

## RESEARCH CONFERENCES

ESF-UB Conference in Biomedicine in association with Eurovisionnet and Fondation Voir et Entendre

# Rare Diseases II: Hearing and Sight Loss

Hotel Eden Roc, Sant Feliu de Guixols (Costa Brava) • Spain  
22 - 27 November 2009

Chairs:

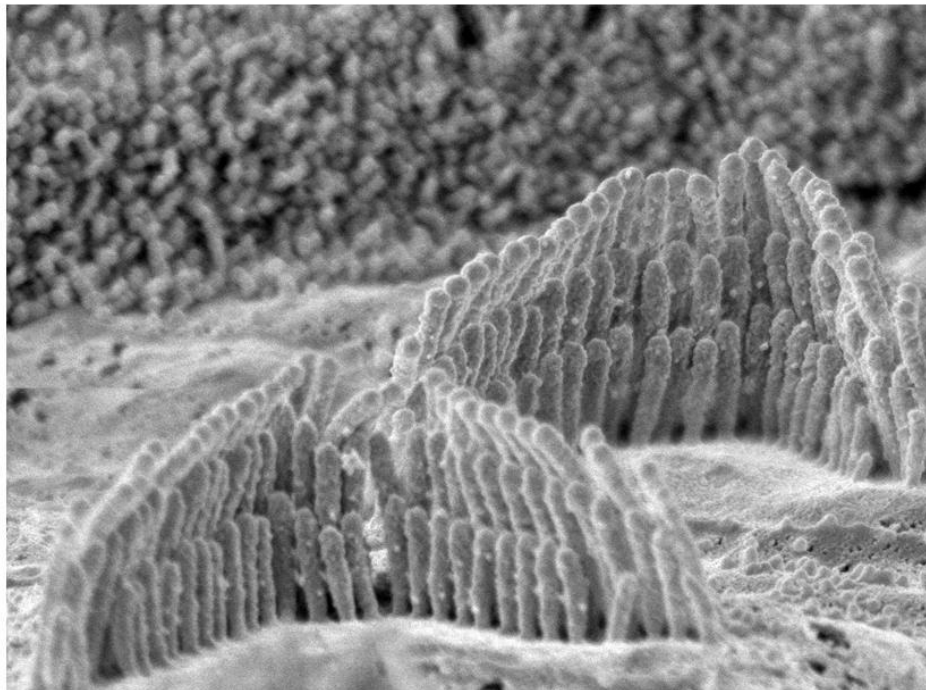
**Christine Petit**, Institut Pasteur, Paris, FR &  
**José-Alain Sahel**, Institut de la Vision, Paris, FR

Organising Committee:

**Olivier Lorentz**, Institut de la Vision, Paris, FR  
**Michèle Roa**, Institut Pasteur, Paris, FR

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# Final Programme

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## Sunday, 22 November

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Late afternoon /  
early evening

19.00

20.00

Registration at the ESF-RC desk

Welcome Drink

Supper

## Monday, 23 November

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08.30-09.00

**Conference Opening**

**Christine Petit**

Institut Pasteur, Paris, FR

**Presentation of ESF**

**Isabel Varela-Nieto**

Universidad Autonoma de Madrid, ES

ESF Rapporteur

**Chairs:** **Adam Sillito**, Institute of Ophthalmology, London, UK, **Robert V. Shannon**, House Ear Institute, Los Angeles, US

09.00-10.20

**Process of the sensory information in the two sensory systems**

**Yves Fregnac**

Institut de Neurobiologie Alfred Fessard, Gif-sur-Yvette, FR

*Reading out the neural code by listening to the synaptic echoes of the visual world in a single cell*

**Paul Avan**

University of Auvergne, Clermont-Ferrand, FR

*From key stages of acoustic signal processing to the clinical impact of their disruption*

10.20-10.50

Coffee break

10.50-12.30

**Visual and auditory neurophysiology**

**Adam Sillito**

Institute of Ophthalmology, London, UK

*Role of motion linked feedback in visual processing*

**Sophie. K. Scott**

Institute of Cognitive Neurosciences, London, UK

*Acoustic speech and voice signal neural processing*

12.30

Lunch

**Chairs:** **Alain Chedotal**, Institut de la Vision, Paris, FR, **Paul Avan**, University of Auvergne, Clermont-Ferrand, FR

