

# Ketotic hypoglycemia in patients with Down syndrome

Danielle Drachmann<sup>1,2,3</sup>, Austin Carrigg<sup>1</sup>, David A. Weinstein<sup>1,4</sup>, Jacob Sten Petersen<sup>1,5</sup>, Astrid Janssens<sup>2,3,6</sup>, Henrik Thybo Christesen<sup>1,7,8</sup>

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.

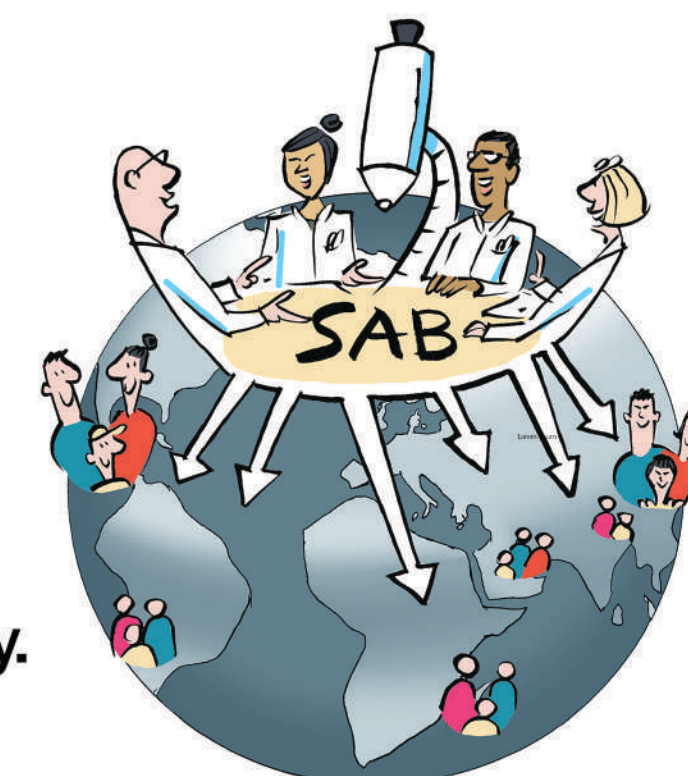
### 1. The Beginning

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.



