ANGELMAN SYNDROME ASSOCIATION

www.angelmansyndrome.org

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National Conference Postponed until 2016

Thank you to John Hosie from Victoria for the work that he has already done in preparation for the Conference in Melbourne. However, in an effort to organise a Conference that is beneficial to all our members and that offers specialists and experts from overseas and within Australia, we have made the decision to postpone our Conference until 2016. Further information about the conference will be communicated through future editions of our Newsletter. Stay tuned!

National Committee for 2015

Thank you to the following people who continue to provide their support for all our members:

Committee:	President:	*No nominations at present*
	Secretary:	Sally Shackcloth
	Treasurer:	Kevin Kennedy
	VP'S: SA:	Kerri Monaghan
	WA:	Leticia Grant
	TAS:	Chloe Simons
	NSW:	Anne Funke
	VIC:	John Hosie
	QLD:	Lysandra Warren
	ACT:	Lynne Cousins
General Committee:		Liz Stanley
		John Hannaford
		Kellie Wild
		Mary Bills

At this stage we do not have a President, but the National Committee will continue to function as normal through the year. If there is anyone who would like to become involved in the Association at whatever level, please do not hesitate to contact me at kevin.kennedy@angelmansyndrome.org

Kevin Kennedy (Treasurer)

The Committee would like to wish all our families and supporters a happy and safe Easter!

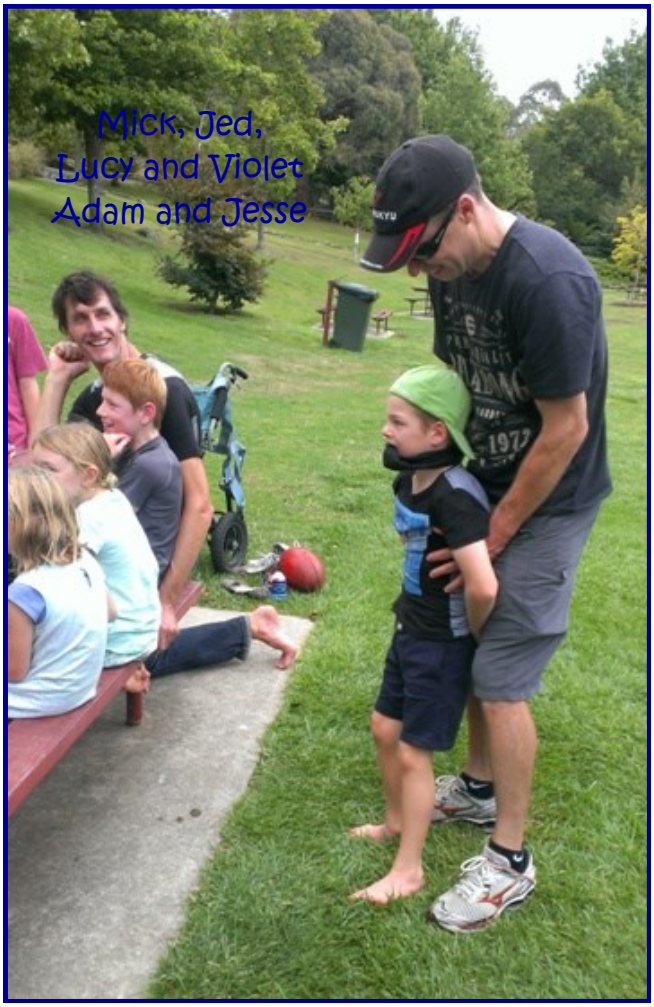
International Angelman Syndrome Day, 15 February 2015

Six families from across Tasmania gathered at the Punchbowl Reserve, Launceston to celebrate IAS Day. Sally made a delicious cake to celebrate. It was great to catch up and share stories, highs and lows, discuss research and of course watch the children get soaking wet playing with the water pump in the park!

Chloe Simons.


Samantha, Sally (support worker), David and Hannah


David and Hannah


Mick, Jed,
Lucy and Violet
Adam and Jesse


ANGELMAN
DAY


Samantha, Larna and Hannah


Charlotte, Oliver
and Emma


Mick, James
and Adam

International Angelman Syndrome Day

15 February Sydney 2015

Quite a number of NSW families gathered on Sunday February 15 at Livvi's Place, All Ability Playground, in Yamble Reserve, Ryde for a picnic day.

