

# SSIEM newsletter

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## Secretary's Column

Mention quality control and most people remember that they have an urgent telephone call to make! But they should show more interest. Quality issues are increasingly relevant to us all. Mistakes are not only costly but tragic if avoidable. In the UK clinical governance is the phrase being used to describe the whole process of ensuring better standards of patient care by defining responsibility more clearly. See the DoH website ([doh.gov.uk](http://doh.gov.uk)) for more details. Better training of staff at all grades is part of this and initiatives being promoted by the SSIEM are going some way to improve things within Europe. Another very important initiative is improvement of laboratory practice through external quality control. This is where samples are analysed 'blind' and results returned to an organising centre to be returned as part of a performance report. The importance of this cannot be over emphasised. The only way that users of any service can have confidence is if there is some system of independent scrutiny or audit. Within Europe the initiative for QC of IEM laboratories has been taken by the ERNDIM organisation. The SSIEM is fully supportive of the ERNDIM initiatives. To find out more take a look at the ERNDIM pages on the SSIEM website.

Although there won't officially be an SSIEM meeting this year we hope to see as many members as possible at Brisbane. A hearty Australian welcome is guaranteed and supported by an excellent scientific programme. See you there,

Kind Regards,

*Mick Henderson*

Department of Clinical Biochemistry  
St James's University Hospital  
Leeds, UK

## ICIEM 2003

**Brisbane Australia. 2- 6th  
September**

If you haven't already done so, it is not too late to register for the IX International Congress on Inborn Errors of Metabolism. The meeting is

World sure to be one of the social highlights. There is a wide range of day tours available for accompanying persons so there is no excuse for leaving the family behind.

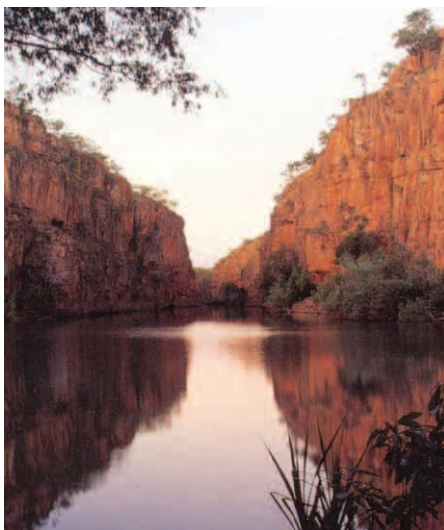
Discounted registration rates still apply until 26th August so get on



River bank beside the conference centre, Brisbane

shaping up very well with excellent keynotespeakers and a very good response to the call for abstracts. It also offers the chance to sample a little Australiana with a visit to Sea

the web and visit [www.iciem.org](http://www.iciem.org) to get all the information and book your place in the sunshine state where you can even do some "real" surfing!



Post Conference Tours - A Wide Variety on offer  
Katherine Gorge, Northern Territory



Satellite meetings in Sydney

## HUMAN GENETICS NETWORK IN LATIN AMERICA



After a great effort by a group of researchers, lead by Dr. Jose Maria Cantu (Mexico), RELAGH (Latin American Network of Human Genetics) was launched last April. The planning initially took place during the Human Genome Organization meeting held in Cancun. Prof. Roberto Giugliani (Brazil), who was elected President for the 2003-2005 term, is strongly inviting scientists and professionals who work in this field in Latin America to join the network.

RELAGH intends to be a powerful tool to increase cooperation among Latin American professionals and services involved in the vast field of Human Genetics (including genetic metabolic diseases).

More information on RELAGH and affiliation instructions are available at the website: [www.relagh.ufrgs.br](http://www.relagh.ufrgs.br)

### Vth Spanish Congress on Inborn Errors of Metabolism

**Madrid, 5-7th of November 2003**

Neurometabolic Diseases

President of the organizing committee: Prof. Magdalena Ugarte

More information is available from the website: <http://www.tilesa.es/ecm2003>

### IXth International Congress of Pediatric Laboratory Medicine Italian Society of Clinical Biochemistry

"bringing the best in pediatric laboratory medicine to the children of the world"

**September 13 - 16th, 2003**

For more information contact: Rosemary Carr, e-mail: [RCARR@cnmc.org](mailto:RCARR@cnmc.org).

### After the symposium, the book

#### PEROXISOMAL DISORDERS AND REGULATION OF GENES

Edited by Frank Roels, Myriam Baes and Sylvia De Bie

Fully updated chapters by Myriam Baes & Paul Van Velthoven, Johannes Berger & Sonja Forss-Petter, Maurice Bugaut, Annamaria Cimini, Marianne Depreter, Martine De Rycke, Osman El-Maarri, Lloyd Horrocks, Jutta G%oortner, Marisa Giros & Montse Ruiz, Johanna Grabarek & Magdalena Zernicka-Goetz, Guy Haegeman, Ann Heinzer & Kirby Smith, Axel Imhof, Jukka Kallijarvi, Werner Kovacs & Skaidrite Krisans, Norbert Latruffe, Hanna Mandel & Stanley Korman, Hugo Moser, Claus Rodemer & Wilhelm Just, Frank Roels, Christiane Van den Branden & Joseph Vamecq, Ronald Wanders, Anna Zomer, and many others. Special chapters on RNA interference, histone modifications and DNA methylation. Illustrated. Preface by Hans Galjaard.

