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Universidad de Buenos Aires
Facultad de Ciencias Exactas y Naturales
Departamento de Química Biológica

## DEPARTAMENTO DE QUÍMICA BIOLÓGICA

## CURSO DE POSTGRADO O SEMINARIO

## AÑO: 2008

1) NOMBRE DEL CURSO/SEMINARIO: Diagnóstico Molecular en Medicina. Su Aplicación a Enfermedades Genéticas, Tumorales e Infecciosas.

2) NOMBRE Y APELLIDO DEL RESPONSABLE: Dra. María Victoria Rossetti, Dra. Victoria Estela Parera

3) DOCENTES QUE COLABORAN EN EL DICTADO DEL CURSO: Dras Ana María Buzaleh, Esther Gerez y Jimena Lavandera, Lic. Federico Colombo y la participación de destacados especialistas.

4) FECHA DE INICIACIÓN: 2/12 FECHA DE FINALIZACION: 19/12

5) CANTIDAD DE HORAS TOTALES DE DICTADO: 90
   a) TEORICAS: 35 horas
   b) LABORATORIO: 55 horas

6) FORMA DE EVALUACIÓN: Examen final escrito

7) LUGAR DE DICTADO: Departamento de Química Biológica, FCEN, UBA

8) PUNTAJE QUE OTORGA PARA EL DOCTORADO: 3

9) Nº DE ALUMNOS: Mínimo: - Máximo: 10

10) ARANCEL PROPUESTO: 300 módulos: 100 teóricos y 200 prácticos

11) PROGRAMA ANALÍTICO Y BIBLIOGRAFÍA DEL CURSO

PROGRAMA

- Técnicas básicas en Biología Molecular: PCR, Secuenciación, PCR en tiempo real, PCR inversa, PCR larga distancia, Microarreglos.

- Expresión de proteínas. Mutagénesis. Minigenes.

- Bases moleculares de diversas patologías: cáncer, fibrosis quística, retinoblastoma, porfirias, hemocromatosis, talasemia, distrofia muscular de Ducheme, hemofilia.

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- Clínica, bioquímica, diagnóstico molecular de HIV y virus de hepatitis. Sida: situación actual.

- Isoenzimas del citocromo P450. Polimorfismos y su implicancia clínica

BIBLIOGRAFIA

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- High prevalence of the H63D HFE mutation in italian patients with Porphyria Cutanea Tarda. Sampietro M, Piperno A, Lupica L, Arosio C, Vergani A, Corbetta N, et al. Hepatology 1998; **27**: 181-4.

- Familial Porphyria Cutanea Tarda: Characterization of seven novel Uroporphyrinogen Decarboxylase gene mutations and Frequency of Common Hemochromatosis Alleles. Manuel Méndez; Lonnie Sorkin; María Victoria Rossetti; Kenneth H. Astrin; Alcira M.del Carmen Batlle;

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Victoria E. Parera; Gerardo Aizencang & Robert J. Desnick. American J Human Genetics 1998; **3:** 1365-1375

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- Identification and characterization of Hydroxymethylbilane Synthase mutations causing Acute Intermittent Porphyria: Evidence for an ancestral founder of the common G111R mutation De Siervi A.; M.V. Rossetti; V.E. Parera; K.H. Astrin; G.I. Aizencang; I.A. Glass; R.J.; A.M. del C. Batlle & R.J. Desnick. Am. J Med. Genet. 1999; **86:** 366-375

- Variegate Porphyria in Western Europe: identification of PPOX gene mutations in 104 families, extent of allelic heterogeneity, and absence of correlation between phenotype and type of mutation. Whatley SD, Puy H, Morgan, RR, Robreau AM, Roberts AG, Nordman Y, Elder GH, Deybach, JC. Am J Hum Genet 1999, **65**: 984-994

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- Co-inheritance of mutations in the uroporphyrinogen decarboxylase and hemochromatosis genes accelerates the onset of porphyria cutanea tarda. Brady J, Jackson H, Roberts A, Morgan R, Whatley S, Rowlands G, et al. J Invest Dermatol 2000; **115**: 868-74.