Families came from far and wide to enjoy the day, and as always our AS kids had a great time. It was great to catch up with new and old families alike, and the kids were also treated to face painting as well as Superwoman who wowed the children with magic, bubbles and balloons. Many thanks to **Fly by Fun** for providing this entertainment for us.

A huge thank you to Joanne Caracoglia and Michaela Townsend who organised the day for us, as well as a cake to celebrate the special event.

Kevin Kennedy

Correspondence to **Secretary:**
PO Box 364 MARGATE Tas 7054

Payments to **Treasurer**
PO Box 554 SUTHERLAND NSW 2232

Newsletter articles to
sallyshackcloth@live.com.au

Deadline for next issue: 30 June 2015

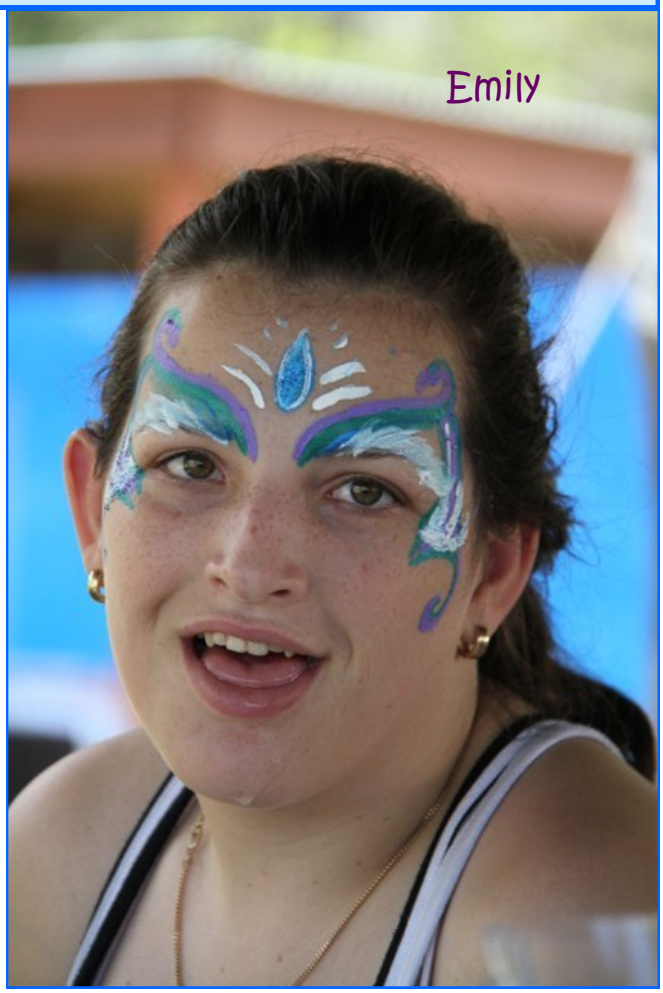
QUEENSLAND NEWS!!!!

See below some photos of the IAS day, 15 February 2015, hosted by **Janet and Chris Taylor** at their home this year. It was a wonderful day with a fairy floss machine, snow- cones, bouncy castle all donated by local businesses.

Lysandra Warren.

Joel` and Lucy

Emily



Joel and Caitlin

The Prospect of Molecular Therapy for Angelman Syndrome and Other Monogenic Neurologic Disorders.

Authors : Barbara Bailus and David Segal ; from California, USA

I would like to summarise here this new paper, recently published in BMC Neuroscience.

David Segal came to the day on Angelman Syndrome (AS) held in Melbourne, at the Children's Hospital, a couple of years ago. He spoke about recent research in AS. David Segal is a highly respected and progressive genetic researcher and what he says is always worth hearing and reading.

In the current paper, his team review recent progress and the prospect for gene therapy in AS.

The authors start by describing the syndrome and the gene, UBE3A. UBE3A is established as the gene, which, when not active from the maternal chromosome, leads to the AS phenotype. Thus, we know the genetic target in AS (and it is essential to know the target for gene therapy) and this allows for genetic treatment options. This is a big plus for AS - in disorders where the genetic target is not known, gene therapy is still a way off.

It is also essential to know if postnatal intervention would be beneficial. This is an important question, because the brain has been without UBE3A expression throughout development - until diagnosis in fact. The studies by Weeber et al on AS mice showed some beneficial effects, suggesting that the major defects were in signalling and not in development. So that is another big plus for AS.

