

1st South American Workshop on Genomics and Community Genetics

Florianopolis, Brazil

Provisional Programme

Fri Sept 9

All day *Arrival and Registration of Workshop Fellows*

Sat Sept 10

All day *Arrival and Registration continues*

07.00 pm *Evening Reception* with Welcome Cocktails and snacks
followed by dinner at the Coral Plaza Hotel

Sun Sept 11

- 08.30 am *Welcome Introduction*
Professor Eliana Ternes-Periera, Florianopolis
- 08.45 am *The Workshop Development Programme, Objectives and Intentions*
Professor Alan Bittles, Perth, Australia
- 09.00 am *A welcome to Florianopolis*
The Rector of the Federal University of Santa Catarina
Professor Lucio Jose Botelho
- 09.15 am *Introduction to the Workshop, Course Structure and Orientation*
Professor Eliana Ternes-Periera, Florianopolis
- 09.45 am *Introduction to Community Genetics*
Professor Alan Bittles, Australia
- 10.15 am *Discussion*
- 10.20 am *Inborn errors of metabolism in Southern Brazil*
Dr Christina Netto, Porto Alegre, Brasil
- 11.00 am *Discussion*
- 11.05 am *Clinical Genetic Services and malformation syndromes in Santa Catarina, Brasil*
Professor Eliana Ternes-Periera, Florianopolis
- 11.45 am *Discussion*
- 11.55 am **Lunch**
- 02.00 pm *Student presentations* *Followed by discussion*
- 02.15 pm *Student presentations* ‘ ‘ ‘
- 02.30 pm *Student presentations* ‘ ‘ ‘
- 02.45 pm *Student presentations* ‘ ‘ ‘
- 03.00 pm *Student presentations* ‘ ‘ ‘
- 03.15 pm *Break for Refreshments*
- 03.30 pm *Student presentations* ‘ ‘ ‘
- 03.45 pm *Student presentations* ‘ ‘ ‘
- 04.00 pm *Student presentations* ‘ ‘ ‘
- 05.30 pm **Open Lecture**
Community Genetics Worldwide
Professor Alan Bittles, Perth, Australia
(venue to be confirmed)

Mon Sept 12

- 08.30 am *Epigenetics and human disease*
Dr Philip Avner, Paris, France
- 09.15 am *Discussion*
- 09.30 am *Inherited renal diseases*
Dr Anand Saggar, London, UK
- 10.15 am *Discussion*
- 10.30 am *Break for Refreshments*
- 11.00 am *Congenital and syndromic heart disease*
Dr Diana Baralle, Cambridge, UK
- 11.50 am *Student presentations* *Followed by discussion*
12.05 pm *Student presentations* ‘ ‘ ‘
12.20 pm *Student presentations* ‘ ‘ ‘
12.35 pm *Student presentations* ‘ ‘ ‘
- 01.00 pm **Lunch**
- 02.00 pm *Clinical cytogenetics*
Dr Juan Llerena, Rio de Janeiro, Brasil
- 02.40 pm *Discussion*
- 02.50 pm *Genetics of ageing*
Dr Bernard Lakowski, Paris, France
- 03.30 pm *Discussion*
- 03.40 pm *Break for Refreshments*
- 04.00 pm *Student presentations* *Followed by discussion*
04.15 pm *Student presentations* ‘ ‘ ‘
04.30 pm *Student presentations* ‘ ‘ ‘
04.45 pm *Student presentations* ‘ ‘ ‘
- 05.30 pm **Open Lecture**
Neurofibromatosis: Recent Advances in Understanding Splicing
Dr Diana Baralle, Cambridge, UK
(venue to be confirmed)

Tues Sept 13 Infectious Diseases Theme

- 08.30 am *Progress and Challenges in infectious diseases*
Dr. Olivier Neyrolles, Paris, France
- 09.10 am *Discussion*
- 09.20 am *Genetic susceptibility to infectious disease*
Dr Lluís Quintana-Murci, Paris, France
- 10.00 am *Discussion*
- 10.10 am *Pathogen recognition receptors: host microbial interface*
Dr. Olivier Neyrolles, Paris, France
- 10.40 am *Discussion*
- 10.50 am *Break for Refreshments*
- 11.15 am *Title to be announced*
Professor Edmundo C. Grisard, Florianopolis, Brasil
- 11.45 am *Discussion*
- 12.00 pm *Student presentations* *Followed by discussion*
12.15 pm *Student presentations* ‘ ‘ ‘
12.30 pm *Student presentations* ‘ ‘ ‘
12.45 pm *Student presentations* ‘ ‘ ‘
- 01.00 pm **Lunch**

AFTERNOON OUTING TO RAINFOREST OR BOAT RIDE

Wed Sept 14 Multifactorial Diseases and Introduction to Genetic Databases

08.30 am **Bioinformatics Center**

Mutation nomenclature and what makes a good mutation database
Dr. Ted Edkins, Perth, Australia

