

SECOND ANNUAL VIRTUAL CONFERENCE

IDIOPATHIC KETOTIC HYPOGLYCEMIA

11-12 JUNE 2022

SCIENTIFIC ADVISORY BOARD



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Clinical Professor
H.C. Andersen Children's Hospital



Dr. David Weinstein
Former Director, GSD Program
The University of Connecticut



Dr. Pratik Shah
Pediatric Endocrinologist
The Royal London Children's Hospital



Dr. Terry Derks
Pediatric Metabolic Diseases
Beatrix Children's Hospital



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Dr. Mary-Elizabeth Patti
Medical Director
Joslin Diabetes Center, Harvard Medical School



Dr. Joseph Wolfsdorf
Clinical Director
Boston Children's Hospital, Harvard Medical School

ketotichypoglycemia.org

Register now:
ketotichypoglycemia.org/register

WELCOME

We would like to personally welcome each of you to the second annual conference of Ketotic Hypoglycemia International. We are hosting a virtual conference to make sure everyone has a chance to join, no matter where in the world they live. All of the presentations will be available after the conference for educational purposes. Please feel free to share this conference invitation with whoever could be interested in learning more about idiopathic ketotic hypoglycemia – colleagues, family members, friends, doctors, medical students, industry partners – everyone is welcome!

We are beyond excited to have this opportunity to come together, patients, caregivers, healthcare professionals, leading medical experts, pharma- and biotech companies to share current knowledge, ongoing studies, best practices, patient stories, and our collective vision for the future. Our goal with the conference is to unite the families affected by idiopathic ketotic hypoglycemia, the experts, the healthcare professionals, and the industry so we together can take the first step towards an enhanced understanding of idiopathic ketotic hypoglycemia. We have a line-up of presentations made by our scientific advisory board and guest speakers, mixed up with patient stories from our KHI families from around the world. Our speakers will be ready to answer questions after each scientific presentation and in the “Meet the Experts” sessions. We hope you leave the conference inspired and informed on the latest knowledge in the field of idiopathic ketotic hypoglycemia.

If you are a patient, a caregiver, a family member, a nurse, a dietitian, a medical student, a researcher, a pharma- or biotech company, or something completely different: Your interest is truly our greatest asset today and tomorrow. We could not accomplish what we do without your support. Do you have a burning question for the experts? Please e-mail danielle.drachmann@ketotichypoglycemia.com, or submit your question in the registration form. See you virtually on June 11-12 2022

With gratitude on behalf of the entire staff team,



D. Drachmann

Danielle Drachmann
Executive Director



Nick Rasmussen

Nick Rasmussen
Marketing Lead

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TIME ZONES

The following timeframes, specify when the conference will be happening in each particular country and zone.

Time slots listed on the program are in CEST

| | |
|---------------------------------|---------------------------|
| DENMARK (CEST) | 04:00PM - 08:30PM |
| WEST COAST USA (PDT) | 07:00AM - 11:30AM |
| MOUNTAIN ZONE USA (MDT) | 08:00AM - 12:30PM |
| CENTRAL USA (CDT) | 09:00AM - 01:30PM |
| EAST COAST USA (EDT) | 10:00AM - 02:30PM |
| UNITED KINGDOM (BST) | 03:00PM - 07:30PM |
| SOUTH/CENTRAL AFRICA (SAST/CAT) | 04:00PM - 08:30PM |
| MOSCOW, RUSSIA (MSK) | 05:00PM - 09:30PM |
| UNITED ARAB EMIRATES (GST) | 06:00PM - 10:30PM |
| INDIA (IST) | 07:30PM - 12:00PM |
| CHINA (CST) | 10:00PM - 02:30AM(+1) |
| JAPAN (JST) | 11:00PM - 03:30AM(+1) |
| SYDNEY, AUSTRALIA (AEST) | 00:00AM(+1) - 04:30AM(+1) |
| NEW ZEALAND (NZDT) | 02:00AM(+1) - 06:30AM(+1) |



