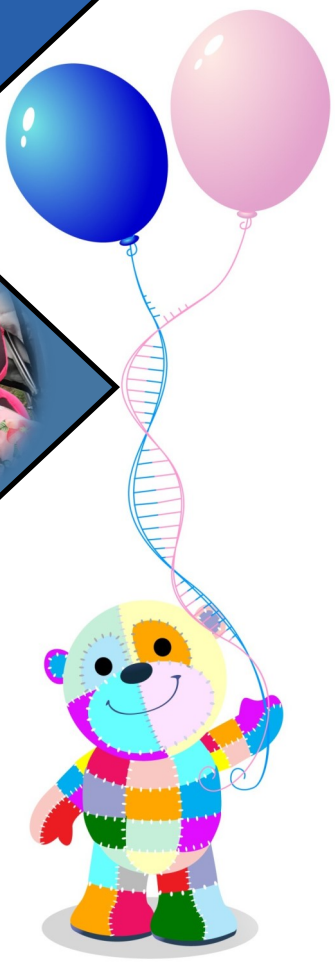


1st & 2nd November

De Vere Wotton House Dorking

4th AADC Conference 2018

12 YEARS RESEARCH

2006 - 2018

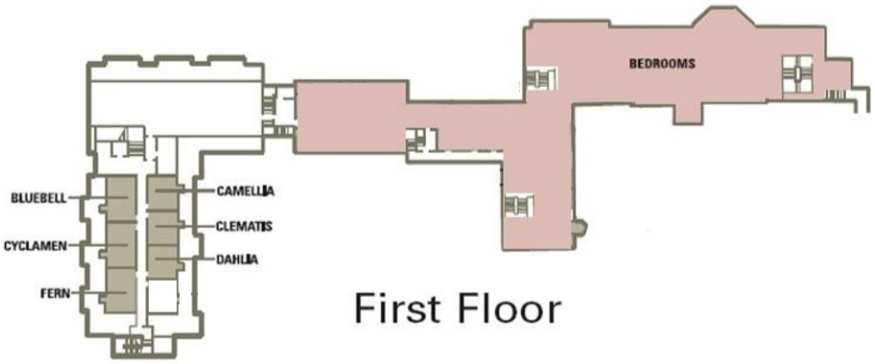
the aadc research trust

making a rare child healthy

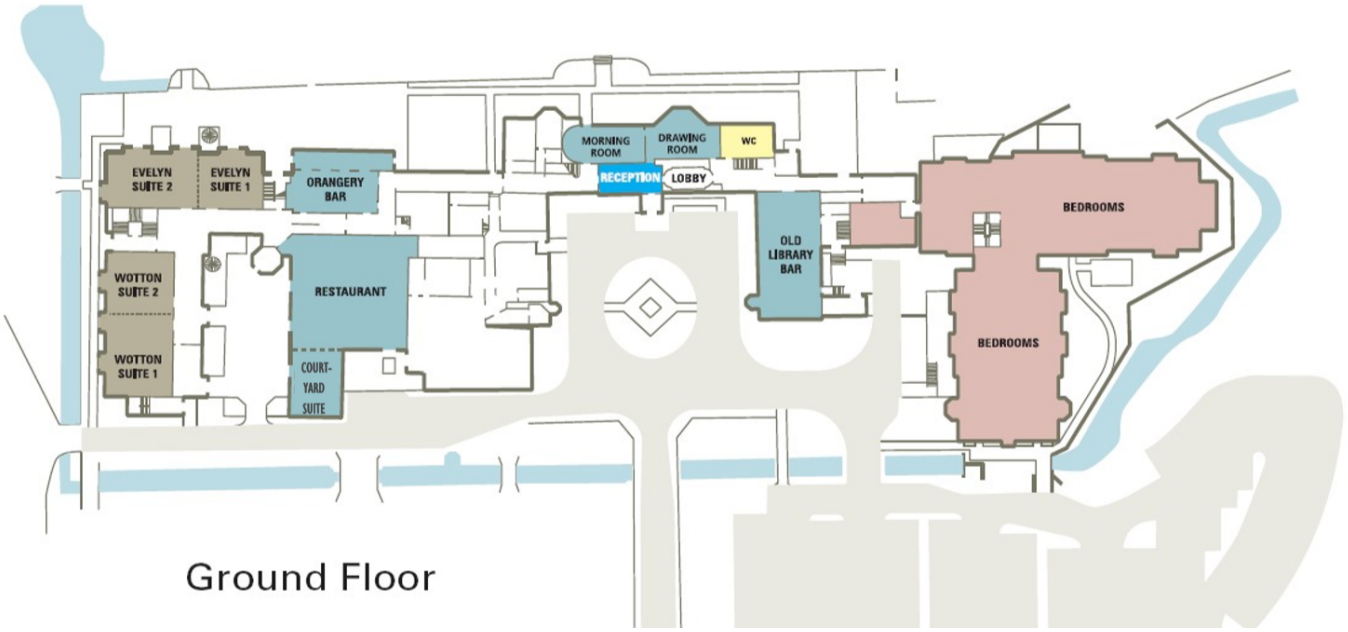
Map

KEY

- Reception
- Bedrooms
- Meeting Rooms
- Bar/Lounge/Restaurant
- Leisure Facilities
- Toilets

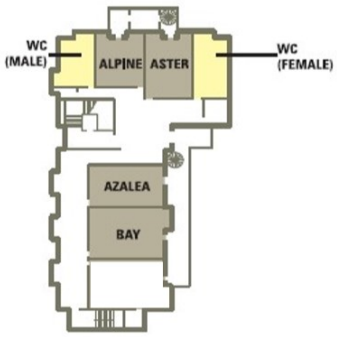


First Floor

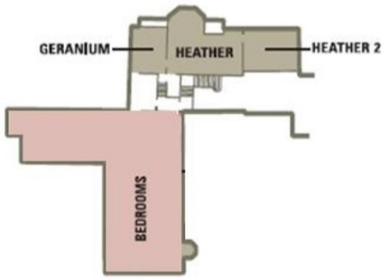


Ground Floor

Lower Ground East Wing



Mezzanine Floor



Lower Ground West Wing

We would like to dedicate this Conference to every child who has lost their battle with AADC deficiency … before we had a chance to SAVE them!

Table of Contents

Dear All,

Thank you for joining us for our AADC 4th World 2-Day Conference. It is with absolute pleasure that I welcome each and every one of you to this incredible event.

The Conference will bring together our AADC families, renowned Medical & Scientific experts in the field from around the world to discuss the latest research updates and future proposals.

An important aim of the Conference is to offer networking opportunities and foster debate among over 1oo professionals and families from many different countries & cultures, united by the same disease AADCd. So please make the most of the Conference and enjoy this very special occasion.

