Welcome to the 1st GRI-UK Conference

Bringing UK families and science together for the first time

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Institute for Neuroscience and Cardiovascular Research

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The Patrick Wild Centre

WELCOME TO THE INAUGURAL GRI-UK CONFERENCE

Thank you for being here to share in this special and significant event. Just two years ago, GRI-UK was founded by parents and grandparents to build a strong, supportive community across the UK, and raise awareness of GRI disorders in our efforts to find better treatments and cures.

This conference marks an important milestone for us, and we are delighted to see the scientists advancing research and families living with GRI disorders under one roof. Over the coming days, we hope you will connect, listen, and collaborate.

We're deeply grateful to Professor David Wyllie and his team at the Institute for Neuroscience and Cardiovascular Research at the University of Edinburgh for making this conference possible.

We would also like to extend our sincere thanks to our sponsors – GRIN Therapeutics, Scrumconnect, Simons Initiative for the Developing Brain, Simons Foundation, Edinburgh Medical School 300 and the Patrick Wild Centre – whose generous contributions have allowed us to make attendance at the event free, and our Trustee Colette Dufficy for designing all the wonderful artwork.

Thank you for being part of our growing community. We hope your conversations inspire new ideas, strengthen partnerships, and bring fresh momentum to the work ahead. By combining scientific and clinical expertise with lived experience of GRI disorders, we hope to accelerate progress in ways that none of us could achieve alone.

Alyson, Jillian, Toni and the GRI-UK Team

Welcome to the inaugural GRI-UK Conference. The origins of this event began at the GRIN Europe meeting held in Barcelona in November 2024 where, after a few beers, I was persuaded to host a meeting for GRI-UK. The ambition was to provide an opportunity for the GRI community to meet, interact and learn from each other. The key element would be a need for "family presentations" in addition to traditional "research presentations" so that everyone could gain knowledge and understanding, whether a parent, grandparent, sibling or friend of a GRI child or a researcher at an early-stage in their career or, like me, someone who has studied glutamate receptors for over 35 years. I very much hope that the programme fulfils this remit.

I am hugely indebted to my research colleagues who have come for near and far to showcase how all of us working in this research area seek knowledge that will ultimately lead to therapeutic benefit. Indeed, I had no concept when starting out all those years ago and very much focused on "discovery research" that insights from this type of work would shape our understanding of what underlies the devastating consequences of mutations in GRI receptor subunits. It is the combination of discovery, translation and clinical research that will allow us to understand GRI, and many other disorders.

I, too, am deeply appreciative of the very generous support we have received from all our sponsors. I'd also like to acknowledge the efforts of so many who have brought this GRI-UK Conference to fruition. It has been a delight to work with GRI-UK Trustees; their drive and passion is a reminder to all of us of what needs to be central to our research ambitions. Huge thanks to Jess Lonsdale who complied the glossary to demystify terminologies for our GRI community. Special thanks to our Professional Services Staff and Technical Support Teams who tolerated my requests for help and assistance, with particular thanks to Laura Thomas, James Griffiths, Krisztina Vinko and Mike Kerr for their outstanding support.

I very much hope you enjoy GRI-UK 2026 and have time to explore the beautiful city of Edinburgh!

David Wyllie
co-Director, Institute for Neuroscience and Cardiovascular Research, University of Edinburgh

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PROGRAMME

Research presentations – Session 1

Thursday 14 May

Anatomy Lecture Theatre, Old Medical School, Teviot Place, Edinburgh EH8 9AG

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Session Chair: David Wyllie

12:30-13:10	**Lunch available – Teaching Rooms 8 and 9**		
13:15-13:30	**Welcome address**	**GRI-UK and David Wyllie**	
13:30-14:00	**Research talk**	**Lonnie Wollmuth**	**Stony Brook University, USA**
	Calcium signalling and NMDA receptor variants		
14:00-14:00	**Research talk**	**Katherine Roche**	**NIH, Bethesda, USA**
	NMDA receptor expression trafficking and *GRIN* disorders		
14:30-15:00	**Research talk**	**Graham Collingridge**	**University of Toronto, Canada**
	Characterisation of disease-relevant *GRIN1* variant mouse models		
15:00-15:30	**Coffee and tea break – Teaching Rooms 8 and 9**		
15:30-16:00	**Research talk**	**Derek Bowie**	**McGill University, Canada**
	Restoring inhibitory synapse plasticity in intellectual disability		
16:00-16:30	**Research talk**	**Emma Wood**	**University of Edinburgh, UK**
	Characterisation of hippocampal function in a rat model of *GRIN2B*-related neurodevelopmental disorder		
16:30-18:00	**Posters (Teaching Room 7) and drinks reception**		

See **Further details – access to venues** at end of the programme showing location of the Anatomy Lecture Theatre. Enter via Doorway 3 and take the stairs on the left to the first floor.

Research presentations – Session 2

Friday 15 May

Anatomy Lecture Theatre, Old Medical School, Teviot Place, Edinburgh EH8 9AG

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Session Chair: Katherine Roche

09:00-09:25	**Coffee and tea available in Teaching Rooms 8 and 9**		
09:30-10:00	**Research talk**	**Hongjie Yuan**	**Emory University, USA**
	Truncating *GRIN2B* variants: molecular insights and behavioural deficits		
10:00-10:30	**Research talk**	**Carole Torsney**	**University of Edinburgh, UK**
	Characterisation of tactile and pain phenotypes in a rat model of *GRIN2B* haploinsufficiency		
10:30-11:00	**Research talk**	**Ingo Greger**	**LMB Cambridge, UK**
	Structure and bi-functional role of a peripheral AMPA receptor auxiliary subunit		
11:00-11:30	**Research talk**	**Jelena Baranovic**	**University of Edinburgh, UK**
	Mechanism and subunit-dependence of disease-causing Lurcher mutation in AMPA-type glutamate receptors		
11:30-11:45	**Closing discussion**		

See **Further details – access to venues** at end of the programme showing location of the Anatomy Lecture Theatre. Enter via Doorway 3 and take the stairs on the left to the first floor.

Family presentations – Session 1

Friday 15 May

Lecture Theatre, Hugh Robson Building, George Square, Edinburgh EH8 9XD

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Session Chair: Ian Coombs

12:15-13:15	**Lunch available – Hugh Robson Common Room**		
13:15-13:30	**Welcome address**	**GRI-UK and David Wyllie**	
13:30-14:05	**Family presentation**	**Johannes Lemke and Maike Karnstedt**	**University of Leipzig, Germany**
	The global *GRIN* registry: advancing genotype-phenotype correlations and therapeutic insights		
14:05-14:30	**Family presentation**	**Stephen Traynelis**	**Emory University, USA**
	Functional analysis of *GRIN* variants: how does it work and what does it tell us?		
14:30-14:55	**Family presentation**	**Sameer Zuberi**	**University of Glasgow, UK**
	Participating in research trials		
14:55-15:25	**Coffee and tea break – Hugh Robson Common Room**		

Session Chair: Stephen Traynelis

15:25-15:50	**Family presentation**	**David Wyllie**	**University of Edinburgh, UK**
	Receptors, synapses and signalling in the brain		
15:50-16:15	**Family presentation**	**Andy Penn**	**University of Sussex, UK**
	Probing the consequences of *GRIN2* variants on signal transmission between neurons		
16:15-16:40	**Family presentation**	**Julia Vitarello**	**Mila's Miracle Foundation**
	Mila to Millions: a new era of individualised medicines		
16:40-17:05	**Family presentation**	**Bruce Leuchter, Hillary Savoie and Anne-Marie Li-Kwai-Cheung**	**GRIN Therapeutics**
	Developing Investigational Radiprodil for Targeted Treatment of GRIN-Related Neurodevelopmental Disorder (GRIN-NDD)		
17:05-17:20	**Closing discussion**		