SATURDAY 11 JUN

| Time CEST | Description | Speaker | |
|-------------------|--|---|---|
| 04:00pm-04:15pm | Opening session | | |
| 04:00pm-04:05pm | Welcome |  | Danielle Drachmann Executive Director of Ketotic Hypoglycemia International |
| 04:05pm-04:15pm | Opening session Update from Danielle Drachmann, Executive Director, and Jacob Petersen, chairman | | |
| 04:15pm-07:05pm | Understanding ketotic hypoglycemia | | |
| 04:15pm - 04:25pm | The roadmap to diagnosis: Establishing incidence, prevalence, subtypes, and a diagnostic protocol for ketotic hypoglycemia |  | Dr. Anne Benner Odense University Hospital, Denmark |
| 04:25pm-04:55pm | What is hypoglycemia? - 10 min. Patient story or/and Q&A |  | Dr. Paul Thornton Cook Children's Hospital, US |
| 04:45pm-05:15pm | What is ketotic hypoglycemia? - 10 min. Patient story or/and Q&A |  | Dr. David Weinstein Passage Bio, US |
| 05:15pm-06:05pm | Adaptation to fasting in children - 10 min. Patient story or/and Q&A |  | Dr. Joseph Wolfsdorf Boston Childrens' Hospital, US |
| 06:05pm-06:35pm | The role of fasting studies in the diagnosis and management of ketotic hypoglycemia - 10 min. Patient story or/and Q&A |  | Dr. Paul Thornton Cook Children's Hospital, US |
| 06:35pm-07:05pm | Idiopathic ketotic hypoglycemia in adults - a case presentation - 10 min. Patient story or/and Q&A |  | Dr. Mary-Elizabeth Patti The Hypoglycemia Clinic, Joslyn Diabetes Center, US |
| 07:05pm | 15 min. break | | |
| 07:20pm-08:40pm | Following up on projects from last years conference | | |
| 07:20pm-07:45pm | GSDIXa and KH - 10 min. Patient story or/and Q&A |  | Dr. Henrik Thybo Christesen, Odense University Hospital, Denmark |
| 07:45pm-07:55pm | Ketotic hypoglycemia in patients with Down syndrome |  | Dr. Henrik Thybo Christesen, Odense University Hospital, Denmark |
| | |  | Austin Carrigg Ketotic Hypoglycemia International |
| 07:55pm-08:05pm | Learning from the field of Patient Advocacy |  | Bastian Hauck #DeDoc Diabetes Online Community |
| 08:05pm-08:40pm | Closing session | | |
| 08:05pm-08:35pm | Meet the experts: - Your questions answered by the Scientific Advisory Board |  | Danielle Drachmann Executive Director of Ketotic Hypoglycemia International |
| 08:35pm-08:40pm | Closing remarks | | |

SUNDAY 12 JUN

| Time CEST | Description | Speaker | |
|-----------------|---|---|--|
| 04:00pm-04:15pm | Opening Session | | |
| 04:00pm-04:05pm | Welcome |  | Danielle Drachmann Executive Director of Ketotic Hypoglycemia International |
| 04:05pm-04:15pm | Opening session Introduction to boardmember, Jovana Vlajkovic | | |
| 04:15pm-07:10pm | Living with ketotic hypoglycemia | | |
| 04:15pm-04:45pm | Children presenting with hypoglycemia in the setting of an intercurrent illness - a retrospective study - 10 min. Patient story or/and Q&A |  | Dr. Elizabeth Rosenfield Childrens Hospital of Philadelphia, US |
| 04:45pm-05:15pm | Eating difficulties: PROMs and PREMs - 10 min. Patient story or/and Q&A |  | Dr. Annieke Venema University Medical Center Groningen, The Netherlands |
| 05:15pm-05:45pm | The role of CGM - 10 min. Patient story or/and Q&A | Alessandro Rossi University of Naples Federico II | |
| 05:45pm-06:15pm | Emergency protocols - a initiative from MetabERN - 10 min. Patient story or/and Q&A |  | Dr. Terry Derks Beatrix University Hospital, The Netherlands |
| 06:15pm-06:40pm | Symptoms and signs of ketotic hypoglycemia - 10 min. Patient story or/and Q&A |  | Dr. Pratik Shah The Royal London Children's Hospital, UK |
| 06:40pm-07:10pm | From hyperinsulinism to ketotic hypoglycemia - 10 min. Patient story or/and Q&A |  | Dr. Paul Thornton Cook Children's Hospital, US |
| 07:10 | 10 min. break | | |
| 07:20pm-08:30pm | Closing session: Münchausen by Proxy / Factitious Induced Illness | | |
| 07:20pm-07.40pm | Münchausen by Proxy and idiopathic ketotic hypoglycemia |  | Dr. Henrik Thybo Christesen, Odense University Hospital, Denmark |
| | |  | Dr. Paul Thornton Cook Children's Hospital, US |
| 07:40pm-07.50pm | Living in fear - a familys experience | Anonymous | |
| 07:50pm-08.25pm | Meet the experts: - Your questions answered by the Scientific Advisory Board |  | Danielle Drachmann Executive Director of Ketotic Hypoglycemia International |
| 08:25pm-08.30pm | Closing Remarks | | |