Lisa Flint

Founder & Managing Director

The AADC Research Trust Children's Charity 2006—2018

Below are some useful numbers in case you need assistance or in case of an emergency:

The AADC Research Trust—Event Organisers

Lisa Flint / Managing Director
M: 07778 356533

Julie Ramsay / Charity Manager
M: 07703 110945

Diane Augustin / Event Coordinator
M: 07709 944800

HOTEL / De Vere Wotton House, Guildford Road, Dorking, RH5 6QQ
T: 01306 730000
W: www.phcompany.com/de-vere/wotton-house

Taxi's / Magnum Cars
T: 01483 281111

Nearest Hospital / Epsom Hospital, Dorking Road, Epsom, Surrey, KT18 7EG
T: 01372 735735

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#CureAADC
#CureAADC

Thursday 1st November 2018

De Vere Wotton House, Dorking, Surrey / Wotton Suite (9:00am)

06:45-08:45	Breakfast served in the 1877 Restaurant - For all Delegates (At your Lesiure)
07:30-08:45	Breakfast for AADCd Families ONLY served in 'The Courtyard' within the 1877 Restaurant ***(From 7:30am families have an opportunity to spend some time together to discuss the day ahead)***

An Introduction to AADC Deficiency
Conference Chair: Prof Simon Heales

09.00-09.10	Lisa Flint – Welcome
09.10-09:25	Prof Simon Heales / The Biochemistry of AADCd Deficiency (UK)
09:25-09:40	Dr Manju Kurian / A Clinical Introduction to AADCd

AADCd - Session 1
Chair: Prof Simon Heales

09:40-10:00	Prof Mita Bertoldi / An Overall Picture of the Structure-Function Relationship of AADC Variants Causing AADC Deficiency
10:00-10:20	Karolin Kramer / Generation of Patient-Derived Dopaminergic Cell Model of Aromatic L-Amino Acid Decarboxylase Deficiency (AADCd)
10:20-10:40	Prof Marcelo Coba / Modelling Human Disease with iPSC Derived Neurons
10:40-10:50	Q & A – Audience and Online

10:50-11.10 **Refreshment Break**

AADCd - Session 2
Chair: Dr Keith Hyland

11:10-11:30	Dr Mario Mastrangelo / Successful Pregnancy in A Patient With L-Amino Acid Decarboxylase Deficiency: Therapeutic Management & Clinical Outcome
11:30-11:50	Dr George Allen / CSF Findings in a Carrier of AADC deficiency
11:50-12:10	Dr Wang-Tso Lee / Probiotics in DDC Mouse Model
12:10-12:30	Dr Paldeep Atwal / Metabolomic Profiling in AADCd
12:30-12:40	Q & A - Audience and Online

12:40–14:00 **Working Lunch (to be served in the Conference Room)**

12:50-13:30 **AADC Meeting** - Invitation ONLY (The Boardroom)

AADCd - Session 3
Chair: Dr Roser Pons

14:00-14:45	Prof Krystof Bankiewicz / Update on AAV2-hAADC Gene Therapy Trials in Paediatric Patients (Target Area VTA & SNpc)
14:45-15:05	Dr Toni Pearson / The Natural History of AADC deficiency: A Retrospective Study
15:05-15:25	Prof Stanley Satz / Non-Invasive PET Visualisation of AADCd Manifestations, Therapy Planning, Outcomes Monitoring and Theranostics/Artificial Intelligence for Underserved and Rare Paediatric Conditions
15:25-15:45	Kaili Ngadiman – Sellina's GT Journey / Family Experience
15:45-15:55	Q & A – Audience and Online
16:00-17:00	Professionals around the table with the AADCd families

17:00 **Conf Summary & CLOSE – Prof. Simon Heales**

18.15 – 19:00 Drinks Reception in The OLD LIBRARY Bar *(followed by)*
19:00 Onwards Formal Dinner in the Evelyn Suite (Smart Dress)

FOR INFO **Contact: Lisa Flint – 07778 356533**

Geographical Epidemiology…

- Approx. 130-150 Children/Young Adults are known to suffer Aromatic Amino Acid Decarboxylase deficiency (AADCd) Worldwide
- Spanning 30+ countries
- At least 3 children die every year rom AADCd
- IN 2018 alone 6 children have so far lost their battle with AADCd

Friday 2nd November 2018

De Vere Wotton House, Dorking, Surrey / Wotton Suite (9:30am)

06:45-09:15 Breakfast served in the 1877 Restaurant - For all Delegates (At your Lesiure)
Breakfast for AADCd Families ONLY is served in 'The Courtyard' within the 1877 Restaurant
08:15-09:15 ***(From 7:30am families have an opportunity to spend some time together to discuss the day ahead)***

Welcome Back
09.30-09.40 Prof. Simon Heales / Welcome Back

AADCd - Session 1
Chair: Dr Manju Kurian

09:40-10:10 Prof Shin-ichi Muramatsu / AADCd Gene Therapy in Japan
10.10-10:40 Dr Karin Kojima / GT Case Studies
10:40-11.00 Todd Berner / PTC Acquisition & AADCd GT update
11.00-11:20 Tom Goss / Boston Healthcare / AADCd BOI Study
11:20-11:40 Q & A – Audience and Online

11:40–12:00 Refreshment Break

AADCd – Session 2
Chair: Dr Marcel Verbeek

12:00-:12:20 Dr Riccardo Montioli / Biochemical Advances in AADC deficiency: an approach to understand the molecular pathogenesis of heterozygous patients and protein engineering for the enzyme replacement therapy
12:20-12:40 Dr Tessa Peters / Metabolomic Screening (PhD – Marcel Verbeek)
12:40-13:00 Dr Roser Pons / Case Presentation: AADC deficiency PLUS
13:00-13:10 Q & A – Audience and Online

13:15-14:30 Lunch is served in the 1877 Restuarant

AADCd – Session 3
Chair: Dr Toni Pearson

14:30-14:50 Dr Keith Hyland / 3OMD - A Reliable Biomarker for AADCd
14:50-15:10 Dr Roser Pons / Dystonia Scale in Paediatric Neurotransmitter Disorder: A Pilot Study
15:10-15:30 Dr George Allen / Urinary Sulphatoxymelatonin
15:30-15:50 Dr Charles Laurenco / Against All Odds - Diagnosis & Presentation of AADCd Brazilian Patients
15:50-16:00 Q & A – Audience and Online

16:00-16:45 Professionals around the table with the AADCd families

16:45-16:55 Conf Summary & CLOSE – Prof. Simon Heales / Lisa Flint

17:00-18:00 AADC Meeting - Invitation ONLY (The Drawing Room)

19:00 Onwards Buffet Dinner in The OLD LIBRARY Bar **(Smart/Casual)**

Contact: Lisa Flint – 07778 356533

Current Research Projects...

AADCd Induced Pluripotent Stem Cells (iPSC) (UK)

create a cell model to study disease variations

explore the possible benefits of individualised treatment strategies

Study the longevity of disease, current medications and their effects

explore the effects of new medications as well translational scientific discoveries in an attempt to find a cure for AADCd

AADCd Genetic Variations (Italy)

Looking at how genetic variations play a role in symptomology (mild vs moderate vs severe)

Scaling AADCd Disease Severity (Greece)

An equivalent scale to that used in Parkinson's Disease diagnosis and treatment to measure severity and changes disease

Some Past AADCd Research Projects...

PhD Project on the basic understanding of AADCd disease mechanisms (UK)

Importance of the co-factor Vitamin B6 and AADCd (UK)

Dopamine in the Kidneys (Netherlands)

JAKEdb – 1st Global AADCd Clinical Database, to help identify disease trends and common features (Switzerland)

OUTREACH - Creation of a Diagnostic Facility in KL Malaysia to aid diagnosis of AADCd & other neurotransmitter diseases.