Publication date: fall 2003. Special Prepublication Offer of Euro 85, GBP£53.50 valid until 30 September 2003. Orders to be sent with prepayment by sterling cheque (payable to Plenum Publishing Company) or credit card to: Joanne Duggan, Kluwer Academic/Plenum Publishers, 100 Borough High Street, London SE1 1LB, Email: [Joanne.Duggan@wkap.com](mailto:Joanne.Duggan@wkap.com)

## 41st Annual SSIEM Symposium, Amsterdam, 31st of August to the 3rd of September 2004



Dear colleagues,

On behalf of the SSIEM Council and the Local Organizing Committee I would like to invite you to attend the 41st Annual Meeting of the SSIEM. The topic of the meeting will be 'metabolic encephalopathies'

Following a decision by Council the SSIEM annual meeting officially starts now on Tuesday. This means that the ERNDIM meetings, the dieticians meeting and the mini-symposium become fully included in the program. The mini-symposium on Tuesday is devoted to 'Dominant expression of metabolic diseases'.

The main programme, which is rather clinical, has 4 plenary sessions dealing with leucoencephalopathies, movement disorders, congenital malformations of the brain, and acute cerebral presentation of inherited metabolic disorders.

A fifth plenary session will cover 'late breaking news' chosen from the submitted abstracts.

In a large number of parallel sessions with free communications selected from the submitted abstracts, new clinical, but mainly biochemical and molecular findings related to inherited metabolic diseases can be presented. We plan parallel sessions on fatty acid oxidation defects, cholesterol

biosynthesis defects, glycosylation defects, creatine deficiency syndromes, respiratory chain defects, peroxisomal and lysosomal disorders, disorders of neurotransmission and neuroreceptors, and disorders of homocysteine and B vitamin metabolism. However, the final choice of the topics of these parallel sessions will depend on the number and quality of the abstracts.

The space available for poster presentation is large and we will accommodate up to 300 posters.



Amsterdam

The Symposium venue is the main building of the Vrije Universiteit (VU) in the southwestern part of Amsterdam, close to Schiphol International Airport. There are good

railway, tram and bus connections between the VU, the hotels, central train stations and the airport.

Amsterdam, the Dutch capital, is known as 'the gateway to Europe' and through centuries has been characterized by its open mind and flexibility. Although Amsterdam is one of Europe's leading capitals, it still has the charm of a village. Amsterdam has a dazzling array of cultural attractions with something for everyone. There are 42 museums, including the Rijksmuseum, van Gogh Museum, the Maritime Museum, the Stedelijk (modern art) Museum, and the Anne Frank House. It has numerous theaters, cosy bars and inviting pubs, more than 800 outdoor terraces, and a wide variety of restaurants where you can eat at prices and levels of comfort to suit everyone. Moreover the Dutch are multilingual which makes getting around so much easier.

We look forward to welcoming you to Amsterdam and we hope that each delegate will find much of interest in the scientific programme, and much to enjoy in the social events we have planned, and in those you intend to discover yourself.

*Cornelis Jakobs*

Chairman Local Organizing Committee

## The Dutch Society for Inborn Errors of Metabolism (ESN)



The ESN was founded in 1988 by Prof Wadman and Dr van Gennip, but its origin actually was the 24-th SSIEM symposium in Amersfoort, where the idea to found the Dutch Society was born. The close relationship with the SSIEM has been ever since. Not only was a detail from the cover of the SSIEM abstractbook chosen to be the societies logo, one can only become a member of the Dutch society if one is a

paying member of the SSIEM.

Although the ESN started as a small society, over the years the number of members stays at around a 100. The members are clinicians and biochemists and there is an equal distribution between the two groups. During the last ten years colleagues from Belgium and Germany joined our society.

We have two annual meetings, one one-day meeting in spring and a two-day meeting in autumn. The meetings are usually focussed at special themes and host free-communications. The autumn meetings are organised by one of the Dutch or Belgium centers for inborn errors. Recently, we started with a educational day aimed at trainees and

specialists in general pediatrics and clinical chemistry to raise awareness for metabolic diseases. Furthermore, the ESN encourages young people interested in inborn errors to come to our meetings and be able to present research data or get in touch with the society.

In contrast with many other societies, you won't find the ESN at the net. If you want to contact us, please send us an e-mail.

*Bert Dorland* (l.dorland@wzk.azu.nl)

*Tom de Koning* (t.dekoning@wzk.azu.nl)

## Obituary

On May 23rd Prof. Paolo Durand, leading figure in Pediatrics and Metabolism, suddenly deceased.



Prof. Durand

His long career as a scientist is well known worldwide. We like to remember mainly two of his areas of outstanding achievement, the Fucosidosis and the lactose

intolerance. Prof. Paolo Durand was one of the founders of the SISECM, the first Italian Society for the Study of Inherited Metabolic Diseases. From which, in 1995, the SISMME evolved. His contribution, as Scientific Director to the development of the G. Gaslini Institute in Genoa and of the Bambino Gesù Hospital in Rome, was enormous. Also in the last years, after retirement, he has still been active in the fields of Metabolism and Genetics, a great promoter of the need to co-operate.

We like to remember, here however, mainly the man, his culture, the wisdom, the common

sense, the kindness, the smile. The fondness addressed to all of us pupils, co-workers, colleagues, and care and love he gave to his patients and their families.

We have lost a scientist and a teacher, but we will miss mainly the man and, for some of us, the friend.

*Roberto Cerone,*

President SISMME

*Carlo Dionisi-Vici,*

SSIEM Corresponding Member

*Ubaldo Caruso,*

Secretary SISMME and SSIEM Council Member.

## ETAC, Education and Training Advisory Committee

As previously reported the inaugural meeting of this Committee took place during last year's SSIEM symposium at Dublin. Essentially this committee will encourage and accredit training centers for Paediatric metabolic medicine within Europe. The first inspections are being planned for the Autumn. Prof James Leonard is leading this initiative and will write a more detailed progress report for the next Newsletter. In the meantime, documents describing the work of the committee and in particular the criteria to be met for successful accreditation will be posted on the SSIEM website.

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