13.30-14.50

**Evolution of the two sensory systems**

**Don Zack**

Johns Hopkins Hospital, Baltimore, US

*Role of transcription factors in the evolution of the visual system*

**Hernan Lopez-Schier**

Centre de Regulació Genómica, Barcelona, ES

*Hair-cell function through evolution*

14.50-16.10	<p><b>Development of the sensory organs</b></p> <p><b>Valeria Marigo</b> University of Modena and Reggio Emilia, IT <i>Development of the vertebrate retina</i></p> <p><b>Donna Fekete</b> Purdue University, West Lafayette, US <i>Development of the vertebrate inner ear</i></p>
16.10-16.40	Coffee break
16.40-17.20	<p><b>Neural development and wiring</b></p> <p><b>Alain Chedotal</b> Institut de la Vision, Paris, FR <i>Genetic dissection of hindbrain commissures</i></p>
17.20	<p><b>Inherited genetic development disorders of the retina and the cochlea</b></p> <p><b>Shomi Bhattacharya</b> Institute of Ophthalmology, London, UK <i>Genetics of Retinitis pigmentosa : focus on splicing factors</i></p> <p><b>Karen Avraham</b> Sackler School of Medicine, Tel Aviv, IL <i>MicroRNAs in the vertebrate inner ear</i></p>
20.00	Dinner

## Tuesday, 24 November

**Chairs:** José Cunha-Vaz, AIBILI, Coimbra, PT, Hannie Kremer, Radboud University Nijmegen, NL

08.30-09.50	<p><b>Visual and auditory sensory transduction</b></p> <p><b>Eberhart Zrenner</b> University of Tübingen, DE <i>Signal transduction in photoreceptors and related diseases</i></p> <p><b>Maryline Beurg</b> University Bordeaux II, FR <i>Mechanotransduction of mammalian cochlear hair cells</i></p>
09.50-10.20	Coffee break
10.20-11.40	<p><b>Signal transduction diseases</b></p> <p><b>Christina Zeitz</b> Université Pierre et Marie Curie Paris 6, FR <i>New insights towards the understanding of night blindness</i></p> <p><b>Christine Petit</b> Institut Pasteur, Paris, FR <i>Mechanosensation in auditory sensory cells: what did we learn from deafness genes?</i></p>
11.40-12.30	<p><b>From rare to complex disease</b></p> <p><b>Philip J. Luthert</b> UCL Institute of Ophthalmology, London, UK</p>
12.30	Lunch

**Chairs: Hélène Dollfuss**, CHU de Strasbourg, FR, **Ping Chen**, Emory University, Atlanta, US

13.30-14.30

**Role of the cilium**

**Philip Beales**

Institute of Child Health, London, UK

*Sensory deficits and disease mechanisms of Bardet-Biedl syndrome*

14.30-15.50

**Cilium in pathogenesis**

**Hélène Dollfuss**

CHU de Strasbourg, FR

*Clinical and biological overview of the retinal degeneration in ciliopathies*

**Ping Chen**

Emory University, Atlanta, US

*Shaping the mammalian hearing organ by the planar cell polarity pathway*

15.50-16.20

Coffee break

16.20-17.00

**Usher syndrome pathogenesis**

**Hannie Kremer**

Radboud University Nijmegen, NL

*Usher interactome*

17.00-18.30

**Genotype/phenotype correlation issues in rhodopsin and connexion paradigms**

**Isabelle Audo**

Université Pierre et Marie Curie, Paris, FR

*Mutation spectrum of rhodopsin mutations in French autosomal dominant rod-cone dystrophy patients and their phenotype/genotype correlation*

**Francisco J. del Castillo**

Hospital Ramón y Cajal and CIBERER, Madrid, ES

*Role of connexins in sensorineural hearing impairment*

18.30

Poster session

21.00

Dinner

**Wednesday, 25 November**

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**Chairs: Arne Holmgren**, Karolinska Institute, Stockholm, SE, **Karen Avraham**, Tel Aviv University, IL

08.00-08.30

**Bernd Fritsch**

University of Iowa, US

*Molecular conservation of eye and ear development*

08.30-09.30

## Function and dysfunction of ribbon synapses

Johann H. Brandstätter

University of Erlangen-Nuremberg, DE

*Function and dysfunction of photoreceptor ribbon synapses*

Paul Fuchs

Johns Hopkins University, Baltimore, US

*Synaptic physiology of cochlear hair cells*

09.30-10.30

## Oxidative stress

Arne Holmgren

Karolinska Institute, Stockholm, SE

*Defence against oxidative stress and redox signaling*

Thierry Leveillard

Institut de la Vision, Paris, FR

*RdCVF signaling*

10.30-11.00

Coffee break

11.00-13.00

## 8 Short Talks:

Paulo Ferreira

Duke University Medical Center, Durham, US

*RanBP2-mediated neuroprotection is associated with the modulation of functionally diverse but linked pathways in response to oxidative stress*