The Creation of an AADCd Mouse Model (Taiwan)

iNTD - International Neurotransmitter Database created with 1st published AADCd Consensus Guidelines for the Diagnosis and Treatment of AADCd

Standardised the AADCd Enzyme Assay Test in the UK (UK)

Please #CureAADC

Speaker's

Presentations & Biographies

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Speaker's Presentations & Biographies

Lisa Flint

Presentation Title:

A Welcoming Introduction to our 4th International AADCd Conference

Lisa Flint Biography:

After hundreds appointments, Lisa's youngest son was diagnosed with Aromatic Amino Acid Decarboxylase deficiency (AADCd), a rare Parkinsonism brain disease. The AADC Research Trust, which she founded in 2006, has an expert Medical and Scientific Advisory Board and serves as a lifeline for families and children with rare brain diseases. The Trust has driven the development of an international database. It is working with a team of genetic scientists to understand the various mutations and their impact on disease severity. Lisa was an author on the Consensus Guidelines for Diagnosis and Treatment of AADC, and her expertise is sought from patients and medical institutions worldwide.

Professor Simon Heales

(Conference Chairman)

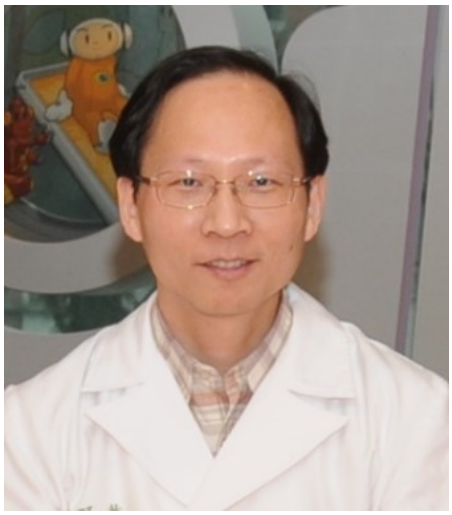
Presentation Title:

The Biochemistry of AADC Deficiency

Professor Simon Heales Biography:

Simon obtained his PhD from Aston University in 1987 and was made a Fellow of the Royal College of Pathologists in 2003. He is the Chief of Service for Paediatric Laboratory Medicine at Great Ormond Children's Hospital in London. As well as being Head of Service, he holds the UCL Chair of Clinical Chemistry and has a strong interest in the diagnosis and monitoring of patients with inherited metabolic disorders. This work is underpinned by a number of basic and translational research projects that are carried out in conjunction with the UCL Great Ormond Street Institute of Child Health. He has published over 150 papers in the area of mitochondrial, neurotransmitter and lysosomal disorders. Simon is also the Director of the Neurometabolic Unit at the National Hospital, Queen Square (UCLH Foundation Trust).

Speaker's Presentations & Biographies



Dr Wang-Tso Lee

Presentation Title:

Probiotics in AADCd

Previous studies had shown the change of gut microbiota to be possible pathogenic mechanisms for some neurological diseases. Therefore, treatment with probiotics may be beneficial in many neurological diseases. Lactobacillus plantarum strain PS128 (PS128) was found to change the dopamine and serotonin turnover in previous study. Therefore, in the present study we investigate whether it was also helpful in an animal model of dopamine deficiency. We found that treatment with PS128 could have some help in the animal model of dopamine deficiency, and may improve the neuronal differentiation. We further conduct a double-blind randomized control trial in another disease, which may have severe dystonia when growing up. Preliminary results showed that treatment with PS128 may have some help in improving dystonia in patients. Therefore, it is potentially applicable in human neurological diseases.

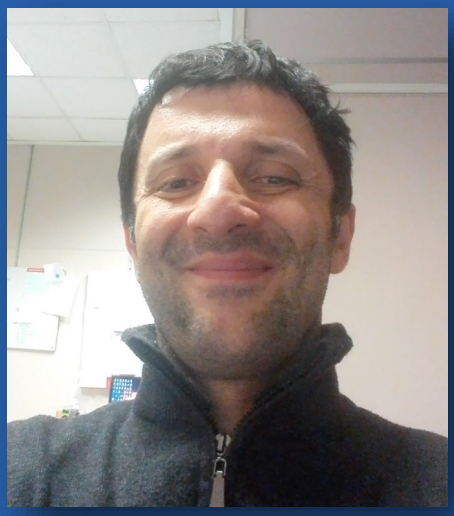
Dr Wan-Tso Lee Biography:

Dr. Wang-Tso Lee is now professor and chief of Department of Pediatric Neurology, National Taiwan University Children's Hospital and professor of Department of Pediatrics and Graduate Institute of Brain and Mind Sciences, College of Medicine, National Taiwan University, Taipei, Taiwan. He finished University medical education in College of Medicine, National Taiwan University, Taiwan in 1988, and got the PhD degree from Graduate Institute of Clinical Medicine, College of Medicine, National Taiwan University in 2000. Dr. Lee did neurology research in Department of Neurology, Children's Hospital of Philadelphia, USA in 1999. He also went to several institutes in USA and Canada as visiting scholar in the past. He is interested in movement disorders, neurometabolic and neurotransmitter diseases. His Lab is focused on movement disorders and neurotransmitter research.

Dr Mario Mastrangelo

Presentation Title:

Successful Pregnancy in a patient with L Amino Acid Decarboxylase deficiency : Therapeutic Management and Clinical Outcome.



The talk will be focused on the case report of a 26 year's old patient with AADC deficiency who delivered a healthy new-born (carrier) without complications (this is the first reported cases of pregnancy in AADC deficiency. Therapeutic implications and clinical management of the pregnancy will be discussed.

Dr Mario Mastrangelo Biography:

Pediatric neurologist of the Pediatric Neurology Division in The Department of Human Neurosciences at "Sapienza University of Rome". He is among the staff of the Regional Referring Centre for Rare Neurological Disease and with special area of interested being represented by neurotransmitter disorders and genetic epileptic encephalopathies. He is actually involved in the clinical management of 30 patients with inherited disorders of biogenic amine metabolism (5 patients with AADC deficiency) and is a member of iNTD network.

Speaker's Presentations & Biographies

Prof. Manju Kurian

Presentation Title:

Clinical Introduction to AADC Deficiency

Professor Manju Kurian Biography:

Dr Manju Kurian is a UCL Professor of Neurogenetics and NIHR Research Professor at UCL-Great Ormond Street Institute of Child Health. She is also a Consultant Paediatric Neurologist at Great Ormond Street Hospital. After graduating from the University of Cambridge (1998), she trained in Paediatrics before subspecialising in Paediatric Neurology. At the end of her clinical training, she undertook a PhD with Professor Eamonn Maher (University of Birmingham) investigating the molecular genetic basis of childhood neurological disorders (2007-2011). During this time, she identified mutations in the gene encoding the dopamine transporter (*SLC6A3*) as the cause of infantile parkinsonism-dystonia, as well as other genes causing movement disorders and epilepsy in children.She moved to UCL after her PhD, and has since established herself as an independent Principal Investigator at the Institute of Child Health. She was awarded a Wellcome Intermediate Fellowship in 2013, NIHR Professorship in 2017 and The Jules Thorn Award for Biomedical Research in 2017. Her working pattern is an 80:20 academic/clinical divide. Clinically, she looks after children with neurogenetic conditions, including a group of patients with AADC deficiency and other neurotransmitter disorders. The remainder of her time is spent in academic work, managing her research group, which comprise a mixture of post doctoral fellows, clinicians, PhD students and research assistants.

Her current research encompasses gene discovery for childhood neurological disorders. Her lab uses a number of different models to investigate the underlying pathological basis of disease. Her lab has developed expertise in induced pluripotent stem cell models for studying neurological diseases of childhood, including AADC deficiency. She works closely with UCL Gene Therapy groups to develop novel therapeutic strategies for children with pharmacoresistant movement disorders. Her long term goal is to translate her research for patient benefit, through improved clinical diagnosis and better therapies.