IDIOPATHIC KETOTIC HYPOGLYCEMIA

MEET OUR EXPERTS

Ketotic Hypoglycemia International aims to challenge current perceptions and attitudes to expand the recognition that IKH can be more than a normal variation and establish an international platform for future research collaboration. Our mission is supported by leading medical experts within the field of inherited metabolic diseases, pediatric endocrinology, and endocrinology united in our Scientific Advisory Board.



Dr. Henrik Christesen
Odense University Hospital, Denmark

Henrik Thybo Christesen is a clinical professor at Hans Christian Andersen Children's Hospital, Odense University Hospital, Denmark and a pediatric endocrinologist with special clinical and research interest in hypoglycemia. He leads the multidisciplinary national team in complex hypoglycemia at the hospital, also receiving international patients. Through international collaboration, his research has led to many published papers in the field, mostly on congenital hyperinsulinism, but also, more recently, on ketotic hypoglycemia. Of more than 110 papers, he has published over 40 papers on glucose disorders and beta cells. He functions as the head of the scientific advisory board in Ketotic Hypoglycemia International.



Dr. Paul Thornton
Cook Children's Hospital, US

Paul Stephen Thornton is a Medical Director, Distinguished Consultant of the Endocrinology/Diabetes Clinic and the Hyperinsulinism Center at Cook Children's Medical Center, in Fort Worth, Texas. His recent awards include the Cook Children's Clinical Scholar Award 2012-2014 and recipient of the 2012 Cook Children's Health Care System Endowed Chair Award. He has coauthored more than a dozen book chapters and over 50 journal articles. Paul is the lead author of the recent Pediatric Endocrine Society's Recommendations and Management of Persistent Hypoglycemia in Neonates, Infants, and Children. Dr. Thornton is also a member of the Pediatric Endocrine Society, Society for Pediatric Research, and The Endocrinology Society.

“

Through my work with children with idiopathic ketotic hypoglycemia, it struck me that many of them really have a disease being much more than normal variation. This has prompted us to dive into various genetic explanations and improved management and treatment “

- Dr. Henrik Christesen



Dr. David Weinstein
Passage Bio, US

Following his graduation from Trinity College (CT) and Harvard Medical School, Dr. Weinstein did a residency, chief residency, and fellowship in pediatric endocrinology at Boston Children's Hospital. He subsequently obtained a Masters in Clinical Investigation from Harvard and MIT, and became Director of the Glycogen Storage Disease Program at Boston Children's. In 2005, Dr. Weinstein moved to the University of Florida where he directed the Glycogen Storage Disease Program and became a tenured professor. He and his team moved to the University of Connecticut and Connecticut Children's Medical Center in 2017 to perform gene therapy for GSD. In August 2020, he left the academic world to serve as the medical lead for the GM1 gangliosidosis gene therapy trial at Passage Bio.

Dr. Weinstein has published over 100 manuscripts and 28 textbook chapters in the field of disorders of carbohydrate metabolism. He was named as one of the inaugural Goldwater Scholars in 1989. He is a former Jan Albrecht Award winner from the American Association for the Study of Liver Diseases, and he was the George Sacher Award winner from the Gerontological Society of America. In 2013, Dr. Weinstein was honored with the Order of the Smile international humanitarian award, and he was knighted in Poland as part of this recognition. He has also been inducted in the Rare Disease Research Hall of Fame.



