PROGRAM

SENSGENE
ANNUAL DAY

Rare Diseases & Malformations of Sensory Organs

Imagine Institute, Paris

25 / 01

19

SENSGENE
Maladies Rares Sensorielles

FILIÈRE DE SANTÉ MALADIES RARES

PROGRAM

Moderation :

1st session : Pr Patrick Calvas
2nd session : Dr Jean-Michel Rozet
3rd session : Dr Anne-Françoise Roux
4th session : Pr Hélène Dollfus

INTRODUCTION

From 8:45 **Welcome coffee**

9:15-9:20 Opening of the first SENSGENE annual clinics & research day
Pr Hélène Dollfus, coordinator of SENSGENE, CHU Strasbourg, France

1st SESSION - GENETICS IN RARE EYE MALFORMATIONS

09:20-10:05 Anterior Segment Dysgenesis, an update
Pr Elena Semina, Medical College Wisconsin, USA

10:05-10:50 Animal models in gene discovery and understanding of eye malformations
Pr Anne Slavotinek, University of California, San Francisco, USA

10:50-11:15 Coffee break

11:15-12:00 Genetics and Micro-Anophthalmia
Pr Nicola Ragge, Birmingham Women's and Children's NHS Foundation Trust, Birmingham and Oxford Brookes University, Oxford UK

2nd SESSION - FRENCH STATE OF THE ART FOR RESEARCH IN RARE EYE MALFORMATIONS

12:00-12:15 Genotype/phenotype correlation in aniridia
Pr Dominique Brémond-Gignac, Necker Enfants-Malades Hospital, Paris, France

12:15-12:30 The French cohort RaDiCo-Ac-Œil
Pr Patrick Calvas, CHU Toulouse, France

12:30-12:45 About non coding mutations in rare developmental diseases
Dr Nicolas Chassaing, CHU Toulouse, France

12:45-13:00 The challenges of rare iridal malformations: the microcoria example
Dr Jean-Michel Rozet, Imagine Institute, Paris, France

13:00-14:00 Lunch

3rd SESSION - FRENCH STATE OF THE ART FOR RESEARCH IN RARE EAR MALFORMATIONS

14:00-14:45 Malformation of the outer ear and mandibulofacial dysostosis
Pr Jeanne Amiel, Necker Enfants-Malades Hospital, Imagine Institute, Paris, France

14:45-15:15 Inner ear malformations
Dr Sandrine Marlin, Necker Enfants-Malades Hospital, Imagine Institute, Paris, France

15:15-15:30 Genetic basis of non-syndromic hearing loss with enlarged vestibular aqueduct
Dr Fiona Alvin, Necker Enfants-Malades Hospital, Imagine Institute, Paris, France

4th SESSION – SHORT COMMUNICATIONS FROM SENSGENE MEMBERS

15:30-16:30 Molecular characterization of a series of 990 index patients with albinism
Dr Eulalie Lasseaux, CHU Bordeaux, France

Linear naevus sebaceous syndrome with ocular involvement
Dr Laura Mauring, East Tallinn Central Hospital, Estonia (ERN-EYE fellow at CHU Strasbourg, France)

Characteristics of the management of patients with a Geyser malformation of the inner ear
Dr Isabelle Rouillon, Hôpital Necker, Paris, France

Malformations of the outer ear
Dr Marie-Cécile Cochet, Hôpital Necker, Paris, France

CONCLUSION

16:30- 16:35 General conclusions
Pr Hélène Dollfus, coordinator of SENSGENE, CHU Strasbourg, France

INVITED SPEAKERS

Rare Eye Malformations:

Pr Elena V. Semina

Medical College Wisconsin, USA

Pr Elena Semina is the Chief of the Division of Developmental Biology within the Department of Pediatrics, and a Professor in the Departments of Pediatrics, Ophthalmology and Visual Sciences, and Cell Biology, Neurobiology, and Anatomy (CBNA) at the medical College of Wisconsin.
She has significant expertise in vision research with over 90 publications and continuous NIH funding since 2002. She serves as an Ad Hoc Reviewer for numerous granting agencies, on the Editorial Board of *Molecular Vision*, and as Vice President, Americas, for the International Society for Eye Research (ISER).
Pr Semina has mentored over 50 undergraduate, graduate, and postdoctoral students at MCW. She is a member of the Executive Evaluations Committee, regularly teaches PhD and MD students, and has been a member of many PhD thesis committees.