Karolin Kramer

Presentation Title:

Generation of Patient Derived Dopaminergic Cell Model of Aromatic Amino Acid Decarboxylase Deficiency (AADCd)

Background: Aromatic L-Amino Acid Decarboxylase (AADC) deficiency is a severe pharmacoresistant neurological disorder due to inherited autosomal recessive loss-of-function mutations in the *DDC* gene. The resultant impairment of AADC enzyme activity severely impacts on monoamine synthesis, leading to reduced levels of dopamine and serotonin. Affected patients present with marked neurodevelopmental delay, hypotonia, oculogyric crises and autonomic dysfunction. Currently, there are few truly disease-modifying therapies.

Aims: To generate AADC patient-derived induced pluripotent stem cells (iPSC) for subsequent differentiation into midbrain dopaminergic neurons, and to utilise this model to better define disease mechanisms and test novel therapeutic strategies.

Methods: Patient and age-matched control fibroblasts were reprogrammed into iPSC using Sendai virus methods. A modified dual SMAD inhibition protocol was then utilised for differentiation of all iPSC lines to day 70 of maturation. The generated neuronal model was then analysed for mature mDA neuronal identity and AADC disease-specific features.

Results: We have generated iPSC lines from skin fibroblasts derived from two patients with AADC deficiency and one age-matched control subject. One patient harboured a homozygous missense mutation (p.R347G) and the other was a compound heterozygote for a nonsense variant (p.Arg7*) and missense mutation (p.C100S) in *DDC*. Generated iPSC lines were confirmed as being truly pluripotent, then successfully differentiated into midbrain dopaminergic (mDA) neurons, with characteristic neuronal morphology, expressing tyrosine hydroxylase (TH) and microtubule-associated protein 2 (MAP2). There was no evidence of neurodegeneration in the cell model. However, we detected a number of disease-specific features, including significantly marked reduction of AADC enzyme activity and dysregulation of the dopaminergic system in patient mDA neurons when compared to the age-matched control. Lentiviral rescue of the patient-derived mDA cell model is underway.

Conclusion: Our iPSC-derived mDA neuronal model represents an ideal platform to further elucidate disease mechanisms, as well as to screen novel pharmacological agents to treat AADC deficiency.

Karolin Kramer Biography:

Karolin studied Molecular Life Science in her Bachelor's and Master's at the Friedrich-Alexander-University in Erlangen, Germany. She joined the lab of Dr Manju Kurian in 2015 for her PhD. In her project she generates a dopaminergic cell model for Aromatic L-Amino Acid Decarboxylase (AADC) Deficiency working with induced pluripotent stem cells (iPSCs).

Speaker's Presentations & Biographies

Dr George Allen

Presentation Title:

CSF findings in a carrier of AADC deficiency.

A case report of a sibling of an AADC deficient patient will be presented, including clinical, biochemical and genetic findings. Importantly, in this case CSF analysis demonstrated some abnormalities and in particular a low 5-HIAA. The potential consequences of these findings will be discussed and the results placed in context with other reports from carriers of AADC mutations.

Second Presentation Title:

Sulphatoxymelatonin, a new urinary marker of serotonin status in AADC deficiency

Melatonin is a hormone produced from serotonin in the pineal gland. In the liver, melatonin is broken down to 6-sulphatoxymelatonin (aMT6s). This waste product is removed from the body in urine. Measurement of aMT6s in urine may allow detection of serotonin deficiency. This talk will summarise the latest developments in the use of aMT6s and its potential role in diagnosis and treatment of AADC deficiency.

Dr George Allen Biography:

George undertook a PhD studentship funded by the AADC Research Trust from 2007 to 2010. Studying at the UCL Institute of Neurology, this work included establishing the plasma AADC enzyme assay as a diagnostic test in the UK. Following this, he performed postdoctoral research at the MRC Protein Phosphorylation Unit working on mitochondrial quality control and Parkinson's disease. He then trained as a clinical biochemist at the Royal Devon and Exeter NHS Foundation Trust, where he currently works.

Dr Keith Hyland

Presentation Title:

30MD : A Reliable Biomarker for AADCs

Presentation will describe the Utility of 3-O-methyldopa analysis for the diagnosis of aromatic L-amino acid decarboxylase deficiency. Included will be discussion of methodology, sensitivity and specificity of the methodology.

Dr Keith Hyland Biography:

Dr. Hyland is an internationally recognized researcher and educator in clinical chemistry, child neurology, and inherited metabolic disorders. In a career spanning 35 years, he has led numerous grant funded research teams in the UK and the US in the study of inherited disease and the neuroprotective molecular medicines used to treat them. He is a leading expert in the area of inherited disorders affecting serotonin, catecholamine, folate, and pyridoxal phosphate metabolism. With Professor Peter Clayton he described the first cases of aromatic L-amino acid decarboxylase deficiency. In 1989 Dr. Hyland moved from the Institute of Child Health in London to the Baylor Research Institute in Dallas, Texas. During his time in Texas, he held the positions of Senior Research Scientist at the Baylor Research Institute, Adjunct professor of Neurology at the University of Texas, Southwestern Medical Center and Professor of Biomedical Sciences at Baylor University. In 2004, he moved to Atlanta to join Horizon Molecular Medicine, which eventually became Medical Neurogenetics LLC in 2008. Dr. Hyland has over 130 peer reviewed publications and regularly presents to the medical community.. He has supervised numerous PhD candidates and post-doctoral fellows in their research and maintains professional membership in societies relevant to his work. Dr. Hyland has been honored several times for his work in the area of Neurometabolic Disease, including the Noel Raine Award from the Society for the Study of Inborn Errors of Metabolism, in both 1989 and 1998.

Speaker's Presentations & Biographies

Dr Roser Pons

Presentation Title:

Parkinsonism– Dysonia Scale in Paediatric Neurotransmitter Disorder : A Pilot Study.

In this presentation I will show the first results of a pilot study on thedevelopment of a scale for infants and young children with parkinsonism-dystonia. This scale has been designed to be used inpatients with primary neurotransmitter disorders associated with congenital dopamine deficiency, and also other acquired conditions an neurogenetic disorders leading to parkinsonism early in life. The ultimate goal of this work is to establish the baseline status of patients with parkinsonism, to monitor their response to treatment and to qualitatively evaluate disease progression over time.

Second Presentation Title:

Case presentation: AADC deficiency plus

In this presentation we will describe the case of a child with severe AADC deficiency and a form of mucopolysaccharidosis. The occurrence of two rare
neurometabolic disorders is exceptional. In this case the occurrence of 2 neurometabolic disorders probably explains the severity of her phenotype

Dr Roser Pons Biography:

Dr Roser Pons is a graduate of the Pediatric Neurology training program at Columbia Presbyterian Medical Center, New York. Prior to that, she had s everal years of training in the genetics and biochemistry of mitochondrial and fatty acid oxidation disorders that manifest with neuromuscular and cerebral syndromes. Subsequent to her residency, she underwent further training in the subspecialty of Movement disorders at Columbia University. She is mainly interested in the diagnosis and management of rare neurological conditions of childhood that manifest themselves through abnormalities of motor function and motor control. Currently she is working in the First University Clinic at Children's Hospital Agia Sofia in Athens where she is the responsible for the Special Unit of Pediatric Neurology. She runs several outpatient clinics including "Movement disorders and Rare Neurological Disorders", "Neurocutaneous disorders" and "Dystonia Clinic"

Prof Marcelo Coba

Presentation Title:

Modeling human disease with iPSC derived neurons.