Linkage analysis, sib pair analysis/LOD scores and others
(to be confirmed)

10.30 am *Break for Refreshments*

10.45 am *Clinical Genetic databases on the internet*

(OMIM/ECURUCA/LMD/ECLAMC/others)

Dr. D. Baralle, Prof. E. Ternes-P, Dr. A. Saggarr, Prof. M. Patton,
Dr. K. McElreavey, Dr. L. Quintana-Murci and others

Cytogenetic databases

Professor Eliana Ternes-Periera, Florianopolis, Brasil

12.30 pm **LUNCH**

01.30 pm *Childhood cancer syndromes*

Dr. Diana Baralle, Cambridge, UK

02.00 pm *Discussion*

02.15 pm *Later onset cancer syndromes*

Dr. Anand Saggarr, London, UK

02.45 pm *Discussion*

03.00 pm *Genetics of diabetes*

Dr. Philip Avner, Paris, France

03.40 pm *Discussion*

03.50 pm *Break for Refreshments*

04.15 pm *Student presentations* *Followed by discussion*

04.30 pm *Student presentations* “ “ “

04.45 pm *Student presentations* “ “ “

05.30 pm **Open Lecture**

Using Genetics to Understand Population Origins and Migrations
Professor Lluís Quintana-Murci, Paris, France

(Venue to be confirmed)

Thurs Sept 15 Free Day

Frid Sept 16 **Bioinformatics Centre**

09.00 am *All day course on Bioinformatics and Population Genetics*

05.30 pm **Open Lecture**
Prospective for Commercial Biotechnology in Brazil
Dr Carlos Moriera-Filho, Rio de Janeiro, Brasil
(venue to be confirmed)

Sat Sept 17

08.30 am *Model organisms to study human disease*
Dr. Bernard Lakowski, Paris, France

09.10 am *Discussion*

09.20 am *Understanding human disease through comparative genomics*
Professor Luismar Marques Porto, Florianopolis, Brasil

10.00 am *Discussion*

10.10 am *Break for Refreshments, with sandwiches/empanadas as late lunch*

10.45 am *Brazilian Cancer Genome Project*
Dr. Carlos Moriera-Filho, Rio de Janeiro, Brasil

11.15 am *Genetic services in Cuba –An integrated approach*
Dr. Lidia Ines Novoa-Perez, Havana, Cuba

12.00 pm **Close for afternoon barbeque**
(Coaches depart direct from the University)

Sun Sept 18 Genetics of Complex Traits

- 08.30 am *Genetics of sport*
Professor Michael Patton, London, UK
- 09.15 am *Discussion*
- 09.30 am *Cross cultural and ethical issues*
Dr. Anand Saggarr, London, UK
- 10.00 am *Discussion*
- 10.15 am *Cancer genes related to chromosomal abnormalities*
Dr Jose Carlos Cabral de Almedia, São Paulo, Brasil
- 10.45 am *Discussion*
- 11.00 am *Syndromes with deafness*
Professor Michael Patton, London, UK
- 11.30 am *Discussion*
- 11.40 am *Break for Refreshments*
- 11.45 am *Student presentations* *Followed by discussion*
- 12.00 pm *Student presentations* ‘ ‘ ‘
- 12.15 pm *Student presentations* ‘ ‘ ‘
- 12.30 pm *Student presentations* ‘ ‘ ‘
- 12.30 pm **Lunch**
- 02.40 pm *Errors in human gonadal development*
Dr. Ken McElreavey, Paris, France
- 03.10 pm *Discussion*
- 03.20 pm *Genetics of human infertility*
Dr. Ken McElreavey, Paris, France
- 03.50 pm *Discussion*
- 04.00 pm *Student presentations* *Followed by discussion*
- 04.15 pm *Student presentations* ‘ ‘ ‘
- 04.30 pm *Student presentations* ‘ ‘ ‘
- 04.45 pm *Student presentations* ‘ ‘ ‘

Mon Sept 19

09.00 am *Genetic mutation screening: An update*
Dr. Ted Edkins, Perth, Australia

09.40 am *Discussion*

10.00 am *Nimblegen*

11.00 am *ABI*

12.30 pm **Lunch**

02.00 pm *Practical demonstrations/tutorials*

05.30 pm **Open Lecture**
Huntington's Disease, a Paradigm for Neurogenetics
Professor Michael Patton, London, UK
(venue to be confirmed)

Tues Sept 20

- 08.30 am *Genetics of murder*
Professor Michael Patton, London, UK
- 09.00 am *Lecture to be confirmed*
Professor Alan Bittles, Perth, Australia
- 09.30 am *Case presentations*
Professor Michael Patton, London, UK
- 10.30 am *Distribution of Workshop Questionnaires*
- 11.30 am *Summing up*
- 12.00 pm **Close of Workshop**