Pr Anne Slavotinek, MD

Medical Genetics and Genomics Clinic, San Francisco, USA

Pr Anne Slavotinek is a pediatric medical geneticist practicing at UCSF Medical Center in San Francisco, California.

She's specialized in clinical genetics. Her interests include dysmorphology - the study of patterns of congenital anomalies - and her research is directed towards the discovery and understanding of genes that cause multiple congenital anomaly syndromes.

Slavotinek earned her medical degree from the University of Adelaide in South Australia. After moving to the United Kingdom and completing her doctorate, she did a residency in medical genetics at the Churchill Hospital in Oxford and a fellowship at St. Mary's Hospital in Manchester. Then she went to the National Institutes of Health where she completed a fellowship in medical genetics.
She is a professor in the Division of Medical Genetics in the Department of Pediatrics at UCSF.

Pr Nicola Ragge

Oxford Brookes University & Birmingham Women & Children hospital, UK

Pr Nicola Ragge trained in Paediatrics before entering Ophthalmology. After her general Ophthalmology training, she undertook Fellowships in Paediatric Ophthalmology, Neuro-ophthalmology, Adnexal Surgery and Ophthalmic Genetics. She started to specialise in understanding the genetics of congenital eye anomalies, mainly anophthalmia and microphthalmia in 1999 whilst working in the Adnexal Dept at Moorfields Eye Hospital with Professor Richard Collin and set up a joint multidisciplinary clinic. In 2003, she was awarded a Senior Surgical Scientist Fellowship to establish a research programme in developmental eye genetics in Oxford whilst working as Consultant Paediatric Ophthalmologist in Moorfields and Birmingham Children's Hospital. She undertook specialist training in Clinical Genetics to become dual accredited in Genetics and Ophthalmology, and now works as a consultant geneticist, with a special interest in developmental eye genetics and runs a national laboratory research programme on Eye Genetics in Oxford. She has identified new genes and syndromes involving eye anomalies and has established new rapid diagnostic testing for these genes.

Rare Ear Malformations:

Pr Jeanne Amiel

Necker Enfants-Malades Hospital, Paris, France

Pr Jeanne Amiel is Professor of Developmental Biology and Clinical Geneticist at Imagine Institute, University Paris Descartes, Hôpital Necker in Paris, France.

She has conducted a number of studies aiming at gene identification of neural crest cell derived congenital malformations and tumour predisposition, in particular in Hirschsprung disease, congenital central hypoventilation, neuroblastoma, conotruncal heart defects and mandibulofacial dysostoses. She is or has been the coordinator or associate investigator in a number of research programs of the French National Agency for Research (ANR) and the Fondation pour la Recherche Médicale (FRM).

SENSGENE SPEAKERS

Rare Ear Malformations:

Dr Sandrine Marlin

Necker Enfants-Malades Hospital, Paris, France

Dr Sandrine Marlin is a pediatric geneticist, clinician and researcher in the field of genetic deafness. Coordinator of a national reference center and a European Network on the same theme, she works at the Necker Hospital (AP-HP) and the Imagine Institute. She has published extensively in the field as well as more than 90 articles in peer-reviewed journals.

Dr Fiona Alvin

Necker Enfants-Malades Hospital, Paris, France

Dr Fiona Alvin is a Doctor of Medicine, specialised in Otorhinolaryngology and Cervico-Facial Surgery, registered with the Order of Physicians of Val-de-Marne in November 2017.
She's currently in Internship of Master's degree at the Laboratory INSERM UMR 1163-Laboratory «Embryology and Genetics of Malformations», at Imagine Institute.

She graduated from a Specialized Diploma in Otorhinolaryngology and Head and Neck Surgery, Paris VII-Diderot University, in 2017.

