Welcome to the 2015 Kleefstra syndrome conference summary which follows the success of last year’s event held in Coventry, UK on 15th August. Once again we were fully booked with 36 families attending from around the world to learn more about Kleefstra syndrome.

The weekend offered a unique chance for families to learn from the expertise of our guest speakers during the formal presentations while being able to ask questions relating to their own individual circumstances. Throughout the weekend families took part in 1:1 sessions, video interviews and most importantly were able to network with other families affected by Kleefstra syndrome to learn from each other’s experiences.

We would like to take this opportunity to thank all those who attended and helped to make the weekend a complete success.

Chris & Fiona Heslehurst.
We celebrated the 3rd Kleefstra syndrome awareness day on 17th September, the 9th month of the year is September and the 17th letter of the alphabet is Q, together this makes 9Q.

We like many families rushed home on diagnosis day and took to the internet in search of answers, what we found were old medical journals which were difficult to understand and very negative. Together we set up the website in May 2008 because we felt passionate about providing a network of support for families and the opportunity to share positive experiences.

We hosted our first gathering of 9 families in August 2011 and our first conference with 27 families attending in August 2013.

When Leah was diagnosed with Kleefstra syndrome we were told she was number 35 in the world. We were overwhelmed and amazed to be able to welcome 36 families to the 2015 conference.

Families came from Canada (1), US (2), China (1), Poland (1), Switzerland (1), Denmark (1), Sweden (1), France (4), The Netherlands (3), Italy (1), Ireland (1) and UK (19). Individuals from the age of 1 to 23 years were represented by their families.

During the conference weekend many of our families had the opportunity to have 1-1 consultations with Professor Chris Oliver and Dr Jane Waite from Birmingham University. The appointments focused on behavioural aspects of Kleefstra syndrome and changes with age.

A big thank you to Donna Solomon and Rachel Leak who organised a fabulous care facility (crèche) for our young people during the conference. The theme for the day being “Under the sea”.

INTRODUCTION

Chris and Fiona Heslehurst are parents to Leah (10) and Lucas (6). Leah was born in 2006 and diagnosed with Kleefstra syndrome in 2007, or 9q34.3 deletion syndrome as it was then known.

Chris works full time in the IT department for a local council and Fiona works as a teaching assistant within a local special school.

Kleefstrasyndrome.org was able to help subsidise the cost to families attending the conference with the help of a small grant awarded to FIND from the Economic and Social Research Council.

Money was also kindly raised by families which helped to subsidise the crèche, children’s entertainer and conference materials.

We celebrated the 3rd Kleefstra syndrome awareness day on 17th September, the 9th month of the year is September and the 17th letter of the alphabet is Q, together this makes 9Q.
The children had the opportunity to take part in lots of lovely activities including singing, dancing, crafts, stories, messy play, activities in the garden, music therapy and a magic act which incorporated Makaton sign language. Activities for the siblings were organised too with a special time set aside to share experiences and feelings associated with having a brother or sister with Kleefstra syndrome. Thank you to Rachel and all her volunteers for their amazing efforts.

Two professional photographers captured many amazing pictures and memories from our conference. This was made possible by a generous donation from one of the attending families. We hope to get those images onto the website and Facebook community page very soon!

To access our social networking sites please see page 14 for the links.

**FUTURE AIMS**

Our future aims include investigating the feasibility of gaining charity status along with a refresh of our website, so keep your eyes peeled for our new look. We also aim to continue momentum with our social media pages on Facebook and Twitter, making them easily accessible for those needing support in the future. Many prefer Facebook these days so we will look to make better use of social media, although as always privacy and security are of utmost importance. We also intend to start work on the 2017 Kleefstra syndrome conference in the coming months!

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**WRISTBANDS**

We now have Kleefstra syndrome awareness wristbands for sale. The bands are £1 each + postage and available in adult size only.

