



## INAUGURAL SCIENTIFIC MEETING

JUNE 21-22, 2021

**MEET • SHARE • LEARN**

(virtually)

The Glut1 Deficiency Foundation is a nonprofit patient advocacy organization dedicated to improving lives in the Glut1 Deficiency community through its mission of increased awareness, improved education, advocacy for patients and families, and support and funding for research.

We are honored to be members of the [Chan Zuckerberg Initiative Rare As One Network](#), where we are receiving support, tools, training, and funding to help develop a collaborative research network. As part of these efforts, we are planning our first scientific meeting June 21-22, 2021. We had originally planned [this meeting to be held in San Diego](#), but the decision was made to move to a virtual event in 2021 with plans to hold a follow-up scientific convening in person in 2022 in conjunction with our 9<sup>th</sup> family conference.

The goals of this first scientific convening will be to:

- Share key areas of current research in Glut1 Deficiency and identify critical gaps
- Establish connections and identify overlaps and synergies among related research areas
- Identify potential researchers for participation in a collaborative research network
- Identify potential projects for a strategic research plan to drive progress

We are full of excitement and optimism about the impact of this meeting, and we are grateful for your interest in joining us. The scientific meeting is designed to provide a forum for researchers and clinicians to share and collaborate, and lay summaries of the meeting will be provided for family members at our [virtual family conference in July](#).

Registration Information [here](#)

Poster Session Information [here](#)

Monday, June 21, 2021

10:00 AM to 5:00 PM Eastern Daylight Time



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## Opening Session

10:00-10:05 Opening Welcome  
Glenna Steele, Jason Meyers, Sandra Ojeda Glut1 Deficiency Foundation, USA

## Glut1 Deficiency Clinical Insights

Chair: Dr. Toni Pearson, MD, MBBS

10:05-10:25 Glut1 Deficiency Syndrome 1991-2021: Lessons Learned from a Quintessential Rare Disease  
Darryl De Vivo, MD Columbia University Irving Medical Center, New York City, USA

10:25-10:45 Symptoms and Biomarkers  
Juan Pascual, MD, PhD UT Southwestern Medical Center, Dallas, USA

10:45-11:00 Natural History  
Prof. Dr. Michèl Willemsen Radboud University Medical Center, Nijmegen, Netherlands

11:00-11:15 Current Treatment Options: What Works, What Doesn't, What Might  
Prof. Dr. Jörg Klepper: Aschaffenburg Children's Hospital, Aschaffenburg, Germany

11:15-11:35 Q&A

11:35-11:50 **break**

## Glut1 Deficiency Current Research Session 1

Chair: Prof. Dr. Jörg Klepper

11:50-12:05 Delineating the Spatial and Temporal Requirements for Glut1 in Glut1DS  
Umrao Monani, PhD: Columbia University Irving Medical Center, New York City

12:05-12:20 Gene Therapy for Glut1 Deficiency Syndrome  
Hitoshi Osaka, MD, PhD: Jichi Medical University, Tochigi, Japan

12:20-12:35 RNA Therapy  
Federico Zara, MD: University of Genoa, Genoa, Italy

12:35-12:45 Novel Genetic Interactions  
Paolo Grumati, PhD: Telethon Institute of Genetics and Medicine, Milan, Italy

12:45-12:55 Learning from Other Rare Genetic Diseases and the Non-coding Genome  
T.S. (Stefan) Barakat, MD, PhD: Erasmus MC/Sophia Children's Hospital Rotterdam

12:55-1:15 Q&A

1:15-1:30 **break**

# Monday (continued)

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Glut1 Deficiency Current Research Session 2		Prof. Dr. Michél Willemsen
1:30-1:45	Identifying New Treatments for Glut1 Deficiency Jason Park, MD, PhD: UT Southwestern Medical Center, Dallas, USA	
1:45-2:00	Precision Medicine Prof. Dr. Matthias Selbach: Max Delbrück Center for Molecular Medicine, Berlin, Germany	
2:00-2:20	Summary of Current Research Juan Pascual, MD, PhD: UT Southwestern Medical Center, Dallas, USA	
2:20-2:40	Q&A	
<b>2:40-3:00</b>	<b>break</b>	
Patient and Family Perspectives		Chair: Kris Engelstad, MS, CGC
3:00-3:15	Glut1 Deficiency Collaborative Voices Project Report Glenna Steele and Sandra Ojeda: Glut1 Deficiency Foundation, Owingsville, Kentucky USA	
3:15-4:00	Patient and Family Panel family experience stories, Q&A	
<b>4:00-4:10</b>	<b>break</b>	
Poster Session Lightning Round		Chair: Matthew Gentry, PhD
4:10-5:00	Selected Poster Presentations	

Tuesday, June 22, 2021

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## Glut1 - The Transporter

Chair: Juan Pascual, MD, PhD

### Structure, Function, Regulation

10:00-10:10	Structure and Function <a href="#">Prof. Catherine Etchebest</a> : Paris Diderot University
10:10-10:20	Role of Glut1 in Endothelial Cells <a href="#">Prof. Dr. Katrien De Bock</a> : Swiss Federal Institute of Technology (ETH), Zurich, Switzerland
10:20-10:30	Glut1 Regulation in Astrocytes <a href="#">Prof. L. Felipe Barros</a> : Centro de Estudios Científicos CECs, Valdivia, Chile
10:30-10:45	Q&A