Advances in human induced pluripotent stem cells (iPSCs) provides an exceptional platform to model human disease. Differentiated cells derived from iPSCs in monolayers have proven to be an extraordinary tool for exploring the underlying mechanisms of disease pathogenesis. I will discuss progress and limitations of iPSC disease-modeling platforms, as well as recent advancements in the development of different CRISPR-Cas9 applications to model human disease. I will discuss the use of enhanced expression systems, fluorescent and protein isolation tags and the use of disease models to study protein function.

Dr Marcelo Coba Biography:

My laboratory at the Zilkha Neurogenetic institute of USC seeks to understand signaling mechanisms associated to rare disease and complex brain disorders. Many mutations associated with these disorders encode proteins found within the postsynaptic density (PSD) of excitatory synapses. We try to understand how the synaptic signaling machinery is organized in protein interaction networks (PINs) and their role in disease. We pioneered the use of a systems biology approach in synaptic signaling, and developed new methods for its study, including mass spectrometry, computational biology, peptide arrays, CRISPR-Cas technology and mini-CA1 slices-proteomics. I developed hiPSC editing methods using CRSIPR technology that allow us the systems study of signaling networks in patient derived iPSC neurons. My laboratory is actively developing analytical and bioinformatics tools for the systems biology study of cellular processes involved in neurological disorders. Recently, my laboratory defined for the first time the spatio-temporal map of the PSD protein interaction network their modulation by synaptic activity, how it is affected by mutations in their components and the distribution of developmental risk factors within networks As a research associate at the Wellcome Trust Sanger Institute (UK), I developed proteomics technologies to study the network properties of signaling complexes at the synapse. I developed large-scale approaches that have produced the first global maps of synapse signaling defining for the first time the signaling networks interactions between different neurotransmitter receptors. We determined how the Induction of LTP modifies the PSD phosphoproteome and interactome. Thus establishing a framework for the large-scale study of signaling mechanisms modulating different forms of synaptic plasticity.

Speaker's Presentations & Biographies

Prof Mita Bertoldi

Presentation Title:

An Overall Picture of the Structure-Function Relationship of AADC Variants Causing AADC deficiency.

The talk will present all collected biochemical data regarding AADC variants, both the old and the new reported ones (in the last 2 years) causing AADCd with a particular focus on protein localisation of the pathogenic change and structure-function of each variant. The aim is to contribute in giving an help in prediction and treatment.

Professor Mita Bertoldi Biography:

Associate Professor of Biochemistry at the Department of Neuroscience, Biomedicine and Movement of the University of Verona, Italy. Her scientific interest spans from protein chemistry to enzymology. Even if she is involved in multiple research topics, from bovine pancreatic ribonuclease as a model for protein aggregation to peroxiredoxin-2, a novel essential redox enzyme for erythrocytes and central nervous system, her main scientific interest is the chemistry of vitamin B6 dependent enzymes, in particular aromatic amino acid decarboxylase. She gave important contribution to the understanding of the catalytic mechanism of the enzyme together with the unraveling of a pecularіar oxygen-consuming side reaction. In the last years, she is interested in elucidating the structure-function relationships and the kinetics of variants of aromatic amino acid decarboxylase causing AADC deficiency. In her researches she undertakes multiple approaches: a classsical biochemical approach using a combination of biophysical and biochemical methods together with a bioinformatics approach in collaboration with experts in the field. Her research activity is witnessed by several publications on international medium-high ranked journals.

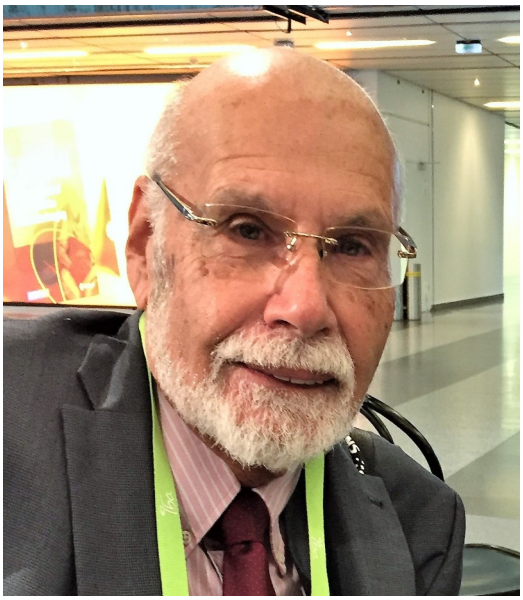
Until now, only the molecular effects of homozygous mutations of the DDC were analyzed. However, although several heterozygous carriers of AADC deficiency were identified, the molecular aspects of their enzymatic phenotypes are not yet investigated. We focused our attention on the R347Q and R358H mutations because both have been found in homozygous patients and are born together by compound heterozygous patients. We purified in recombinant form either the R347Q and R358H homodimeric variants or, for the first time, the R347Q/R358H heterodimeric variant of DDC. The comparison of the biochemical features of the heterodimer with those of the relative homodimers provided evidence for a positive interallelic complementation between the R347Q and the R358H mutations.

Speaker's Presentations & Biographies

Prof. Stanley Satz

Presentation Title:

Non –invasive PET Visualization of AADCd Manifestations, Therapy Planning, Outcomes Monitoring and Theranostics/Artificial Intelligence for Underserved and Rare Paediatric Conditions.



Dr. Satz will lecture on the specific application of positron emission tomography (PET) for non-invasive monitoring of treatment outcomes at an early stage,
visualizing disease manifestations and aiding in clinical management, prognosis and treatment planning of pediatric patients with aromatic l-amino acid decarboxylase (AADC) deficiency and other rare pediatric diseases. PET is a nuclear medicine molecular imaging technique that creates detailed, computerized pictures of organs and tissues inside the body. A PET scan is used to diagnose certain health conditions, to plan treatment, to find out how an existing condition is developing, and to monitor or see how effective a treatment is. Other advantages demonstrated by this novel technology are very short imaging time required, and ease of preparation using automated equipment and superior imaging compared to other modalities.

Advance Innovative Partners has developed Fluorine-18 L-3,4-Dihydroxy-6-[18F] fluorophenylalanine. PET demonstrates a profile of unrivaled radiation safety; no adverse events in patients imaged and is a major advance demonstrating clinical efficacy as a diagnostic. The short half-life and scan time allows for increased detection sensitivity without posing additional radiation risk to the pediatric patient. A specific diagnostic and follow-up tool to

monitor, say, the effectiveness of AAV2-hAADC treatment enables imaging the brain. Positron-emission tomography (PET) imaging with this amino acid radiotracer has several advantages in AADC patients including differentiation, elimination of the need for many of invasive, traumatic brain biopsy

procedures, and improved detection of disease recurrence after treatment.

It is a non-invasive functional imaging technique that measures the distribution and uptake of a positron emitting radiopharmaceutical for optimized targeting. PET enables measurement of changes in standardized uptake values (SUVs) and when performed after the completion of therapy might also help determine which patients are trending toward improved survivals.

Dr Stanley Satz Biography:

Stanley Satz, Ph.D. is Chairman, Chief Scientific Officer and co-founder of Bio-Nucleonics, Inc. a Miami, Florida based manufacturer of diagnostic and therapeutic radiopharmaceuticals, nanoengineered molecular imaging agents and medical devices. A former educator, Satz comes from the 'death sciences' to the 'life sciences'. He is a scientist and radiation physicist with more than 25 years of research and development and production experience, manufacturing and marketing of products for medical and industrial use. In addition, Satz is Chairman of the G8 International Working Group on Nanosafety and Nanosecurity, headquartered in Como, Italy.