If you wish to order some wristbands bands please contact us by e-mail:

info@kleefstrasyndrome.org

Please let us know the number of bands you wish to purchase, your country of residence and your PayPal e-mail address.
Dr Kleefstra works at the Department of Human Genetics, Radboud University, Nijmegen Medical Centre, The Netherlands.

Kleefstra syndrome - previously known as 9q34.3 deletion syndrome was re-named in April 2010 after Dr Kleefstra identified the gene responsible for the syndrome - EHMT1. The identification of Kleefstra syndrome was possible after the introduction of micro chromosomal sequencing, when smaller deletions were able to be seen. Dr Kleefstra noted that access to facilities to identify the deletions may still be an issue in certain areas.

Although the number of diagnosed cases is not known due to the fact there is no central registry database, it is very apparent that there is an increasing number of patients, in fact recent figures from the Deciphering Developmental Disorders (DDD) study in the UK found that EHMT1 is in the top 15 of WES (Whole exome sequencing) results.

Dr Tjitske Kleefstra

Dr Kleefstra also noted that the reprogramming of somatic cells opens up a new field of study. It was stated that the team may be looking for families to take part in this research in the future, which involves the taking of a skin biopsy under local anaesthetic.

An information sheet with further information will be available shortly so once we have further details on how to take part we will place an update on the website and Facebook pages.
Dr Kleefstra spoke about the challenges of obtaining funding for research of rare ID/autism disorders and that as a result there is a real need for a patient registry and an international collaboration study with a network of dedicated professionals.

There is however pressure from the European Union to have more research into rare disorders, with recognised expert centres, so there is hope for Kleefstra syndrome.

Dr Kleefstra shared past and ongoing research and findings of particular interest were the ongoing research on mice models.

EMHT1 +/- mice show reduced exploration and increased anxiety, decreased social play, memory deficits and fear.

“Thank you for your great investment in bringing the families and knowledge together, I realise I have two Kleefstra families!”

Dr Tjitske Kleefstra.

RADIANT Survey

At the end of Dr Kleefstra’s presentation, families were handed an optional survey on behalf of RADIANT. RADIANT are an international team of experts in rare diseases who’s aim is to conduct surveys asking individuals and families affected by such diseases what treatments they would prefer in the future for their respective conditions. They intend to initiate and coordinate clinical trials for rare developmental disorders, develop treatment guidelines and educational support for health care professionals.

The overall surveys are ongoing but we have preliminary results from the Ks conference which shows families believe it’s more important for professionals to concentrate their efforts on both Intellectual and Behavioural difficulties.
Hira and Andy explained the motivation behind the foundation had been their respective children Nikhil Verma age 3 and Natalaya Klump age 2, both who have Kleefstra syndrome.

Hira explained that the GeneSpark.org has an aggressive approach to their goals, preliminary project discussions have already taken place with formal proposals expected in soon. There is a $1 million immediate funding goal to launch 2-3 key projects.

GeneSpark.org are asking families to get involved by signing up to the newsletter, submitting data to the patient database and to volunteer for specific roles within the organisation. For further information please visit GeneSpark.org
Cindy qualified as a Music Therapist in 2006 and is HCPC registered. She initially started working for a music therapy charity in North Yorkshire offering a service at special schools, mainstream schools, medium secure hospital units, day centres and family homes.

Considerable experience has also been gained in the NHS working with a number of individuals with Rett syndrome. Cindy is now Music Therapy Advisor to Rett UK, attending conferences and regional days and has a paper published about her work with Rett syndrome – A retrospective practice based evaluation of music therapy: Rebecca’s Story.

Alongside this Cindy provides music therapy into the Autism Service offering individual and group sessions and is qualified to carry out Autism Diagnostic Assessments.

“Music is a powerful emotional medium, which can affect us all deeply. In music therapy sessions, interactive music is spontaneously created by the client and therapist” Cindy-Jo Morison.

FURTHER RESOURCES

If you are interested in trying Music Therapy for yourself, Cindy has created a document to help. It also includes a handy list of iPad apps which she uses during her sessions.