So, what has been tried ?

1. Restore UBE3A expression.

Gene therapy could in principle replace the missing components. Replacing the large 5Mb region that is deleted (in most cases) is not really a viable option due to size limitations of the viral vectors (agents for putting the gene in) - but it could be possible to replace the causative part (UBE3A) rather than the whole region. This has been done in a mouse model, using an adenovirus as vector and did show some localised restoration of Ube3a cDNA and improvement in associated learning and memory defects in the mice used. However, this sort of treatment is difficult in humans, due to the widespread expression of UBE3A in the human brain.

2. Reactivate the intact but silent paternal copy of UBE3A

The paternal UBE3A is silenced by a long antisense RNA transcript [UBE3A-ATS] – this is the usual or normal situation. The maternal allele is not affected by the antisense, and so is expressed normally – but when something happens to it eg deletion, there is no or null expression of UBE3A resulting in AS. Progress towards reactivating the paternal UBE3A was made in mice, using various drugs eg topotecan which acts by inhibiting the transcription of the antisense [Ube3a –ATS]. Topotecan is an approved anti-cancer drug. However, various complications (related to specificity, toxicity, biodistribution, and delivery) have prevented human trials of the drug for AS treatment.

Another approach in mice was to use a Ube3a with an early termination signal ie the final ATS was shorter than the usual one and so was not active. Great idea, and there were some improvements recorded in these mice – eg in long term memory and motor skills. However, as this mouse was created from a single modified cell, the same method could not be used for therapy in humans.

3. Try ATFs – artificial transcription factors

The authors consider this the most efficacious gene therapy. ATFs are engineered DNA-binding proteins - they can be programmed to act as activators or repressors of specific gene expression. Such ATFs could be used to reactivate the paternal UBE3A that is silenced by imprinting or inactivate the paternal UBE3A – ATS antisense transcript. He talked about 3 main ATFs - Zinc Finger (ZNF) , TALEs and CRISPR. A single ZNF module can recognise approx 3 base pairs of DNA. TALEs contain a series of repeat modules, each repeat recognises one base pair of DNA. CRISPR, a new (2012) powerful technology, uses a large protein [Cas9], in a complicated way to make manifest specific mutations in cells – these cells can then be assessed for various effects of for example, drugs or other medications. One of the main problems of ATFs is any off-target effects , but these will be studied !

Conclusion

I have given here a brief summary of this review paper. However, my reading of this paper gives me the "best feeling" to date that progress is coming on a genetic basis for an eventual therapy for AS. In other words, advances may allow for the expression of paternal UBE3A which may give some phenotypic rescue. Great stuff !!

Assoc. Prof. Ellie Smith [Patron of ASA]

David Segal

Thankyou,Ellie, for providing this succinct report. (Editor).

CLINIC CHAT
with Dr Antonia Milner
Angelman Clinic
Co-ordinator

Dear All,
The clinic has been a little quiet so Anne Funke suggested I chat about a few recent research articles.

I wanted to talk about 3 recent publications, first by our dear friend and patron Dr Bernard Dan. This is a commentary called Maternal UBE3A in Angelman syndrome: "The rest is silence"? published in the European Journal of Paediatric Neurology in 2012. He talks about Dr Weeber's work in his lab and whether there is some tiny UBE3A production by the "silenced" father's gene. He also talks about the drug topotecan which may activate the father's "silenced" gene and wonders whether it will be effective and if so, if the effect will be maintained. Also when and how it should be administered.

The other was published in Research in Developmental Disabilities in 2014 by Danish researchers. This is exciting because they have done follow up developmental tests on some AS children 12 years after the initial tests. The article is entitled: Neurodevelopmental outcome in Angelman syndrome: Genotype–phenotype correlations. They looked at 42 children with varying mechanisms (deletion, UPD and mutation) and found as has been previously found that the children with deletion have lower developmental age, especially those with a larger deletion. They also used a test for autism (ADOS) and found that around 70% of the children with deletion scored above the autism cut off on the ADOS, meaning a diagnosis of Autism Spectrum Disorder could be made if history and observation was corroborative as compared with 10% of the kids with UPD and mutation. They commented however that it is very difficult to assess children with developmental delay for autism and children with AS generally are socially engaged and seek out adults with positive vocalisation and facial expression.

