Christmas 2014
Newsletter

The ECHO study is growing!

ECHO STUDY

(Experiences of people with copy number variants)

Contents

Dear ECHO study families,

It is hard to believe it is almost Christmas again and time for our Season's newsletter. Many things have happened since our last newsletter. There are now 239 ECHO study families. As many of you know, we are re-contacting families about two years after the first visit to schedule a second interview. We have now seen 70 families twice and this is greatly helping us understand the changes that take place throughout childhood and adolescence. Furthermore, we are now also interviewing participants aged 18 years and older. This will allow us to gain valuable insights into the experiences of adults with Copy Number Variants (CNVs).

Parents have told us about their children's lives and older ECHO study participants have given us insights into their own experiences. We feel very privileged that you have shared your stories with us.

We have learned a great deal from meeting and talking with you and we will make sure that we keep informing the clinical, scientific and other communities about what we have learned.

Below, we will give more information about what has happened since we were last in touch with you by newsletter. I hope you will enjoy reading our update.

I am wishing you and your families a Merry Christmas and a very happy New Year.

Marianne van den Bree

Professor of Psychological Medicine

ECHO study Lead Investigator

THANK YOU!

239

We have now seen an astonishing 239 families!

We would like to say a special thank you to all of our ECHO families who have given up their time to help us reach this milestone!

We are now aiming for 250 families.

If you know anyone who would like to take part in our study please tell them about us and help us reach our goal! (contact details are at the end of the newsletter)

Knighthood for Professor Michael Owen

We are pleased and very proud to announce that Professor Michael Owen, one of our Principal Investigators, has received a knighthood in recognition of his services to Neuroscience.

Professor Owen is the Director of the Institute of Psychological Medicine and Clinical Neurosciences where the ECHO study is based and joins us for weekly meetings where everyone is updated about the study's progress.

He has contributed to over 800 publications and has made a huge impact in neuroscience and psychiatry.

Congratulations Professor Owen!

Meet our new team members

Prof. Jeremy Hall

Dr Stefanie Linden

Prof. David Linden

Dr Andrew Cuthbert

Ffion Evans

Sophie Andrews

Meet our new team members

Adam Cunningham

Hayley Anne Moulding

Alys Glover, Jessica Townsend & Josh Roberts

The ECHO team has grown significantly this year with new members bringing their skills and strengths to our team.

Professor Jeremy Hall has expertise in psychiatry and learning disability, Professor David Linden has expertise in psychiatry and brain imaging and Dr Stefanie Linden is an adult psychiatrist. Dr Andrew Cuthbert is a Genetic Counsellor. Ffion and Sophie are Psychology Assistants. Adam and Hayley Anne are our new PhD students and last but not least Alys, Jessica and Josh are our placement students.

2015 looks like it is going to be a very busy year!

The ECHO study continues to grow!

2014 has been a very busy year for the ECHO study.
We have been very lucky this year as not only have lots of new families helped us with the study, we have also visited many families for a second time as part of our 'longitudinal study', where we are following children as they grow older.

The ECHO study has also branched out into new areas this year.
Over the next few pages we will take a quick look at our progress…

The Adult Study

We are excited to welcome Professor Jeremy Hall, Professor David Linden, Dr Stefanie Linden and Ffion Evans who have joined the ECHO team as part of the new adult study.

The adult study is looking at the experiences of people aged 18 to 89 years who have been diagnosed with a copy number variant (CNV) (or chromosome disorder).

As part of this study, adults are visited at home and complete interviews and puzzles which are similar to the ones in the child study. Participants are also invited to Cardiff where they can take part in brain scans.

The study has got off to a flying start, the team have visited fifteen adults and four adults have travelled to Cardiff to take part in brain scans.

If you would like to know more about the adult study, please don't hesitate to get in touch with us.

Email: echo@Cardiff.ac.uk

Telephone Ffion: 02920 688 242

A Brand New Study!

Experiences of families living with rare chromosome disorders

Where do parents discover information about their child's condition?

As part of a new study initiative, we would like to invite parents and guardians of children with chromosome disorders to complete an online questionnaire about their experiences (this takes approximately 20 minutes).

With your help we would like to know:

- How you found out about your child's chromosome disorder.
- Your experiences of services (for example, the medical genetics service).
- How you think the services you received could be improved.

The survey can be found here:

https://cardiff.onlinesurveys.ac.uk/informationsources

In the future we hope to also interview GPs, geneticists, genetic counsellors and support group representatives. The overall aim is to improve service provision.

Please do not hesitate to get in tough if you would like to find out more

echo@Cardiff.ac.uk
Email

02920 688 358
Phone Aimée

Spreading word about the study

The ECHO team have been attending conferences throughout the UK and beyond to let people know about our research

16p Family Meet-up in Manchester

We were very kindly invited to a 16p family meet-up in Manchester. This was organised by a great Facebook support group for families with 16p11.2 deletion and duplication syndromes. Hayley, Maria and Sam gave a presentation about the study and some preliminary findings. It was really great to meet so many families and children! We had a lovely day, and the cake was spot on!

If you're not already aware of this support group on Facebook, please search for *'16p11.2 deletion we are family xx'* , you'll be welcomed by all of the other families. Family support groups on social media are a great way of meeting people and sharing your experiences with people who really know and understand the issues your children may face.