The document can be downloaded here:

http://bit.ly/1Qzu8Mh
The iPad is a useful tool used in conjunction with instruments such as Mini Egg Shaker Maracas, Egg Shakers, tambourines and bells. Cindy produced a list of frequently used iPad apps which contribute to her sessions.

- Beatwave
- MelodicaFree
- Air Guitar
- Bubble Harp
- Virtuoso Piano
- Relax Melodies HD
- Magma
- Electra
- iMeba
- Beamz
- Bloom
- iKaoscillator
- Aquasonic
- 12 String
- Bubl Draw
- ThumbJam

Another useful accessory is the Wowee One Classic portable speaker which increases volume from the iPad and vibrates at the same time.

Music therapists help find a voice – a means of communication through the use of improvised music. Music has a physical effect on us and often offers a way to express “that which cannot be put into words”. Through music we communicate at different levels. Everyone can respond to music irrespective of disability or illness. Therapy sessions can bring about changes in listening and communication skills, sound tolerance, vocalisation, playful interaction and confidence. Whatever the level of participation the client is always in charge. Feelings of joy and satisfaction and equally of anger and frustration can emerge in any session. Music therapists are trained to explore and contain these difficult feelings and in doing so the individual is able to show emotions that cannot be easily expressed in other ways.

During the conference some video footage was shown of music therapy sessions with Chris & Fiona’s daughter Leah (age 10) who has Kleefstra syndrome. The videos showed the progress made by Leah throughout her sessions which have been taking place for around a year. Leah has grown in confidence and developed skills in a number of areas owing to music therapy.

A huge thank you to Cindy for giving up her time to speak at the conference, for providing a group session for the children within the crèche and for being on hand to discuss music therapy with parents throughout the weekend. Having seen her expertise work for Leah first hand we hope others found this session useful.

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Professor Chris Oliver and team from the University of Birmingham are interested in furthering research into Kleefstra syndrome with a view to developing support and intervention for diagnosed individuals and their families. Their team are launching a new website called FIND in March 2016 which is a novel online resource aiming to improve the exchange of knowledge about rare genetic conditions and difficulties experienced by people with Intellectual Disability. This year the site will be expanding to include Kleefstra syndrome. The site focuses on behaviour and its purpose is to summarise research studies using a range of interactive and engaging formats, for example, parent stories, professional talking heads, bite-sized written information and an interactive database.

FIND is driven by the aim of providing current research findings to families and professionals in a bite-sized format. Learn more at www.findresources.co.uk

During the weekend Chris and his team conducted 1:1 consultations. These sessions were designed to allow parents an opportunity to discuss any concerns they may have around challenging behaviour whilst allowing the staff from the university to collect and collate information to further their studies and research. Those families who took part in an individual consultation received direct feedback, advice and signposting which proved to be very helpful. Those who completed the questionnaire will receive feedback based on upon their responses.


During the conference, some parents were filmed describing their experiences of having a child with Kleefstra syndrome. Topics included characteristics of autism spectrum disorder, social skills, communication, mood and behaviours that challenge. We are very excited to share with you the first in a series of six videos on the FIND website here: [http://bit.ly/1Spcrkk](http://bit.ly/1Spcrkk) with the rest to be published very soon.

A full overview of the presentation given by Prof Chris Oliver can be found on page 10.
Overview of Presentation by Prof Chris Oliver:

Understanding and Changing Behaviour in Kleefstra Syndrome

Chris Oliver, Alice Welham, Jane Waite, Tracey Grandfield and Jo Moss

Everyone with Kleefstra Syndrome is an Individual

It is important to remember that in many ways children and adults with Kleefstra syndrome are just the same as everyone else. They have the same needs, wants and rights.

However, in some ways, children and adults with Kleefstra syndrome differ from people who do not have this syndrome. It can be important to know about these differences when supporting a person with Kleefstra syndrome.

There are lots of factors that influence our behaviour including our culture, our experiences and our genes.

Is Behaviour Hard-Wired?

