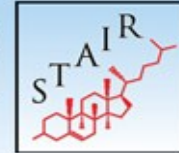




STEROL & ISOPRENOID  
RESEARCH CONSORTIUM



**THE UNITED LEUKODYSTROPHY FOUNDATION  
AND THE STAIR COMMITTEE**

*CTX (Cerebrotendinous Xanthomatosis) 2012*

Celebrating 30 years of service to  
Leukodystrophy Families

Friday  
July 20, 2012

Best Western Hotel  
DeKalb, Illinois

**The United Leukodystrophy Foundation  
would like to express our sincere  
thanks to our sponsors:**

**Manchester Pharmaceuticals, Inc.  
And  
Sigma-Tau Pharmaceuticals, Inc.**

**Manchester**   
**Pharmaceuticals, Inc.**™



## **CTX SCIENTIFIC & FAMILY MEETING**

THURSDAY, JULY 19, 2012

3:00 pm Registration  
6:00 pm Buffet Dinner and Hospitality

FRIDAY, JULY 20, 2012

6:45a.m – 8:00am Registration & Breakfast

8:00am - 8:20am Welcome to CTX 2012  
Paula K. Brazeal President  
William Rizzo, M.D. ULF Medical &Scientific Advisory Board Chairman  
Shailesh Patel, M.D., Division of Endocrinology, Metabolism and Clinical Nutrition, Medical College of Wisconsin, Milwaukee

8:25am - 8:55am CTX: From Bed to Bench and Contrary in the Siena Experience  
Professor Antonio Federico, Direttore, U.O. Neurologia e Malattie Neurometaboliche, Universita degli Studi di Siena

9:00am – 9:30am CTX: Update on Genetic Data and Methods  
Maria-Jesus Sobrido, M.D., PhD, Neurogenetics Group, Fundacion Publica Galega de Medicina Xenomica, Santiago de Compostela, Spain

9:35am-10:05am The Mouse Model for CTX  
Sandra Erickson, PhD, Professor, Medicine/Endocrinology and Metabolism, Department of Medicine, University of California, Veterans Affairs Medical Center, San Francisco, California

10:05am-10:25pm BREAK

10:30 pm - 11:00pm Diagnosis of CTX  
Kenneth Setchell, PhD, Pathology and Laboratory Medicine, Department of Pediatrics, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio.

11:05pm – 11:35pm Improved Blood Tests For CTX  
Andrea DeBarber, PhD, Physiology & Pharmacology Department (L334), Oregon Health & Science University, Portland, Oregon

11:40pm - 12:10pm Mechanisms Behind Development of Xanthomas in Cerebrotendinous Xanthomatosis  
Ingemar Bjorkhem, Senior Professor, Division of Clinical Chemistry, Department of Laboratory Medicine, Karolinska University Hospital Huddinge, Sweden

12:10pm - 1:40pm LUNCH

## **CTX SCIENTIFIC & FAMILY MEETING**

- 1:40pm - 1:55pm Topics of Interest to CTX Families  
William Rizzo, M.D., ULF Medical & Scientific Advisory Board Chairman
- 2:00pm - 2:30pm “Summary Current CTX Research/Future Research Directions/Funding Opportunities”  
Q & A Speaker Panel Session
- 2:35pm - 2:55pm Long Term Treatment in CTX  
Gerald Salen, M.D., Department of medicine, University of Medicine and Dentistry, New Jersey Medical School, Newark, New Jersey
- 3:00pm - 3:30pm Cholic Acid Treatment for CTX  
James Heubi, M.D., Department of Pediatrics, University of Cincinnati College of Medicine, Cincinnati, Ohio
- 3:35pm - 4:05pm “Ask the Doc” Questions & Answers Speaker Panel Session  
William Rizzo, M.D., ULF Medical & Scientific Advisory Board Chairman
- 4:10pm - 4:30pm CTX Treatment Update  
Paula Brazeal, President  
Brian Schreiber and GianFranco Fornasini, Sigma-Tau Pharmaceuticals, Inc., Gaithersburg, Maryland
- 4:35pm - 4:55pm John Wolf, ULF Board of Directors, Family Liaison, Aloha, Oregon
- 5:00pm - 5:30pm Break for Dinner and Poster Session Set-up
- 5:30pm DINNER
- 6:30pm Evening Poster Session & Hospitality