Social event - Ceilidh

Friday 15 May, 18:45-21:00
Binks Hall, 138-140 Pleasance, Edinburgh, EH8 9RR

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Family presentations – Session 2

Saturday 16 May
Lecture Theatre, Hugh Robson Building, George Square, Edinburgh EH8 9XD

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Session Chair: Jillian Hastings Ward

09:30-10:00	**Coffee and tea available – Hugh Robson Common Room**		
10:00-10:10	**Welcome address**	**GRI-UK and David Wyllie**	
10:10-10:25	**Family presentation**	**Alyson Koopman**	**GRI-UK**
	GRI-UK: How we got here, who we're here for, and where we're going		
10:25-10:40	**Family presentation**	**Keith McArthur and Lauren Williams**	**CureGRIN**
	The Global View: CureGRIN, the GRI Census and the 'How to Treat and Cure GRI Disorders Guide'		
10:40-10:55	**Family presentation**	**Gilianne Nelissen**	**GRIN Europe**
	GRIN Europe's initiatives to advance research, patient care and family support		
10:55-11:05	**Discussion / Q&A with these patient advocacy groups**		
11:05-11:30	**Family presentation**	**Danielle Veenma**	**Erasmus MC, The Netherlands**
	The Dutch Brain Lab Facility: adult GRI patients as a window into natural history and trial outcome measures across the lifespan		
11:30-11:55	**Family presentation**	**Narjes Rohani**	**University of Oxford, UK**
	Genes, Brains, and Breakthroughs: carer-centred accessible educational videos on neurodevelopmental conditions and therapy		
11:55-12:20	**Family presentation**	**Ian Coombs**	**University College London, UK**
	Loss- or gain-of function in GRIA disorder: why it matters and how we find out		
12:20-13:30	**Lunch – Hugh Robson Common Room**		

Family presentations – Session 3

Saturday 16 May

Lecture Theatre, Hugh Robson Building, George Square, Edinburgh EH8 9XD

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Session Chair: Lindsay Mizen

13:30-13:55	**Family presentation**	**Lonnie Wollmuth**	**Stony Brook University, USA**
	Those beautiful but complicated NMDA receptors		
13:55-14:20	**Family presentation**	**Katherine Roche**	**NIH, Bethesda, USA**
	An update on mouse models to study *GRIN* disorders		
14:20-14:45	**Family presentation**	**Graham Collingridge**	**University of Toronto, Canada**
	Characterisation of disease-relevant *GRIN1* variant mouse models		
14:45-15:10	**Family presentation**	**Carole Torsney**	**University of Edinburgh, UK**
	Understanding sensory processing in a rat model of *GRIN2B* haploinsufficiency		
15:10-15:45	**Coffee and tea break – Hugh Robson Common Room**		

Session Chair: Emma Wood

15:45-16:10	**Family presentation**	**Alfredo Gonzalez-Sulser**	**University of Edinburgh, UK**
	Sleep and brain activity in a rat model of *GRIN2B* disorder		
16:10-16:35	**Family presentation**	**Derek Bowie**	**McGill University, Canada**
	Finding common ground in intellectual disability		
16:35-17:00	**Family presentation**	**Peter Kind**	**University of Edinburgh, UK**
	Convergence in neurodevelopmental disorders		
17:00-17:15	**Closing discussion**		

Social event – Garden Party

Saturday 16 May, 17:15-19:00

George Square Gardens, George Square, Edinburgh EH8 9XD

what3words: ///lower.views.gifts

Family fun in Edinburgh

Sunday 17 May

Some suggestions of places to visit or activities to do:

Places to visit

- Edinburgh Castle Esplanade – great views over the city and beyond (because it's nearly at the top of the Royal Mile)
- Walk down the Royal Mile, from the castle esplanade to Holyrood (Parliament & Palace) (some cobbled sections)

Free entry

- National Museum of Scotland, Chambers Street – big central museum with lots of interactive exhibits, very child friendly, Changing Place - National Museum of Scotland | National Museums Scotland
- National Art Galleries (4 sites) – all free to enter; pay for some exhibitions -National Galleries of Scotland
- Museum of Childhood, Royal Mile - Museum of Childhood | Culture Edinburgh

Turn up and pay

- City sightseeing bus tours - Edinburgh Bus Tours: Open Top Hop-On Hop-Off | City Sightseeing©

Advised to book well ahead

- Edinburgh Castle - Official Edinburgh Castle Website
- Dynamic Earth, bottom of the Royal Mile – Dynamic Earth | Edinburgh's world-class science centre and planetarium
- Holyrood Palace, bottom of the Royal Mile - Palace of Holyroodhouse

Classic Edinburgh sights (but not good for wheelchairs / limited mobility)

- Edinburgh Dungeon - Best Edinburgh Attractions | The Edinburgh Dungeon (not sensory friendly, only 1 wheelchair allowed at once)
- Camera Obscura - Camera Obscura and World of Illusions Edinburgh (historic 5-storey building, no lift)
- Edinburgh Zoo - Welcome to Edinburgh Zoo | Edinburgh Zoo (on a steep hillside)

Poster Presentations – Teaching Room 7, Teviot Building

GluA3 AMPA Receptors: Structure, biogenesis and disease-association
Authors: Josip Ivica, Aditya Pokharna, Imogen Stockwell and Ingo Greger
Laboratory for Molecular Biology, Cambridge

Charge at the Q/R editing site shapes Gria2 biogenesis and synaptic transmission
Authors: Sengupta N., Ivica I., Dou X., Cais O., Zhang D., Krieger J.M., Wu H., Hegde R.S., Greger I.H.
Laboratory for Molecular Biology, Cambridge

Distinct targeting of GluN2D: implications in disease
Authors: Detlef Vullhorst and Katherine W. Roche
National Institute of Neurological Disorders and Stroke, NIH, Bethesda, MD, USA

Disease-associated variants destabilize GluN2B NMDA receptors by disrupting the PDZ binding motif
Authors: Hye Young Ryu[1], Marta Vieira[1], Saerom Lee[1] Sehoon Won[1], Kai Chang[1], John D Badger 2nd[1], Shuxi Liu[2], Colin L Sweeney[3], Unimook Choi[3], and Katherine W. Roche[1]
[1]Receptor Biology Section, National Institute of Neurological Disorders and Stroke, NIH, Bethesda, MD
[2]Clinical Genetics Department, GeneDx, 207 Perry Parkway, Gaithersburg, MD
[3]Genetic Immunotherapy Section, National Institute of Allergy and Infectious Diseases, National Institutes of Health, Bethesda, Maryland, USA

Potential advantages of endogenous neurosteroids in the treatment of GRIN disorders
Authors: Abdel Rahman FE[1,3], Korinek M[1], Kysilov B[1,4], Hrcka Krausova B[1], Vyklicky V[1], Vyklicky L[1], Balik A[1,2] and Smejkalova T[1]
[1] Institute of Physiology, Czech Academy of Sciences, Videnska 1083, 142 00 Prague 4, Czech Republic [2] Institute of Physiology, Czech Academy of Sciences, BIOCEV, Prumyslova 595, 252 50 Vestec, Czech Republic [3] Faculty of Science, Charles University, Albertov 2038, 128 00 Prague 2, Czech Republic [4] Present Address: Stony Brook University, Stony Brook, 100 Nicolls Road, NY 11794, USA

Spikeling: An open-source hardware implementation of spiking neurons for neuroscience teaching
Authors: Paul Rignanese[1], Artemis Koumoundourou[1,2,3] Lucia Zanetti[1,4] Maxime Zimmermann[1,5]
[1]OpenSourceNeuro, Paris, France, [2]VIB-KU Leuven Center for Neuroscience, Belgium, [3]TreND in Africa, Kenya, [4]SISSA Trieste, Italy, [5]University of Sussex, UK

