



## **Guidelines**

# **Molecular Genetics of Blood Disorders**

## **A. Facility Description**

The Molecular Genetics of Blood Disorders Unit was created in 1996 to support basic and clinical research on genetics.

The goal of the Molecular genetics of Blood Disorders Unit is to establish a shared facility for basic and clinical research on genetics markers of human diseases at the University of Puerto Rico Medical Science Campus. The Unit will provide research and technical support of human genetics projects in the areas of molecular biology and genetics. Several basic and clinical science faculties with overlapping research interest are collaborating in this endeavor.

## **B. Physical Location**

The Unit is located in the department of Biochemistry on the 6<sup>th</sup> floor of the main Medical Science Campus building, and occupies an area of approximately 1,400 square feet. One of the laboratory areas contains all the necessary instrumentation to perform molecular genetics methodologies, such analysis of point mutations, DNA sequencing, PCR analysis, etc. , and another is a Tissue culture and cold room facilities with room numbers A-639 and A-640 respectively.

**Our postal address is as follows:**

Molecular Genetics of Blood Disorders Core Facility  
Department of Biochemistry room A-639  
P.O. Box 365067  
San Juan, Puerto Rico 00936-5067

## **C. Staff and Contact information**

Carmen L. Cadilla-Vázquez, Ph.D  
Molecular Genetics of Blood disorders Core Facility Coordinator  
Office A-640 6<sup>th</sup> floor Dept. of Biochemistry  
Tel: 1-787-758-2525 Ext 1638  
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Molecular Genetics of Blood disorders Core Facility Technician  
Office A-640 6<sup>th</sup> floor Dept. of Biochemistry  
Tel: 1-787-758-2525 Ext 1638 or 1372  
E-mail: [Jestable@hotmail.com](mailto:Jestable@hotmail.com)

#### D. Service hours

The Molecular genetics of blood disorders Core facility is available to users from Monday to Friday from 7:30 a.m. to 5:00p.m.. In special cases like holidays and closing hours special agreements are often made for users to access the lab facilities after service hours.

#### E. Services provided

<b>Instrumentation</b>	<b>Function and Use</b>
2 Nuair biosafety cabinet hood	To prepare sterile samples and procedures like PCR, and cells handling
UV-transluminator	For agarose and acrylamide gels
Mettler AT analytical balance	For microweights reagents
2 MJ research thermal cyclers	PCR reactions and incubation periods for other procedures
Ultrarefrigerator Revco	Stocks -80 degrees samples
Beckman avanti centrifuge	
2 microcentrifuge lite 5420	For microtubes centrifugation
Dcode system Bio Rad	Equipment for point mutations detection like, SSCP, DGGE techniques
I Cycler Bio Rad	PCR reactions (with temperature gradient feature)
2 PE 2700 Thermal cyclers	For PCR reactions
Beckman spectrophotometer	DNA, RNA and proteins concentrations
Fluorometer	For DNA concentrations more sensitive instrumentation
ABI 377 Sequencer	Automated DNA sequencing (with previous agreements)

**The most important service is provided by Dr. Carmen Cadilla on Genetics and molecular biology advising, and technical support by Jessica Renta.**

## **F. Operation Rules**

### **. User registration and eligibility criteria:**

Users should fill designated **Registrations forms** localized on each instrumentation spot in service. **(Appendix A)** For eligibility each interested person has to be authorized to use our instruments by the facility coordinator or the technician in charge.

### **. Priorities**

RCMI investigators/ graduated students / technicians  
MSC investigators/ technicians/ graduated students  
Investigators outside MSC

### **. Instrumentations Logs**

Its very important the existence of log books for each instrument, first, to see the importance of money inversion on it, second, to track the use and quality of the instrument and; finally, if the instrument suffer any damage we have by these logs books any idea who was the responsible of it.

### **. Security**

\*Our security measures consist only in supervised each person during the usage process, and the use of our logs books.

\*When a problem arises with the equipment, the user should notify it immediately and/or fill out a **Problem Report Sheet** and handle it to the technician in charge of the facility, users should not attempt to solve the problem by himself. **(Appendix B)**

. **Acknowledgement of RCMI support**

Users of services or resources of the Molecular Genetics of Blood Disorders Core Facility should acknowledge RCMI for the support in their research by including the following sentence in their publications, abstracts or presentations:

*“ This investigation was supported, in part, by a Research Centers in Minority Institutions Award, G12RR-03051, from the National Center for Research Resources, National Institutes of Health”.*

Your cooperation in this respect is vital to the success of the program and the support of this facility.

Copies of publications acknowledging support to the RCMI Program should be sent to our office to include the reference in our progress report:

RCMI PROGRAM  
OFFICE 621-A  
6th FLOOR  
UPR MEDICAL SCIENCE CAMPUS  
Tel: (787) 763-9401  
Fax: (787) 758-5206  
E-mail: efernandez@rcm.upr.edu

## G. References

DNA thermal cycler 4800 Users manual. 1992

PTC 100 operations manual version 7.0. 1997 MJ research Inc.

I cycler Instruction manual. Bio Rad

Microfuge lite Instruction Manual. 1996 Beckman Instruments

Labconco 48,60 & 72 hoods Instruction Manual Protector Laboratory Hoods. 1992

Protector Acid storage cabinets and protector solvent Storage cabinets Instruction Manual. 1992. Labconco Corporation

Geneamp PCR system 2700 For amplification of nucleic Acids software. Applied Biosystem.

The Dcode Universal Mutation Detection System. 1996 Bio Rad Laboratories.

## H. Approval of Guidelines

Approved by:



Dr. Emma Fernández-Repollet  
Director, RCMi Program

Date:

11-18-02

# Appendix

## APPENDIX A

<i>Perkin Elmer Thermal Cycler/ PE 2700 (Genie I, Genie II)</i>					
<i>Name</i>	<i>PI</i>	<i>Ext</i>	<i>Date</i>	<i>Instrument used</i>	<i>Comments</i>

APPENDIX B

**Molecular Genetics**

Problem Report Form

Date: \_\_\_\_\_

Time: \_\_\_\_\_

Operator: \_\_\_\_\_

Instrument: \_\_\_\_\_

Description of Problem: \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

Send to : \_\_\_\_\_

Rcvd/date: \_\_\_\_\_

Action Taken: \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

## APPENDIX C



1. Thermal cyclers – PE 2700



2. Thermal cyclers