He is a leading authority on the development of radiopharmaceuticals required for effective targeted gene therapy, stem cell therapy, radiation therapy and imaging chemistry. His studies have led to initiation of multiple small molecule clinical trials. Basic coordination chemistry studies have also addressed the synthesis and in vivo validation of novel agents that have been translated from pre-clinical studies to clinical use onwards to commercial products for both imaging and therapy of cancer and rare diseases including amino acid dehydrocarbolase deficiency, neuroblastoma, acute lymphoblastic leukemia and diffuse intrinsic pontine glioma.

Satz founded Bio-Nucleonics after leaving Mount Sinai Medical Center of Miami Beach and a Professorship the University of Miami. A graduate of the University of Miami, he is former Chairman and of the Scientific Advisory Board of Florida Atlantic University, and is on the Board of the U.S. Industry Coalition, and a publicly traded biotechnology company. He developed a U.S. Food and Drug Administration approved therapeutic radiopharmaceutical Strontium Chloride Sr-89 Injection, for treatment of bone pain arising from metastatic prostate and breast cancer, and Theranost™, a nanoparticle engineered drug for peptide receptor radionuclide targeted therapy of cancerous tumors, currently in clinical trials. Satz has authored numerous journal articles and holds five issued and three pending medically related U.S. patents.

Satz concentrates his scientific and technical leadership on radiopharmaceutical and receptor-avid peptide development and strategic planning. He is also co-founder of Boca Raton, Florida-based Advanced Imaging Projects, Inc. (AiP), a clinical stage specialty biopharmaceutical company focused on the discovery, development and commercialization of molecular medicines for prevention, diagnosis and treatment of oncological, neurological and infectious disorders; childhood rare diseases in particular. AIP GmbH, located near Erfurt, Germany was established in 2017. In the Company's product portfolio are small molecules that demonstrate safety and efficacy in clinical management of neuroblastoma. Pediatric rare disease designations were granted by the FDA for multiple indications including aromatic-L amino acid decarboxylase deficiency neuroblastoma, diffuse intrinsic pontine glioma and acute lymphoblastic leukemia.

Speaker's Presentations & Biographies

Prof. Krys Bankiewicz

Presentation Title:

Update on AAV2-Haadc Gene Therapy Trials in Primary AADCd in Paediatric Patients (Target Area VTA and SNpc)

In 2017 we started a clinical trial to test a new gene therapy approach for AADC deficiency. The objectives of this clinical trial is to determine the safety and efficacy of 2 doses of adeno-associated virus serotype 2 (AAV2)-hAADC delivered to the substantia nigra pars compacta (SNc) and ventral tegmental area (VTA). Our MR-guided AAV delivery platform permits AAV delivery to the target with submillimetre accuracy and for monitoring of vector delivery in real time. To our knowledge, this is the first prospective study that utilizes axonal transport to deliver a transgene to projection regions (striatum and nucleus accumbens), which control the motor and mesolimbic systems. Here I will present a summary of our data to date and future steps.

Professor Krystof Bankiewicz Biography:

Dr. Krystof Bankiewicz earned his MD at the Jagielonian University, and his PhD from the Institute of Neurology and Psychiatry, both in Poland. He received a NIH-NINDS post-doctoral fellowship and later held the Chief of CNS Implantation Unit and Acting Chief of the Molecular Therapeutics Section positions, also at NIH-NINDS. Currently, he is the Kinetics Foundation Chair in Translational Research and Professor in Residence of Neurological Surgery and Neurology at UCSF. Dr. Bankiewicz is also Vice-Chair for Research in the Department of Neurological Surgery and Director of Interventional Neurology Center at UCSF.

Dr. Bankiewicz has both industry and academic experience, being an inventor on numerous patents and published more than 180 peer-reviewed research articles. He has considerable experience in supervising multi-investigator translational programs and is a Principal Investigator on several research grants and clinical trials. Along the years, he has performed preclinical work that yielded 4 clinical trials both in brain cancer and gene therapy for adult and pediatric neurological diseases. He is a Special Expert in the NIH Grant Review Registry, member of the Editorial Board of numerous journals as well as *ad hoc* referee for scientific journals in the neurology, neuroscience, neurosurgery and molecular therapeutics fields.

Dr. Bankiewicz's research interests are focused on the development of translational approaches to gene and cell replacement therapies, and he has displayed the ability to synthesize distinct technologies into powerful new approaches to the treatment of serious diseases, including brain cancer, Parkinson's disease, Huntington disease, Alzheimer's disease, pediatric neurotransmitter deficiency and lysosomal storage disorders.

Speaker's Presentations & Biographies

Dr Riccardo Montioli

Presentation Title:

Biochemical advances in AADC deficiency: an approach to understand the molecular pathogenesis of heterozygous patients and protein engineering for the enzyme replacement therapy.

Until now, only the molecular effects of homozygous mutations of the DDC were analyzed. However, although several heterozygous carriers of AADC deficiency were identified, the molecular aspects of their enzymatic phenotypes are not yet investigated. We focused our attention on the R347Q and R358H mutations because both have been found in homozygous patients and are born together by compound heterozygous patients. We purified in recombinant form either the R347Q and R358H homodimeric variants or, for the first time, the R347Q/R358H heterodimeric variant of DDC. The comparison of the biochemical features of the heterodimer with those of the relative homodimers provided evidence for a positive interallelic complementation between the R347Q and the R358H mutations. Our findings shed some light on the molecular complexity of the heterozygous condition revealing that the enzymatic phenotype of a combined heterozygous patients could exhibit features substantially different from those are expected. A second project we have recently undertaken concerns the engineering of the human DDC in order to move the first steps toward an enzyme replacement therapy of AADC deficiency. In the first place our efforts are aimed to fix the enzyme "weaknesses" to achieve a more stable and active variant of AADC that will be employed in the development of a delivery strategy.

Dr Riccardo Montioli Biography:

In 2004 Dr. Montioli gained master's degree in Agro-Industrial Biotechnologies and in 2009 achieved a doctorate in Biochemistry at the University of Verona (Italy). Since January 2010 to date he worked as post-doc researcher at the Dep. of Neurosciences, Biomedicine and Movements sciences of University of Verona. The scientific activity of Dr. Montioli is focused on the pyridoxal 5'-phosphate(PLP)-dependent enzymes involved in human diseases. By means of biochemical, molecular biology, and computational techniques he contributed to define the structural/functional relationships of several enzymes and the molecular defects of their pathogenic variants. In particular, the most recent projects he carried out concerned the characterization of the human ornithine δ-aminotransferase, whose deficit causes the Gyrate Atrophy and of the human Dopa decarboxylase, whose deficiency causes the AADC deficiency syndrome and contribute to the Parkinson's disease symptoms.

Dr Toni Pearson

Presentation Title:

The Natural History of AADC Deficiency: a Retrospective Study

Dr Toni Pearson Biography

Dr. Pearson obtained her medical degree from the University of Adelaide Medical School in Australia, and then pursued postgraduate residency and fellowship training in child neurology and movement disorders at Columbia University in New York. She is currently Associate Professor of Neurology at Washington University in St. Louis, where she directs the program for Pediatric Movement Disorders and Cerebral Palsy.

Speaker's Presentations & Biographies

Dr Todd Berner

Presentation Title:

PTC Acquisition and AADCd GT Update.

Update on PTC acquisition of Agilis Biotherapeutics and Update on AGIL-AADC Clinical trial data.