Rare Eye Malformations:

Pr Dominique Brémond-Gignac - MD, PhD

Necker Enfants-Malades Hospital, Paris, France

Pr Dominique Brémond-Gignac is Professor of Ophthalmology specialized in Pediatric Ophthalmology. She is currently Head of Ophthalmology Department with pediatric subspecialty at University Hospital Necker-Enfants Malades and Paris V René Descartes University in Paris.
Head of Paris V Orthoptic Departement, affiliated to the CNRS FR3636 Research Unit, in binocular vision, her activity is distributed in clinical practice, teaching and research (eye development). Current practice includes pediatric anterior segment, strabismus and oculo-plastic surgery as she is also graduated in maxillo-facial surgery. She contributed over a hundread peer review publications in the ophthalmic literature and more than fifty book chapters. Involved in visual health in children, she is Executive member of WSPOS *(World Strabismus and Pediatric Ophthalmology Society).* She is also expert for the French Health and Higher Education Ministries about Orthoptic training, head of CLAIROP Research Clinical Center accredited by Europe EVI-CT and head of OPHTARA Rare Eye Diseases Center accredited by French Health Ministery and ERN-EYE (accredited by European Commission).

Pr Patrick Calvas - MD, PhD

CHU Toulouse, France

Pr Patrick Calvas is Professor of Medical Genetics at the University of Toulouse - UPS-Toulouse III.
He is Head of Medical Genetics Department in Toulouse University Hospital, Research group leader on Ocular Developmental Defects at INSERM/UPS UMR1056-UDEAR - Toulouse, and Vice-Chair of the French Human Genetics Society (SFGH).

Dr Nicolas Chassaing - MD,PhD

CHU Toulouse, France

Medical geneticist working in the Medical Genetics Department of Purpan's Hospital (Toulouse, France), Dr Nicolas Chassaing has both clinical genetics and molecular genetics activities (molecular diagnosis of ocular developmental defects mainly).
He also obtained a PhD and an agreement thesis to supervise research. His research topic concern ocular developmental defects such as microphthalmia, anophthalmia, aniridia, and anterior segment dysgeneses. These researches are conducted in Toulouse (UMR 1056 INSERM - Université de Toulouse laboratory).
He is the principal investigator of the rare disease cohort RaDiCo-AC-Oeil which aims to better understand natural history of patients with ocular developmental defects by recruiting patients among 65 investigative centres during 10 years.

Dr Jean-Michel Rozet

Imagine Institute, Paris, France

Dr Jean-Michel Rozet is a PhD in Genetic Sciences from Paris Descartes University and Research Director INSERM. He trained in genetics with Professor Arnold Munnich before becoming a pupil of Dr. Josseline Kaplan, who he succeeded in 2009 as head of the Ophthalmic Genetics Laboratory of the Institute of Genetic Diseases of Paris, Imagine. His research focuses on degenerative diseases of the retina and optic nerve as well as abnormalities of ocular embryonic development. The Ophthalmic Genetics Laboratory has to its credit the identification of many genes of iconic diseases such as Leber congenital amaurosis, Stargardt's disease, Kjer's disease and other optic neuropathies, congenital microcorie, Gillespie syndrome ... In addition to the genetic decoding of these diseases, the laboratory is developing a therapeutic approach to retinal diseases based on the use of antisense oligonucleotides.

SENSGENE NETWORK

A network at the service of Rare Eye & Ear Diseases

SENSGENE carries out national missions around rare sensory diseases with the aim of improving the care of patients, coordinating and encouraging research and developing training and information.

The network brings together 15 reference centers and 52 competence centers. These centers are labeled by the Ministry of Health.

4 main objectives:
- Guide patients and health professionals
- Developing successful clinical and molecular diagnostic strategies
- Facilitate access to medico-social care
- Allow the establishment of new clinical trials and make access possible to all patients in the territory

SENSGENE takes part in two European reference networks: ERN-EYE and ERN-Cranio.

Pr Hélène Dollfus, coordinator of SENSGENE

www.sensgene.com

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