## INVITED SPEAKERS AND FACILITATORS

Ingemar Bjorkhem  
Senior Professor  
Division of Clinical Chemistry  
Department of Laboratory  
Medicine  
Karolinska University Hospital  
Huddinge, Sweden

Paula K. Brazeal  
ULF  
President  
Sycamore, IL

Andrea DeBarber, PhD  
Research Assistant Professor  
Physiology & Pharmacology  
Department  
Oregon Health & Science  
University  
Portland, OR

Sandra Erickson  
Department of Medicine  
University of California  
Veterans Affairs Medical Center  
San Francisco, CA 94121

Professor Antonio Federico  
Direttore  
U.O. Neurologia e Malattie  
Neurometaboliche  
Universita degli Studi di Siena  
Siena, Italy

James Heubi, M.D.  
Department of Pediatrics  
University of Cincinnati  
College of Medicine  
Cincinnati, OH

Ann Moser, B.A.  
Kennedy Krieger Institute  
Peroxisomal Diseases Lab  
Baltimore, MD

Shailesh Patel, M.D.  
Division of Endocrinology  
Metabolism & Clinical Nutrition  
Medical College of Wisconsin  
Milwaukee, WI

William Rizzo, M.D.  
Department of Pediatrics  
University of Nebraska Medical  
Center  
Omaha, NE

Jean-Baptiste Roulet, PhD  
Oregon Health & Science  
University  
Portland, OR

Gerald Salen, M.D.  
Department of Medicine  
University of Medicine &  
Dentistry  
New Jersey Medical School  
Newark, NJ

Brian Schreiber  
GianFranco Fornasini  
Sigma-Tau Pharmaceuticals, Inc.  
Gaithersburg, MD

Kenneth Setchell, PhD  
Pathology & Laboratory  
Medicine  
Department of Pediatrics  
Cincinnati Children's Hospital  
Medical Center  
Cincinnati, OH

Maria-Jesus Sobrido, M.D., PhD  
Neurogenetics Group  
Fundacion Publica Galega de  
Medicina Xenomica  
Santiago de Compostela  
Spain

Robert Steiner, M.D.  
Professor  
Oregon Health & Science  
University  
Portland, OR

John Wolf  
ULF Board of Directors &  
Family Liaison  
Aloha, OR

## HOTEL GENERAL INFORMATION:

- Complimentary coffee and iced tea available 24 hrs., in Hospitality Center; coffee makers in rooms; cold drink vending machines.
- Pool Hours: 9:00 am - 10:00 pm
- Incidental expenses: Telephone calls, TV, extra hotel nights, etc., are the responsibility of the participants and must be paid at check out time.
- Business Center: Fax and photocopy machines, printer for laptop computer.
- Fitness Center: 6:30am - 9:00 pm
- Check-in 3:00pm and Check-out 11:00am

## HOSPITALITY HOURS BEST WESTERN INN

Thursday Registration: 3:00pm  
Buffet Dinner & Hospitality: 6:00pm  
Welcome Reception for Speakers & STAIR Guests: 6:00pm- 10:00pm  
Friday Lunch and Dinner Buffet

## BREAKFAST:

Friday Buffet 6:45am - 8:00am  
Saturday Continental Breakfast

## EMERGENCY TELEPHONE NUMBERS:

- 1) Best Western (815) 758-8661
- 2) ULF Office, Sycamore, IL: (answering machine only):  
(815) 895-3211 or (800) 728-5483