They then reassessed 7 deletion clients who had been assessed 12 years earlier with the developmental test and the ADOS. They found that there was no significant change in cognitive skills but there was improvement in language skills. There was no change in the ADOS scores.

The last article I want to talk about is a commentary on a case study by WH Tan published this year in Clinical Chemistry. Dr Tan has published many articles on AS. He mentioned that Tegretol and Phenobarbitone can worsen seizures in AS and that Epilim and clonazepam and low GI diets can be helpful.

All the best,

Antonia

Newsletter by email

If you would like your newsletter emailed (PDF for-mat) to you instead of being posted send a note to Kevin Kennedy at kevin.kennedy@angelmansyndrome.org requesting that your future newsletters be emailed to you. This is not only cheaper for the association but faster for our current Treasurer who volunteers his time while, like us all, juggling work, family and life with an angel.

NDIS Quality & Safeguarding Framework Consultation

Have your say on NDIS quality and safeguards

Are you interested in the National Disability Insurance Scheme (NDIS)?

The Australian and state and territory governments are working together to develop a new national quality and safeguarding framework for the NDIS. It's about making sure people with disability are provided with quality support, choice and control to pursue their goals and aspirations.

What do you think are the best ways to ensure that people have access to high quality supports?

Take a look at the consultation paper to see what issues are being considered. Think about how the different quality and safeguarding options affect you or those you care for. Perhaps you have some new ideas for maintaining quality and safety

Have your say on any of the key topic areas:

- building capacity
- monitoring and oversight
- National Disability Insurance Agency provider registration
- systems for handling complaints
- ensuring staff are safe to work with participants
- safeguards for participants who manage their own plans
- reducing and eliminating restrictive practices in NDIS funded supports.

How to get involved

You can get involved by making a submission, joining the discussion in the online forum, completing a questionnaire or attending a public meeting.

Visit **engage.dss.gov.au** to download the consultation paper and fact sheets and have your say today.

Thanks to Mary Bills for contributing this article.(Editor)

For News and inspiring stories relating to the NDIS go to the website:

www.everyaustraliancounts.com.au

Mums weekend away

February

Glenelg, South Australia

A wonderful weekend away was enjoyed by 29 AS mothers and a grandmother, as well as a mother of two children with AS. Mothers came from all over Australia including Broome WA, country Victoria and Queensland. There was laughter aplenty and dancing by some (who shall remain nameless) until the wee hours of the morning. The weekend enabled many mothers to let their hair down, relax, network, share and most of all, to not feel alone in many of their thoughts which were shared openly on this amazing weekend. It was a cathartic feeling for many. On the Sunday a lovely mini tour of Adelaide occurred with a stop at Mt Lofty (pictured) with amazing views. This has been the second successful getaway thanks to the amazing Michaela Townsend and she is planning to make it a yearly event. Watch this space.....

Lysandra.

I would like to say it was very daunting for me as being the oldest mum of an AS, as all the mums were in their 20's 30's and 40's - would I fit in? Well I learnt SO MUCH from the younger mums as medicines, technology plus disposable nappies have made their path a lot easier than the unknown.

Every moment of the weekend was fully organised by the wonderful Michaela from breakfasts, lunches and dinners to excursions for the in-between times. Here a bunch of normal looking mums who ache on the inside about the unknown and what the future holds were enjoying dinners full of laughter and the most unusual stories. Face-to-face friendships were formed rather than FaceBook friends and a big thankyou to Emily in Australia for her role as Moderator of FaceBook. Now, back home, life continues and I'm eagerly awaiting the next AS weekend away.

Jenny.

SOURCE kids

SourceKids is a free magazine distributed throughout Australia and also available online. It is Australia's first magazine for children with special needs. However, the information provided can be very useful for parents of adults as well. Bikes and trikes, advocating for your child in a medical minefield, nutrition and Fragile X syndrome are just a few of the articles published in the December issue.

Go to:
www.sourcekids.com.au

QANTAS CARER CONCESSION

Nican administers the Qantas Carer Concession Card on behalf of Qantas.

The Qantas Carer Concession Card is issued to people with a disability and high level support needs who require the full-time assistance of a carer whilst they are on the plane. A person is eligible if they need to have one-on-one support when seated on the plane for assistance with meals/drinks, transferring to the bathroom, orientation, communicating with the flight staff etc. A person would not be eligible if they only need assistance boarding the plane, or when they arrive at their destination.