At present, there is very little research evidence to suggest that any specific behaviour is ‘hardwired’ in the brain; however, we do see different patterns of behaviour depending on which genetic syndrome a person is diagnosed with. Therefore, a person’s genetic syndrome can make it more or less likely that certain behaviour will develop.

Behaviour in Kleefstra Syndrome

Our research suggests that individuals with Kleefstra syndrome are very sociable with both familiar and unfamiliar people.

Around 6 out of 10 individuals with Kleefstra syndrome show self-injurious behaviour. This is higher than people with intellectual disability generally; however, it is lower than some other genetic syndromes; for example, over 9 out of 10 people with Smith-Magenis syndrome will show this behaviour.

Around 6 out of 10 individuals with Kleefstra syndrome will engage in behaviours that could cause physical injury to another person (e.g. pushing, hitting). However, it is important to remember that this behaviour is not necessarily intentional.

Around 9 out of 10 individuals with Kleefstra syndrome reach cut-off for an Autism Spectrum Disorder on a screening measure, and 6 out of 10 reach cut-off for Autism. However, this screening measure is not the same as a diagnosis of Autism, which requires a much in-depth assessment.
Profile of Autism Spectrum Disorder in Kleefstra Syndrome

While many individuals reach cut-off for an autism spectrum condition on a screening tool, the profile of autism spectrum disorder characteristics may be different in Kleefstra syndrome to idiopathic autism. This is because children with Kleefstra syndrome have high levels of sociability. Further research needs to be done to fully understand these differences.

Behaviours that Challenge and Pain

Pain from an undiagnosed medical condition (e.g. ear infection, gastroesophageal reflux) can increase the likelihood that a person will show challenging behaviour.

We have produced a pain guide that can be downloaded from:


This guide explains how pain can lead to challenging behaviour and suggests ways that parents can monitor pain over time.

Behaviour is a Form of Communication

Challenging behaviour can serve a function for an individual. Behaviours do not necessarily start out with a function, but acquire this over time due to the consequences that typically follow the behaviour.

One example is when behaviour starts to serve the function of obtaining social attention. When a child shows self-injurious behaviour this is typically, and of course, naturally, followed by a parent trying to intervene to reduce the behaviour. Over time the behaviour begins to serve a function of obtaining attention from the parent. In this example, the child is most likely to start showing this behaviour in the future when they are bored and no one is interacting with them. This is not to say that either the child or parent is to blame; simply that the behaviour and the positive consequence get paired together over time.

Which Functions can Behaviours Serve?

Not all behaviours function to access social attention. There are a number of functions that behaviour can serve, and this can differ from child to child. Common functions are: escape from non-preferred activities, access to social interaction, access to sensory stimulation, access to preferred objects/items. In addition, sometimes the same type of behaviour can have different functions for a child, depending on the situation they are in. As behaviours are typically rewarded by external consequences, interventions can be effective at reducing behaviours by changing these consequences.

Remember, Behaviours are a Form of Communication

It is important to remember that challenging behaviours are more likely to develop in children with limited communication strategies. This is because it may be difficult for a person with a communication impairment to make requests in other ways. Therefore, working on developing your child’s communication (i.e. learning a sign or picture to request a break or access to preferred activities) can help to reduce behaviours that challenge.
Resources
To learn more about behaviour and how to intervene, visit the challenging behaviour foundation: http://www.challengingbehaviour.org.uk/

There are also good resources on FIND. These resources were not developed for Kleefstra syndrome specifically, however, the principles for understanding behaviour are the same: www.findresources.co.uk

Who Can Help?
If your child engages in challenging behaviour, a referral to a Clinical Psychologist with expertise in behaviour interventions may be appropriate. Usually referrals to these services can be made by your G.P. or paediatrician.

