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17th Deafblind International World Conference 2019
AUGUST 12-16, 2019 | GOLD COAST, AUSTRALIA

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Usher Pre-Conference

DBI USHER NETWORK WORKSHOP PROGRAM

Sunday 11th August 2019

Monday 12th August 2019

Verandah Room, Level 2

Surfers Paradise Marriott Resort

Facilitator: Emma Boswell

Event hashtags

#dbi2019australia

#deafblindworldconference

Welcome to the Deafblind International Usher Network Pre-Conference.

Welcome to the Gold Coast and a very warm welcome to the 16th Usher Network Pre-conference 2019.

It was previously called the 'Usher Study Group' and was last held in Aalborg, Denmark in 2010 and we're very proud to be hosting a newly named and formed Network for 3rd time, now 'Down Under' in Australia.

I hope this will be a great opportunity to meet people from many different countries and walks of life and find opportunities to spread our communications all across the globe. I also hope it will be an opportunity to share expertise and unique experiences in the field of Usher syndrome, our knowledge is our power and working together can truly make us stronger.

I must say a huge thank you in advance and anticipation to all the speakers, delegates and particularly the Usher Network Committee members for their ongoing fantastic commitment in helping to deliver this year's packed programme.

I hope you go home full of new ideas and most of all that you have enjoyed your experience with us.

Enjoy the pre-conference and also Australia!

Best wishes

Emma Boswell, Chair.

Usher Network Pre-Conference

PROGRAM - Sunday 11th August 2019

9:30am Registration & Communication Check

9:50am Welcome & Introduction Emma Boswell Usher Network Chair & Sense, UK

10:00am "Usher Syndrome: Genetics, Personalised Medicine and the Australian Usher Project" Dr Tina Lamey, Australian Inherited Retinal Disease Registry and DNA Bank, Sir Charles Gairdner Hospital, Western Australia

10:50am "Evaluating Biomarkers in Usher Syndrome: Identifying Outcome Measures for Trials" John Grigg, Save Sight Institute

11:30am Morning Tea Break

11:50am "Helping Children with Usher Syndrome Thrive" Emily Shepard, UsherKids Australia

12:40pm "Connecting Usher Kids in the UK" Chloe Joyner (via video teleconference), Usher Kids UK

12:50pm Lunch Break

1:50pm "Research and Patient Collaboration"
Dr Nicole Lo-A-Njoe-Kort, The Netherlands

2:40pm "The Importance of Interdisciplinary Studies in Health and Usher Syndrome" Dr Moa Wahlqvist, Örebro University, Sweden

3:30pm Afternoon Tea Break

3:50pm "The Five Pillars of Well-Being for Deafblind"
Christopher Woodfill, Helen Keller National Centre

4:40pm Questions About the Day? Emma Boswell, Sense UK

5:00pm End of Conference - Day 1

Usher Network Pre-Conference

PROGRAM - Monday 12th August 2019

9:30am Registration & Communication Check

9:40am Welcome & Introduction Karen Wickham, Senses Australia

9:50am "Advances for Stem Cell Research in Retinal Diseases" Dr Raymond Wong, Centre for Eye Research Australia

10:40am "Better Practice in the Real World" Alana Roy-Psychologist, Dr Annmaree Watharow, General Practitioner with Usher Syndrome

11:30am Morning Tea Break

11:50am Mental Health Panel facilitated by Alana Roy

1:10pm Lunch Break

2:10pm "Helping Bridge the Gap between Children and Adults with Usher Syndrome" Hollie Feller, UsherKids Australia & Genetic Cures Australia

3:00pm "Getting on With Life" Jennifer Weir, Allied Health professional, Perth, Western Australia

3:50pm Pre-conference wrap up Emma Boswell, Usher Network Chair & Sense, UK

4:00pm End of Pre-conference – Day 2 "afternoon tea will be served in the foyer"

Abstracts – Usher Network Preconference, DbI World Conference, Gold Coast 2019

SPEAKER: Dr Tina Lamey, Australian Inherited Retinal Disease Registry and DNA Bank, Sir Charles Gairdner Hospital- Centre for Ophthalmology and Visual Science, Faculty of Health and Medical Sciences, UWA

ABSTRACT**: Usher Syndrome: Genetics, Personalised Medicine and the Australian Usher Project**

Usher syndrome is a clinically and genetically diverse, inherited deafblind condition. Twelve Usher-causing/associated genes have been identified, and it is likely that others are yet to be discovered.

