is another topic altogether in finding the right people to work with a person with AS.) Joshua's House is totally designed for Joshua to live freely and fully in the community, including having a business. Joshua has what is called a microenterprise. This is a small business based on Joshua's loves. Example: Time Savers Concierge Service is the name of Joshua's company. Even though he does a small portion of the job, it is still his company, not mine! Joshua runs errands: Takes dry cleaning to and from the dry cleaner, picks up and returns tapes to Blockbusters, does simple grocery store shopping (that gallon of milk and loaf of bread that you don't have time to stop for) picks up cards and gifts for people, will pick up home office supplies for you, movie tickets, etc. Joshua loves to drive in the car, push carts (like grocery carts) loves being around people, loves to be the center of attention. We built the business around Joshua's loves. (Note we worked through our Division of Rehab Services and the school system to set this up) Joshua's business plan actually became his IEP when he was in school. So when the bus stopped coming to Joshua's House in June, Joshua had his business waiting for him. DORS (Division of Rehab Services)

Paid for equipment that Joshua needed for his job. His direct support care staff assist him in doing the business, they drive and do most of the work, but Joshua does what he can and has a wonderful life built doing all the things he enjoys.

Joshua is invited to community events, has lots of friends with and without disabilities and just plain loves life. Some people have thought that we have set up this system of supports to be in a more "controlling" mode over Joshua, but this is far from true. Our goal in life is for Joshua's life to be his own, not his parents life, but truly his. That he can be a young man living life that he has designed. Our role was to help him facilitate this. Actually I have finally earned the right to die, a hard topic to discuss among families of children with disabilities (young and old). You see I want Joshua's life to be the fullest with or without me and I had to truly educate myself and push the system to make this happen.

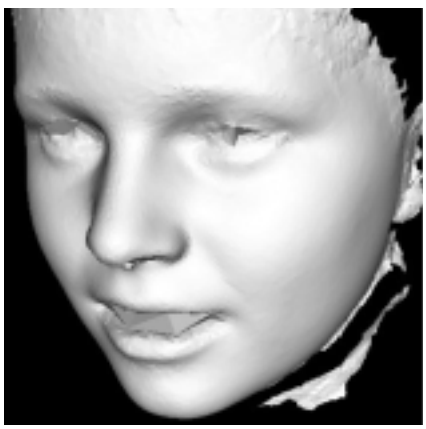
Some questions that people have asked are: What happens when you are gone? Do you worry about Joshua's life continuing the way it has been set up? My response is that there is no guarantee in life period. That the way we have set up Joshua's life to be his own has a better chance of survival than anything else out there because in the structure of Joshua's network. These are people that truly care about Joshua and his life outcomes. This assures me that someone will always be there to make Joshua's life his own.

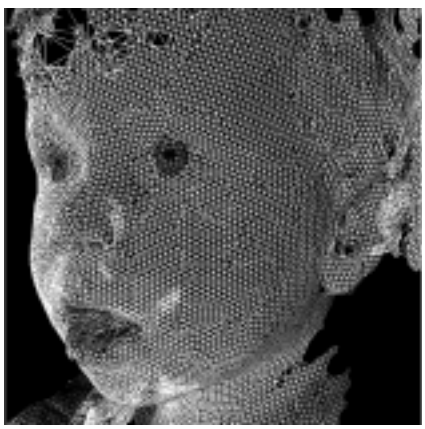
Jackie Golden (Joshua 21, del+)
Baltimore, MD

3D photography at Loughborough

3D photography is a new technology and has previously been used for taking pictures of the earth's terrain from satellites. Professor Peter Hammond attended the ASSERT family day and brought with him a prototype 3D face scanner developed by 3dMD Ltd, a company based near Harefield in Middlesex. Peter leads a group of computer scientists in the Eastman Dental Institute at University College London and has a special interest in the development and growth of the face. His group has collected over a thousand 3D face images in the past two years and has developed computer software to study how a young child's face alters from birth, through puberty and on to adulthood. The age range of his subjects is from two weeks to 80 years old.

While at Loughborough he took 3D photographs of 19 children and adults with Angelman syndrome and about 44 family members and volunteers. Each 3D image contains as many as 10,000 points on the face as can be seen below from the mesh of points for one of the children scanned at Loughborough. One of his team, Tim Hutton, has developed new computer software that can calculate the average of a collection of faces as well as the major ways in which each face differs from the average. The average surface of the Angelman images is also shown below. Professor Hammond will work with Dr Jill Clayton-Smith to analyse the images in detail. Any findings will be reported in a future edition of the newsletter. Professor Hammond is extremely grateful to all those who agreed to have their faces captured in 3D. By now everyone who took part should have received a CD with their images. Anyone who hasn't yet received their images should contact him on 020 7915 \2303 or p.hammond@eastman.ucl.ac.uk.





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.

HELPLINE ASSERT
Families/New Diagnosis/ Support Line

01268 415940