Characterising region- and sex-specific synaptic plasticity in *Grin2b*$^{+/-}$ rats
Authors: Lysandra Fryer-Petridis, Daniel W. Hunter, David J. A. Wyllie, Emma R. Wood
Institute for Neuroscience and Cardiovascular Research and Simons Initiative for the Developing Brain, University of Edinburgh, Edinburgh, UK

Optogenetic Stimulation to Modulate Absence Seizures in GRIN2B Knockout Rats -
Authors: Mehmet Taskiran, Alfredo Gonzalez-Sulser
Institute for Neuroscience and Cardiovascular Research and Simons Initiative for the Developing Brain, University of Edinburgh, Edinburgh, United Kingdom

Diverging seizure properties in rat models of GRIN2B and SYNGAP1 neurodevelopmental disorders

Authors: Natalie Ling Sum Hung, Alfredo Gonzalez-Sulser

Institute for Neuroscience and Cardiovascular Research and Simons Initiative for the Developing Brain, University of Edinburgh, Edinburgh, UK

Characterisation of pain and tactile phenotypes in a rat model of *Grin2b* haploinsufficiency

Authors: Ying Sze, Ganre Akpubi, Charlotte Westlake, Carole Torsney

Institute for Neuroscience and Cardiovascular Research and Simons Initiative for the Developing Brain, University of Edinburgh, Edinburgh, UK

Loss of the NMDA Receptor Subunit GluN2A Selectively Alters Synaptic Integration in CA1 Somatostatin Interneurons

Authors: Daniel W. Hunter, Peter C. Kind, David J.A. Wyllie

Institute for Neuroscience and Cardiovascular Research and Simons Initiative for the Developing Brain, University of Edinburgh, Edinburgh, UK

Absence seizures in a rat model of GRIN2B neurodevelopmental disorder.

Authors: Emma Perkins, Katerina Hristova, Darren Walsh, Alfredo Gonzalez-Sulser, Peter Kind

Institute for Neuroscience and Cardiovascular Research and Simons Initiative for the Developing Brain, University of Edinburgh, Edinburgh, UK

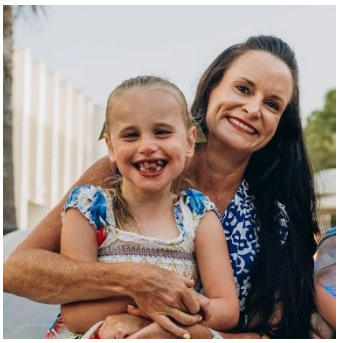
Activation of AMPA-type glutamate neuroreceptors in health and disease

Authors: Alexander Edwards[1], Chigdem Arslan[1], David Wyllie[2], and Jelena Baranovic[1]

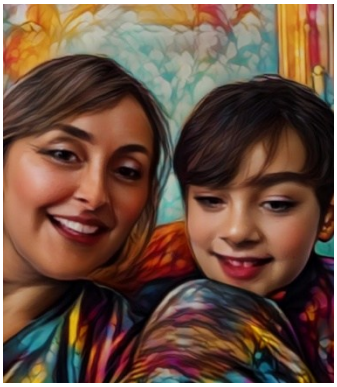
[1]Institute of Quantitative Biology, Biochemistry and Biotechnology, University of Edinburgh, Edinburgh, UK, [2]Institute for Neuroscience and Cardiovascular Research and Simons Initiative for the Developing Brain, University of Edinburgh, Edinburgh, UK

Biographies of Speakers and Conference Team

Patient Advocates

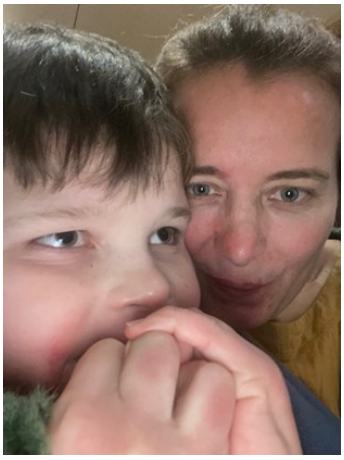


Toni Clarke, mother to Solana, who has a GRIN2A variant, serves as the Vice-Chair of GRI-UK. She has a professional background in finance and previously managed her own small business, having stepped away from the corporate sector to support her life with Solana. Toni is deeply passionate about sport and has undertaken numerous challenging trail races to raise essential funds for GRI-UK. She brings a dynamic and energetic presence to the charity and remains committed to pursuing ambitious fundraising initiatives to support GRI-UK's mission of advancing research toward treatments and, ultimately, a cure for all GRI disorders.



Colette Dufficy I am a software engineer and board member of GRI UK, a charity supporting families affected by GRI-related conditions. With a background in graphic design, I bring a creative approach to the charity's communications and events. As a mum to Aaron who has a GRIN2B variation, I'm passionate about connecting families, clinicians, and researchers, and helping to ensure that no parent or carer feels alone on their journey.

Jillian Hastings Ward I am a patient advocate and mother of Sam (12), who has a GRIN1 disorder and is profoundly disabled. I was recently appointed as Co-Chair of GRI-UK and have helped to put this conference together. I'm also currently studying to become a Fellow of the European Patients' Academy on Therapeutic Innovation (EUPATI). I've been active in the GRI community since Sam's diagnosis in 2017, and was a founding trustee of CureGRIN. Between 2016 and 2024, I was a member and later Chair of the Participant Panel at Genomics England. I've published articles in Nature Medicine and the European Journal of Human Genetics, and I'm about to lead a re-run of the massive open online course (MOOC) I was commissioned to produce for Wellcome Connecting Science in 2025, 'The Power of Patient Advocacy in Genomics'. I've spoken at conferences around the world, and am delighted to help bring this one to my home town!



Alyson Koopman Being mum to Evan led to my involvement with GRI-UK, where I became Chair at the start of 2024. As we continue to grow, I now share this responsibility as Co-Chair with Jillian. Alongside my role with GRI-UK – where we have worked hard to bring together families affected by GRI disorders across the UK, raise the organisation's profile within the rare disease landscape, and develop the foundations of a sustainable organisation – I work as a veterinary practice administrator and are a mum and wife. Managing a busy household comes on top of caring for Evan who has GRIN1 and profound and complex needs and disabilities. Prior to joining GRI-UK, I spent 20 years as a Humanitarian Child Protection Advisor with Save the Children, the International Labour Organization, and UNICEF. In these roles, I worked in challenging environments to design and implement programmes supporting vulnerable children and families affected by crisis, advocate for policy change, and develop tools to reduce risks and improve outcomes. I bring a diverse skill set grounded in both strategic and practical leadership in complex contexts, alongside lived experience as a caregiver. I am deeply committed to building a strong, supportive community, raising awareness, and improving support for families across the UK.

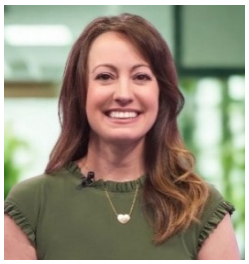
Keith McArthur I am Executive Director of CureGRIN Foundation, where I lead global efforts to accelerate research and drive treatments and cures for GRI Disorders. I have a wealth of senior leadership experience from the business and non-profit sectors, including a career as a newspaper reporter, author and communications executive. I graduated with a Master's degree in Journalism from the University of Western Ontario. It was my son Bryson's diagnosis with GRIN1 disorder in 2015 that triggered my commitment to the science of ionotropic glutamate receptors. In 2018, my wife and I organized a GRI family conference where the consensus from the world's top researchers—that a cure for GRI Disorder is possible—led me to commit my life to this mission. My current interests in glutamatergic synaptic function span the full spectrum of genes that encode these receptors, including GRIN, GRIA, GRIK, and GRID. To advance this field, I oversaw the development and launch of the GRI Census to better understand our global patient population and drafted the "How to Develop Treatments and Cures for GRI Disorders Guide" to provide a strategic roadmap for biopharma and the GRI community. My vision is to drive an integrated, patient-led partnership with families, researchers, clinicians, and industry, with the ultimate goal of finding treatments and cures for all those living with GRI Disorders.