Usher genes are involved in the development, maintenance and/or function of specialised sensory cells found both in the inner ear and retina. Errors in these genes ultimately result in cell dysfunction, leading to the combined vision and hearing impairment that characterises this condition.

Significant progress has been made in the development of animal models and therapeutic strategies for Usher and other inherited retinal diseases (IRDs), and the number of human clinical trials is increasing.

Further, recent development of genetic screening methods has enabled rapid and simultaneous screening of large numbers of

genes. Though not without challenge, this has significantly expedited the identification of disease-causing gene-changes in IRD-affected individuals. Importantly, identification of the genetic basis for disease in an individual is a requirement for their participation in gene-based trials and resulting therapies and is central to 1) the further development of diagnostic and prognostic tools and treatment strategies, and 2) the establishment of clinical trial-ready cohorts of Usher-affected Australians.

To this end, the Australian Usher Project is underway. As one of several core projects undertaken by the Australian Inherited Retinal Disease Registry and DNA Bank (AIRDR), this work aims to ascertain the genetic basis of disease in approximately 150 Australian Usher families with expected completion in 2020.

This presentation will outline the genetics of Usher syndrome, genetic screening of IRDs and its challenges, emerging treatments for Usher syndrome, and an update on the Australian Usher Project.

SPEAKER: Professor John Griggs, Save Sight Institute

Professor John Grigg is passionate about helping children and adults with blinding eye conditions. He is involved in caring for patients, researching new ways to save sight and teaching the next generation of eye doctors.

John is Head of the Discipline of Ophthalmology at The University of Sydney's Save Sight Institute. He also consults at Sydney Eye Hospital and The Children's Hospital Westmead.
With sub-specialties in glaucoma, cataract, paediatric and genetic ophthalmology and clinical electrophysiology, John completed his training at Sydney Eye Hospital and undertook fellowships in Australia and the UK. His research interests are genetic eye disease, glaucoma management and electrophysiology of the visual system.

ABSTRACT: **Evaluating Biomarkers in Usher syndrome: Identifying outcome measures for trials**

John R. Grigg Alessandro Invernizzi Nina Mustafic, Federica Ristoldo, Clare Fraser Robyn V. Jamieson
Save Sight Institute, The University of Sydney, Sydney, New South Wales, Australia;
Department of Biomedical and Clinical Science 'L. Sacco', Luigi Sacco Hospital, University of Milan, Milan, Italy;
Children's Medical Research Institute, The University of Sydney, Sydney, New South Wales, Australia;

Purpose
To investigate the functional and structural biomarkers in Usher syndrome patients. By correlating visual acuity (VA), full field electroretinogram (ffERG) and pattern electroretinogram (PERG) with ultra-widefield autofluorescence (UW-FAF) and optical coherence tomography (OCT).

Methods - Usher syndrome patients attending the Save Sight Institute between 2012 and 2017 were reviewed. UW-FAF images were qualitatively graded to identify hypo/hyper fluorescence patterns in the peripheral fundus. OCT scans assessed Macular thickness (MT), linear extent of preserved outer retinal layers (macular island) and presence of cystoid macular edema (CME).

Results - Thirty-six eyes from 18 subjects were included. UW-FAF identified 4 patterns (granular 55%, annular 11%, bone spicule 17% and patchy 17%). Patients with patchy peripheral fundus appearance demonstrated worse VA, in comparison to those with granular ($p<.0001$) and bone spicule ($P=0.0179$) patterns, but less than those with annular.

