OCA PAD INITIATION - PROJECT HEADER INFORMATION 09/28/90 15:55:26 Active Project #: G-32-611 Cost share #: Rev #: 0 Center # : 10/24-6-R7061-0A0 Center shr #: OCA file #: Work type : RES Document : AGR Contract#: AGREEMENT DTD 9/17/90 Mod #: Prime #: Contract entity: GTRC Subprojects ? : N Main project #: BIOLOGY Unit code: 02.010.134 Project unit: Project director(s): MATHIS J N (404)894-8432 BIOLOGY Sponsor/division names: UNIV OF SOUTH CAROLINA / COLUMBIA, SC Sponsor/division codes: 400 / 109 900417 Award period: 910416 (performance) 910416 (reports) to New this change Sponsor amount Total to date Contract value 4,200.00 4,200.00 4,200.00 Funded 4,200.00 0.00 Cost sharing amount Does subcontracting plan apply ?: N Title: DEVELOPMENT OF A RAPID CLINICAL SCREENING TEST FOR THE CYSTIC FIBROSIS GENE PROJECT ADMINISTRATION DATA OCA contact: Kathleen R. Ehlinger 894-4820 Sponsor technical contact Sponsor issuing office DR. ROBERT G. BEST DAN CHRISTMUS (803)765-7316 (803)777-7093 SCHOOL OF MEDICINE UNIVERSITY OF SOUTH CAROLINA TWO RICHLAND MEDICAL PARK, SUITE 301 OFFICE OF SPONSORED PROGRAMS UNIVERSITY OF SOUTH CAROLINA COLUMBIA, SC 29208 COLUMBIA, S.C. 29208 Security class (U,C,S,TS) : U ONR resident rep. is ACO (Y/N Defense priority rating : N/A N/A supplemental sheet Equipment title vests with: Sponsor X GIT Administrative comments -INITIATION OF PROJECT. NO INDIRECT COSTS ALLOWED. SEE PAGE 2 OF AGREEMEN

## GEORGIA INSTITUTE OF TECHNOLOGY OFFICE OF CONTRACT ADMINISTRATION

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NOTICE OF PROJECT CLOSEOUT

	Closeout Notice Date 04/16/91	
Project No. G-32-611	Center No. 10/24-6-R7061-	
Project Director MATHIS J N	School/Lab BIOLOGY	
Sponsor UNIV OF SOUTH CAROLINA/COLUMBIA, SC_	<u></u>	
Contract/Grant No. AGREEMENT DTD 9/17/90	Contract Entity GTRC	
Prime Contract No.		
Title DEVELOPMENT OF A RAPID CLINICAL SCREEN	ING TEST FOR THE CYSTIC FIBROSIS	
Effective Completion Date 910416 (Performance	e) 910416 (Reports)	
Closeout Actions Required:	Date Y/N Submitt	
Final Invoice or Copy of Final Invoice Final Report of Inventions and/or Subcon Government Property Inventory & Related ( Classified Material Certificate Release and Assignment Other	Certificate Y N ; N ;	
Comments		
Subproject Under Main Project No.		
Continues_Project No.		
Distribution Required:		
Project Director Administrative Network Representative GTRI Accounting/Grants and Contracts Procurement/Supply Services Research Property Managment Research Security Services Reports Coordinator (OCA) GTRC	Y Y Y Y N Y Y Y	

## Final Report Grant 32-611 - A Subcontract from the University of South Carolina School of Medicine

Development of a Rapid Clinical Test for the Most Common Deletion Resulting in Cystic Fibrosis ( $\Delta$ F508)

The primary objective of this grant was to set up a facility to perform DNA testing for the  $\Delta$ F508 deletion responsible for 70% of cystic fibrosis (CF) mutations.

During June and July 1990, we developed polymerase chain reaction (PCR) methods for identification of the  $\Delta$ F508 deletion. This involved synthesis and purification of a variety of oligonucleotide primers, determination of optimal reaction conditions, and isolation of DNA from the blood of CF carriers.

During July - September 1990, we set up a laboratory facility at the University of South Carolina, School of Medicine in Columbia, S.C. We adapted previously developed procedures for DNA isolation and PCR to fit with the equipment available in this facility. We set up the DNA thermocycler for PCR, gel electrophoresis equipment for separating DNA molecules and transilluminator and photosystem for detecting and identifying DNA in gels. During the start-up period of this project, we received 150 blood samples from children affected with CF and their immediate family members (parents and sibs).

From September 1990 - March 1991, diagnoses were developed on each of the patients by two techniques. One of the techniques involved the generation of a

Sau3A restriction site by a nucleotide mismatch (Friedman K. and J. Stoerker, Clin. Chem. 36:1702-1703) during PCR and the second involved hybridization of PCR products from a known homozygous normal individual with DNA from individuals of unknown status (Rommens et al., Am. J. Hum. Genet. 46: 396-397). Three other techniques were developed; one of these failed to be robust enough (Ballabio et al., Nature 343:220) and the other two are still in development (Golden et al., unpublished). Screening of blood samples revealed the following results from 43 affected CF patients 23 are homozygous for  $\Delta$ F508, 15 are heterozygous and 5 are homozygous for other deletions. The carrier status of individuals related to these affected children has also been determined. All cases have been verified by at least two separate replicates with each of the two primary tests described above. None of the initial 150 patients whose blood was collected during the start-up phase of this project has been (or will be) billed for the laboratory work. The total value of services provided during this start-up phase will be approximately \$22,500.00 (based on 150) patients with a test cost of \$150.00). We are currently continuing studies to optimize testing methodology and test alternative methodologies. The two methodologies still being developed are different from other methods being used currently and may provide new technology for CF screening.