Qantas Carer Concession cardholders and their nominated carer will receive the following discounts for Qantas domestic travel within Australia.

For Qantas Carer Concession Cardholders: 1

10% discount on domestic Economy Class fares

For Nominated Carers:

50% discount on domestic Economy Class fares

Business Class Travel For Qantas Carer Concession Cardholders:

50% off J class Business Class Fares when D class are available

For Nominated Carers:

50% off J class Business Class Fares when D class are available

Notes:

All discounts are subject to booking class availability.

Bookings for the cardholder and their nominated carer must be made at the same time and both bookings must be made using the Card in order to obtain the discount.

Fare conditions for community fares apply and not the fare rules of the published fare type.

The Qantas Booking Fee is not payable on bookings made using the Qantas Carer Concession Card.

Discounts are not available:

- on international travel;
- on flights operated by any of Qantas' alliance partners; or
- in conjunction with any other concessional airfare eg airfares for children and seniors

This card is a photo ID card which is valid for three years and has an administration fee of $27.50 including GST.

For further information and an application form contact Nican.

http://nican.com.au/about/qantas-carer-concession

(Thanks to Anne Funke for providing this article.)

Up, up and away with fairer service

Thanks to some great work by the Public Interest Advocacy Group and the Anti-Discrimination Board of NSW, people with a disability flying with a carer can now access discount online fares with Virgin Australia.

The issue was taken up when Jenny Brown tried to book a flight online and because she didn't know the name of her son's carer in at the time of flying, she couldn't book the flights. Virgin required the names of the people flying for the booking to be made.

The fact is, her family books carers through a nursing agency and they rarely know who the individual carer will be very far in advance as it depends on the carer's rosters at that time.

As we all know, the closer you book your flight to the time you want to fly, the more expensive it is likely to be so that means Ms Brown was stuck with expensive flights or missed out on seats altogether.

Thankfully, Virgin Australia has now changed its policy and people travelling with a carer can make a reservation over the phone up to 331 days in advance at the internet rate. They then can confirm the name of the carer any time up to 72 hours before the flight.

Great work Ms Brown, the Public Interest Advocacy Group and the Anti-Discrimination Board of NSW! And Virgin Australia for stepping up and changing the policy.

From the latest NDIS newsletter.

www.everyaustraliancounts.com.au

orphanet

You may be interested in reading the newsletter from Orphanet which is a reference portal for information on rare diseases and orphan drugs. Orphanet's aim is to help improve the diagnosis, care and treatment of patients with rare diseases. The Australian site for Orphanet is:

www.orpha.net/national/AU.EN/index/homepage/

The newsletter is titled:
OrphaNews—The Newsletter of the Rare Diseases Community.
In the newsletter dated 17 March, I found this mention of a paper:

Angelman syndrome: partial restoration of UBE3A protein in mouse model ameliorates some cognitive deficits

Consult the Pubmed abstract

To read more about "Angelman syndrome"

Nature ; 518(7539):409-12 ; February 2015

Disclaimer

The views expressed in this newsletter and any enclosures are not necessarily those of the Angelman Syndrome Association. Information is presented in the interest of providing a range of alternatives. Inclusions in this newsletter does not imply endorsement by the Angelman Syndrome Association.

Angelman sites on Facebook:

Australian families with AS
Angelman Research & Trials
Angelman Syndrome Foundation
Angelman Connections
Angelman Directory
Angelman Grandparents
International Angelman Day
Buy/Sell Disability products Australia New & Used

Some useful Apps:

