The 5th Florence-Utah-Symposium on the Genetics of Male Infertility was held September 19-21 in Florence. The Program Committee included Drs. Douglas Carrell, Csilla Krausz, Gianni Forti and Paolo Sassone-Corsi.

The symposium has been attended by a total of 147 participants from 35 different countries working in the fields of Andrology, Genetics and Urology, representing basic science research as well as clinical work.

The main sessions included: Clinical genetics, Genetics of Aging Male gamete, Epigenetics and Proteomics, Pharmacogenetics and Evolutionary aspects of the sex chromosomes (see detailed program below).

A special focus has been given to the new EAA/EMQN Y chromosome-microdeletion guidelines, which were intensively discussed during a large round table session (the Guidelines are "Open access" published in “Andrology”, Krausz et al, 2013).

An entire selected oral presentation session has been devoted to the "Reprotrain” Marie Curie Network and data on epigenetic aspects and meiotic regulation of male gamete have been presented by young fellows of the Network (http://reprotrain.eu/).

Both the oral and the poster sessions were very vivacious allowing to exchange ideas and discuss relevant topics for the future of male infertility and the role of genetics/epigenetics in this area of medicine.

The three ISA award winners presented their data during the Poster sessions. The winners were as follows: Evguenia Alechine from Argentina, Paulo Navarro-Costa from Portugal and Katerina Popovska-Jankovic from Macedonia (see photo enclosed at the end of the report). E. Alechine presented data on “Native American haplogroup Q-M3 might protect against AZFc microdeletions”; P. Navarro-Costa presented a poster on “The Drosophila melanogaster homolog of the AZFb gene KDM5D is required for female meiotic
progression” and K. Popovska-Jankovic discussed her data on “Microarray microRNA profiling in patients with male infertility”.

The next meeting should be held in Salt lake City, USA in the fall of 2014.

Dr. Csilla Krausz
Co-Organizer
Florence-Utah Symposium on the Genetics of Male Infertility

Drs Carrell and Krausz with the three ISA awardee: Evguenia Alechine from Argentina, Paulo Navarro-Costa from Portugal and Katerina Popovska-Jankovic from Macedonia.

The Programme

19th September 2013

Registration from 14:00 to 16:45
17:00-20:00 **Opening Ceremony** at City Hall (Palazzo Vecchio)

**20th September 2013**

**Clinical Genetics I**  
Chairpersons **D. Carrell** (USA) and **G. Forti** (Italy)  
9:00-9:30 Whole-genome studies: from arrays to next generation sequencing  
**D. Conrad** (USA)  
9:30-10:00 Genetic susceptibility to male infertility: news from genome wide association studies  
**K. Aston** (USA)  
10:00-10:30 Sex chromosome-linked Copy Number Variants  
**C. Krausz** (Italy)  
10:30-10:50 **Coffee Break**

**Genetics of the Ageing Male Gamete**  
Chairpersons: **B. Robair** (Canada) and **E. Nieschlag** (Germany)  
10:50-11:20 The ageing male germ cell: causes and consequences for human diseases  
**A. Wilkie** (UK)  
11:20-11:50 Telomere length of the human sperm: the bright side of aging  
**A. Aviv** (USA)  
11:50-12:20 Germ Cells: Sensing Nutritional and Circadian Pathways  
**P. Sassone-Corsi** (USA)

**Selected Oral Presentations I**  
Chairpersons: **S. Rousseaux** (France) and **S. Lewis** (UK)  
12:20-12:50 1) Newborns of obese fathers have lower methylation levels at imprinted genes  
**A. Soubry** (Belgium)  
2) Sperm DNA oxidative damage targets histone-rich and nuclear matrix-attached domains.  
**J. Drevet** (France)  
12:50-14:20 **Poster tour + lunch**
Clinical Genetics II and Pharmacogenetics
Chairpersons: M. Maggi (Italy) and R. Oliva (Spain)