Dr. Terry Derks
Beatrix Children's Hospital, The Netherlands

Terry Derks has been certified as consultant pediatric metabolic diseases and he is appointed as associate professor at the University Medical Center Groningen, University of Groningen, the Netherlands. Since his PhD thesis defense in 2007 (title: "MCAD deficiency: clinical and laboratory studies"), he developed a special interest, both clinically and scientifically, in inherited disorders of glycogen metabolism (i.e. hepatic glycogen storage diseases) and fatty acid oxidation disorders. Dr. Derks has more than 60 international publications, 2 book chapters and is a SSIEM Advisory Council Member. In MetabERN Dr. Derks is a Medical Executive Board member and the Work package leader for the "Virtual Counseling Framework".



Dr. Pratik Shah
The Royal London Children's Hospital, UK

Pratik Shah is a Consultant in Pediatric Endocrinology/Diabetes and Honorary Senior Lecturer at The Royal London Children's Hospital (Barts Health NHS Trust) and Queen Mary University of London (QMUL), London. He did his PhD in the field of Congenital Hyperinsulinism and specializes in children with all forms of glucose disorders and has led the highly specialized service for Congenital Hyperinsulinism. His main research includes understanding molecular basis various forms of non-diabetic hypoglycemia in children and identifying novel therapies in hypoglycemia/hyperinsulinism. He has been a speaker at various international and national meetings and has been primary investigator for various investigational studies and clinical trials.

SAY HELLO TO OUR LATEST ADVISORS

We are beyond excited to give a warm welcome and applause to our latest Scientific Advisory Board members; two outstanding world-class experts in complex hypoglycemia:

Dr. Mary-Elizabeth Patti and Dr. Joseph Wolfsdorf are dedicated to enhance the understanding of idiopathic ketotic hypoglycemia.



Dr. Mary-Elizabeth Patti
Joslin Diabetes Center, Harvard Medical School, Boston, US

Dr. Mary-Elizabeth Patti is a physician-scientist, serving as a Principal Investigator at Joslin Diabetes Center, Director of the Hypoglycemia Clinic, Co-Director of the Molecular Phenotyping Core, and Associate Professor of Medicine at Harvard Medical School.

Dr. Patti’s NIH-funded lab focuses on identification of molecular/epigenetic mechanisms by which nutritional exposures during early life increase diabetes risk in subsequent generations.

Translational studies are focused on mechanisms contributing to diabetes resolution and hypoglycemia after bariatric surgery.

Dr. Patti received her MD from Jefferson Medical College magna cum laude, internal medicine residency at the University of Pittsburgh, and endocrinology fellowship at Harvard. She is board-certified in endocrinology and metabolism.

Dr. Patti has held numerous leadership roles in the diabetes scientific community, including service as organizer of a diabetes-focused Keystone Symposium and chair of the American Diabetes Association Scientific Sessions Planning Committee.

She was elected to the American Society of Clinical Investigation in 2009 and to Fellowship in both the American College of Physicians and Obesity Society in 2014.



Dr. Joseph Wolfsdorf
Boston’s Children’s Hospital, Harvard Medical School, Boston, US

Professor of Pediatrics at Harvard Medical School and the first incumbent of the Boston Children’s Hospital Chair in Endocrinology. He served as the Director of the Diabetes Program at Boston Children’s Hospital for 35 years until June 2018.

From 1986-1997 he was also Chief of Pediatrics at Joslin Diabetes Center and Medical Director of the Pediatric Diabetes Treatment Unit of the New England Deaconess Hospital. Was a member of the Joslin Diabetes Center DCCT research team from 1984-1993. Served as Clinical Chief of the Division of Endocrinology, Boston Children’s Hospital from 1994-1998 and from 2006-2016. Dr. Wolfsdorf continues to be active in the practice of pediatric endocrinology. He has a special interest in disorders of carbohydrate metabolism (hypoglycemia disorders in children, glycogen storage disease) and, especially, the care of children, adolescents and young adults with diabetes mellitus. Professor Wolfsdorf is an author of more than 250 publications and has edited and contributed to 5 monographs and textbooks. He serves on the editorial board of Diabetes Care and Pediatric Diabetes, is an Associate Editor for Hormone Research in Pediatrics, and is the pediatric endocrinology section editor for all topics related to diabetes mellitus and hypoglycemia in UpToDate.

Dr. Wolfsdorf Is the immediate Past President of the International Society of Pediatric and Adolescent Diabetes (ISPAD) for the period 2016-2018 and currently chairs the ISPAD eLearning committee.