Mean MT was 271 ± 35 µm. Mean foveal island was 2405 ± 1528 µm. Mean VA was 0.22 ± 0.3 LogMAR. Macular island extent correlated with VA ($r=-0.69$, $p<0.0001$), PERG P50 ($r=0.49$, $p= 0.004$) and PERG N95 ($r=-0.47$, $p=0.007$). MT showed a strong correlation with VA ($r=-0.78$, $p<0.0001$). CME was present in 41.7% of eyes. VA was not different in patients with and without CMO.

Conclusions - Structural changes identified on OCT and UW-FAF strongly correlated with the functional measures of VA and PERG. This study identified a set of structural and functional parameters that could provide strong measures for outcome

biomarkers in therapeutic clinical trials for patients diagnosed with Usher syndrome.

SPEAKER: Dr Nicole Lo-A-Njoe-Kort

ABSTRACT: **Research and Patient Collaboration**
Presentation about the collaboration between people who live with Usher syndrome and researchers, both medical as well as other researchers. It is a new way of working side by side, where people with Usher syndrome help raise money for research, but also get to discuss the direction of research. An important role, where we can help each other to get better results. There are connections between medical doctors, both eye and ENT, genetic researchers and patients, who work side by side to inform and support another. We were able to finance gene-therapy lab-research on Usher type 2a in The Netherlands this way true a foundation called SWODB of which I was one of the founders.

It is a process I started in 2011 when I was a medical student with my doctor who guided me trough the final thesis. At the moment there are other experienced Usher's who work side by side with doctors, orthopedagogy and revalidation centra in The Netherlands.

If we all see the possibilities then we can change the quality of life, as well as future perspectives. I was able to graduate and

find work as a doctor with deafblindness, I could take my guide dog with me to work. Sky is the limit and please let people with Usher syndrome try to follow their dreams, let's think in possibilities instead of restrictions.

SPEAKER: Moa Wahlqvist, Örebro University
Moa Wahlqvist, PhD Disability science, Researcher at the Audiological research centre, Örebro University hospital. Affiliated researcher to the Faculty of Medicine and Health, The Swedish Institute for Disability Research, Örebro University, Sweden. Coordinator at the Swedish National Resource centre for deafblindness, Lund, Sweden.
Co- author: Mattias Ehn, clinical psychologist, PhD student, Audiological research centre, Örebro University Hospital.
Affiliated to the Faculty of Medicine and Health, The Swedish Institute for Disability Research, Örebro University, Sweden.

ABSTRACT: **The Importance of Interdisciplinary Studies in Health and Usher Syndrome**
Current research and previous studies - The importance of interdisciplinary studies in Health and Usher syndrome

At the Audiological Research Centre Örebro University hospital, research about Usher syndrome and biopsychosocial health has been conducted for the last ten years.

In the presentation we want to share our results from some of the studies that have been conducted focusing on psychosocial health, working life and life strategies in adults with Usher syndrome. The presentation will address the need for an interdisciplinary approach within research of the field and of how it is to live with Usher syndrome.

In previous studies we have described the health problems that people with Usher syndrome have reported, such as headache, pain in shoulder and neck, major problems with fatigue as well as sleeping problems, refraining from going out alone and feelings of not being able to solve problems that occur. Further studies have addressed work as a factor for maintaining health for people with Usher syndrome type 1 and 2. A recently submitted article, describes the strategies used by people with Usher syndrome type 2 to handle their life situation. The study reveals strategies such as remaining active, telling others about their situation, finding ways to be present in the here and now. An overall theme of "being at the helm" emerged in the study which will be further elaborated at the presentation.

