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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: 2006

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, Susana Afonso y Esther Gerez y la participación de destacados especialistas.**

4) FECHA DE INICIACIÓN: **20/11** FECHA DE FINALIZACION: **7/12**

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 Duchenne, 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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- A Batlle (1997). Porfirias y Porfirinas. Aspectos clínicos, bioquímicos y biología molecular. Acta Bioquímica Clínica Latinoamericana, Supl. 3.

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

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Sampietro M, Fiorelli G, Fargion S
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- The physiological and pharmacological roles of cytochrome P450 isoenzymes.
Chang, G.W.M. & Kam, P.C.A.
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- Cytochrome P450 enzyme system: genetic polymophisms and impact on clinical pharmacology
van der Weide, J. & Steijns, L.S.W.
Ann. Clin. Biochem. 1999; **36:** 722-729

- 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.
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- 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
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- HFE mutations analysis in 711 Hemochromatosis probands: Evidence for S65C implication in mild form of Hemochromatosis.
Mura C, Raguenes O, Ferec C.
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Gouya L, Puy H, Lamoril J, Da Silva V, Grandchamp B, Nordmann Y, Deybach JC.
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Brady J, Jackson H, Roberts A, Morgan R, Whatley S, Rowlands G, et al
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- Homozygous variegate Porphyria: 20 y follow-up and characterization of molecular defect.
Kauppinen R, Timonen K, von und zu Fraunberg M, Laitinene E, Ahola H, Tenhunen R, Taketani S, Mustajoki P.
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K. Mehrany, L. Drage, D. Brandhagen & M. Pittelkow.

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- Autosomal recessive erythropoietic protoporphyria in the United Kingdom: prevalence and relationship to liver disease.
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-Actualización a cargo de los docentes del curso y especialistas invitados.

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V°B° Del Departamento

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Firma del Responsable

MU - ROSSETTI

VICTORIA PARERA

.....
V°B° de la Subcomisión de Doctorado

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