What is ketotic hypoglycemia?

“Ketotic hypoglycemia may be unexplained, or idiopathic (IKH). This is a challenge and should urge for more research”

For the patients

In a normal person, fuel for the brain and the general cell metabolism primarily comes from the burning of sugar deposits (glycogen). When the glycogen stores are depleted, the body will switch to burn fat deposits. The fat burn lead to two fuels for the brain, both glucose (sugar) and ketone bodies. However, ketones in the blood will lead to nausea and eventually vomiting. This will lead to a vicious circle, where you cannot eat or drink sugar-rich items, which again leads to further fat burn and production of ketone bodies.

In a KH-patient, the glycogen stores are somehow insufficient. This leads to decreased fasting tolerance with earlier onset of fat burn and hence ketone bodies. In most patients, the hypoglycemia is relatively mild, and the ketone bodies help to provide fuel to the brain, which prevents loss of consciousness and convulsions. However, in relatively few patients, the condition is more severe, but still without an identified cause despite intense investigations in hormones and cell metabolism. Such patients are said to have “idiopathic” KH, or IKH, which simply means KH without any known cause.

Source: Professor Henrik Christesen, Odense University Hospital, Chair of the Scientific Advisory Board in KHI, Denmark



Illustration by Illulines

For the doctors

Ketotic hypoglycemia can be seen in children because of growth hormone deficiency, cortisol deficiency, metabolic diseases with intact fatty acid consumption, including glycogen storage diseases (glycogenosis; GSD) type 0, III, VI, and IX, or disturbances in transport or metabolism of ketone bodies. When these diagnoses are excluded, ketotic hypoglycemia can be categorized as unexplained or idiopathic (IKH), otherwise known as accelerated starvation.

The primary treatment is dietary prevention with long carbohydrates (e.g. uncooked cornstarch) and frequent meals. In some more severely affected patients, continuous feeding may be needed through a gastrostomy tube.

Emergency treatment constitutes of oral or i.v. glucose, eventually i.m. glucagon, to raise the plasma glucose, which will prevent further lipolysis. However, the ketones can take hours to be eliminated. In more severely affected patients, the ketone production can be significant before hypoglycemia occurs, why recognition of increased ketones above approximately 1.5 mol/L is important, sometimes already when the glucose concentration goes below 3.9 mmol/L (70 mg/dL).

Source: Professor Henrik Christesen, Odense University Hospital, DK

What is this organization about?

Ketotic Hypoglycemia International (KHI) is a global reaching family organization, uniting more than 1500 families from all over the world affected by idiopathic ketotic hypoglycemia. KHI aims to globally unite leading hypoglycemia experts with patients, caregivers, and healthcare providers, and to establish a greater understanding of IKH in the hopes to facilitate improved treatment options and greater quality of life for individuals suffering from IKH. The organization aims to challenge current perceptions and attitudes to expand the recognition that IKH is more than a normal variation and establish an international platform for future research collaboration. We are committed to supporting and advocating for those in the KH community through our international KHI Parents Support Group by utilizing the contributions of general medical guidance made by our scientific advisory board, supporting research on the general well-being of KH-families, and furthering research into KH etiology, diagnosis, and management.

Since last year’s conference, we have published 2 scientific papers, presented and won a poster prize at the Engaging Citizen Science Conference in Århus, Denmark. The publications can be found below, and the poster can be found on page 14-15.

Article 1

 **BMC** Part of Springer Nature

 **Orphanet Journal of Rare Diseases**

Published: 13 April 2021 | <https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-01797-2>

“Towards enhanced understanding of idiopathic ketotic hypoglycemia: a literature review and introduction of the patient organization, Ketotic Hypoglycemia International”

[Danielle Drachmann](#), [Erica Hoffmann](#), [Austin Carrigg](#), [Beccie Davis-Yates](#), [Valerie Weaver](#), [Paul Thornton](#), [David A. Weinstein](#), [Jacob S. Petersen](#), [Pratik Shah](#) & [Henrik Thybo Christesen](#)



Article 2

 **JIMD** REPORTS

 **SSiEM**

Published: 28 July 2021 | <https://doi.org/10.1002/jmd2.12241>

“Ketotic hypoglycemia in patients with Down syndrome”

[Danielle Drachmann](#), [Austin Carrigg](#), [David A. Weinstein](#), [Jacob Sten Petersen](#), [Henrik Thybo Christesen](#)



Where to find us

Our families are united through a variety of social media channels. Ketotic Hypoglycemia International are active on LinkedIn, Instagram, Twitter, and Facebook. The Facebook Support Group is where the families are most active, and where we are sharing experiences and support each other to navigate the everyday-IKH-rollercoaster ride.

