



## RESEARCH CONFERENCES

ESF-UB Conference in Biomedicine

# Rare Diseases: Channels and Transporters

Hotel Eden Roc, Sant Feliu de Guixols (Costa Brava) • Spain

8-12 March 2008

Chair: **Manuel Palacín**, Institute for Research in Biomedicine and University of Barcelona, ES

Vice-Chair: **Thomas Jentsch**, Leibniz-Institut für Molekulare Pharmakologie and Max-Delbrück-Centrum für Molekulare Medizin, Berlin, DE

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Centro de Investigación Biomédica En Red  
de **Enfermedades Raras**



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# Final Programme

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## Saturday 8 March

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Late afternoon / early evening	Registration at the ESF-RC desk
19.30	Welcome Drink
20.00	Dinner

## Sunday 9 March

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08.30-08.45	Conference Opening - on behalf of the European Science Foundation <b>Isabel Varela-Nieto</b> Consejo Superior de Investigaciones Científicas, Madrid, ES
08.45-09.30	Special Talk <b>Bernard Rossier</b> Université de Lausanne, CH

## Session 1: SLC25 Mitochondrial Transporters

Chair: **Manuel Palacín**, Institute for Research in Biomedicine and University of Barcelona, ES

09.30-10.00	<b>Ferdinando Palmieri</b> Universita' di Bari, IT <i>Mitochondrial membrane transport proteins in health and disease</i>
10.00-10.30	Coffee break
10.30-11.00	<b>Edmund R.S. Kunji</b> Medical Research Council, Cambridge, UK <i>Substrate recognition and transport mechanism of mitochondrial carriers</i>
11.00-11.15	<b>Javier Traba</b> Universidad Autónoma de Madrid, ES <i>Transport activity and calcium regulation of the yeast ATP-Mg/Pi mitochondrial carrier</i>

## Session 2: Heteromeric Amino acid Transporters (HATs)

Chair: **Ferdinando Palmieri**, Universita' di Bari, IT

11.15-11.45	<b>Manuel Palacín</b> Institute for Research in Biomedicine and University of Barcelona, ES <i>Structure of HATs: 4F2hc ectodomain</i>
11.45-12.15	<b>Josep Chillarón</b> University of Barcelona, ES <i>Molecular basis of type I cystinuria</i>
12.30	Lunch

## Session 3: TRP Channels

Chair: **Bernd Nilius**, KU Leuven, BE

15.30-16.00	<b>Bernd Nilius</b> KU Leuven, BE <i>TRP channels: Entering the age of TRPpathies?</i>
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16.00-16.30	<b>Patrick Delmas</b> CNRS, Marseille, FR <i>TRPP2 (polycystin) and polycystic kidney disease</i>
16.30-17.00	Coffee Break
17.00-17.30	<b>Gideon Bach</b> Hadassah University Hospital, Jerusalem, IL <i>TRPML and lysosomal Function</i>
17.30-18.00	<b>Michelle P. Winn</b> Duke University Medical Center, Durham, US <i>TRPC6 and Familial Focal and Segmental Glomerulosclerosis</i>
18.00-18.15	<b>Miguel Valverde</b> Universitat Pompeu Fabra, Barcelona, ES <i>Cross-talk between TRPV4 channel and IP3 receptor signalling in ciliated epithelia</i>
18.15-18.30	<b>Silvia Vergarajauregui</b> NIH, National Heart, Lung and Blood Institute, Bethesda, US <i>Mucolipin-1 channel activity is regulated by protein kinase A mediated phosphorylation</i>
19.00	Dinner
20.30-22.00	Poster Session

## Monday 10 March

### Session 4: CLC Channels and Transporters

**Chair: Thomas Jentsch**, Leibniz-Institut für Molekulare Pharmakologie and Max-Delbrück-Centrum für Molekulare Medizin, Berlin, DE