SPEAKER: Emily Shepard, UsherKids Australia

Emily Shepard is a co-founder and Director of UsherKids Australia which is a leading advocacy and support group for children in the Australian Usher syndrome community. Emily is passionate and committed to making the lives of children with Usher syndrome and their families better. She has brought her

perspective as a mother of a child with Usher syndrome, as well as skills from working in a commercial environment and combined them with her studies in Auslan and Health Promotion to successfully develop the not-for-profit organisation.

ABSTRACT: **Helping Children with Usher Syndrome Thrive**
After her son was diagnosed with Usher syndrome Type 1 at age 3, Emily Shepard and her family found themselves extremely isolated and lonely. There was no support group to turn to and many of their doctors knew little of the condition. The support services all concentrated their efforts on adults with the condition. After meeting another family in a similar situation, Emily, along with Hollie Feller, set out to improve the path for families that followed them by establishing an organisation dedicated to families of children diagnosed with Usher syndrome in Australia.

Historically the diagnosis for Usher syndrome came in the second decade after the onset of Retinitis Pigmentosa, but with genetic testing advancements, it allows conditions such as Usher syndrome to be diagnosed at a much earlier age, some in their first years of life. The earlier diagnosis of children with Usher syndrome in Australia created a gap in the knowledge and support services available. UsherKids Australia was established with the main purpose of bridging this gap by:
-providing information and support to families

-educating clinicians and support services of the various needs of children with Usher syndrome
-advocating for improve clinical management of children with Usher syndrome, including funded genetic testing
-raising awareness of the condition through various social media campaigns
-assisting to disseminate research findings
-collaborating with various organisations in Australia and globally

Since it was established in 2016, UsherKids Australia has had a significant impact in uniting the Australian Usher syndrome community and ensuring that newly diagnosed families never feel the isolation and helplessness felt by Emily and Hollie when their children were diagnosed. Prior to their awareness-raising efforts, many service providers were not experienced with this age group, many clinicians were unfamiliar with the condition and parents had no way of connecting with other parents for support.

SPEAKER: Chloe Joyner, Usher Kids UK (via video conference)
Chloe is the mother of two children, the youngest of which was diagnosed profoundly deaf when she was 2 weeks old, and with Usher Syndrome (1B) via ERG test and genetic testing at 18 months old. Chloe's learning curve since each diagnosis has been steep, and she is now determined to make a difference for children and families living with the unique challenges

posed by Usher syndrome. In addition to her work with Usher Kids UK, Chloe is a Registered Associate Nutritionist and teaches on nutrition programmes at Sheffield Hallam University.

ABSTRACT: **Connecting Usher Families in the UK**
Chloe Joyner from Usher Kids UK will provide via video teleconference an update on the work she has done to establish a support group for families of children with Usher syndrome in the UK. Chloe is also a parent of a young child with Usher syndrome Type 1 and met Hollie Feller and Emily Shepard from UsherKids Australia at an International Usher Symposium in Boston in 2014. After realising that many of the hurdles they faced after diagnosis were similar in their respective countries, they decided to join forces and share resources and ideas on their sister organisations.

Chloe will also provide a summary of the "Connect Usher" event taking place in June 2019 in Birmingham, UK in conjunction with RP Fighting Blindness. For the first time, the Usher Kids UK community will come together and experience a one-stop event designed to address the specific needs of families and children living with Usher syndrome in the UK.

SPEAKER: Christopher Woodfill, Helen Keller National Center
Chris is DeafBlind with Usher Syndrome type 1. He has worked at Helen Keller National Center for seven years and has been

its Associate Executive Director for five. He is currently serving a second term as the North American region representative for the World Federation of the DeafBlind. He served on the board of American Association of the DeafBlind for almost a decade until last year. He is one of the administrators of the DeafBlind Thoughts, a Facebook group open to only the DeafBlind. Before working at Helen Keller National Center, he worked for thirteen years as a high school teacher at Wisconsin School for the Deaf. He also worked for three years as English as Second Language instructor at Gallaudet University. He holds a Master's degree in Latin American Studies from the George Washington University and a Master degree in Bilingual-Bicultural Deaf Education from McDaniel College.