Gilianne Nelissen I graduated from Maastricht University in 1996 with a master's degree in tax law, specializing in European and international tax law. For the next two decades, I worked at leading tax firms, focusing on cross-border taxation, social security, and labour-related emigration law. In my wildest dreams, I never imagined becoming involved in rare disease advocacy. That changed when our eldest daughter, Charlotte, was diagnosed at age 8 with GRIN2B neurodevelopmental disorder. The journey to her diagnosis and to securing appropriate care and support was long, challenging, and often very lonely, clearly exposing the lack of awareness and support for families like ours. Motivated by this experience, my husband Nino and I, together with six other European GRIN parents, co-founded GRIN Europe in 2019, where I currently still serve as board member and Community Liaison. GRIN Europe is dedicated to improving the lives of individuals affected by GRIN-related neurodevelopmental disorders and their families. We promote research, collaboration, and advocacy to improve diagnosis, access to care, and the development of effective treatments, while connecting families, clinicians, researchers, and industry partners across borders. Additionally, in my home country of the Netherlands, I founded GRIN in Nederland, an informal organisation for Dutch and Belgian GRIN families, focused on peer support and on sharing knowledge about GRIN disorders in the broadest sense.

Hillary Savoie PhD, is Head of Advocacy, Director of Community Engagement and Communication at Neurvati Neurosciences and GRIN Therapeutics. She is an internationally recognized leader in patient advocacy in the rare disease community, with extensive experience as a rare disease advocate, patient engagement consultant, and writer on patient advocacy issues. She has a deep personal understanding of the role of patient advocacy as the mother of daughter Esmé, who is medically and developmentally complex. She is the founder of The Cute Syndrome Foundation (TSCF), an advocacy organization focused on support for individuals and families affected by a rare genetic and developmental encephalopathy. She served as Executive Director at TCSF for nine years and now serves on the organization's board of directors. Her essays and reported features focused on life with medically complex children have been published in news outlets including Romper.com, New York Times, Rolling Stone, and the podcast Story Collider, among others. She holds a doctorate in Communication and Rhetoric from Rensselaer Polytechnic Institute (RPI).

Sandra Silva-Arrieta I am the mother of a 17-year-old girl with a GRIN2B-related disorder. I am a co-founder of GRIN Europe (formerly GRIN2B Europe) and the Spanish GRINpatías association, and for the past eight years have helped coordinate the European GRIN conferences. I hold a doctorate in veterinary medicine and surgery from Tufts University in the USA, and from 2011-2023 I have worked in biomedical research, with a focus on infectious diseases and cancer. Over the past three years, I have served as an active ePAG member within ERN EpiCARE and, together with fellow ePAGs, co-founded CREA-E+, an alliance of associations supporting patients and families affected by developmental epileptic encephalopathies (DEEs). Alongside my biomedical consulting work, I am a passionate patient advocate and have helped raise awareness of GRIN disorders and other DEEs at international conferences—speaking to clinicians, researchers, and pharmaceutical companies about the unmet needs of patients and caregivers living with rare and complex neurodevelopmental disorders. This past year, I have also participated with GRIN experts in two ERDERA grants and one EU Horizon grant, which aim to expand treatment options and improve understanding of the non-seizure manifestations affecting our children.

Julia Vitarello is Mila's Mom and founder of Mila's Miracle Foundation. In 2016, Julia's daughter, Mila, was diagnosed with a rare fatal genetic condition called Batten disease. In a race against time, Julia drove an unprecedented collaboration between scientists, clinicians and regulators which led to the development of her daughter's treatment, milasen—the first ever drug tailored to a single person. Since then, Julia has led an international movement to get from 'Mila to Millions,' turning her daughter's ground-breaking story into an entirely new field of individualized medicines. Julia speaks regularly around the world, engaging academics, biotechs, governments, regulators, payers and families alike.



Lauren Williams brings her passion for personal growth and most importantly her son to the team of CureGRIN. Lauren has 15 years public relations and fundraising experience, 5 years of that with an international non-profit organization. She is a graduate of Louisiana Partners in Policymaking, a national leadership training program with the goal of creating effective systems change agents. Lauren was also appointed to the Louisiana Rare Disease Council in 2022, advocating on behalf of Louisianan's living with rare diseases. Lauren's love for her son, Carter, who was born with a mutation of the GRIN1 gene, has ignited a new ambition and purpose in life…finding a cure for GRIN.

Researchers

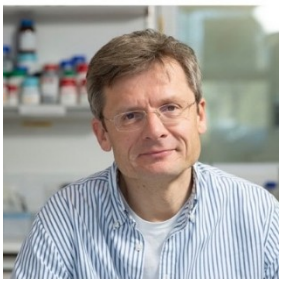
Jelena Baranovic I am a lecturer in biochemistry at the School of Biological Sciences at the University of Edinburgh. After my undergraduate studies in molecular biology at the University of Zagreb, Croatia, I did my PhD at the Department of Physics at the University of Oxford. In 2012, I went on to do my postdoctoral research in the lab of Prof. Andrew Plested at the Leibniz Institute for Molecular Pharmacology in Berlin, followed by the lectureship in Edinburgh. My whole scientific career revolves around AMPA-type glutamate receptors. I want to understand the intricate, molecular details of their activity, how it is changed by mutations and drugs and how we can control it. As part of this, I study peptides produced by predatory sea snails that specifically target AMPA receptors. I believe this vast, unexplored, nature's pharmacy holds great potential to develop new modulators of AMPA receptors, that can advance AMPA receptor biology and inspire new drugs.

Derek Bowie is the Endowed University Chair in Medicine at McGill University and Co-Director of the Cell Information Systems. His laboratory focuses on understanding the building blocks of the mammalian brain, with an emphasis on several major ion channel families, including ionotropic glutamate receptors. His research examines the role of these ion channels in autism and intellectual disability. In 2022, he co-founded Nospharma, a preclinical- and clinical-stage therapeutics company developing innovative treatments for rare genetic neurological disorders. Dr. Bowie earned his Ph.D. from the University of London after completing his undergraduate studies at the University of Strathclyde in Scotland. He subsequently undertook postdoctoral training in France (Université Louis Pasteur), Switzerland (Universität Zürich), and the United States (National Institutes of Health). He held a faculty position at Emory University before joining McGill. He has served as a Visiting Professor at the National Institute for Physiological Sciences in Okazaki, Japan, and is a recipient of a Canada Research Chair award. He also serves on numerous national and international advisory panels and editorial boards and is currently a member of the Society for Neuroscience Government and Public Affairs Committee as the Canadian representative.

Graham L. Collingridge, FRS, CBE, graduated with a BSc in Pharmacology at the University of Bristol (UK) in 1977. After postdoctoral positions in Vancouver (Canada) and Sydney (Australia) he returned to Bristol in 1983 to become a lecturer (Assistant Professor) in the Department of Pharmacology. From 1990 to 1994 he was the Chair of Pharmacology at the University of Birmingham (UK), and in 1994 he became a Professor of Neuroscience in Anatomy at the University of Bristol (currently Professor Emeritus). He served as Chair of the Department of Anatomy from 1997 to 1999 and then became the founding Director of the MRC Centre for Synaptic Plasticity from 1999 to 2012. In 2016 he was a co-recipient of The Brain Prize (with Tim Bliss FRS and Richard Morris FRS, CBE), and in 2019 received a CBE for services to Biomedical Sciences. Collingridge is currently the Director of the Tanz Centre for Research in Neurodegenerative Diseases, the inaugural holder of the Krembil Family Chair of Alzheimer's Research, and an Affiliated Scientist at the Krembil Research Institute. He is also a Professor in the Department of Physiology at the University of Toronto, Canada, and a Senior Investigator at the Lunenfeld-Tanenbaum Research Institute at Mount Sinai Hospital, Toronto, Canada.