Free: **Sounding Board** - you can use either compic with the computerised voice

Or choose your own pictures and own voice,

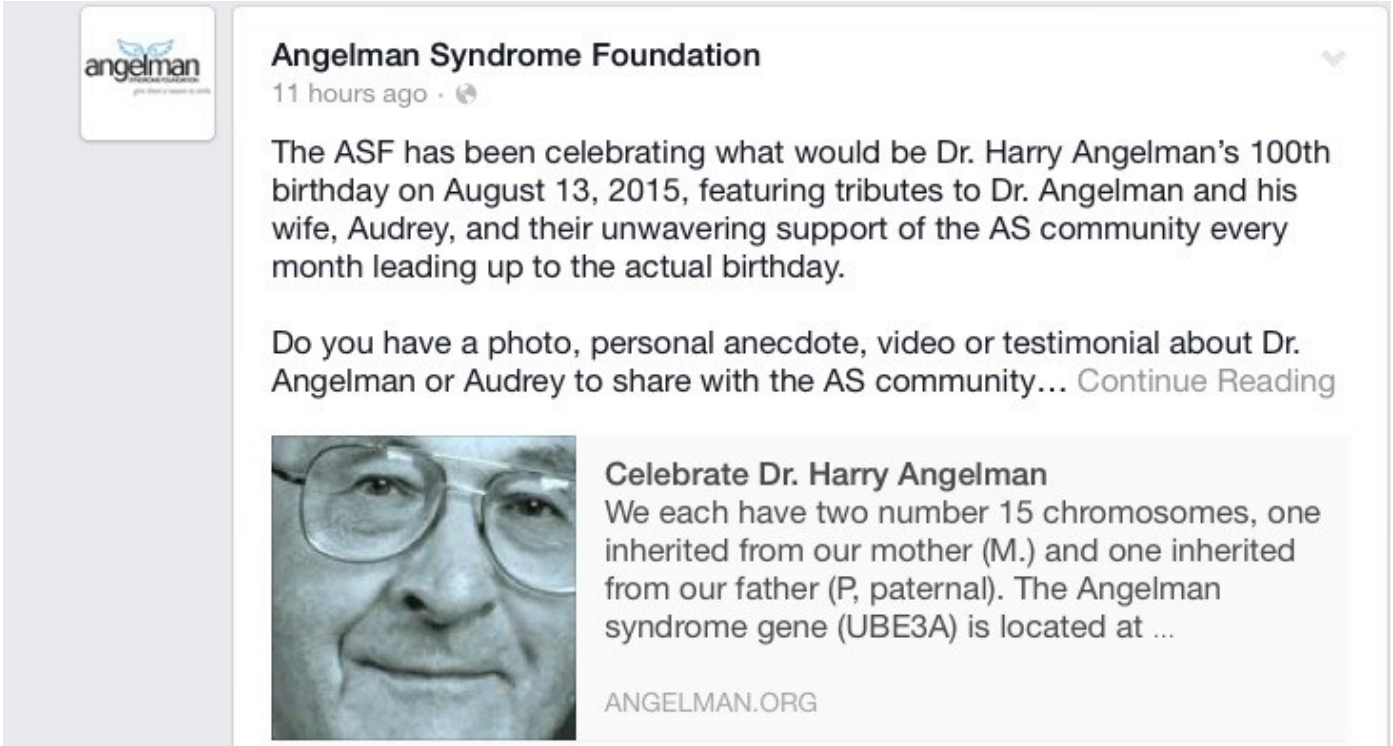
Great for using family/friends and personal photos.

Balloon pop

Fireworks

Fingerprint

Many of the Autism Apps may be useful too.



Thanks to Jenny Chippindale, NSW, for these contributions. (Editor).

The Cerebra Centre for Neurodevelopmental Disorders at the University of Birmingham in the United Kingdom are conducting research into sleep in children with neurodevelopmental disorders and are looking for interested families to complete an online questionnaire about their child's sleep quality.

Who can take part?

Parents/carers of children with Angelman syndrome aged between 2 – 15 years (inclusive).

Why is this research being done?

We are interested in working with individuals with Angelman syndrome because sleep disorders are particularly common in this syndrome. The findings from this research will be very important in helping us to develop ways of helping individuals Angelman syndrome in the future.

What will it involve?

The project is called 'Sleep in children with neurodevelopmental disorders,' and aims to further our understanding of sleep quality in children with neurodevelopmental disorders. We are inviting parents to complete a questionnaire about their child's sleep quality, behaviour and health, and any impact on parent/carers' well-being.

Who should I contact to take part/for more information?

There is an information sheet which can be accessed via the link below that gives you more details about why the research is being carried out and what participation will involve.

After having reading the information sheet, if you think that you and your child you care for are interested in taking part in this study, you may complete the consent form and an online questionnaire.

Alternatively, if you have further questions about the study or would like a paper copy of the questionnaire, please either telephone Jayne Trickett on +0044 121 414 2855 or email her at jxt292@bham.ac.uk

Please follow this link to read an information sheet about the study:

http://tinyurl.com/pt2enjs

If the above tiny URL does not work, please enter the full link:

https://lesweb2.bham.ac.uk/surveys/index.php?r=survey/index/sid/293485/lang/en

The password is Cerebra