Ketotic hypoglycemia in patients with Down syndrome

Danielle Drachmann^{1,2,3}, Austin Carrigg¹, David A. Weinstein^{1,4}, Jacob Sten Petersen^{1,5}, Astrid Janssens^{2,3,6}, Henrik Thybo Christesen^{1,7,8}

1. Ketotic Hypoglycemia International, Skanderborg, Denmark | 2. University of Southern Denmark, Unit of User Perspectives, Odense, Denmark | 3. Center for Research in Partnership with Patients and Relatives, Odense University Hospital, Odense, Denmark | 4. University of Connecticut, Connecticut, United States | 5. Novo Nordisk A/S, Bagsvaerd, Denmark | 6. University of Exeter Medical School, United Kingdom | 7. Department of Clinical Research, University of Southern Denmark, Odense, Denmark | 8. Hans Christian Andersen Children's Hospital and Steno Diabetes Centre Odense, Odense University Hospital, Odense, Denmark

Conclusion

We present a case of co-created and family-organization driven research in the family organization Ketotic Hypoglycemia International, from the establishment of the organization to the scientific publication. This is the first demonstration of a possible high prevalence of ketotic hypoglycemia (KH) in Down syndrome (DS). Even though this finding needs to be confirmed in other research settings, identification of KH in DS could have a dramatic impact, as simple treatments with cornstarch, protein and frequent meals may prevent KH attacks and, analogous to other conditions with KH, improve growth, stamina and prevent overeating and obesity. GYG2 deletion may contribute to KH in DS, resembling glycogen storage disease type 0. Our research is an example of a family-run patient organization driven and co-produced research, where novel observations may arise, not firstly caught by the health care system. This story can inspire others to adopt this approach to health research within research and patient groups.

1. The Beginning

Danielle Drachmann and her two kids, Noah and Savannah: Diagnosed with low blood glucose and high ketones, without any known genetic cause: idiopathic ketotic hypoglycemia (IKH).



2. The Organization

Danielle established Ketotic Hypoglycemia International (KHI), established a scientific advisory board (SAB) with leading medical experts from all over the world - expanded with many IKH-families united in KHI rapidly.



3. The Discovery

Melanie is a girl with DS who got diagnosed with KH. Her mother entered the support group and contacted Danielle with suspicion about a correlation between DS and KH after seeing posts in the online support group on social media.



4. The Survey

No association between DS and KH was ever reported in the literature - SAB members supported Austin and Danielle to draft out a survey. SAB consulted on survey draft.



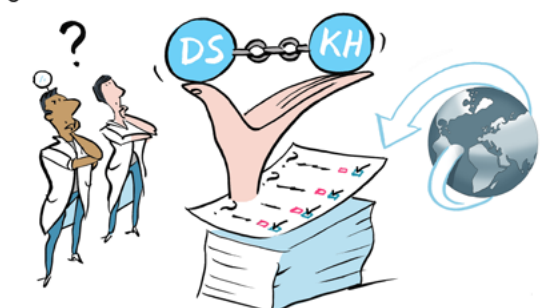
5. The Data

A survey was sent to DS organisations and families worldwide.



6. The Findings

The results indicated an association. Want to know about the findings? Scan QR code 2 below.



7. The Mice Study

A SAB member found a mice study (1), giving a medical explanation (2) for why patients with DS could have KH. Want to know more about the mice-study? Scan QR code 1 below.