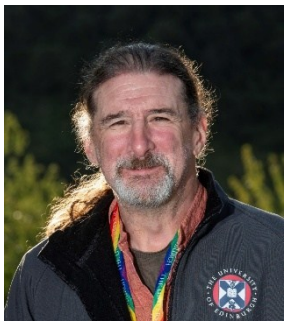
Ian Coombs is a Lecturer in Pharmacology at University College London. He got his degree in Biochemistry and a PhD in Biophysics from Imperial College. Following his doctoral studies, he moved to UCL to investigate the molecular pharmacology of AMPA-type glutamate receptors. Since 2021, his research has focused on GRIA disorders, studying how *de novo* genetic variants in the GRIA genes alter AMPA receptor function and endeavouring to identify targeted treatments to counteract the impact of these variants. His laboratory additionally provides hands-on advanced pharmacology training for third-year pharmacology students with a focus on the electrophysiological characterisation of previously unstudied *GRIA1-4* variants associated with GRIA disorder.

Alfredo Gonzalez-Sulser I am a researcher in the Institute of Neuroscience and Cardiovascular Research focused on rodent models of epilepsy and neurodevelopmental disorders. I studied Biology at the University of Pennsylvania and then undertook my PhD in Neuroscience at Georgetown University in Washington DC. I have been focused on epilepsy for a long time but, as the rest of my field, I am focusing more and more on epilepsy rodent models with genetic aetiologies similar to those seen in patients. This has led to overlapping interests and multiple collaborations with the Simons Initiative for the Developing Brain (SIDB). Neurodevelopmental disorders studied by the SIDB result in severe autism and intellectual disability often are comorbid with epilepsy. My lab specializes in EEG recordings in rodents, where in collaboration with engineers and informaticians we have developed advance analyses including artificial intelligence algorithms for automated seizure detection, sleep scoring and biomarker identification.



Ingo Greger I am a Senior MRC Investigator at the MRC Laboratory of Molecular Biology in Cambridge, UK. Prior to this, I conducted postdoctoral research at the NYU School of Medicine, HHMI, USA (1999-2003). I graduated with a D.Phil. from the University of Oxford, UK (1994-1998), followed by a short post-doc there (1998-1999). My interest in glutamate-gated ion channels and their central role in learning and memory emerged after my Ph.D, during my post-doc, and developed throughout my early years as an independent group leader at the LMB, Neurobiology Division. Our research primarily focuses on the AMPA-subtype, encompassing its biogenesis, molecular structure as well as its supramolecular organisation and regulation at glutamatergic synapses.

Maike Karnstedt I studied Biology for my Bachelor's degree at the University of Mainz and the University of Göttingen from 2012 to 2016. I then completed my Master's degree in Biodiversity, Biology and Evolution between 2016 and 2018, with a specialization in forensic biology. After graduation, I worked for one year at Evotec in Göttingen, focusing on transcriptomics and RNA sequencing (RNA-seq) data analysis. I subsequently moved to Leipzig, where I worked as a technical assistant at the Institute of Human Genetics. Since December 2024, I have been pursuing my PhD on GRIN-related disorders. My current research focuses on therapeutic approaches involving memantine, the definition and evaluation of VCEPs (variant curation expert panels), and a study investigating facial phenotypes in affected patients to identify potential common features.



Peter Kind is Director of the Simons Initiative for the Developing Brain and the Patrick Wild Centre. He is also a Senior Advisor to the SFARI Autism Rat Consortium. Professor Kind received his PhD from Oxford University in 1993, after which he completed his postdoctoral training with Professor Colin Blakemore (Oxford University) and Professor Susan Hockfield (Yale University). He moved to Edinburgh in 2000 where he has been Professor of Developmental Neuroscience since 2007. His laboratory uses electrophysiological, imaging and behavioural techniques to examine the pathophysiology associated with rodent models of single gene causes of neurodevelopmental disorders (NDDs) such as autism spectrum disorders and intellectual disabilities. His group investigates whether there is convergence of developmental trajectories for different genetic forms of ASDs/IDs and whether there are critical periods during which therapeutic strategies are more effective.

Johannes Lemke I am the director of the Institute of Human Genetics and head of the Centre for Rare Diseases at University of Leipzig, Germany. During my training to become a clinical geneticist, I spent a stage at the Swiss Epilepsy Centre Zurich, which became the basis of my long-standing research interest in epilepsy genetics. Since that, I was involved in gene discovery and first description of several genetic epilepsies. Right after its first description in 2010, the GRIN genes kept fascinating me in particular. I collected data and established the GRIN database in 2017 and within an international collaboration I was involved in building up of the GRIN Portal. My particular interest lies in the correlation of genetic, clinical and functional data as well as in precision medicine approaches in GRIN-related disorders, comprising both pharmacologic as well as genetic therapy approaches.

Bruce Leuchter is President, CEO, and board member of Neurvati Neurosciences and GRIN Therapeutics. A physician by training and neuropsychiatrist by specialty, he brings deep expertise in clinical research, patient care, and the development of novel therapies for neurological and psychiatric disorders. Prior to his work as a biotech executive, Dr. Leuchter held senior roles for over a decade in biotechnology equity research at Goldman Sachs, healthcare investment banking at Credit Suisse, and mergers and acquisitions at PJT Partners, where he provided strategic and capital markets advisory to leading biopharmaceutical companies. Dr. Leuchter served as the Director of Clinical Neuropsychiatry at Weill Cornell Medical College, where he currently holds a voluntary faculty appointment. He is the founding neuropsychiatrist of Click Therapeutics. He serves on the board of Arrivo BioVentures, is a member of the Scientific Advisory Committee for the Daedalus Fund for Innovation at Weill Cornell, and sits on the Life Sciences Institute Leadership Council at the University of Michigan.

Anne-Marie Li-Kwai-Cheung (MChem, MTOPRA, RAPS) is the Chief Development Officer for Neurvati Neurosciences and GRIN Therapeutics. A medicinal chemist by training, Anne-Marie has more than 25 years of experience leading development of medicines for neurological and rare diseases like MS, AD, PD, RLS, epilepsy, ALS, FTD, DMD, HD and GRIN. Originally focused on regulatory affairs, Anne-Marie started her career in large to mid-sized pharmaceutical companies (GSK, Wyeth, Genzyme, Sanofi) later moving into development and product leadership (Roche) and eventually bringing her passion for developing impactful medicines for diseases with high unmet need to biotech (Wave, Neurvati).

Andrew Penn I am currently an Assistant Professor in Neuroscience at the School of Life Sciences at the University of Sussex (UK). I have worked at the University of Sussex since 2015, when I joined with an MRC Career Development Award to start up my independent research group. Prior to this, I worked and lived in Bordeaux (France) for a period 5 years as a post-doctoral researcher (2010-2015) funded by a string of fellowships from European funding agencies, having previously graduated with a PhD in Biological Sciences (in the theme of Neuroscience) from the University of Cambridge in 2009. My research was, and continues to be, on the subject of a particular family of proteins, called glutamate receptors, which are crucial for conveying and tuning signal transmission between nerve cells in the brain. Since being at the University of Sussex, my research has steered from fundamental research questions to lines of enquiry that have more translational relevance, in particular, the consequences of human mutations in glutamate receptors on neurophysiology and pharmacology. I have recently been awarded grants from CureGRIN Foundation and GRIN2B Foundation to enable my ongoing research into GRIN disorders.

Katherine Roche is a Senior Investigator in the Intramural Research Program at the National Institute of Neurological Disorders and Stroke (NINDS) at the National Institutes of Health (NIH) in Bethesda, Maryland. Over the last two decades, Dr. Roche has studied the molecular mechanisms that underlie synaptic plasticity at excitatory synapses. Her lab investigates glutamate receptors and the mechanisms that dictate their expression at synapses. In recent years, her research program has focused on using human genetics to better understand the synaptic dysfunction underlying neurodevelopmental disorders. In particular, the Roche lab investigates rare variants in NMDA receptor genes (GRINs) and the neuroligin family of adhesion molecules. Using molecular and biochemical approaches, her lab characterizes mechanistic dysfunction and generates related mouse models to study in an in vivo context.

