



# 7TH INTERNATIONAL SCIENTIFIC CONFERENCE ANGELMAN SYNDROME



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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 15<sup>th</sup> until the 17<sup>th</sup> 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 sleeping problems, augmented communication and information on Expertise Centres 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	20.30	Scientists + Students Medical professionals Pharmaceutical professionals ASA members	Scientific talks Angelman Syndrome Plenary sessions (Scientists, AS Expertise centres, ASA meeting), 10-minute talks
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

Social program (125 €): On Thursday evening after the conference there will be city tour with a traditional street car with a guided tour of Vienna followed by a dinner at a wine tavern.

- **Saturday only (17.9.2022) :** 99 Euro\* (snacks, lunch and drinks included)
  - Scientists
  - Medical professionals
  - Pharmaceutical professionals
  - ASA members
  - Parents
  - Caretakers

On Saturday Translators are present to translate from English to German.

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

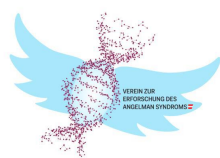


Internet / Wlan / Wifi access code:

➔ Choose Wifi "Gast Wlan" then access Internet and use credential to login:

Username (Benutzername): V\_1126

Password (Passwort): 3wnpt



## Our speakers

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



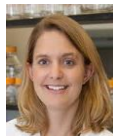
- Ugo Mayor, Dr. (University of the Basque Country UPV/EHU, Department of Biochemistry, Spain)  
"UBE3A-induced ubiquitination changes in the brain elucidated by proteomics"



- 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"



- Mattijs Punt is a PhD student in the lab of Ype Elgersma (see above) and Ben Distel, Ph.D. (Amsterdam UMC, Netherlands) at the Erasmus Medical Centre in Rotterdam, the Netherlands.  
"The effect of UBE3A on the function of the proteasome"



- Hanoch Kaphzan, Ph.D. (University of Haifa, Sagol Department of Neurobiology, Israel)  
"Angelman syndrome, learning from bioinformatics analyses and the role of redox homeostasis in early brain development"



- 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-de Heus (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 (Registered doctor)  
“CBD anticonvulsant - Hype or real possibility”

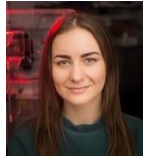


- Michaela Zöbl (Assista soziale Dienste GmbH, Austria)  
“Unterstützte Kommunikation“ (“Augmentative and Alternative Communication”)



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

“Angelman syndrome biomarkers and treatment opportunities”



- 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”



- Cesar Ochoa-Lubinoff, MD (Rush University Medical Center, USA)  
“A Prospective, Longitudinal, Observational Clinical Trial in Individuals with Angelman syndrome to Enable Endpoint Development for Interventional Trials (FREESIAS)”



- Short presentations by:
  - Matteo Fossati (Humanitas Research Hospital, Rozzano, Italy)



- Carina Maranga (Universidade de Lisboa, Portugal)



- Eugenie Suter (Roche)



- Adriana Vieira (iMM-JLA (Portugal))



- Doesjka Hagenaar (Erasmus MC Sophia, Netherlands)



- Alberto Velez-van-Meerbeke (Universidad del Rosario, Bogotá, Kolumbien)



- Amanda Moore (ASF, USA)



- Betty Willemsen (Nina Foundation & ASA, Netherlands)



- Liz Jalazo (ASF, USA)



- Anne Wheeler (RTI International, USA)



- 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.





## 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	<b>Speaker 1: Ype Elgersma</b>
14:20	Novel mouse mutants to address emerging questions
14:30	
14:40	
14:50	<b>Speaker 2: Stormy Chamberlain</b>
15:00	Coordinated Regulation of UBE3A-ATS and UBE3A
15:10	
15:20	
15:30	<b>Speaker 3: Sebastian Camillo Holst (Roche)</b>
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	<b>Speaker 4: Evguenia Bekman</b>
16:40	Recapitulating Angelman Syndrome
16:50	using iPSC-derived cerebellar organoids
17:00	<b>Speaker 5: Simão Da Rocha</b>
17:10	Stem cell toolkit for modelling cerebellar
17:20	dysfunction in Angelman Syndrome
17:30	<b>Speaker 6: Mattijs Punt</b>
17:40	The effect of UBE3A on the function of the proteasome
17:50	
18:00	
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:**

