

# Friday, July 27th

## Scientific Forum

7:30 am – 8:30 am Breakfast, Continental Ballroom I & II

8:30 am Introduction and Welcome | *Whitnie Strauss*

### Session 1: CCDS Science of Creatine Metabolism

8:35 am – 9:00 am Physiology & Pathophysiology | *Dr. Gajja Salomons*

9:00 am – 9:45 am Discoveries of CCDS Mice Models | *Dr. Matthew Skelton, Dr. Alex Kuan*

### Session 2: CCDS Introduction and Indicators

9:45 am – 10:15 am Clinical Presentation | *Dr. Ton DeGrauw*

10:15 am – 10:30 am Break

10:30 am – 11:00 am Identifying Patients and Patient Challenges | *Dr. Sarah Young, Carine Schneider, Carole Chehowah*

11:00 am – 11:30 am Brain Changes with MRI/MRS | *Dr. Kim Cecil*

11:30 am – 12:00 noon CCDS Seizures | *Dr. Denise Morita*

12:00 noon – 1:00 pm Lunch, Continental Ballroom I & II

### Session 3: CCDS Corrective Measures

1:00 pm – 1:30 pm Qualifying Newborn Screening | *Dr. Nicola Longo, Kim Tuminello*

1:30 pm – 2:00 pm GAMT Novel Therapeutics | *Dr. Andreas Schulze*

2:00 pm – 2:30 pm CTD and AGAT Novel Therapeutics | *Dr. Saadet Andrews*

2:30 pm – 2:45 pm Break

2:45 pm – 3:40 pm Investigational Treatments

*Dr. Aloïse Mabondzo*  
*Dr. Bruce Barshop*  
*Dr. Arjun Natesan (Ultragenyx)*  
*Dr. Maurizio Balestrino*

3:40 pm – 4:00 pm Development of Consensus Guidelines for Treatments Through Patient Registries and Natural History Studies | *Dr. Nicola Longo*

4:00 pm – 4:30 pm Observational Trial of Males with CTD (Vigilan Study)  
Panel Participants | *Dr. Judith Miller, Dr. Simona Bianconi, Dr. Aleksandra Bruchey, Carol Dutch, Regina Bogar*

4:30 pm – 5:00 pm Open Panel Discussion  
Moderator | *Dr. Yiumo Chan*

6:30 pm – 7:00 pm Cocktail Reception

7:00 pm BBQ Dinner, Continental Ballroom

# Saturday, July 28th

## Patient Forum

7:30 am – 8:30 am	Breakfast, Continental Ballroom I & II
8:30 am	Introduction and Welcome   <i>Whitnie Strauss</i>
8:35 am – 9:00 am	The Role and Importance of Patient Advocacy Groups ACD Board   <i>Whitnie Strauss, Heidi Wallis, Linda Cooper, Kim Tuminello, Melissa Parker, Laura Trutoiu</i>
9:00 am – 9:30 am	A Deeper Understanding of CCDS   <i>Dr. Sarah Young</i>
9:30 am – 10:00 am	CCDS, Defining the Genetic Components   <i>Dr. Nicola Longo</i>
10:00 am – 10:30 am	Break
10:30 am – 11:00 am	Recognizing Gene Sequencing and Gene Therapy   <i>Dr. Matthew Skelton</i>
11:00 am – 11:30 am	FDA and Drug Development   <i>Dr. Dave Weiner</i>
11:30 am – 12:00 noon	Understanding Clinical Trials   <i>Dr. Bruce Barshop</i>
12:00 noon – 1:00 pm	Lunch, Continental Ballroom I & II
1:00 pm – 1:30 pm	Planning for the Future Transitions and Special Needs Trust   <i>Julia Nickerson, Esq.</i>
1:30 pm – 2:00 pm	Assistive Technology for Speech and Language   <i>Maggie Kay Gibbs, MS, CCC-SLP</i>
2:00 pm – 3:00 pm	Patient Family Round Table Moderators   <i>Melissa Parker, Dr. Judith Miller</i> Round Table Participants   <i>Dr. Nicola Longo, Dr. Judith Miller, Dr. Sarah Young, Dr. Denise Morita</i>
3:00 pm – 3:30 pm	Break
3:30 pm – 5:00 pm	Applied Behavior Analysis (ABA) Parent Training First Leap Pediatric Therapy Group   <i>LeAnn Peacock</i>
6:30 pm – 7:00 pm	Cocktail Reception
7:00 pm	Awards Dinner, Continental Ballroom

# Sunday, July 29th

## Family Time

10:00 am – 12:00 pm

Let's Have Fun!

Patients and Families Are Invited to a Family Brunch at the Westin Pool Side Garden

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## Kids Camp Hours

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### Friday

Drop-Off

8:00 am – 8:30 am

Pick-Up

5:00 pm – 5:30 pm

Deep in the Heart of Texas!

*You've made the trip. Now let us give you a warm Texas welcome. In true Austin fashion, kids will create one of a kind bandanas, participate in rodeo-themed games, and enjoy Texas themed snacks and treats. Y'all, it's going to be fun!*

### Saturday

Drop-Off

8:00 am – 8:30 am

Pick-Up

5:00 pm – 5:30 pm

Imagine, Create, Invent

*Kids will have the opportunity to create unique art pieces, collaborate with friends on inventions, and use their imaginations across a variety of pretend play opportunities. There will even be a technology petting zoo to spark the interest of our older campers!*

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## Registration Hours

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### Thursday

6:30 pm – 7:30 pm

### Friday

7:30 am – 8:30 am

6:00 pm – 6:30 pm