Narjes Rohani is a Postdoctoral Research Associate at the University of Oxford in the Sanders and Rinaldi labs, where her research focuses on the genetic, functional genomic, and epigenetic mechanisms underlying neurodevelopmental and psychiatric disorders. Her work aims to understand when and how these conditions arise by integrating diverse biological datasets, including gene expression (bulk and single-cell RNA sequencing), chromatin accessibility (single-cell ATAC sequencing), and DNA sequencing from brain tissues across developmental stages. A key objective of her research is to identify gene regulatory elements, particularly enhancers, that may serve as potential therapeutic targets for brain disorders. In parallel, she is actively involved in Patient and Public Involvement and Engagement (PPIE), developing accessible animations to communicate complex concepts in genetics and emerging therapies to patient families. This work helps bridge the gap between research and public understanding. She completed a PhD in Precision Medicine at the University of Edinburgh and holds a Master's degree in Computer Science and Bioinformatics from Shahid Beheshti University. She is also an Associate Fellow of the Higher Education Academy (HEA).

Carole Torsney is a Reader at the University of Edinburgh and is based in the Institute for Neuroscience and Cardiovascular Research. She is a member of the Simons Initiative for the Developing Brain and the Simons Foundation Autism Rat Models Consortium. She received her PhD, under the supervision of Professor Maria Fitzgerald, at University College London. She then did postdoctoral training at Columbia University, New York before establishing her lab at the University of Edinburgh. Her research investigates nervous system processing of pain and touch and how this can differ between the sexes. She is specifically interested in the

plasticity that underlies chronic pain conditions and the altered pain and tactile reactivity that manifests in neurodevelopmental disorders.

Stephen Traynelis received a BS in Chemistry from West Virginia University in 1984 and a PhD in Pharmacology from the University of North Carolina in 1988. He completed postdoctoral fellowships at University College London and the Salk Institute before joining the faculty in the Department of Pharmacology and Chemical Biology at Emory University School of Medicine in 1994, where he is a Professor and Dean's Eminent Investigator and the founding Director of the Center for Functional Evaluation of Rare Variants. Dr. Traynelis served as the Editor-in-Chief of Molecular Pharmacology and on advisory boards for private foundations and multiple pharmaceutical companies. He was elected to the National Academy of Medicine, and is a Fellow of AAAS and ASPET, and a John Merck Scholar. He received a Javits Neuroscience Investigator Award from the NIH, the Pharmacia-ASPET Award (2025), and was selected to deliver the Hodgkin-Huxley-Katz Prize Lecture sponsored by the Physiological Society. He is a co-author of over 250 publications, book chapters, and invited commentaries, and has given over 200 invited lectures. He is a co-inventor on more than a dozen issued and pending patent applications, co-founded NeurOp Inc. and Sensa Neurosciences. His lab has run a drug discovery program in collaboration with Dr. Dennis Liotta in the Department of Chemistry for 25 years, which has identified multiple first-in-class subunit-selective allosteric positive and negative allosteric modulators of glutamate receptors, including GluN2D-containing NMDA receptors. He has also developed software to model and analyze ion channel gating, and worked on acute CNS injury, glial biology, GPCR regulation of neuronal function, genetics, Parkinson's disease, and epilepsy.

Danielle Veenma is a genetic and developmental pediatrician, who specializes in the complex care of children with multiple congenital defects and/or ultra-rare genetic conditions for which she serve as the multi-disciplinary team coordinator in a tertiary academic hospital setting. Her ultimate goal is to improve the daily lives of my patients and their families, and serve the unmet treatment needs of those that are most vulnerable in our society. She is a member of the research steering committee of the Erasmus MC Child Brain Lab, which aims to build developmental curves across medical diagnosis focusing on adaptive functioning (link). Output from this facility will not only help construct natural history cohorts for rare disease but will also create an environment for fast implementation and evaluation of new therapies. Close collaboration and tuning in with fundamental scientists is key to this process. She serves as PI for the worldwide only genotype-phenotype study of CAMK2-GRI related neurodevelopmental disorders (NDD). CAMK2 is abundantly present in our brain and is a protein that is central to synapse function and therefore crucial for learning and memory. In 2019, she successfully adapted the "ENCORE-modus operandi" to make it suitable for international patient inclusion with collection of deep phenotyping data via online methods. As a result, we have built a unique patient-variant registry which in 2020 was expanded to include GRI disorders, another key synapse protein and downstream of CAMK2 in the calcium signalling pathway. For both disorders, we also collect blood for iPSC biobanking that will serve the development of our pre-clinical pipeline for drug & gene-therapy development. She strongly believes in caregiver involvement in rare disease research.

Lonnie P. Wollmuth. I am a Professor in the Departments of Neurobiology & Behavior and Biochemistry & Cell Biology and Director of the Center for Nervous System Disorders at Stony Brook University. I earned my PhD in Physiology and Biophysics in 1992 from the University of Washington and subsequently was a Senior Fellow in Cell Physiology at the Max Planck Institute for Medical Research in Heidelberg, Germany, from 1993 to 1998. I joined the Department of Neurobiology and Behavior at Stony Brook in 1998. Since I have started my laboratory, I have had a strong interest in mechanisms of synaptic transmission and the function of ionotropic glutamate receptors, the major excitatory ligand-gated ion channel in the brain. My lab has made a major push to study disease-associated variants in the *GRIN* genes encoding NMDA receptor subunits. At present, we study these variants in several contexts: We express them in a heterologous expression system to understand how they alter receptor function (gain- or loss-of-function); we study variants in human embryonic cells to understand how they disrupt molecular mechanisms of cell-to-cell signaling; and we introduce variants in zebrafish to study how they generate clinical phenotypes and to carry out high-throughput drug screens.

Emma Wood graduated with a 1st class Honours degree in Psychology from the University of St Andrews before going to Canada on a Commonwealth Scholarship to complete her PhD in Neuroscience at the University of British Columbia. She then spent 6 years in the USA as a postdoc - first in the lab of Janis Weeks at the University of Oregon, studying neural mechanisms of non-associative learning in the caterpillar Manduca sexta, and next in the lab of Howard Eichenbaum at Boston University,investigating how hippocampal place cells contribute to memory. She was recruited to a Lectureship in Neuroscience at the University of Edinburgh in 1999, and now holds a Personal Chair in Behavioural Neuroscience in the Institute for Neuroscience and Cardiovascular Research and Simons Foundation for the Developing Brain at the University of Edinburgh. We study the neural circuits mediating spatial cognition, memory, and cognitive flexibility, and how they are affected in rat models of neurodevelopmental disorders (NDDs). We probe this using high density in vivo electrophysiological recording techniques for measuring the activity of individual neurons and neuronal populations in awake freely behaving rats, combined with behavioural tasks designed to measure specific cognitive abilities. We are particularly interested in how spatially tuned neurons in the brain such as place cells and head direction cells support spatial cognition, memory, and cognitive flexibility, and how the juvenile development and function of these circuits are affected in rat models of NDDs.