Time	FRIDAY
8:30	
8:40	
8:50	
9:00	Opening day 2 by organizers
9:10	<b>Speaker 7: Hanoch Kaphzan</b>
9:20	Angelman syndrome, learning from bioinformatics
9:30	analyses and the role of redox homeostasis
9:40	homeostasis in early brain development
9:50	<b>Speaker 8: Hanna Vihma (member of Ben Philpot's lab)</b>
10:00	Angelman syndrome biomarkers and treatment
10:10	opportunities
10:20	
10:30	Coffee / Tea break
10:40	
10:50	<b>Speaker 9: Ilaria Tonazzini + Laura Baroncelli</b>
11:00	Innovative brain-targeting nano-tools and imaging
11:10	methods for therapeutic development in AS
11:20	
11:30	<b>Speaker 10: Gaia Novarino</b>
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	<b>Short talks I:</b>
13:10	Matteo Fossati, Carina Maranga, Eugenie Suter (Roche)
13:20	& Adriana Vieira
13:30	
13:40	<b>Speaker 11: Narayanan Ramanathan (Roche)</b>
13:50	Characterizing mICD mouse model for AS
14:00	<b>Speaker 12: Rajini Rao</b>
14:10	Is Angelman Syndrome a disorder
14:20	of organellar ion homeostasis?
14:30	Insights from Christianson Syndrome.
14:40	<b>Speaker 13: Ugo Mayor</b>
14:50	UBE3A-induced ubiquitination changes
15:00	in the brain elucidated by proteomics
15:10	
15:20	Refreshments
15:30	
15:40	<b>Speaker 14: Ramiro Almeida</b>
15:50	The role of UBE3A in synapse formation and function
16:00	
16:10	
16:20	<b>Speaker 15: Karen Bindels-de Heus</b> Sleep problems in children with
16:30	AS: The effect of a behavioral intervention program
16:40	<b>Speaker 16: Rob Komorowski (Ionis Pharmaceuticals)</b>
16:50	Detecting UBE3A in Cerebral Spinal Fluid of AS Patients
17:00	<b>Speaker 17: Cesar Ochoa-Lubinoff</b> - Clinical Trial in Individuals
17:10	with AS to enable Endpoint Developm. f. interventional trials
17:20	<b>Short talks II</b>
17:30	(10 minutes + 8 minutes Q&A at the end)
17:40	Doesjka Hagenaar, Alberto Velez-van-Meerbeke,
17:50	Amanda Moore, Betty Willemsen
18:00	<b>Announcements</b>
18:10	Dinner on location: Traditional Austrian food + Cheese & Wine
18:20	Split into three groups.
18:30	<b>Three plenary sessions in parallel:</b>
18:40	<b>1. Scientists:</b> 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	<b>2. Doctors:</b> Why start an AS clinic? Which specialisations
19:40	needed? Which facilities needed? Program?
19:50	Chaired by Karen Bindels-de Heus & Marie-Claire de Wit
20:00	<b>3. ASA members:</b> 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**

Morning:

Time	SATURDAY	
8:30	Registration to conference	
8:40	@ Van Swieten Saal	
8:50		
9:00	Welcome from the organizers	
9:10	<b>Speaker 18: Harald Sitte</b>	
9:20	Overview on the latest AS research presentations	
9:30		
9:40		
9:50	<b>Speaker 19: Da Rocha / Bekman</b>	
10:00	Layman explanation of new research project	
10:10	<b>Speaker 20: Tonazzini/Baroncelli</b>	
10:20	Layman explanation of new research project	
10:30	Coffee / Tea break	
10:40		
10:50	<b>Speaker 21: Brenda Vincenzi (Roche)</b>	
11:00	Roche: AS research on therapy & study status	
11:10		
11:20	<b>Speaker 22: Anne Wheeler / Elizabeth Jalazo</b>	
11:30	Ladder database & learning network	
11:40	<b>Speaker 23: Michaela Zöbl</b>	
11:50	Augmentative and Alternative Communication /	
12:00	Unterstützte Kommunikation	
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	<b>Speaker 24: Becky Crean (Ionis Pharmaceuticals)</b>
13:10	Update on the Ionis clinical trial of HALOS for
13:20	Angelman syndrome
13:30	<b>Speaker 25: Karen Bindels-de Heus &amp; Marie-Claire de Wit</b>
13:40	Information about the Rotterdam
13:50	Expertise Centre "Encore" & information on
14:00	how to improve sleep
14:10	
14:20	Refreshments
14:30	
14:40	<b>Speaker 26: Christel Kannegiesser Leitner</b>
14:50	CBD anticonvulsant - Hype or real possibility
15:00	
15:10	<b>Speaker 27: Ifode Ajari (PTC Therapeutics)</b>
15:20	Industry update AS program
15:30	
15:40	<b>Short talks III:</b>
15:50	Anna Kyrychenko, Stan Krolczyk (Ultragenyx)
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)

