



7TH INTERNATIONAL SCIENTIFIC CONFERENCE ANGELMAN SYNDROME



Platinum sponsor



The 7th international conference on the rare disorder Angelman Syndrome organized on behalf of the Angelman Syndrome Alliance by the Medical University of Vienna and the Austrian association supporting Angelman Syndrome Research brings together leading researchers in the field, as well as scientists from adjacent fields in order to discuss latest research results and future directions.

In addition, members of Angelman Syndrome Expertise Centres will attend to share their experiences in both treatment and care of individuals affected by Angelman Syndrome.

For the first time, the conference will take place in Vienna, Austria, from the 15th until the 19th of September 2022. The first two days are focussed on the scientific community and on medical experts. On the third day of the conference, families and caretakers will join to get a detailed overview of the latest Angelman Syndrome research in layman terms, thereby giving an unique insight into the current status of research. In addition, ongoing clinical studies and their status will be featured. Also, more practical approaches regarding dietary measures, augmented communication and information on Expertise Centers will be shared within the community.



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Overview: Thursday – Saturday (15-17.9.2022), in Vienna, Austria

Date	Day	Start	End	Target group	Content
15.9.2022	Thursday	14.00	18.00	Scientists + Students Medical professionals Pharmaceutical professionals ASA members	Scientific talks Angelman Syndrome
16.9.2022	Friday	9.00	20.00	Scientists + Students Medical professionals Pharmaceutical professionals ASA members	Scientific talks Angelman Syndrome Plenary sessions (Scientists, AS Expertise centers, ASA meeting), 10-minute talks, Poster sessions
17.9.2022	Saturday	9.00	16.00	Parents Caretakers Scientists + Students Medical professionals Pharmaceutical professionals ASA members	Layman talks / ASA meeting Presentations on therapies and trials

Tickets:

- **Thursday – Saturday (15-17.9.2022):** 250 Euro* (snacks, lunch, dinner and drinks included)
 - Scientists
 - Medical professionals
 - Pharmaceutical professionals
 - ASA members

There will also be Social Events organised in the evening, about which we will share information in the next months, as we need to take latest COVID rules into consideration. The registration and the costs for the social event will be charged separately.

- **Saturday only (17.9.2022) :** 99 Euro* (snacks, lunch and drinks included)
 - Scientists
 - Medical professionals
 - Pharmaceutical professionals
 - ASA members
 - Parents
 - Caretakers
- Website link: <https://www.angelman.at/7th-international-scientific-conference-in-vienna/>



Locations

- Two locations:
 - **Thursday & Friday:** Medical University of Vienna, University Clinics for Dentistry, [Sensengasse 2a 1090 in Vienna.](#)



- **Saturday:** Medical University of Vienna, Van Swieten Saal, Van-Swieten-Gasse 1a, 1090 Vienna



Our speakers

- Ype Elgersma, Dr. (Erasmus MC, Department of Neuroscience, Rotterdam, Netherlands)



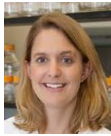
- Ugo Mayor, Dr. (University of the Basque Country UPV/EHU, Department of Biochemistry, Spain)
“UBE3A-induced ubiquitination changes in the brain elucidated by proteomics”



- Rajini Rao, Ph.D. (John Hopkins, Physiology, John Hopkins Medicine, Baltimore, USA)
“Syndrome a disorder of organellar ion homeostasis? Insights from Christianson Syndrome.”