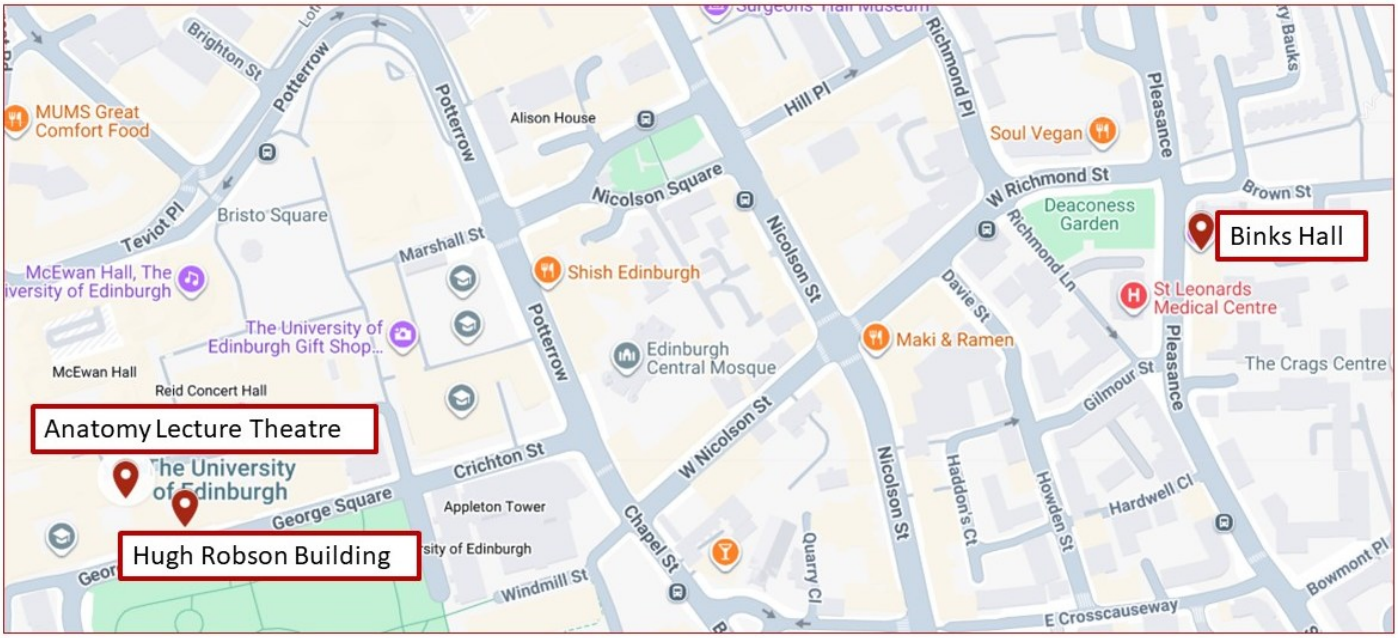
David Wyllie I am currently the co-Director of the Institute for Neuroscience and Cardiovascular Research and co-Head of the School of Neurological and Cardiovascular Sciences at the University of Edinburgh. Previously I was the director of the Centre for Discovery Brain Sciences (2015-24). I graduated with a BSc (Hons) in Pharmacology from the University of Glasgow in 1988 and gained my PhD from University College London in 1992. It was at UCL where my long-standing research interest in ligand-gated ion channels (LGICs), specifically those gated by glutamate was triggered. My current interests in glutamatergic synaptic function LGICs span fundamental structure-function studies through to research in dysfunctional signalling in rodent models of human neurodevelopmental disorders and in particular mutations in or deletions of genes coding for NMDA receptor subunits. My research vision is to develop an integrated approach to research that begins with the study of single protein molecules and synaptic function and extends, through collaboration with colleagues, to whole animal studies with an ultimate goal of the clinical study and treatment of disease.

Hongjie Yuan I am currently an Associate Professor of Pharmacology and Chemical Biology at the Emory University School of Medicine in Atlanta and serve as Deputy Director of the Centre for Functional Evaluation of Rare Variants (CFERV). My research focuses on genetic variation in human glutamate receptors and how these variants contribute to neurological and neuropsychiatric disorders. Using a multidisciplinary approach that integrates *in vitro* functional assays with *in vivo* transgenic animal models, my work aims to elucidate the molecular mechanisms and functional consequences of disease-associated glutamate receptor mutations in both healthy individuals and paediatric patients. My research program also explores novel translational strategies with the potential to inform mechanism-based, targeted therapeutic approaches for specific neurodevelopmental disorders.

Sameer Zuberi is a Consultant Paediatric Neurologist at the Royal Hospital for Children, Glasgow and Honorary Professor in the School of Health & Wellbeing at the University of Glasgow, leading the Paediatric Neurosciences Research Group. He graduated in medicine from the University of Edinburgh in 1989 and undertook postgraduate paediatric and neurology training in Edinburgh, Glasgow and Sydney. His postgraduate MD thesis was titled a Clinical Study of Ion Channel Disorders in Child Neurology. His research group have a broad focus in childhood epilepsies including genetics, epidemiology, outcomes, trials & precision therapies and global health. He is UK Chief Investigator of trials of radiprodil in GRIN related neurodevelopmental disorders. He has developed and researched innovative technologies including smartphone video and AI for diagnosis & management of neurological disorders. He is Neurodevelopment Theme Lead of the Epilepsy Research Institute UK and active in global advocacy work as Past President of the European Paediatric Neurology Society, vice President & President-Elect of the European Brain Council and President-Elect of the British Paediatric Neurology Association.

Maps

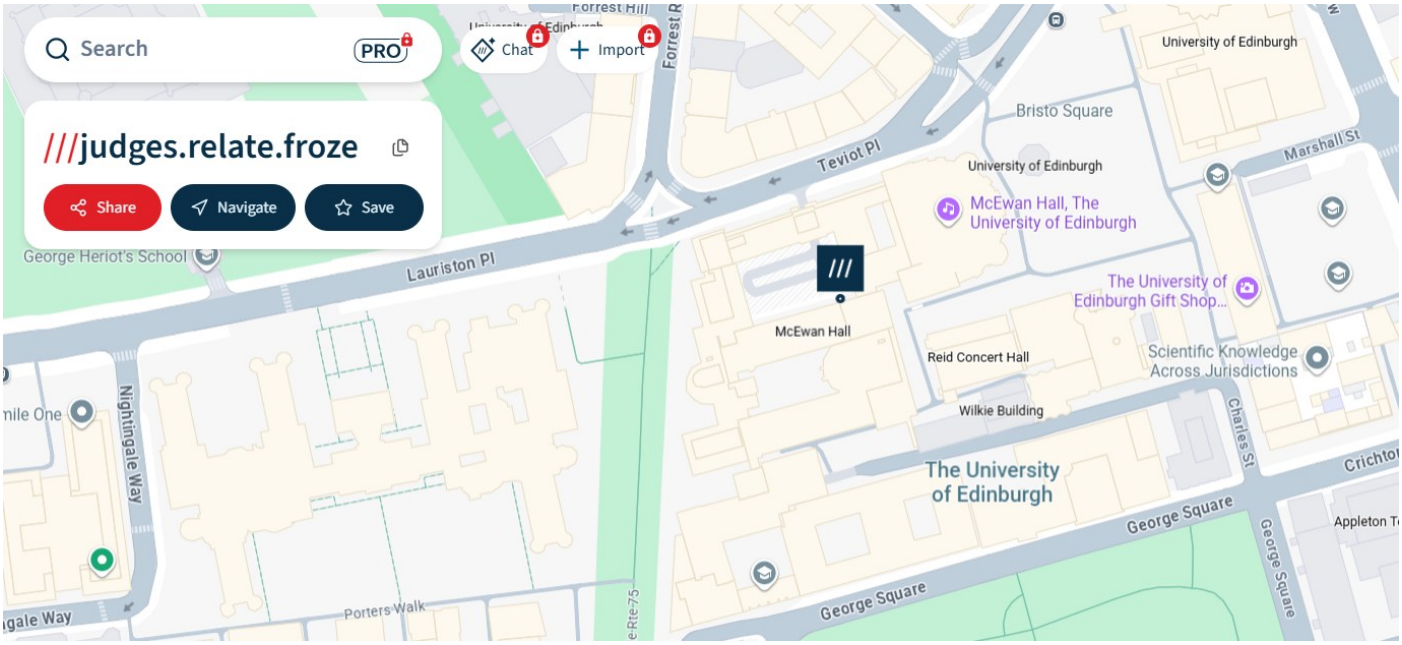
Overview of locations. All venues are within easy reach of each other