International conference for psychiatric genetics

In October Sam travelled to Copenhagen, Denmark to represent the ECHO study at an international meeting and to raise awareness of our work to geneticists from around the world.

Dundee MaxAppeal family meet up

ECHO visits families from the whole of the UK. Whilst Sam & Jess were in Scotland they attended MaxAppeal's 22q11.2 family meet up at Dundee's science centre. They had a great time chatting to families and exploring the science centre!

Professor van den Bree

has been raising NHS geneticists and paediatricians' awareness of chromosome disorders.

RCPsych International Congress 2014

Maria also presented some preliminary findings on the 16p11.2 Deletion Syndrome and the 16p11.2 Duplication Syndrome in the International Congress of the Royal College of Psychiatrists which was held in London on the 24th of June.

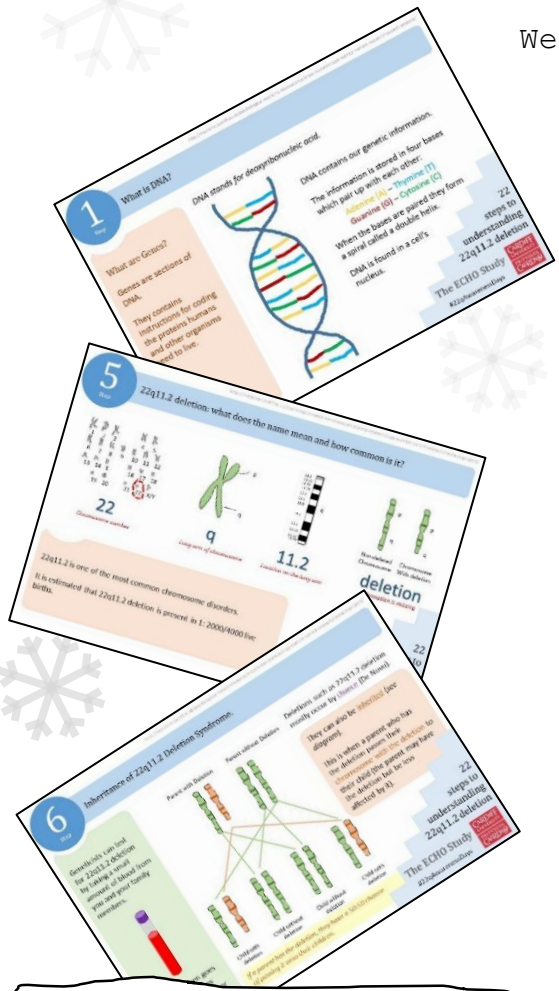
Spreading word about the study

22qAwarenessDays

This year, November was full of activity as we were invited by the Facebook group '22qAwarenessDays' to promote awareness of 22q11.2 deletion syndrome. Each day the ECHO team uploaded a tutorial explaining aspects of 22q from our '22 steps to understanding 22q' series. Our involvement culminated in us taking part in a live Twitter chat, which we really enjoyed! The awareness month was hosted brilliantly by @22qAwarenessDays and @22q11Ireland on Facebook and Twitter. We are extremely thankful to them for organising such an important awareness month and grateful that they let us play a small part in it! We are now turning our 22 steps to understanding 22q slides into leaflets, which we will make available to families through our website.

If any parents would like to organise a similar awareness event about any of the other CNVs we study, please get in touch with the team.

We are more than happy to help!



9th Biennial International 22q11.2 Deletion Meeting

Marianne, Jo, Maria, Sam and Afnan presented our recent findings in the 9th Biennial International 22q11.2 Syndrome Meeting in Palma de Mallorca, Balearic Islands, Spain this June. The meeting is held every two years and is an opportunity for 22q11.2 researchers to share their findings and to work together on trying to understand the syndrome. **Cardiff University is one of five lead sites of the International 22q11. 2DS Brain and Behaviour Consortium. http://www.22qsociety.org/downloads/BBC_Update-Nov2014.pdf**

Cardiff Half Marathon fundraising success!

A HUGE congratulations to Natalie, Sam, Ffion, Laura and Maria who put on their running shoes and completed the Cardiff Half Marathon.

The team set off with over 21,000 other runners to complete the 13.1 mile course around some of Cardiff's most famous landmarks. Thankfully the rain clouds held off and the team stayed dry (apart from a few beads of sweat).

Natalie, Sam, Ffion, Laura and Maria all successfully crossed the finish line and raised an amazing £1000 for Unique, a great charity that supports families affected by chromosome disorders. Unique supports our ECHO study and has been a great help to us over the years, providing input on new study ideas and telling families about our study. If you would like to find out more about their work visit www.rarechromo.co.uk.

Revisiting families: an update

70 families have been revisited as part of our longitudinal study!

It is important to understand how children with genetic syndromes develop and what changes occur in childhood and the teenage years. This is why we are now re-contacting families, approximately 2 years after our first visit.

We would like to thank all the families who have already taken part. We will prepare reports of the valuable information we are learning through this study and spread the word.

Watch this space!

The **ECHO Team**

would like to wish you a very

Merry Christmas

and a

healthy and happy

2015

Contact

If you would like to find out any more information about the ECHO study, or know anyone who would like to take part in our study, please get in touch!

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Adult study
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