Tudor Constantin Badea

Johns Hopkins University & Howard Hughes Medical Institute, US

*Brn3a, Brn3b, and Brn3c define distinct populations of retinal ganglion cell type*

Baerber Rohrer

Medical University of South Carolina, Charleston, US

*Mitochondrial capacity as a biomarker of photoreceptor cell stress*

Enrique De la Rosa

Centro de Investigaciones Biológicas CSIC, Madrid, ES

*Systemic AAV-mediated delivery of proinsulin delays visual loss in a retinitis pigmentosa mouse model*

Aziz El Amraoui

Institut Pasteur, Paris, FR

*The mammalian beta heavy spectrin, an unconventional spectrin defies convention in the auditory and photoreceptor cells*

Catherine Weisz

Johns Hopkins University, Baltimore, US

*Synaptic activity and stimulation of type II cochlear afferents*

Ben Warren

University of Sussex, Brighton, UK

*The dynein motor is the basis of active oscillations of mosquito antennae*

Natalia Caporale

University of California, Berkeley, US

*LigluR-mediated visual responses in the rd1 mouse model of retinitis pigmentosa*

13.00

Lunch

14.00 Half-day excursion to Girona  
20.00 Dinner

## Thursday, 26 November

**Chairs:** **Botond Roska**, Institute Friedrich Miescher, Basel, CH, **Stefan Heller**, Stanford University, US

09.00-09.45 **Spectrum of new therapies for retinal and cochlear diseases**  
**José-Alain Sahel**  
Institut de la Vision, Paris, FR  
*Neuroprotection in rod-cone dystrophies : from bench to bedside*

09.45-10.30 **Yvan Arsenijevic**  
Hôpital Jules-Gonin, Lausanne, CH  
*Gene therapy for retinal dystrophies*

10.30-11.15 **Botond Roska**  
Institute Friedrich Miescher, Basel, CH  
*Targeted expression of light sensitive pumps and channels restores visual function in animal models of Retinitis Pigmentosa*

11.15-11.45 Coffee break

11.45-12.45 **Serge Picaud**  
Institut de la Vision, Paris, FR  
*Electronic retinal implants*

**Robert V. Shannon**  
House Ear Institute, Los Angeles, US  
*Electrical stimulation of the human cochlea, auditory brainstem and midbrain*

12.45 Lunch

**Chairs:** **Yvan Arsenijevic**, Hôpital Jules-Gonin, Lausanne, CH; **Neil Segil**, House Ear Institute, Los Angeles, US

13.30-14.00 **Neil Segil**  
House Ear Institute, Los Angeles, US  
*Development and regeneration of the inner ear*

14.00-14.30 **Stephan Heller**  
Stanford University, US  
*In vitro generation of hair cells from embryonic and induced pluripotent stem cells*

14.30-15.00 **Erik C. Böttger**  
University of Zurich, CH  
*From gene to drug design: mitochondrial deafness alleles and aminoglycoside ototoxicity*

15.00-15.30 **Susanne Trauzettel-Klosinski**  
University of Tuebingen, DE  
*Rehabilitation: reading training in patients with Stargardt's disease - a randomized and controlled study*

15.30-16.00 Coffee break

16.00-16.30 **Christina Fasser**, *Retina Suisse - Retina International*,  
Zurich, CH / **Steffen Suchert**, *FAUN Foundation*,  
Nürnberg, DE  
*Patient expectations*

16.30-17.30 Forward Look Plenary Discussion

17.30

José-Alain Sahel

Institut de la Vision, Paris, FR

Concluding remarks

19.30

Get-together & Conference Dinner

## **Friday, 27 November**

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Breakfast & Departure

## **Abstracts, Posters & Short Oral Presentations**

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Posters will be displayed throughout the meeting. Posters can be fixed with self-adhesive tape, blue-tack or drawing pins onto double-sided poster panels. Recommended poster size is 100 cm high x 70 cm wide.

Short talks will take a maximum of 15 min each. It is suggested to employ most of the slot (~10 min) for the presentation and leave ~5 min for discussion.