



2nd INTERNATIONAL CONFERENCE
ON
UNSTABLE MICROSATELLITES
and HUMAN DISEASE

Co-Conveners:

Jack Griffith, University of North Carolina
David Nelson, Baylor College of Medicine
Robert Wells, Texas A&M University

Sponsored by:

University of North Carolina at Chapel Hill
Program in Molecular Biology and Biotechnology, and
Office of the Vice Provost of Graduate Studies and Research

North Carolina Biotechnology Center

National Institute of Environmental Health Sciences

The Friday center
University of North Carolina at Chapel Hill
April 17-20, 1999

**2nd INTERNATIONAL CONFERENCE
ON
"UNSTABLE MICROSATELLITES AND HUMAN DISEASE"
at
The Friday Center, UNC Chapel Hill
April 17-20, 1999**

Saturday April 17

REGISTRATION 7:30 AM to 9:00 AM Friday Center lobby

Morning Session 9:00 to 12:00 noon

Welcome: Dr. Jack Griffith with Dr. Robert Wells and Dr. David Nelson

First Session Chair: Sam Wilson, Deputy Director, National Institute of Environmental Health Sciences

1. Dr. Robert D. Wells
"Mechanisms of Triplet-Repeat Instabilities"

BREAK 10:10-10:25

2. Dr. Gillian Bates
"Analysis of Transgenic Models of Huntington's disease"
3. Dr. Virginia Zakian
"Consequences of DNA breakage on chromosome behavior in *Saccharomyces*"

12 noon to 1:00 Lunch (provided at the Friday Center) ·

Afternoon workshops and posters

1:15 to 4:15 12 talks each of 13 min with 2 for questions

4:15 to 5:30 Poster Session and wine/beer

5:30 - 6:30 Buffet Dinner: provided at the Friday Center

Evening Session 6:30 - 9:30

Session chair: Dr. David Nelson

1. Dr. David Nelson '~
"Repeat instability and disease: lessons from fragile X syndrome"
2. Dr. Thomas Petes
"Formation and repair of DNA loops during meiotic recombination in yeast: an assay for secondary structure formation for trinucleotide repeats"

BREAK 8:10 - 8:25

3. Dr. Stylianos E. Antonarakis
"Dodecamer repeat expansion in the Cystatin B gene in Progressive myoclonus epilepsy type 1"

Sunday April 18

Morning Session 8:30 AM to 12: noon

Session chair: Dr. Clyde Hutchison III

1. Dr. Keith Johnson
"Genotype to phenotype in DM: The eyes have it!"
2. Dr. Michael Resnick
"Novel genetic factors in yeast affecting stability of At-Risk Motifs (ARMS) formed by DNA repeats common to the human genome"

BREAK 10: 10 - 10:25

3. Dr. Massimo Pandolfo
"The pathogenesis of Friedreich ataxia: from GAA triplet repeat expansions to mitochondrial dysfunction, cardiomyopathy and neurodegeneration"
4. Dr. Steven Tapscott
"CTG expansion in myotonic dystrophy alters adjacent chromatin structure and gene expression"

12 noon to 1:00 Lunch (provided at the Friday Center)

Afternoon workshops and posters

1:15 to 4:15 12 talks of 13 min with 2 for questions

4:15 to 5:30 Poster Session and wine/beer

5:30 - 6:30 Buffet Dinner: provided at the Friday Center

Evening Session 6:30 - 9:30

Session chair: Dr. Berend Wieringa

1. Dr. Stephen Warren
"SNP-Based Haplotypes of FMR1 Suggest That Ancestral state of CGG Repeat Tracts influence Repeat Structure and Predisposition to Fragile X Syndrome"
2. Dr. Thomas Kunke
"Studies of Misalignment-Mediated DNA Synthesis Errors'."

BREAK 8:10- 8:25

3. Dr. Laura Ranum
"An untranslated CTG expansion causes a novel form of spinocerebellar ataxia (SCA8)"

Monday April 19

Morning Session 8:30 AM to 12 noon

Session Chair: Dr. Shelton Earp, Director, Lineberger Comprehensive Cancer Center, UNC-CH

1. Dr. Philip Kantoff
"Androgen Receptor CAG Repeat Sequence and its Role in Health and Disease"
2. Dr. Harry Orr
"Pathogenesis of Spinocerebellar Ataxia Type 1" part I

BREAK 10:10-10:25

3. Dr. Ruda Zoghbi
"Pathogenesis of Spinocerebellar Ataxia Type 1" part II
4. Dr. Jack Griffith
"T-loops, or how our chromosomes end"

12 noon to 1:00 Lunch (provided at the Friday Center)

Afternoon Session 1:15 to 4:15

Session chair: Dr. Thomas Meyer, Vice Provost for Graduate Studies and Research, UNC-CH

1. Dr. Kenneth Fischbeck
"Polyglutamine expansions in Kennedy's Disease"
2. Dr. Richard Sinden
"Slipped Strand DNA Structures formed in (CTG)_n (CAG)_n and (CGG)_n (CCG)_n Triplet Repeats"

BREAK 2:45 - 3:00

3. Dr. Paul Modrich
"Mismatch and large heterology repair in human cells."
4. Dr. Robert Komeluk
"The Myotonic Dystrophy CTG Repeat and "The Myotonic Dystrophy CTG Repeat and Mechanism of Disease"

4:15 to 5:30 Poster Session, Wine/beer

6:00 **Carolina Bar B Que** (provided at the Friday Center)

Tuesday April 20

Morning Session 8:30 AM to 12 noon

Session Chair: Dr. William Marzluff, Director, Program in Molecular Biology and Biotechnology

1. Dr. Michael Hayden
"Cleavage of Huntingtin is Essential for Toxicity in Vitro and in Vivo"
2. Dr. Albert La Spada
"The role of genomic context in trinucleotide repeat instability"

BREAK 10: 10 - 10:25

3. Dr. Ben Oostra
"FMRI: timing of instability and role of the FMRI protein."
4. Dr. Maury Swanson
"Triplet Repeat RNA-Binding Proteins in mRNA Biogenesis and Myotonic Dystrophy"

12 noon to 1:00 Lunch (provided at the Friday Center)