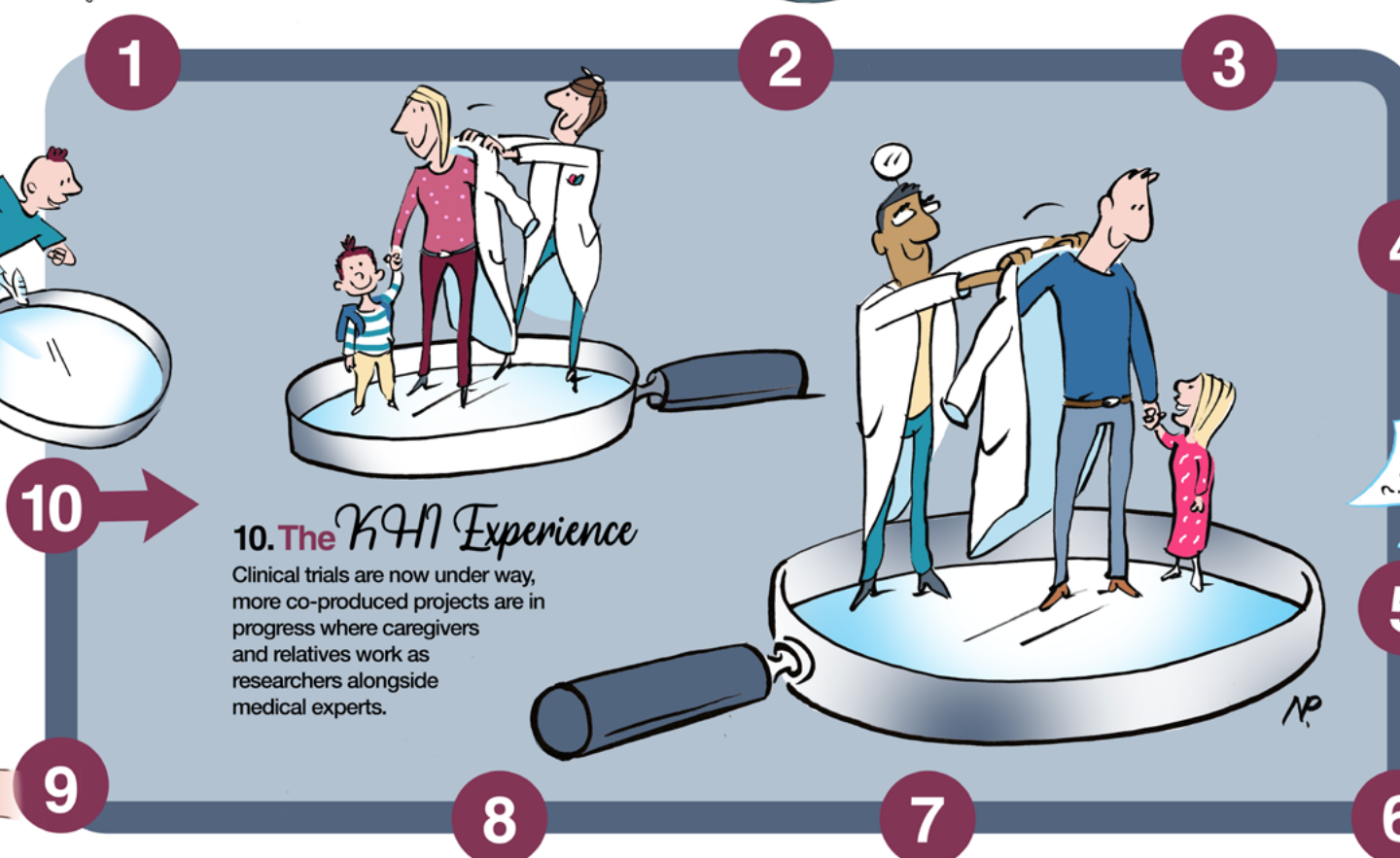
8. The Publication

Scientific paper written by Austin, Danielle and medical experts in the SAB is published alongside a video abstract (3).



10. The KHI Experience

Clinical trials are now under way, more co-produced projects are in progress where caregivers and relatives work as researchers alongside medical experts.



CO-PRODUCED RESEARCH

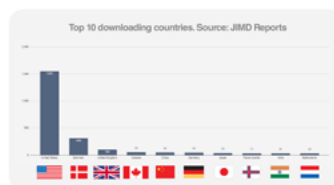
10 steps that shaped a scientific discovery

9. The Dissemination

Worldwide press attention for our findings.

2617 downloads in the first 6 months (average annual downloads for JIMD Reports: 441).

2nd most downloaded article in JIMD Reports in 2021



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D. Drachmann

Danielle Drachmann
Executive Director



Nick Rasmussen

Nick Rasmussen
Marketing Lead