- Stormy Chamberlain, Ph.D. (Roche, Switzerland)
“Coordinated Regulation of UBE3A-ATS and UBE3A”



- Ben Distel, Ph.D. (Amsterdam UMC, Netherlands)
“The effect of UBE3A on the function of the proteasome”



- Hanoch Kaphzan, Ph.D. (University of Haifa, Sagol Department of Neurobiology, Israel)
“The role of cellular redox homeostasis in the early development of Angelman syndrome”



- Ramiro Almeida, Ph.D. (Institute of Biomedicine, Department of Medical Sciences, University of Aveiro, Portugal)
“The role of UBE3A in synapse formation and function”



- Gaia Novarino, Dr. (Institute of Science and Technology, Vienna, Austria)



- Evguenia Bekman, Ph. D. (Instituto Superior Técnico, University of Lisbon)
“Recapitulating Angelman Syndrome using iPSC-derived cerebellar organoids”



- Simão Rocha, Ph. D. (Instituto Superior Técnico, University of Lisbon)
“Stem cell toolkit for modelling cerebellar dysfunction in Angelman Syndrome”



- Ilaria Tonazzini, PhD (Nanoscience Institute - Consiglio Nazionale delle Ricerche (CNR) @ NEST (National Enterprise for nanoScience and nanoTechnology), Pisa (Italy) & Laura Baroncelli, PhD (Neuroscience Institute - Consiglio Nazionale delle Ricerche (CNR), Pisa (Italy))
“Innovative brain-targeting nano-tools and imaging methods for therapeutic development in Angelman Syndrome”



- Dr. Karen Bindels (Erasmus MC) & Dr. Marie Claire de Wit (Erasmus MC)
“Sleep problems in children with AS: the effect of a behavioral intervention program” &
„Information about AS clinics“



- Dr. Christel Kannegießer Leitner
“CBD anticonvulsant - Hype or real possibility”



- Dr. Anastasia Male-Dressler
„Ketogenic diet“

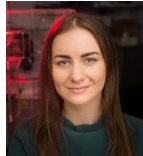


- Michaela Zöbl



“Unterstützte Kommunikation“ (“Augmentative and Alternative Communication”)

- Hanna Vihma, Ph.D. from the Ben Philpot lab. (UNC School of Medicine, Cell Biology and Physiology, Chapel Hill, USA)



- Sebastian Camillo Holst, Ph.D. (Roche)
“Assessments of the sleep abnormalities in AS from the FREESIAS observational study”



- Narayanan Ramanathan, Ph. D. (Roche)
“Characterizing mICD mouse model for AS”



- Rob Komorowski, Ph. D. (Ionis Pharmaceuticals)
“Detecting UBE3A in Cerebral Spinal Fluid of AS Patients”

- Short presentations by:
 - Matteo Fossati
 - Carina Maranga
 - Mia Tichy
 - Doesjka Hagenaar
 - Cesar Ochoa-Lubinoff
 - Alberto Velez-van-Meerbeke

- Presentations from these pharmaceutical companies on their research & ongoing clinical trials
 - Roche

- Dr. Brenda Vincenzi



- PTC Therapeutics
 - Dr. Ifode Ajari (PTC Therapeutics)

- Ionis Pharmaceuticals
 - Becky Crean, Ph. D.



Registration

- Registration for the conference can be done with this link:
<https://univie.ungerboeck.com/PROD/emc00/register.aspx?eid=R0FIODRTL1dBTTA1eUhMWkVnNFFZUT090>

Hotel information

- The hotel we recommend, which is 15 minutes walking from the conference venue and where we have reserved capacity for the conference is:

Hotel Regina - Kreamlehner Hotels, Rooseveltplatz 15, Wien 1090 - [Location](#)



The Hotel Regina - The main building of the Kreamlehner Hotels has been established in 1877 as a city palace.

- For reserving a room at Hotel Regina or a hotel recommendation, please contact Sandra Tronigger (sandra.tronigger@univie.ac.at).

Flight: Special Lufthansa Group fares

The Lufthansa Group airlines bring people together - every day, all around the world. The global route network of Austrian Airlines, Lufthansa, SWISS, Brussels Airlines and Eurowings offers optimal connection and combination options, so you will benefit from quick and direct flights to the event. You will reach the booking platform via this link:
<https://www.lufthansa.com/de/en/meetings-and-events-delegates>

and with the event code **ATZLWAX**. The reduced fares are automatically displayed.

