SATB2 RESEARCH ROUNDTABLE SUMMARY

OCTOBER 7TH 2021

Our goal

is to unite exceptional professionals with dedicated families and thus create a knowledgeable, collaborative and supportive community with the spirit and power to bring treatments and cures for SATB2-associated syndrome.

SATB2 Europe and our global research initiative CureSATB2 hosted the first SATB2 Research Virtual Roundtable on October 7th 2021.

We gathered international SATB2 researchers and SAS clinicians and asked them to introduce themselves, to share their recent and future research projects with us and to listen to our experiences as caregivers.

We also invited affected families and clinicians to simultaneously keep the whole community up to date with what is happening.

At this first event, our panelists were asked to give 6-minute presentations on their work.

This document is a summary of the roundtable.


Registered participants
from 23 countries


117 attending
families, scientists,
clinicians and therapists


14 research
teams represented

Statistics

“It is a great honour and pleasure to host this event and see so many dedicated researchers, clinicians, patient advocacy groups, therapists and numerous parents gathered around the same important topic.

This meeting is the first of many to come and like in life, the first step is to meet, introduce ourselves and our work, then build on collaborations and work on common goals.

SATB2 Europe’s vision is to significantly improve quality of life for individuals with SAS. We aim to do this through our two initiatives: CareSATB2 and CureSATB2.

Today we show that even in rare conditions like SAS, together we are many and we have the power to make a change.”

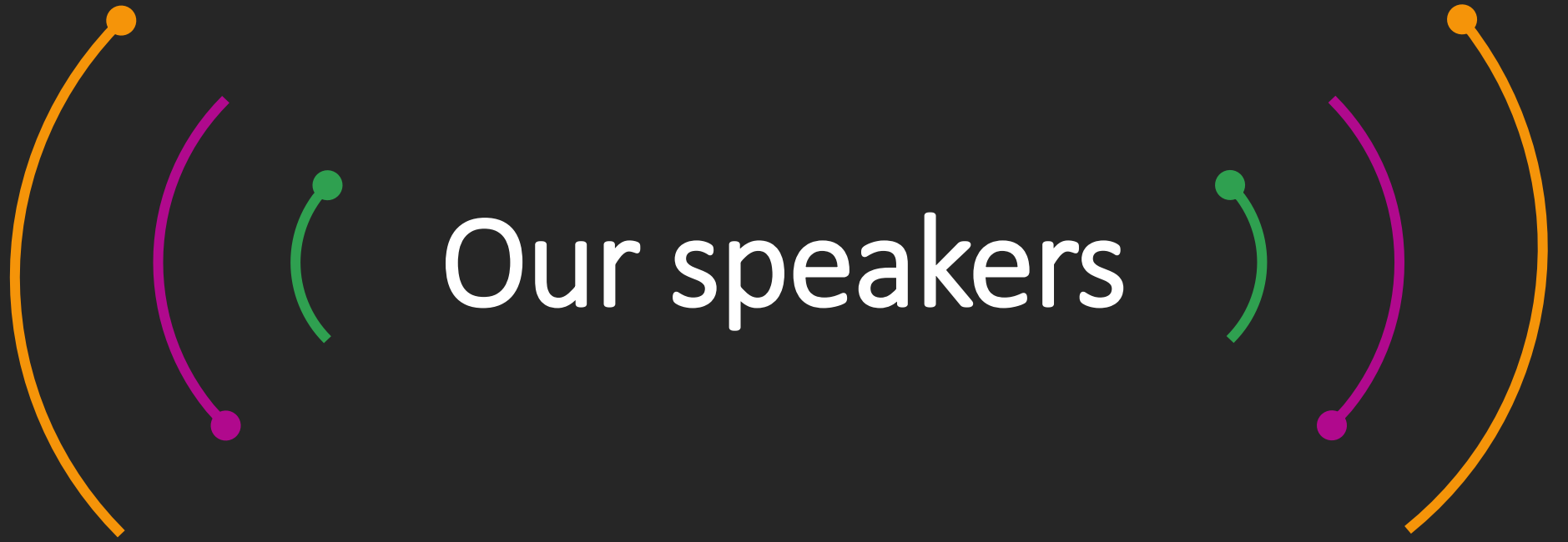
Erika Stariha, President of SATB2 Europe
erika.stariha@satb2europe.org

Jenny-Li Örsell, Head of CureSATB2
jenny-li.orsell@curesatb2.org

"We know that individuals with SATB2-associated syndrome are vulnerable. The spectrum is wide, but in severe cases children are robbed from the ability to talk, to walk, to eat and to be able to play with friends.