Who Are We?
The Cerebra Centre for Neurodevelopmental Disorders (CNDD) is headed by Professor Chris Oliver, and is situated within the School of Psychology at the University of Birmingham. The centre has been funded by Cerebra since 2008 and is the largest of its kind in the UK. At the centre, clinical and academic psychologists, undergraduate and postgraduate students and volunteers conduct high quality research into emotional, cognitive and behavioural difference and disorder in children and adults with neurodevelopmental disorders. In addition to carrying out research, we also translate the latest findings into effective and practical assessments and interventions. This enables the provision of information, advice and support to parents, carers and professionals. The research work conducted at the Cerebra Centre includes the study of numerous different neurodevelopmental disorders. The majority of these are rare genetic syndromes, which have not been the subject of a great deal of research due to their rarity. CNDD believe that research in these groups is crucial in order to raise awareness of these underrepresented groups, and thus enhance the quality of life of affected individuals.

www.findresources.co.uk
Karlijn Vermeulen, child & adolescent psychiatrist and PhD-student at the Radboud University Nijmegen in the Netherlands, spoke at the conference regarding her ongoing research studies.

A pilot study of 5 patients with Kleefstra syndrome was completed in 2013 and further funding was obtained to continue this research. This is an ongoing study of psychopathology in 24 subjects with Kleefstra syndrome.

Karlijn was able to give the conference attendees an insight into some of the preliminary results. She showed a difference in developmental profile between the male and female (ratio 1:2) participants. The female participants in general reached higher levels of development. More research into why there is a marked difference in male and female individuals needs to be carried out.

“This has given me the opportunity to give personalised advice in diagnostics and treatment. Together, we can make a difference”  

Karlijn Vermeulen

All participants showed autistic features which were measured with a qualified observation schedule.

It was also noted that individuals with Kleefstra syndrome score highly for happy moods!

Prevalence of other psychiatric disorders (major depression, generalized anxiety, (manic) psychosis) were high, whereas treatment rates for these disorders were low.

Karlijn highlighted the importance of treatment in general for these disorders, noting that patients with Kleefstra syndrome have a vulnerable brain. She paid special attention to treating sleep disturbances in late adolescence/early adulthood, because these seem to precede regression.

The latest results have recently been published in a paper which is freely accessible online. The paper is entitled “From a Single Gene Defect Towards a Cross Species Neurocognitive Phenotype: The EHMT1 Disruption Example (Kleefstra Syndrome)”. You can download the paper in PDF format here:  http://bit.ly/1N3Ckzw

Karlijn met recently with members of the government in The Netherlands and has been invited to give presentations throughout the country. It is hoped this will create awareness amongst other health care professionals of Kleefstra syndrome, and propose a prevention/treatment plan for some of the psychiatric issues associated with the syndrome.

Karlijn continues her research into Kleefstra syndrome and work on other related papers which she hopes will be published in 2016.
FEEDBACK COMMENTS

“We met great people, everything was perfect. We got so much more information than we expected.”

“Thank you for the opportunity to meet so many different people with Ks, also, being able to speak to knowledgeable doctors/clinicians. Well done!”

“I loved the approach Prof Chris Oliver had. He didn’t shy away from the problems, but he had such an empowering way of talking about it. The feeling was that "we can do something" which was very important to me. I feel empowered as a parent towards the surrounding community; schools, doctors and others.”

“Helpful, empathetic with excellent suggestions how to move forward with several aspects of our child’s health and psychological well-being.”

“The one-to-one was brilliant - again, Jane really knew her stuff, but most important of all she could ask us the right questions and listen. I realised afterwards, that this was the first time a professional has done this in any systematic way.”

“Thanks for a brilliant conference, great to meet the doctors and researchers - it wasn't long enough!”

“We really appreciated and enjoyed the presentation by Cindy, it inspired us to try Music Therapy with our own child.”

“Fantastic organisation and welcoming atmosphere, please, please, please arrange another one!!”

SOCIAL MEDIA RESOURCES

‘Like’ our Facebook page, search Kleefstrasynndrome.org

‘Follow’ us on Twitter @Kleefstrasynndro

Kleefstrasynndrome.org