Dr Todd Berner Biography:

Todd Berner is focused on listening to the Voice of the Patient in Gene Therapy for Rare Diseases and in Immunology as Vice President and Head of Medical Affairs, Americas for PTC Therapeutics, continuing in his responsibilities after the acquisition of Agilis Biotherapeutics by PTC. He has been actively reimagining nimble Rare Disease Medical Affairs functions fit for purpose and relevant to an evolving, value based healthcare environment. He has been engaged in the pursuit of quality and value, first as a clinician leading quality improvement initiatives, and for the past 15 years leading clinical and health outcomes research initiatives at the patient level, as well as from the provider and health system perspective. He formerly was Senior Medical Director and Therapeutic Area Head for Shire's Global Medical Affairs Immunology organization. responsible for leading a team of Medical Affairs professionals, directing all Medical Affairs functions of the division, including clinical trial planning and LCM execution, and safety studies, while providing medical input to cross functional Global teams, including New Product Development. He has made contributions to data generation encompassing Pragmatic, Observational, Comparative Effectiveness, Big Data, and Humanistic studies and has authored numerous publications and scientific presentations on various aspects of the costs and consequences of pharmacy and medical interventions and health policies. He previously was Executive Medical Director, Urology at Astellas, leading the Americas Urology Medical Affairs team and responsible for Clinical Trials, HEOR, and Field Based MSL teams, developed and led many RWE Research Collaborations. Among his proudest accomplishments were, Co-chair ISPOR Patient Centered SIG, and Patient Engagement in Research Working Group, PCORI Grant Reviewer, Sr. Fellow School of Population Health at Thomas Jefferson University, and Chair PQA Palliative Care Pain Management Working Group, Journal reviewer for Value in Health, and for JMCP.

Thomas Goss

Presentation Title:

Measuring the Burden of Illness in AADC Deficiency

Dr. Goss will present a brief overview of a recently initiated observational, multinational study designed to measure the burden of illness associated with AADC deficiency. The purpose of this study is to estimate the total burden of illness (BOI) for patients with AADC-Deficiency in patients in Germany, the United Kingdom, and United States. Specifically, this will be done in two ways: 1) Chart abstraction 2) Caregiver survey.

By systematically measuring the BOI, stakeholders will have a more complete understanding of the burden of the disease – including monetary costs and quality of life - which is essential in benchmarking the clinical and economic value of new treatment options.

Tom Goss Biography:

Thomas Goss has more than 20 years of experience managing and directing healthcare research, including the impact of public and private payer policy on patient access, health-related quality-of-life, patient preference, patient satisfaction, and health economic evaluations. He also directs disease management program evaluations using patient outcomes data. He has published extensively in these areas and has more than 40 peer-reviewed publications and over 75 peer-reviewed abstracts and invited publications. Prior to joining Boston Healthcare Associates, Dr. Goss was Vice President and Director of Consulting Services at Covance Market Access Services, where he had a 15-year career with increasing levels of responsibility in the areas of client management and executive management. Dr. Goss received a Pharm.D. from the State University of New York at Buffalo, and a B.S. from the Albany College of Pharmacy. He completed graduate course work in Epidemiology, and a postdoctoral fellowship in Pharmacoepidemiology and Outcomes Research at the State University of New York at Buffalo. Dr. Goss is a member of the American College of Clinical Pharmacy Outcomes and Economics Practice Research Network, and the International Society of Pharmacoeconomics and Outcomes Research. Dr. Goss is currently an appointee to the U.S. Center for Medicare & Medicaid Services (CMS) Medicare Evidence Development & Coverage Advisory Committee (MEDCAC).

Speaker's Presentations & Biographies

Dr Marcel M. Verbeek

Presentation Title:

Next Generation Metabolic Screening for Elucidation of AADC deficiency and related neurometabolic Disorders.

Timely diagnosis is essential for patients with neurometabolic disorders, as the identification of the deficient step in metabolism is necessary for targeted treatment. At present, neurometabolic disorders are diagnosed by targeted biochemical assays in cerebrospinal fluid (CSF) that are aimed at quantification of a limited number of metabolites. We have developed a new approach, so-called "next-generation metabolic screening" (NGMS), where we use a combination of liquid chromatography and mass spectrometry to semi-quantify an unlimited number of metabolites simultaneously. This approach allows for both simultaneous screening for multiple diseases and obtaining a complete view of disturbed metabolic pathways. Furthermore, it allows identification of yet unidentified disease-related biomarkers, which will improve diagnosis, treatment monitoring and/or understanding of disease mechanisms. NGMS has already shown promising results when applied to plasma. For example, it led to the discovery of a new neurometabolic disorder, N-acetylneuramic acid phosphate synthase deficiency.[1] We now wish to implement this strategy to CSF analysis as well, since this is the biofluid most closely related to the brain. In an NGMS pilot study, using a small number of samples, we have been able to identify several known biomarkers of AADCd (e.g. 3-methoxytyrosine, homovanillic acid) in CSF. Currently, we are investigating additional biomarkers of AADCd that may provide novel information on (personalized) treatment efficiency, prognosis and disease mechanisms. Moreover, we will extend our Investigations to the study of CSF from patients with other known, but also yet uncharacterized, neurometabolic disorders. In conclusion, we believe that the application of NGMS to CSF analysis will greatly improve the understanding, time-to-diagnosis and identification of neurometabolic disorders.

Dr Marcel Verbeek Biography:

I am an Associate Professor in Neurochemistry of Neurodegeneration, at the department of Neurology of the Radboud university medical center, Nijmegen, The Netherlands. Besides, I am principal investigator at the Donders Institute for Brain, Cognition and Behaviour. My research is focused on the neurochemistry of neurodegenerative disorders, specifically movement disorders (e.g. Parkinson's disease, and related disorders, including parkinsonism and pediatric movement neurotransmitter movement disorders) and dementia syndromes (e.g. cerebral amyloid angiopathy [CAA], Alzheimer's disease and related disorders).In my research I aim to obtain a closer understanding of the pathophysiological mechanisms of neurodegenerative disorders. I aim to translate novel insights from pathofysiological studies into biomarkers of disease. Novel candidate biomarkers are identified through screening techniques (such as proteomics and metabolomics) or from new insights into the underlying pathophysiology. The biomarkers mainly comprise proteins, enzymes and metabolites that each have a specific relation to a disease. Therefore, I work on the development and validation of biomarkers in body fluids (especially cerebrospinal fluid) for diagnosis and prognosis of these disorders; I collaborate with clinicians within the Parkinson Centre Nijmegen, the Expertise Center of Inherited Movement Disorders, and the Radboud Alzheimer Centre to reach this goal as well as with many (inter) national researchers. I established a large biobank with cerebrospinal fluid and blood samples from patients with many different types of neurological disorders has been established to support this research. I also lead the national reference centre for specialized CSF diagnostics. This allows me to immediately translate and implement novel biomarker assays in the routine diagnostic work-up of neurological disorders and offer these to clinicians at other institutes. These biomarkers will create personalized value for diagnostic or prognostic purposes. Novel insights into disease mechanisms may also lead to the development of tailor-made therapies, that can be monitored using biomarkers, and therefore, my approach of the combination of basic and translational neurosciences contributes to personalized heath care.