Research Presentations – Thursday 14 and Friday 15 May

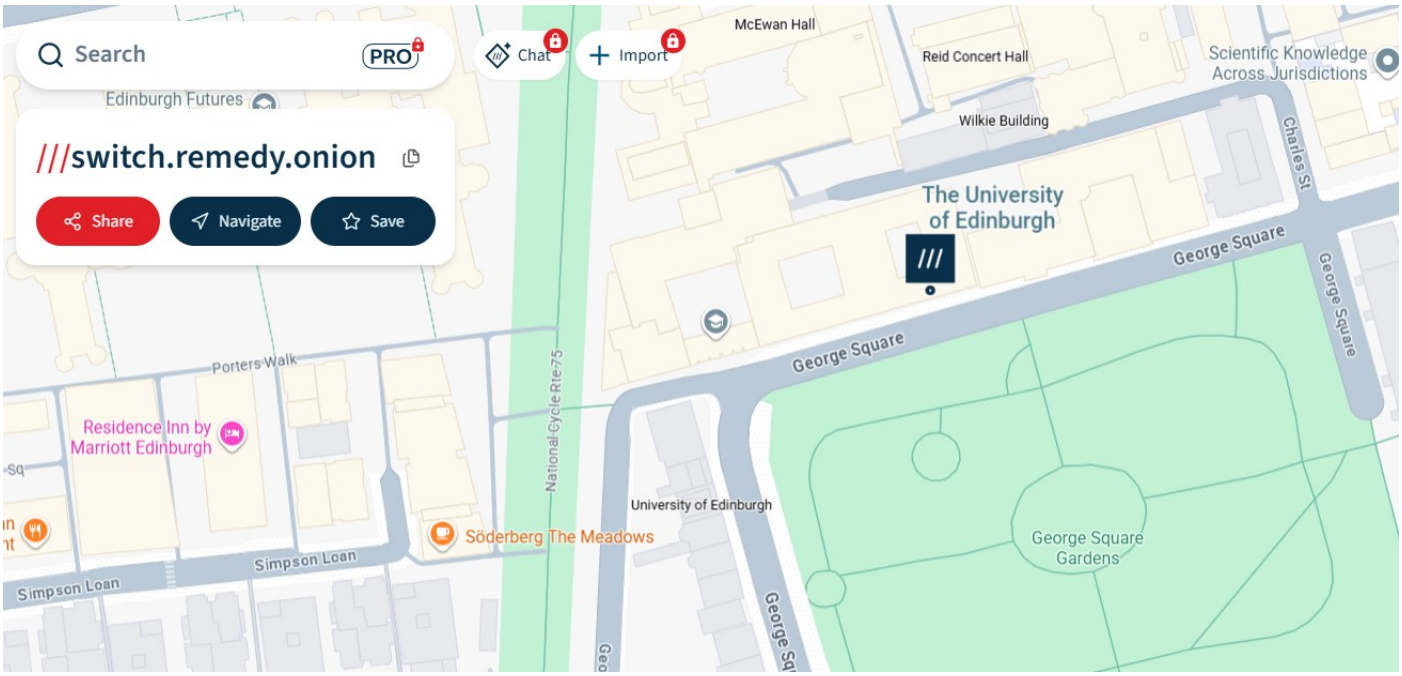
Anatomy Lecture Theatre, Old Medical School, Teviot Place, Edinburgh EH8 9AG

Enter at Doorway 3 from Old Medical School Quad



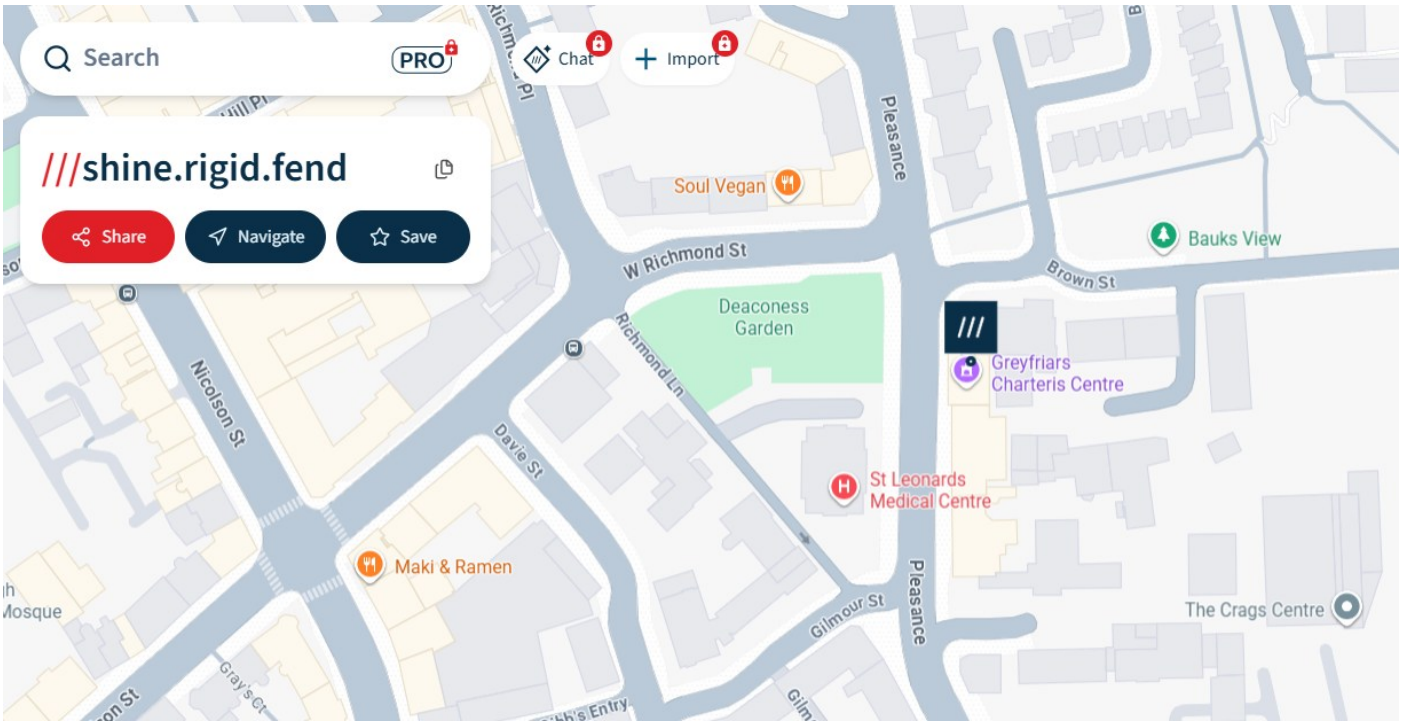
Family Presentations – Friday 15 and Saturday 16 May

Hugh Robson Lecture Theatre, Hugh Robson Building, George Square, Edinburgh EH8 9XD



Ceilidh Venue – Friday 15 May

Binks Hall, 138-140 Pleasance, Edinburgh, EH8 9RR



Further details – access to venues

Thursday afternoon and Friday morning

Anatomy Building – Doorway 3
Old Medical School, Teviot Place
Edinburgh EH8 9AG

More info: AccessAble - Your Accessibility Guide

what3words: ///judges.relate.froze

Friday afternoon and Saturday

Hugh Robson Building,
George Square
Edinburgh EH8 9XD

More info: AccessAble - Your Accessibility Guide

what3words: ///switch.remedy.onion

Friday night ceilidh

Binks Hall, Greyfriars Charteris Centre
138-140 The Pleasance
Edinburgh EH8 9RR

More info: Accessibility | charteriscentre

what3words: ///shine.rigid.fend