### At the Blood Brain Barrier

10:45-10:55	Modeling Glut1 Deficiency Syndrome in a Petri Dish: Stemming a Momentum with Human Pluripotent Stem Cells <a href="#">Abraham Al-Ahmad, PhD</a> : Texas Tech University Health Sciences Center, Amarillo, Texas
10:55-11:05	High Throughput CRISPR Screening to Identify Regulators of Glut1 Expression <a href="#">Ethan Lippmann, PhD</a> : Vanderbilt University, Nashville, USA
11:05-11:15	Blood Brain Barrier on-a-chip: Nanomedicine for Glut1 Deficiency <a href="#">Angels Garcia Cazorla, MD, PhD</a> : Sant Joan de Deu Hospital, Barcelona, Spain
11:15-11:30	Q&A

**11:30-11:45 break**

### In Disease

11:45-12:00	Glucose Transporters in Brain During Health, Diabetes, and Stroke <a href="#">Prof. Hermann Koepsell</a> : University of Würzburg, Würzburg, Germany
12:00-12:10	Lessons from Cancer <a href="#">Etienne Meylan, PhD</a> : Ecole Polytechnique Fédérale de Lausanne, Lausanne, Switzerland
12:10-12:20	Lessons from Alzheimer's Disease <a href="#">Berislav Zlokovic, MD, PhD</a> : University of Southern California, Los Angeles, USA
12:20-12:35	Q&A

**12:35-12:50 break**

## Tuesday (continued)

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Brain Glucose Metabolism		Chair: Matthew Gentry, PhD
12:50-1:00	Overview of Brain Metabolism and Connections to Excitability <a href="#">Gary Yellen, PhD</a> : Harvard Medical School, Cambridge, USA	
1:00-1:10	Central Nervous System Fuels <a href="#">Mary McKenna, PhD</a> : University of Maryland School of Medicine, Baltimore, USA	
1:10-1:20	CSF and Brain Metabolism <a href="#">Maiken Nedergaard, MD, DMSc</a> : University of Rochester, USA, University of Copenhagen, Denmark	
1:20-1:30	Glycogen and Lessons from Lafora Disease <a href="#">Matthew Gentry, PhD</a> : University of Kentucky, Lexington, USA	
1:30-1:40	Role of the Astrocyte Neuron Lactate Shuttle in Glut1 Deficiency Syndrome <a href="#">Pierre Magistretti, MD, PhD</a> : King Abdullah University of Science and Technology, Thuwal, Saudi Arabia	
1:40-1:55	Q&A	
<b>1:55-2:10</b>	<b>break</b>	
Ketogenic Diets		Chair: Jong M. Rho, MD
2:10-2:15	Introduction <a href="#">Jong M. Rho, MD</a> : University of California San Diego, USA	
2:15-2:25	Evolution of Ketogenic Diets Over the Past Quarter Century <a href="#">Elizabeth Thiele MD, PhD</a> : Harvard University, Cambridge, USA	
2:25-2:35	Is the Efficacy of the Ketogenic Diet in Glut1DS Strictly Due to Ketone Body Utilization? <a href="#">Karin Borges, PhD</a> : The University of Queensland, Brisbane, Australia	
2:35-2:45	Neuroprotective Activity of the Ketogenic Diet <a href="#">Robin Williams, PhD</a> : Royal Holloway, University of London, United Kingdom	
2:45-2:55	The Effects of the Ketogenic Diet on Cognition and Behavior <a href="#">Susan Masino, PhD</a> : Trinity College, Hartford, Connecticut, USA	
2:55-3:00	Concluding Remarks <a href="#">Jong M. Rho, MD</a> : University of California San Diego, USA	
3:00-3:15	Q&A	
<b>3:15-3:30</b>	<b>break</b>	

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### Special Interest Roundtables

3:30-4:15

Special Interest Group Breakouts

- small groups in categories below:  
discuss new learnings, overlaps, new ideas, potential research projects
- session moderators listed

1. **Glut1 Deficiency Clinical** *Prof. Dr. Jörg Klepper*
2. **Glut1 Deficiency Research** *Jason Park, MD, PhD*
3. **Gene Therapy** *Umrao Monani, PhD*
4. **Glut1 – the Transporter** *Prof. L. Felipe Barros*
5. **Blood Brain Barrier** *Ethan Lippman, PhD*
6. **Glucose Metabolism** *Matthew Gentry, PhD*
7. **Ketogenic Diets** *Jong M. Rho, MD, Beth Zupec-Kania, RDN, CD, Mackenzie Cervenka, MD*
8. **Patients and Families** *Glenna Steele, Jason Meyers, Sandra Ojeda*

#### Key Questions to Guide Discussion and Sharing/Reporting:

- What are some of the most critical gaps in understanding the disease?
- What are the key gaps in foundational science?
- What additional tools are needed?
- Are any additional models needed?
- What research questions are most important to answer to have the greatest impact for patients?
- What studies are most important to be able to answer these key research questions?

### Closing Session

4:15-4:50

Special Interest Group Reports

*Matthew Gentry, PhD and Glenna Steele*

4:50-5:00

Closing Remarks and Next Steps

*Glenna Steele, Jason Meyers, Sandra Ojeda* Glut1 Deficiency Foundation, USA