Dr Tessa Peters

Dr Tessa Peters Biography:

Tessa Peters is a PhD candidate at the Translational Metabolic Laboratory and the Department of Neurology of the Radboudumc in Nijmegen, the Netherlands. She has a big passion for translational research, wanting to make sure that impressive results from the lab find their way to the clinic, so that not only science, but also patients will benefit. Tessa graduated with honours from Radboud University Nijmegen in 2016, with a master's degree in Biomedical Sciences. She started her career at the Radboud Biobank, a facility for scientists to store their collections of biospecimens, such as blood or cerebrospinal fluid (CSF), to be used in biomedical research. Here, she streamlined laboratory processes and advised researchers on how to set up an optimal collection of biospecimens and clinical data. Furthermore, she set up a biobank of her own for patients with inborn errors of iron metabolism. Inspired by the various research projects she supported, she decided to become a full-fledged researcher herself. In her current project, she focuses on neurometabolic disorders, studying CSF samples to find new biomarkers for diagnosis and treatment of these diseases. To do so, she uses "next-generation metabolic screening": a liquid chromatography/mass spectrometry approach that allows semi-quantitative measurement of ten thousands metabolites at a time. Her goal is to implement this approach in the diagnosis and management of neurometabolic patients, including those with AADC deficiency.

Speaker's Presentations & Biographies

Prof. Shin-Ichi Muramatsu

Presentation Title:

Update on AADCd Gene Therapy in Japan (Target Putamen)

We conducted an open-label phase 1/2 study of population including adolescent patients with different degrees of severity. Seven patients were enrolled, four male (age: 4, 10, 15 and 19 years) patients and 2 female (age: 6 and 12 years) patients with a severe phenotype who were not capable of voluntary movement or speech, and one female patient (age: 5 years) with a moderate phenotype who could walk with support. The patients received a total of 2×10^{11} vector genomes of adeno-associated virus vector harboring *DDC* via bilateral intraputaminal infusions. At up to two years after gene therapy, motor function was remarkably improved in all patients. Three patients with the severe phenotype could stand with support and one patient could walk with a walker, while the patient with the moderate phenotype could run and ride a bicycle. This patient also showed improvement of mental function to be able to converse fluently and perform simple arithmetic. Dystonia had disappeared and occulogyric crisis was markedly decreased in all patients. The patients exhibited transient choreic dyskinesia for a couple of months, but adverse events caused by vector were not observed.

Professor Shin-Ich Muramatsu Biography:

Shin-ichi Muramatsu is currently a Professor of neurology at Jichi Medical University (JMU) in Japan. He received his M.D. and Ph.D. degrees from JMU, Harrisonburg, VA, USA. He completed his neurology residency at both Gunma University and JMU. He was a Visiting Associate at National Institutes of Health in USA from 1995 to 1997. He received the Award for Excellent Research (2001) and the TAKARA Bio Award (2011) from the Japan Society of Gene Therapy. His research group has been developing gene therapies for treating neurodegenerative diseases including Parkinson's disease (PD), Alzheimer's disease, and amyotrophic lateral sclerosis using adeno-associated virus (AAV) vectors.

Dr Karin Kojima

Presentation Title:

Gene Therapy Case Studies in Japan

Dr Karin Kojima Biography:

She is currently an assistant professor of Pediatrics at Jichi Medical University (JMU) in Japan. She completed her Pediatrics and Child neurology residency at JMU. Her research interest is gene and cell therapies for child neurological diseases. She is a member of clinical research group of the gene therapy for AADC deficiency. She received Japan Society of Gene and Cell Therapy Award for Outstanding Research in Gene Therapy (2017) and Journal of Gene Medicine JSGCT Young Investigator Award (2018).

Speaker's Presentations & Biographies

Dr Charles Laurenco

Presentation Title:

Against All Odds : Diagnosis and Presentation of Brazilian AADC deficiency Patients.

The goal of the presentation is to present the current state of diagnosis and management of patients in Brazil with AADC deficiency, challenges and barriers that still persist for those patients.

Dr Charles Laurenco Biography:

Charles M Lourenço is Professor of Clinical Genetics and Applied Medical Research at the Faculty of Medicine, Centro Universitario Estacio in Ribeirão Preto, São Paulo, Brazil. Dr Lourenço is a clinical biochemical geneticist with a special interest in genetic neurodegenerative disorders. He obtained his Medical Degree at the Federal University of Bahia, Brazil, in 2002, and underwent postgraduate training in medical genetics and then neurogenetics at the Clinical Hospital of the State University of São Paulo, and then the Hospital of Ribeirão Preto, University of São Paulo. He holds a PhD in neurogenetics, with his thesis focussing on spinocerebellar ataxia of early onset, especially on a subset of patients with ataxia and hypogonadism. Most recently, he has been involved in a new multidisciplinary clinic at his hospital, which focuses primarily on investigation of childhood neurodegenerative disorders and, in particular, patients with early-onset cerebellar ataxia and genetic white matter disorders. Dr Lourenço's interests include the clinical and molecular aspects of leukodystrophies, hereditary spastic parapareses, metabolic causes of neonatal cholestasis, hereditary spinocerebellar ataxias, genetic epileptic encephalopathies, lysosomal disorders of the brain (neurolipidoses) and inborn errors of metabolism with adult presentation.Dr Lourenço is a member of many professional societies, including the Brazilian Clinical Genetics Society, the American Society of Human Genetics, the International Skeletal Dysplasia Society, the Society for the Study of Inborn Errors of Metabolism, and the Latin American Society of Inborn Errors of Metabolism and Newborn Screening. He has published extensively in journals and books, and serves as a peer reviewer for *Neurology Genetics* and the *Journal of Inherited Metabolic Disease*

Dr Paldeep Atwal

Presentation Title:

Metabolic Profiling in AADCd

Dr Paldeep Atwal Biography

Dr. Atwal is a clinical and medical biochemical geneticist and director of The Atwal Clinic for Genomic & Personalized Medicine, a concierge genetics clinic. Most recently, joined the team at Genome Medical as a medical geneticist. Previously he served as Medical Director for the Center for Individualized Medicine at Mayo Clinic FL on the Jacksonville campus. He received his medical degree from the University of Glasgow, and initially trained in hospital internal medicine with The Royal College of Physicians at Glasgow Royal Infirmary in Scotland. He completed his genetics fellowship at Stanford University in Palo Alto CA and subspecialty biochemical genetics fellowship at Baylor College of Medicine in Houston TX where he was involved in developing a clinical metabolomic profiling test. In addition he holds diplomas in structural molecular biology and forensic medical science and is nearing completion of an MBA program. He has a long-standing interest in rare and undiagnosed disease including the use of multiple concurrent –omics platforms to provide a diagnosis to patients. He conducts translational, (from patient to laboratory to patient) research with the goal of discovering new genetic syndromes and designing new therapies for genetic disease. Dr. Atwal's clinical interests include clinical genomics, undiagnosed diseases following lengthy diagnostic odysseys and inborn errors of metabolism including mitochondrial diseases. Through his work, he has discovered two new genetic connective tissue syndromes that results from defects in the FLNA & AEBP1 genes, co-developed an untargeted metabolic screening test for inborn errors of metabolism, and published extensively on human genetics, with over 50 peer-reviewed publications to date. Dr. Atwal's awards include the 2014-2015 ACMG Foundation/Genzyme Fellowship in Biochemical Genetics Award and The Neurobiology of Disease in Children Young Investigator Award.

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Dear Delegates,

On behalf of The AADC Research Trust and it's team we wish you all a safe journey home and very much look forward to working with you in the future for the benefit of all children, around the World, suffering with Aromatic Amino Acid Decarboxylase Deficiency.

Thank you for your interest in our children's disease….

Best Regards

Lisa Flint

Founder & Managing Director

The AADC Research Trust would like to thank our conference sponsors for their contribution to this important event.

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