09.00-09.30	<b>Raimund Dutzler</b> University of Zurich, CH <i>A structural perspective on CIC channel and transporter function</i>
09.30-10.00	<b>Thomas J. Jentsch</b> FMP (Leibniz-Institut fuer Molekulare Pharmakologie) and MDC (Max-Delbrueck-Centrum fuer Molekulare Medizin), Berlin, DE <i>Overview on physiology and pathology of CLC channels</i>
10.00-10.30	<b>Alessio Accardi</b> University of Iowa, Iowa City, US <i>Breaching the boundaries between ion channels and transporters</i>
10.30-11.00	Coffee Break
11.00-11.30	<b>Michael Pusch</b> Consiglio Nazionale delle Ricerche, Genova, IT <i>Mechanisms of transport and gating in eukaryotic CLC channels and transporters</i>
11.30-12.00	<b>Anselm A. Zdebik</b> UCL, London, UK <i>The role of CIC-K/barttin channels in transepithelial transport</i>
12.00-12.20	<b>Christoph Fahlke</b> Medizinische Hochschule Hannover, DE <i>Modulation of CIC-K channel function by barttin</i>
12.30	Lunch

## Session 5: SLC6 Neurotransmitter Transporters

Chair: Baruch Kanner, Hebrew University, Jerusalem, IL

- 15.30-16.00 **Paul Shaffer**  
Oregon Health and Science University, Portland, US  
*Structural Insights into Neuronal Transporters*
- 16.00-16.30 **Baruch Kanner**  
Hebrew University, Jerusalem, IL  
*Mechanism of chloride interaction with neurotransmitter: sodium symporters*
- 16.30-17.00 **Coffee Break**
- 17.00-17.30 **Gajja Salomons**  
VU University Medical Center, Amsterdam NL  
*X-linked creatine deficiency*
- 17.30-18.00 **Stefan Broer**  
The Australian National University, Canberra, AU  
*Can homology modelling enhance our understanding of neutral aminoacidurias?*
- 18.00-18.15 **Carmen Aragón**  
Universidad Autonoma de Madrid, ES  
*Dynamic properties of neuronal glycine transporter GLYT2*
- 18.15-18.30 **Francisco Zafra**  
Universidad Autonoma de Madrid, ES  
*Mechanisms of endoplasmic reticulum export of the glycine transporter GLYT1*
- 19.00 **Dinner**
- 20.30-22.00 **Poster Session**

## Tuesday 11 March

## Session 6: ABC Transporters

Chair: Kenny Linton, Imperial College, London, UK

- 09.00-09.30 **Kenny Linton**  
Imperial College, London, UK  
*ABC and 'Q' of drug efflux; a mechanistic view of ABC transporter function*
- 09.30-10.00 **Catherine Williamson**  
Imperial College, London, UK  
*The role of ABC transporters in the aetiology of cholestasis of pregnancy*
- 10.00-10.30 **Paola Vergani**  
University College London, UK  
*On CFTR and cystic fibrosis*
- 10.30-11.00 **Coffee Break**
- 11.00-11.30 **Joachim Koch**  
J.W.G. University of Frankfurt, DE  
*The ABC-transporter TAP in physiology and pathophysiology of MHC class I antigen presentation*
- 11.30-12.00 **John Oram**  
University of Washington, US  
*ABCA1: the link between the cardioprotective effects of cholesterol export and anti-inflammation*
- 12.00-12.45 **Business Meeting**  
*Identification of the focus and topics of the next conference. Election of chairs and selection of the organising committee.*
- 12.45 **Lunch**

## Session 7: Connexins

Chair: Klaus Willecke, Rheinische Friedrich-Wilhelms-Universitaet Bonn, DE

- 15.45-16.15      **Klaus Willecke**  
Rheinische Friedrich-Wilhelms-Universitaet Bonn, DE  
*Connexins and disease: overview and Mouse models*
- 16.15-16.45      **Nir Ben-Tal**  
Tel Aviv University, IL  
*Model structure of the connexin channel*
- 16.45-17.00      **Luis Barrio**  
Ramón y Cajal Hospital, Madrid, ES  
*Species specificity of mammalian connexin26 to form open voltage-gated hemichannels*
- 17.00-17.30      Coffee Break
- 17.30-18.00      **Fabio Mammano**  
Università di Padova, IT  
*Dual role of connexin in inner ear calcium signaling*
- 18.00-18.30      **Steven Scherer**  
University of Pennsylvania, Philadelphia, US  
*Why do Connexin Mutations cause Demyelinating Diseases?*
- 18.30-19.00      **David P. Kelsell**  
Queen Mary, University of London, UK  
*Connexins in skin diseases*
- 20.00              Get-together & Conference Dinner

### Wednesday 12 March

Breakfast & Departure

### Abstracts, Posters & Short Oral Presentations

All abstracts submitted are accepted for poster presentation.  
There will be no short talks other than those listed in the programme.