ABSTRACT: **The Five Pillars of Well Being for Deafblind**
Whole person view of the DeafBlind individual is essential for the maximum well-being of the individual. Using myself as a model of leading a well-rounded life full of success, dignity and meaning.

Five pillars of the well-being for the DeafBlind individual are: Education, Employment, Lifestyle, Recreation and Healthcare. All of these five pillars work in unison and in a transdisciplinary way to ensure that the DeafBlind individual is a well-rounded person with dignity and meaning. The presenter will expand on each pillar by referring back to himself as an example. He will also discuss each pillar as it applies to the DeafBlind individuals

in general. The presentation will be uplifting and hopeful in nature with tips, resources and tools for DeafBlind individuals, families and professionals on how to accomplish such a life for the DeafBlind individual.

SPEAKER: Dr Raymond Wong, Centre for Eye Research Australia

Dr Raymond CB Wong, Ph.D., is a Principal Investigator at the Centre for Eye Research Australia (CERA), the University of Melbourne in Australia and a Guest Professor at Shenzhen Eye Hospital in China. He is a stem cell biologist with more than 16 years of research experience, specialising in cellular reprogramming, pluripotent stem cells and neural/retinal differentiation. Dr Wong completed his PhD in stem cell biology at Monash University and was awarded a California Institute of Regenerative Medicine fellowship to pursue overseas postdoctoral training in University of California Irvine (USA), and a Visiting Fellow Award to train in National Institutes of Health (USA). In 2013, Dr Wong joined CERA with the support of a Cranborne Foundation Fellowship and subsequently established the Cellular Reprogramming Unit with the support of a MAWA Fellowship.

ABSTRACT: **Advances for Stem Cell Research in Retinal Diseases**

Dr Wong's current research focuses on understanding the genetic signals that define retinal cells and using cell

reprogramming and stem cell technologies to study and treat retinal diseases. Dr Wong's research has identified new methodologies to culture and generate human pluripotent stem cells, providing important steps to realise the medical potentials of human pluripotent stem cells.

SPEAKER: Alana Roy and Annmaree Watharow

Alana Roy is a Psychologist, Mental Health Social Worker, Counsellor and Advocate who has qualifications in Auslan and is committed to ongoing Auslan training. She has worked in the field of mental health, suicide prevention, trauma and abuse, and disability for the last 12 years. Alana is currently completing her PhD which focuses on Deafblind research methodology and inclusive practices. She works for several universities conducting research and teaching Social Work students. Alana is passionate about spirituality, mindfulness and meditation and how to make these practices accessible and enjoyable for a diverse range of people. Furthermore, she is currently completing a course in Philosophy and has a keen interest in world religion and cultures. Alana has a private practice called The Signs of Life where she works with children, youth, adults, Deaf and Deafblind people. In her spare time, she enjoys travelling with her husband and two young sons and spending time with her French bull dog and British blue cat; she is a lover of nature and animals.

Dr Annmaree Watharow is a retired general practitioner who is now a student, academic, consumer research (SACR). She has Usher syndrome (profound hearing loss combined with one degree of vision). Annmaree is presently at stage three of her doctoral studies. Her topic is hospital communication experiences of people with deafblindness, dual sensory impairments. This interest arose out of both personal experience and the accounts of patients with sensory losses who experience adverse outcomes as a result of communication failures when hospitalised. Annmaree's other research interests are using creative non-fiction to tell the stories of people with deafblindness and exploring how the narrative space is impacted by sensory impairment.

ABSTRACT: **Better Practice in the Real World**
There are substantial gaps in the literature of validated inclusive research practices for Deafblind research participants and researchers. There is also a dearth of material on how a person with dual sensory impairment can do three things: safely attend university and access information, safely research AND provide a safe space for the participants to share their stories.