NB: Please enable pop-ups permanently in your browser while booking, otherwise the window in the booking platform will not open.

Of course, you can also book through your IATA travel agency. The travel agency can request the ticketing instructions by sending an email to lufthansa.mobility@dlh.de, stating the event code ATZLWAX.

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Detail program

Day 1: Thursday 15.9.2022, Location: Zahnklinik

Time	THURSDAY
13:00	Registration to Conference
13:10	
13:20	
13:30	
13:40	Meet & Greet over a cup of coffee/tea
13:50	
14:00	Welcome note from the organizers
14:10	Speaker 1: Ype Elgersma
14:20	Novel mouse mutants to address emerging questions
14:30	
14:40	
14:50	Speaker 2: Stormy Chamberlain
15:00	Coordinated Regulation of UBE3A-ATS and UBE3A
15:10	
15:20	
15:30	Speaker 3: Sebastian Camillo Holst (Roche)
15:40	Assessments of the sleep abnormalities
15:50	in AS from the FREESIAS observational study
16:00	
16:10	Refreshments
16:20	
16:30	Speaker 4: Evguenia Bekman
16:40	Recapitulating Angelman Syndrome
16:50	using iPSC-derived cerebellar organoids
17:00	Speaker 5: Simão Da Rocha
17:10	Stem cell toolkit for modelling cerebellar
17:20	dysfunction in Angelman Syndrome
17:30	Speaker 6: Ilaria Tonnazzini + Laura Baroncelli
17:40	Innovative brain-targeting nano-tools and imaging
17:50	methods for therapeutic development in AS
18:00	Closing Remarks
18:10	Social programme:
18:20	Vienna City Tour with street car
18:30	
18:40	
18:50	
19:00	
19:10	Dinner at a "Heuriger" (wine tavern) "Zum Martin Sepp"
19:20	
19:30	
19:40	
19:50	
20:00	
20:10	
20:20	
20:30	



Day 2: Friday, 16.9., Location: Medical University of Vienna, University Clinics of Dentistry

Morning:

9:00	Opening day 2 by organizers
9:10	Speaker 7: Hanoch Kaphzan
9:20	The role of cellular redox homeostasis
9:30	in the early development of Angelman syndrome
9:40	
9:50	Speaker 8: Hanna Vihma (member of Ben Philpot's lab)
10:00	Angelman syndrome biomarkers and treatment
10:10	opportunities
10:20	
10:30	Coffee / Tea break
10:40	
10:50	Speaker 9: Ben Distel
11:00	The effect of UBE3A on the function of the proteasome
11:10	
11:20	
11:30	Speaker 10: Gaia Novarino
11:40	Studying cortical development through the lens of
11:50	autism spectrum disorders
12:00	
12:10	Lunch
12:20	
12:30	
12:40	
12:50	
13:00	



Friday Afternoon:

Time	FRIDAY
13:00	Short talks I:
13:10	(10 minutes + 8 minutes Q&A at the end)
13:20	Matteo Fossati, Carina Maranga, Mia Tichy,
13:30	& Ana Rodrigues
13:40	Speaker 11: Narayanan Ramanathan (Roche)
13:50	Characterizing mICD mouse model for AS
14:00	Speaker 12: Rajini Rao
14:10	Is Angelman Syndrome a disorder
14:20	of organellar ion homeostasis?
14:30	Insights from Christianson Syndrome.
14:40	Speaker 13: Ugo Mayor
14:50	UBE3A-induced ubiquitination changes
15:00	in the brain elucidated by proteomics
15:10	
15:20	Refreshments
15:30	
15:40	Speaker 14: Ramiro Almeida
15:50	The role of UBE3A in synapse formation and function
16:00	
16:10	
16:20	Speaker 15: Karen Bindels
16:30	Sleep problems in children with AS:
16:40	the effect of a behavioral intervention program
16:50	
17:00	Speaker 16: Rob Komorowski (Ionis Pharmaceuticals)
17:10	Detecting UBE3A in Cerebral Spinal Fluid of AS Patients
17:20	Short talks II
17:30	(10 minutes + 8 minutes Q&A at the end)
17:40	Doesjka Hagenaar, Cesar Ochoa-Lubinoff,
17:50	Alberto Velez-van-Meerbeke
18:00	Announcements
18:10	Dinner on location: Traditional Austrian food + Cheese & Wine
18:20	Split into three groups.
18:30	Three plenary sessions in parallel:
18:40	1. Scientists: What do we NOT know about AS? -
18:50	closed session. Ype Elgersma will chair this discussion
19:00	focussed on emerging research questions that need to
19:10	be addressed concerning UBE3A function, AS
19:20	pathophysiology and development of treatments.
19:30	2. Doctors: Why start an AS clinic? Which specialisations
19:40	needed? Which facilities needed? Program?
19:50	Chaired by Karen Bindels & Claire de Wit
20:00	3. ASA members: Member Meeting-closed session
20:10	Continue working & discussing & exchanging ideas
20:20	
20:30	Drinks at Stiegl Ambulanz in 'Altes AKH'

Day 3: Saturday, 17.9., Location: Van Swieten Saal

Time	SATURDAY	
8:30	Registration to conference	
8:40	@ Van Swieten Saal	
8:50		
9:00	Welcome from the organizers	
9:10	Speaker 16: Harald Sitte	
9:20	Overview on the latest AS research presentations	
9:30		
9:40		
9:50	Speaker 17: Da Rocha / Bekman	
10:00	Layman explanation of new research project	
10:10	Speaker 18:Tonnazzini/Baroncelli	
10:20	Layman explanation of new research project	
10:30	Coffee / Tea break	
10:40		
10:50	Speaker 19: Brenda Vincenzi (Roche)	
11:00	Roche: AS research on therapy & study status	
11:10		
11:20		
11:30	Speaker 20: Michaela Zöbl	
11:40	Augmentative and Alternative Communication /	
11:50	Unterstützte Kommunikation	
12:00		
12:10	Lunch	ASA Meeting (2)
12:20		For ASA members
12:30		
12:40		
12:50		
13:00		

Saturday, Afternoon:

Time	SATURDAY
13:00	Speaker 21: Becky Crean (Ionis Pharmaceuticals)
13:10	Update on the Ionis clinical trial of HALOS for
13:20	Angelman syndrome
13:30	Speaker 22: Christel Kannegiesser Leitner
13:40	CBD anticonvulsant - Hype or real possibility
13:50	
14:00	Speaker 23: Ifode Ajari (PTC Therapeutics)
14:10	Industry update AS program
14:20	Refreshments
14:30	
14:40	
14:50	Speaker 24: Malle Dressler
15:00	Information about Ketogenic diet (Ketogene
15:10	Diät) for AS
15:20	Speaker 25: Karen Bindels & Marie Claire de De Wit
15:30	Information about the Rotterdam
15:40	Expertise Centre "Encore" & information on
15:50	how to improve sleep
16:00	Closing Remarks



Organisation:

- The Hosting organisation is the Medical University of Vienna, Centre for Physiology and Pharmacology, Institute of Pharmacology and the [Angelman Syndrome Alliance \(ASA\)](#)
- The local organising committee:
 - Thomas Schramm (Chairman of [Association for the Research on Angelman Syndrome Austria](#))
 - Sandra Martinz (Deputy [Association for the Research on Angelman Syndrome Austria](#))
 - Pia Schlögl ([Association for the Research on Angelman Syndrome Austria](#))
 - Mirella Karoly ([Association for the Research on Angelman Syndrome Austria](#))
 - Roland Spielhofer ([Association for the Research on Angelman Syndrome Austria](#))
 - Univ.-Prof. Dr. Harald Sitte ([Medical University Vienna](#), Chairman of the scientific advisory board Angelman Syndrome Alliance)