We are convinced that these persons can – and deserve – to have a better future. We are at a point in history where research is opening doors to understand and treat genetic disorders. Our goal is to speed up this process for SAS and bring targeted treatments, and ultimately cures, to those affected.

As we build and nurture this collaborative environment of families, researchers and clinicians, together, we will reach our goals. Faster than ever."


Our speakers

Sarah Vergult, PhD

Department of Biomolecular Medicine,
Center for Medical Genetics, Ghent University, Belgium

I have a background in genetics. I was appointed assistant professor in October 2019 within the RARE-MED consortium at Ghent University, to study the role of the noncoding part of the genome in rare disorders. This consortium brings together clinical and lab experts to spearhead the research regarding rare disorders. Our SATB2 project perfectly fits with the aim of the RARE-MED consortium, as we want to build bridges between the clinic and the research environment. I am always eager to collaborate.

Prof. Dr. Simon Fisher

Language and Genetics Department,
Max Planck Institute for Psycholinguistics, the Netherlands

My research focuses on identification and characterization of genetic pathways that are important for speech, language and reading, by studying developmental disorders as well as variation in relevant skills in the general population. As well as gene discovery using the latest genomic methods, my team investigates functional impacts of gene disruption in a range of model systems, most recently including human brain organoids.

Assoc.Prof. Marcela Buchtová

Institute of Animal Physiology and Genetics,
Academy of Sciences of the Czech Republic, Czech Republic

My background is in the field of normal and pathological morphology. Our lab focuses on morphogenetic processes in organogenesis with special interest in formation of craniofacial structures and limb patterning. Physiological aspects at molecular and cellular levels are investigated and related developmental disorders are examined. Recent effort of our lab is also to contribute to recent knowledge in basic and biomedical research with links to practical applications.

Dr. Ismael Galve-Roperh

Department of Biochemistry and Molecular Biology,
Complutense University of Madrid, CIBERNED, Spain

Our group is focused on the understanding of the molecular mechanisms responsible of cortical development and their regulation. We contributed to the field unraveling the neurodevelopmental role of endocannabinoid signalling in prenatal brain development, that acts as extracellular signalling pathway via CB1 receptors modulating the activity of neurogenic transcriptional program including SATB2.

We are interested to develop novel studies on the consequences of regulating endocannabinoid signalling in neurodevelopmental disorders, such as the SATB2 syndrome characterized by unbalanced generation of layer specific projection neuron populations.

Dr. Luigi Boccuto

School of Nursing, CBSHS, Healthcare Genetics Interdisciplinary Doctoral Program, Clemson, USA

My background is in clinical genetics. My work at the School of Nursing lab (Clemson University) is focused on the characterization of genotype-phenotype correlations in genetic disorders. We study metabolic profiles of cells from individuals with genetic conditions in order to investigate metabolic pathways involved in pathogenic mechanisms, identify potential biomarkers, identify molecular targets for treatment approaches, and assess the efficacy and side effects of candidate drugs.

We are interested in collaborating with clinical centers to collect information on phenotypes, biospecimens for functional studies, and eventually develop novel treatment approaches.

Prof. Victor Tarabykin

Institute of Cell Biology and Neurobiology,
Charité – Universitätsmedizin Berlin, Germany

My background is in molecular biology and mouse genetics. My lab focuses on study genetic basis of the cerebral cortex development. We are interested in identifying genes whose disruption causes brain malformation and investigating molecular functions of proteins encoded by such genes.

Dalal Dawood Baumgartner

Living in Sydney Australia, I am a mother to my beautiful 7-year-old SAS girl, Naomi and an advocate to the SATB2 cause.
Naomi was diagnosed with SATB2 in October 2017

Naomi is an ambassador for all SATB2 children, old and young and will continue to be a beacon of light for future research, awareness and professional education. Throughout her young life she has faced many struggles; surgery, invasive testing, communication barriers with family and educators, health issues, limitations to diet, insomnia and many behavioural rollercoasters, but there is nothing that will dim her smile or light. She is my inspiration and my motivation. Our SAS community will continue to fight and learn for our children and we will become better at it with the support of our researchers and their efforts.

I wouldn't change you for the world, but I would change the world for you.

(Follow Naomi's story on Hope with SATB2 at Facebook)

Dr. Jorge Múnera

Department of Regenerative Medicine and Cell Biology, Medical University of South Carolina, USA.

I have B.S. in Biology from the University of Illinois and a PhD in Molecular Pathology from the University of California San Diego. My expertise is in colonic stem cells and colorectal cancer. I work with colorectal cancer cell lines and human pluripotent stem cell derived colonic organoids.