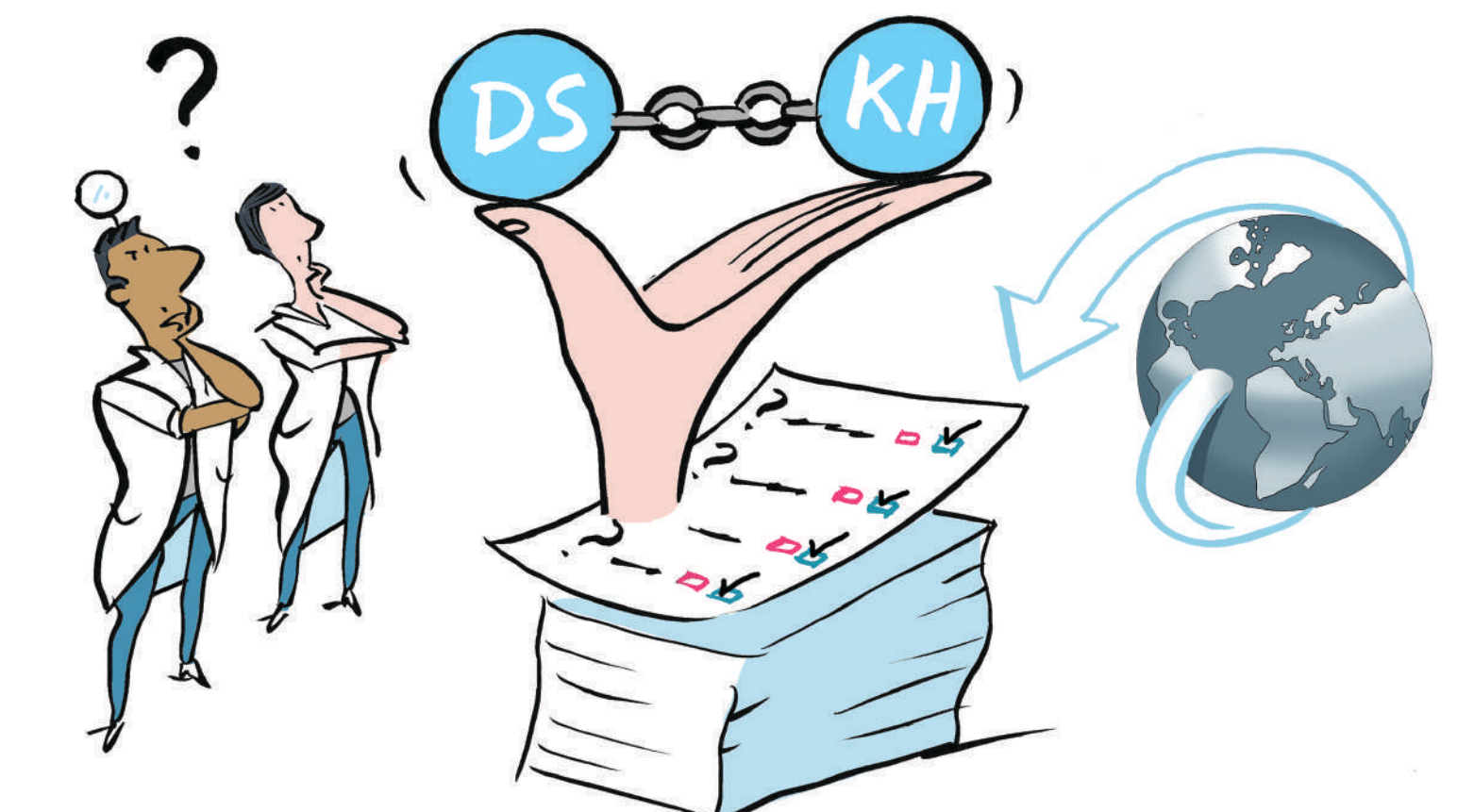
### 5. The Data

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).



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### 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.

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## CO-PRODUCED RESEARCH

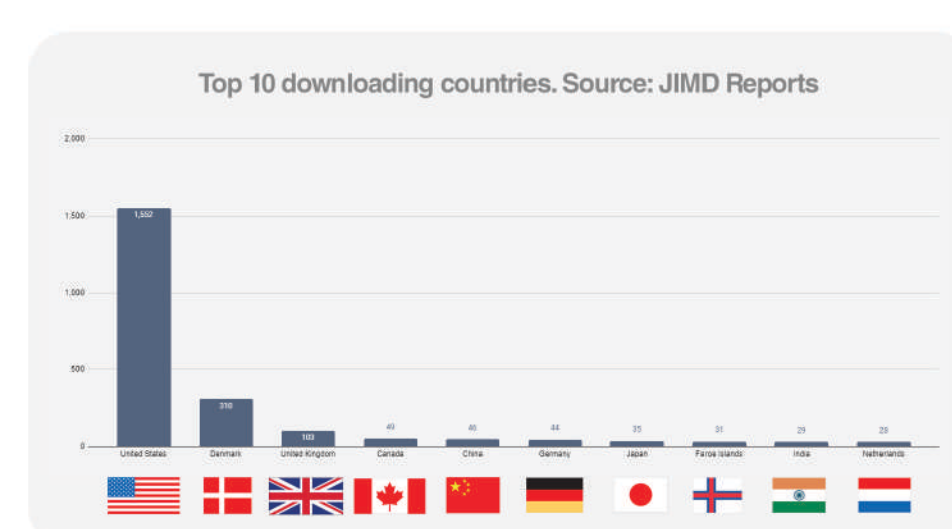
10 steps that shaped a scientific discovery

### 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



1) Arora T, Caviedes P, Sharma SK. Effects of a tripeptide on mitogen-activated protein kinase and glycogen synthase kinase activation in a cell line derived from the foetal hippocampus of trisomy 16 mouse: an animal model of down syndrome. *Neuro-tox Res.* 2020;37(3):714-723  
 2) Drachmann, D, Carrigg, A, Weinstein, DA, Petersen, JS, Christesen, HT. Ketotic hypoglycemia in patients with Down syndrome. *JIMD Reports.* 2021; 62(1): 70- 73. <https://doi.org/10.1002/jmd2.12241>  
 3) Youtube.com/watch?v=ZeA4ZdqGWmo



1. Mice Study  
Hippocampal cell study in the trisomy 21 mice model htk.



2. Paper  
Ketotic hypoglycemia in patients with Down syndrome.



3. Video Abstract  
Extending the reach and visibility of our research, we published a video abstract.