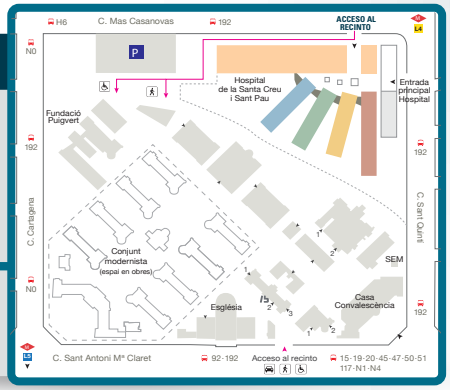
XI JORNADA de ACTUALIZACIÓN en GENÉTICA HUMANA de la Asociación Española de Genética Humana

Barcelona, 4 de abril de 2014

Hospital de la Santa Creu i Sant Pau

SEDE

Salón de Actos del Hospital de la Santa Creu i Sant Pau
C/ Mas Casanova 90
08041 Barcelona



INSCRIPCIONES

Socios AEGH	**80 €**
No socios	**100 €**
Residentes y Estudiantes	**80 €**
EN SEDE	**150€**

La cuota de inscripción incluye la asistencia a la Jornada, documentación, cafés y comida de trabajo. 21% IVA incluido.
Las inscripciones pueden formalizarse online a través de la web: **www.aegh.org**

ALOJAMIENTO

Hotel Amrey Sant Pau **

Doble	**90 €**	Doble Uso Individual	**90 €**

Precios por noche, incluyen alojamiento, desayuno e IVA. Tasa turística no incluida

CANCELACIONES

Con posterioridad al 1 de Marzo de 2014 no se aceptará ningún cambio o anulación en las inscripciones y/o reservas hoteleras efectuadas. Cualquier anulación hecha con anterioridad a esta fecha tendrá unos gastos de gestión del 50%. Todas las cancelaciones deberán ser remitidas a la Secretaría Técnica por escrito. El reembolso de los servicios anulados se efectuará a partir del 1 de mayo de 2014.

COLABORAN

affymetrix Biology for a better world · Durviz · illumina · imegen · IZASA · Roche · Promega · sistemas genómicos

SECRETARÍA TÉCNICA

GEYSECO.es GESTIÓN DE CONGRESOS Y SOCIEDADES
c/ Marina, 27 - 08005 Barcelona / TEL 932212242
FAX 932217005 www.geyseco.es / barcelona@geyseco.es

Comités

Junta AEGH

Juan Cruz Cigudosa
Victor Volpini
Jordi Perez Tur
Contxi Lazaro
Cristina González González
Marta Rodriguez de Alba
Javier García Planells
Guiomar Perez

Comité Organizador
Servicio de Genetica
Hospital de Sant Pau
U-705 CIBERER

Montserrat Baiget
Pia Gallano
Adriana Lasa
Eduardo Tizzano

Presentación

Los avances tecnológicos que se han producido en los últimos años en el campo de la genética humana constituyen un desafío para los genetistas y un motivo de esperanza para los pacientes. Esta Jornada reunirá expertos nacionales e internacionales para debatir la compleja interpretación de la información genómica originada por dichos avances y su aproximación al diagnóstico genético.

Programa

4 de abril de 2014

09:40 **Bienvenida**
Dr. Albert Salazar
Gerente del Hospital de Sant Pau. Barcelona

09:45-10:00 **Inauguración**
Juan Cruz Cigudosa
Presidente de la AEGH

10:00-12:00 **1ª Mesa Redonda: THE GENOMIC FUTURE OF MEDICINE**
Moderador: Eduardo Tizzano
Servicio de Genética, Hospital Sant Pau. Barcelona

10:00 How Genomics change the practice of Medicine
Han Brunner
Human Genetics,Radboud University Medical Center. Nijmegen, The Netherlands

10:30 Personal Genomes
Marjolein Kriek
Center for Human and Clinical Genetics, Department of Clinical Genetics, Leiden University Medical Center. Leiden, The Netherlands

11:00 Long distance regulatory elements
Stanislas Lyonnet
Institut Imagine, Université Paris Descartes, Hôpital Necker-Enfants Malades. París, France

11:30 Discusión

12:00-12:30 Pausa-Café

12:30-13:30 **Asamblea General de la AEGH** (solo socios AEGH)

13:30-14:15 **Premio al Joven Investigador**

14:15-15:00 Comida

15:00-17:00 **2ª Mesa Redonda: ACTUALIZACIÓN DE LOS GRANDES PROYECTOS GENOMICOS**
Moderador: Adriana Lasa
Servicio de Genética, Hospital Sant Pau. Barcelona