Dr Wayland, an academic researcher with a special interest in disability, will interview Drs Roy and Watharow on what better research practices might look like. Asking ‘do people with

Deafblindness want to be researchers and research participants; if so how do they want to participate?'

Dr Roy will be questioned on her recent findings across four studies which investigated good practice approaches to consultation, research, policy and service development. Dr Roy will refer to themes such as; 'acknowledging the deafblind worldview', 'the importance of specialist knowledge, skills and cultural sensitivities', and a focus on 'creating a safe space for research, interaction, and community conversations'.

Dr Watharow will be asked about the special considerations and realities of being a person with deafblindness, a PhD student researcher, researching with participants who are Deafblind.
The session will provide those in attendance with new ways of understanding the lived experience of researchers with sensory impairment in order to provide understanding how to conduct research as a researcher and conduct research with this population group.

SPEAKER: Hollie Feller, UsherKids Australia
Hollie Feller is the mother of Harry Feller and the co- founder of UsherKids Australia; an Australian based support network for families with kids diagnosed with Usher syndrome. She advocates for early diagnosis through genetic testing, education of clinical professionals and support for families

around the country to share research and information about the current generation of USH kids.

ABSTRACT: **Helping Bridge the Gap between Children and Adults with Usher Syndrome**

Usherkids Australia was set up as an organisation to fill a gap that was evident surrounding the management and support of families of children diagnosed with Usher syndrome in Australia who were under 21 years old. The group is the first of its kind in Australia for the under 21 years Usher community.

Since starting their community led organisation in 2016 UsherKids Australia have spent the last 3 years uniting both the families of those diagnosed with Usher syndrome under 21 years old and their Health Professionals and carers alike. They seek to serve this community through greater accessibility of information on the condition and its management including regular events and seminars to bring these two groups together. This results in the best possible outcomes for those children and their families through greater awareness within the medical and support communities. In addition to this it also strives to maintain a community that reaches out in support for the families throughout the diagnosis period and beyond.

Even though the childhood path of the children diagnosed in the last two decades is very different from those before them with the onset of new technologies in communication and

medicine, there still remains the ongoing issues of psychosocial behaviours created by the challenges in sight and hearing and overall communication for those living with Usher syndrome. The social impact of "being different" to your peers is just one example of the psychological challenges these children face that spans all generations and is not necessarily addressed by their increased accessibility through advances in technology.

We discuss the common threads that exist between each of the generations and how young adults with Usher syndrome can be the role models and mentors for younger children within the community providing empathy, experience and foresight to those coming to terms with their condition.

Working as a team and community to support these children through their childhood and teenage years means that real life experiences provide a platform of information for these families to address the reality of their child's diagnosis at its current stage. It is underpinned with the knowledge that there is a network of other families out there to address their concerns with as they occur.

This Conference allows us to bring together the adult community with our newer UsherKids community and share the insights and hindsight of an adult’s experience of Usher syndrome. This combination will allow for everyone to work together and take responsibility for owning the possibilities and

future psychosocial outcomes for these kids within today's world of technology and science.

SPEAKER: Jennifer Weir, Retired Allied Health Care Professional, Usher Syndrome Type 3

ABSTRACT: **Role Model as a professional, experiences working with Usher Syndrome Type 3**

When diagnosed with Retinitis Pigmentosa aged 23 the Consultant told me, "Go and buy a good pair of sunglasses and get on with your life".

That's is exactly what I did I continued studying as a nurse, married and had two beautiful daughters.

When diagnosed as going deaf aged 32 the consultant told me "Go home and be a good mother". I did as I was told BUT I bought hearing aids continued to work in the career that I loved and was a good mother.

I have had a fortunate and privileged life and after 43 years of nursing including Coronary, Intensive Care and management I never let being Deafblind define me.

Usher Network Meeting

Date: Wednesday 14 August 2019

Time: 5.30pm - 6.30pm

Facilitator: Emma Boswell