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- Modulation of penetrance by the wild–type allele in dominantly inherited erythropoietic protoporphyria and acute hepatic porphyrias. Gouya L, Puy H, Robreau AM, Lyoumi S, Lamoril J, Da Silva V, Grandchamp B, Deybach JC. Hum Genet 2004; **114**: 256-262

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- Erythropoietic porphyrias: animal models and update in gene-based therapies. Richard E, Robert-Richard E, Ged C, Moreau-Gaudry F, de Verneuil H. Curr Gene Ther. 2008 **8**:176-86

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- Anticancer therapy in patients with porphyrias: evidence today. Thiery-Vuillemin A, Chaigneau L, Meaux-Ruault N, Villanueva C, N'guyen T, Maurina T, Stein U, Lorgis V, Demarchi M, Pivot X. Expert Opin Drug Saf. 2008 **7**:159-65.

- Actualización a cargo de los docentes del curso y especialistas invitados.

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**VºBº Del Departamento**

ELIDA A. CANDURRA
DIRECTORA ADJUNTA
Dpto. QUÍMICA BIOLÓGICA
F.C.E. y N. - UBA

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**Firma del Responsable**
MARIA V. ROSSETTI

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**VºBº de la Subcomisión de Doctorado**

Universidad de Buenos Aires
Facultad de Ciencias Exactas y Naturales

Referencia Expte. Nº 480.460/04

Buenos Aires, 25 AGO 2008

**VISTO:**

la nota 16/07/2008 presentada por la Dra. Nelida Candurra, Directora Adjunta del Departamento de Química Biológica, mediante la cual eleva la Información y el Programa del Curso de Postgrado **"Diagnóstico Molecular en Medicina. Su Aplicación a Enfermedades Genéticas, Tumorales e Infecciosas"**, que será dictado durante el segundo cuatrimestre de 2008 (desde 02/12/2007 hasta 19/12/2007), por la Dra. María Victoria Rossetti y la Dra. Victoria Estela Parera con la colaboración de la Dra. Ana Maria Buzaleh, Esther Gerez, Jimena Lavandera, Federico Colombo y otros especialistas.

**CONSIDERANDO:**

Lo actuado por la Comisión de Doctorado 23/07/2008,

lo actuado por la Comisión de Enseñanza, Programas, Planes de Estudio y Posgrado

lo actuado por la Comisión de Presupuesto y Administración,

lo actuado por este cuerpo en Sesión Ordinaria realizada en el día de la fecha,

en uso de las atribuciones que le confiere el Artículo Nº 113º del Estatuto Universitario,

**EL CONSEJO DIRECTIVO DE LA FACULTAD DE CIENCIAS EXACTAS Y NATURALES**
**R E S U E L V E:**

**Artículo 1º:** Autorizar el Dictado del Curso de Postgrado **"Diagnóstico Molecular en Medicina. Su Aplicación a Enfermedades Genéticas, Tumorales e Infecciosas"**, de 90 hs. de duración.

**Artículo 2º:** Aprobar el Programa del Curso de Postgrado **"Diagnóstico Molecular en Medicina. Su Aplicación a Enfermedades Genéticas, Tumorales e Infecciosas".**

**Artículo 3°:** Aprobar un Puntaje de 3 (tres) puntos para la Carrera del Doctorado.

**Artículo 4º:** Aprobar un Arancel de 300 Módulos para los alumnos que cursen la totalidad del curso teórico-práctico; aprobar un arancel de 200 Módulos para los alumnos que cursen los Trabajos Prácticos y aprobar un arancel de 100 Módulos para los alumnos que cursen unicamente las clases teóricas. Disponer que los montos recaudados serán utilizados conforme a lo dispuesto por la Resolución CD Nº 072/03.

**Artículo 5º:** Comuníquese a la Dirección del Departamento de Química Biológica, a la Biblioteca de la FCEyN y a la Subsecretaría de Postgrado (con fotocopia del Programa incluida). Cumplido, archívese.

Resolución CD Nº 1958

SP/med / 01/08/2008

Dra. MATILDE RUSTICUCCI
SECRETARIA ACADEMICA ADJUNTA

Dr. JORGE ALIAGA
DECANO