15:00 Human Variome Project
Ángel Carracedo
Fundación de Medicina Xenómica-SERGAS-CIBERER, Universidad de Santiago. Santiago Compostela

15:30 Proyecto "1000 genomes"
Marc Via
Departamento de Psiquiatría y Psicobiología Clínica, Universitat de Barcelona. Barcelona

16:00 Proyecto "Encode"
Roderic Guigó
Coordinador del Programa de Bioinformática y Genómica Centro de Regulación Genómica (CRG), Barcelona

16:30 Discusión

17:00 **Clausura de la Jornada**
Montserrat Baiget
Servicio de Genética, Hospital Sant Pau. Barcelona

How Genomics change the practice of Medicine

Han Brunner
Human Genetics,Radboud University Medical Center. Nijmegen, The Netherlands

Clinical exome sequencing is rapidly becoming indispensible in the analysis of patients with intellectual disability and autism. We have recently used exome sequencing of 100 patients and genome sequencing of a subset of 50 patients with intellectual disability and find that in the majority of these patients, causative de novo mutations can be detected. A number of new ID genes have been identified, some of which have recognizable phenotypes. Some patients had very small copy number variants that were reliably detected by whole genome sequencing. In others, CNVs were detectable by exome sequencing. These observations suggest that sequencing may replace arrays for the detection of structural chromosome variation.

Personal Genomes

Marjolein Kriek
Center for Human and Clinical Genetics, Department of Clinical Genetics, Leiden University Medical Center. Leiden, The Netherlands

We stand today on the threshold of a new era in healthcare. Within the foreseeable future analysis of a complete patients' genome will play a prominent role in the consulting rooms of almost every medical specialist. The major hurdle, however, when implementing full genome analysis is to correctly interpret this complex data and to apply this knowledge effectively in a clinical setting.
Since 2008 onwards, the year in which it was announced that my full genome was sequenced there have been many opportunities for educational exchange with the general public, students, and medical specialists. From these contacts it became clear that while on the one hand there was much curiosity as to the possibilities of the new genetic technologies, on the other hand the general public was uncertain about the applications of these techniques. A general feeling was that genetic techniques impinge upon personal identity, and both ethical dilemmas and psychosocial consequences present significant challenges. During my talk these challenges will be discussed as well as the hurdles and the results of the data analysis of my genome over time.

Long distance regulatory elements

Stanislas Lyonnet
Institut Imagine, Université Paris Descartes, Hôpital Necker-Enfants Malades. París, France

One of the key discoveries of vertebrate genome sequencing was the unexpected amount of evolutionarily conserved non-coding DNA (CNCs). Interestingly, enrichment for CNCs has been demonstrated within gene deserts, close to genes known to be developmental regulators, and likely necessary for appropriate spatiotemporal gene expression during development. In that context, we will discuss a number of genomic alterations suggesting that the non-coding DNA domain to study in malformation syndromes should be much broader than traditionally investigated.

Proyecto "Encode"

Roderic Guigó
Coordinador del Programa de Bioinformática y Genómica Centro de Regulación Genómica (CRG), Barcelona

RNA HETEROGENEITY IN THE EUKARYOTIC CELL: The unfolding of the instructions encoded in the genome is triggered by the transcription of DNA into RNA, and the subsequent processing of the resulting primary RNA transcripts into functional mature RNAs. RNA is thus the first phenotype of the genome, mediating all other phenotypic changes at the organism level caused by changes in the DNA sequence. While current technology is too primitive to provide accurate measurements of the RNA content of the cell, the recent development of Massively Parallel Sequencing Instruments has dramatically increased the resolution with which we can monitor cellular RNA. Using these instruments, the ENCODE project has surveyed the RNA content of multiple cell lines and subcellular compartments. The results of these surveys underscore pervasive transription, as well as great RNA heterogeneity between and within cells. Comparison of RNA surveys with other genome wide epigenetic surveys—such as those of binding sites for Transcription Factors, or of Histone modifications—reveals a very tightly coupling between the different pathways involved in RNA processing, transcription and splicing in particular.

Proyecto "1000 Genomes"

Marc Via
Departamento de Psiquiatría y Psicobiología Clínica, Universitat de Barcelona

Entre los proyectos dirigidos a identificar variabilidad en nuestro genoma, el proyecto 1000 Genomes destaca por la secuenciación del genoma completo de más de 2500 individuos de 26 poblaciones mundiales. Los resultados publicados hasta el momento han duplicado el número de polimorfismos conocidos (SNPs, indels y variantes estructurales). Además, la secuenciación de exomas completos a alta cobertura en estas muestras está evidenciando la elevada carga de variantes raras con implicaciones funcionales de las que somos portadores. Las aplicaciones en biología evolutiva y en genómica médica de este conocimiento son considerables.