I am currently examining the transcriptional and proteomic targets of SATB2 in colorectal cancer cell lines and human pluripotent stem cell derived colonic organoids using CRISPR-Cas9 mediated gene editing and inducible overexpression of SATB2.

Lauren Shelley

Aston University, Birmingham, UK

Lauren is a Doctoral Researcher within the Cerebra Network for Neurodevelopmental Disorders. Her research is focused on behaviours that challenge in SATB2-associated syndrome, with an overarching aim to improve understanding of factors that may be implicated in the development and maintenance of these behaviours.

Univ.-Prof. Dr. Georg Dechant

Institute for Neuroscience, Medical University of Innsbruck, Austria

As a basic researcher I am interested in the molecular and cellular mechanisms of cognitive functions and dysfunctions. My laboratory combines work on transgenic animals as well as human IPSC-based models in order to investigate how experiences influence gene expression in cortical and hippocampal neurons. We have previously shown that SATB2 in the adult forebrain is necessary for memory consolidation.

Dr. Galina Apostolova

Institute for Neuroscience, Medical University of Innsbruck, Austria

My background is in molecular neuroscience. My research contributes to expanding and improving our understanding of the pathogenesis of neuropsychiatric and neurodevelopmental disorders. My research objective is to enable the interpretation of the neurobiological impact of genetic risk loci by making use of functional genomic approaches and experimental validation in relevant in vivo or in vitro model systems.

My current research work is focused on elucidating the molecular functions of SATB2 in the brain. By employing novel conditional knockout lines, we previously demonstrated an important role of SATB2 in the adult mouse CNS as a determinant of long-term memory. In collaboration with Dr. D.W. Morris (National University of Ireland, Galway) we have also recently shown that common variation in the genes regulated by SATB2 or encoding SATB2-interacting proteins influences human cognitive ability and contributes to schizophrenia. We are currently investigating the impact of SATB2-directed three-dimensional nuclear architecture and interactions with nuclear lamina on transcriptional processes that underpin complex behavior. By acutely re-expressing SATB2 in the hippocampus of Satb2 knock-out mice and rescuing long-term memory, we have provided evidence for SATB2 being a candidate gene supporting cognition.

Dr. Jennifer Fish

Department of Biological Sciences,
University of Massachusetts Lowell

My background is in developmental biology and genetics with a focus on craniofacial and bone development.

Dr. Joe Zhou

Division of Regenerative Medicine, Department of Medicine, Weill Medical College of Cornell University, NY, USA

Dr. Zhou received his PhD training in neuroscience at the California Institute of Technology. He then focused on studies of pancreatic islet regeneration first as a postdoctoral fellow, then as a faculty member at Harvard University where he became Associate Professor before moving to Weill Cornell Medicine. His laboratory in New York studies pancreatic islet and intestinal mucosa regeneration. Pertinent to SAS, his lab investigates the role of SATB2 in controlling colonic mucosal biology relevant to diseases including SAS and inflammatory bowel diseases.

Dr. Yuri Zarate

Section of Genetics and Metabolism, University of Arkansas for Medical Sciences, Arkansas Children's Hospital, Little Rock, USA

I am a clinical geneticist and I'm interested in craniofacial disorders. I lead the only SAS multidisciplinary clinic in the US and have evaluated dozens of patients with SAS. I have participated in different research SAS-related efforts for the last several years and helped delineate in greater detail the phenotype.

Lina Granqvist
Åsa Dybeck Alvinge

Senior consultants in paediatric dentistry
Eastman Institute in Stockholm, Sweden

We are clinical working dentists, specialized in paediatric dentistry. The best we know is to meet and help children with usual and unusual syndromes and SATB2 has become our special interest in daily work. We are planning to write a case series report where we will look at oral- and orofacial manifestations caused by SATB2 syndrome. We hope that this project will grow and thus we want to connect with other clinicians and researchers worldwide to collaborate, so that we better can treat these lovely children.

Some of our questions are:
-What do you know about oral manifestations associated with this syndrome?
-How can professionals like us work together optimally to treat these children who have oral and craniofacial symptoms?

Our ambition is to compile all the oral manifestations that are associated with the syndrome so that we better can understand and treat them. We also want to examine all SAS patients in the Stockholm area and do standardized oral examinations over time.

Hope to hear from you and exchange information and thoughts!
Lina Granqvist lina.granqvist@regionstockholm.se & Åsa Dybeck Alvinge

See you at the next roundtable!