14:20-14:50 Genetic aspects of Central Hypogonadism: the missing genotype-phenotype correlation
N. Pitteloud (Switzerland)
14:50-15:20 Defining essential proteins in infertility and for contraception
M. Matzuk (USA)
15:20-15:50 Pharmacogenetics in male infertility: a personalized treatment with FSH?
A. Ferlin (Italy)
15:50-16:20 Disorders of Sex Development – gene discovery and diagnosis using massively parallel sequencing
A. Sinclair (Australia)

16.20-16:40 Coffee Break

Selected Oral Presentations II
Chairpersons: B. Hales (Canada) and P. Piomboni (Italy)

16:40-18:00 1) Loss of H4K12ac interaction and differential DNA methylation pattern within NSD1 promoter in sperm of subfertile patients
A. Paradowska-Dogan (Germany)
2) A paternal influence on the embryonic capacity for implantation observed in a surrogate motherhood program
I. Giakoumakis (Greece)
M.J. Mitchell (France)
4) R31C GNRH1 mutation causes an autosomal dominant form of normosmic congenital hypogonadotropic hypogonadism
Luigi Maione (Italy)
5) Proteomic and genomic distribution of human sperm chromatin
J. Castillo (Spain)

Marie Curie Network: Reprotrain Session
Introduced by Rafael Oliva (Spain)

18:00-19:00 1) Ptl-1, a newly identified chromatin component in Drosophila melanogaster
Z. Eren (Germany)
2) Role of Nut during spermatogenesis
H. Shiota (France)
3) Role of the histone acetyl transferases CBP and P300 during post-meiotic re-organization of the male genome
A. Goudarzi (France)
4) Analysis of meiotic checkpoint activation in azoospermic men
W.M Baarends (The Neetherlands)
21st September 2013

**EAA/EMQN Guidelines for the screening of the Y chromosome microdeletions:**

8:00-8:30 The updated guidelines for AZF deletion screening for AZF deletion screening  
**F. Tüttelmann** (Germany)  
8:30-10:15 Interactive discussion: practical aspects of the routine testing  
**Discussants:** C. Krausz (Italy), S. Kleiman (Israel), F. Tüttelmann (Germany), L.H. Hoefsloot (The Netherlands), H. Skaletsky (USA)

**Selected Oral Presentations III**  
Chairpersons: R. Renkawitz-Pohl (Germany) and J. Gromoll (Germany)

10:15-10:45 1) The mature sperm of *Drosophila melanogaster*: retain nucleosomes that may be involved in the subsequent maintenance of paternal epigenetic marks within genes essential for embryogenesis  
**A.H. Elnefati** (Lybia)  
2) A genome-wide mutation study of familial azoospermia using whole exome sequencing  
**A. Yatsenko** (USA)

10:45-11:00 **Coffee break**

**Selected Oral Presentations IV**  
Chairperson: Navarro-Costa (Portugal) and K. Stouffs (Belgium)

11:00-11:30 1) Overexpression of Full-Length Centrobin Rescues Limb Malformation but not Male Fertility of the Hypodactylous (*hd*) Rats  
**F. Liška** (Czech Republik)  
2) Gene networks during the first wave of spermatogenesis  
**A. Sironen** (Finnland)

**Epigenetics Aspects Of The Male Gamete:**  
Chairpersons: D. Miller (UK) and J. Drevet (France)

11:30-12:00 Chromatin and Transcription Landscapes of the Adult Mouse Germline Reveal Genomic Strategies for Spermatogonial Stem Cells and Gametogenesis  
**B. Cairns** (USA)  
12: 00-12:30 Human Sperm Epigenomes and Transcriptomes Reveal Novel Features of Enhancers, Sex Chromosomes, piRNAs, Gametogenesis, and Inherited Small RNAs  
**S. Hammoud** (USA)
Evolution and Single cell genomes
Chairpersons: W. Baarends (The Neetherlands) and M. Mitchell (France)

12:30-13:00 The dual nature of mammalian sex chromosomes (H. Skaletsky, USA)
13:00-13:30 Single-cell genomics to study DNA-mutation, genetic heterogeneity and disease: current status and beyond (T. Voet, Belgium)

13:30-13:45 Closing remarks
13:45-15:00 Farewell Lunch