

The Start of a Revolution

EURORDIS Open Academy Training



BY DANIELLE DRACHMANN

Where did it begin?

It started with a tragedy that
turned into hope.



A baby was born into a world of
misdiagnosis, trauma and dismissal.
That was later labelled **pathological**
idiopathic **ketotic hypoglycemia**.

Saved by a specialist doctor

PROFESSOR HENRIK CHRISTESEN

HEAD OF THE COMPLEX HYPOGLYCEMIA CENTER
ODENSE UNIVERSITY HOSPITAL, DENMARK



Reliving the nightmare.

Back on the hypoglycemia rollercoaster, this time better prepared and provided vital treatment sooner.



ADHD, bulimia and a bad memory

Identifying 26-years of misdiagnosis



Danielle

Imagining a better future.

WHERE NO FAMILY OR PATIENT IS LEFT BEHIND



MY DREAM

The Mentor and the Mentee

PROFESSOR JACOB S. PETERSEN

ADJUNCT PROFESSOR, UNIVERSITY OF
COPENHAGEN, DENMARK

HEAD OF CELL THERAPY
NOVO NORDISK, DENMARK





Uniting the families

The Medical Mystery



Uniting the experts

The Medical Mystery

The Scientific Advisory Board



Dr. Henrik T. Christesen
SAB Chair, Denmark,
Consultant in
Paediatrics, clinical
professor, leader of a
national center for
complicated
hypoglycemia at H-C
Andersen Children's
Hospital



Dr. Paul Thornton
US, Medical Director at
Endocrinology &
Diabetes,
Hyperinsulinism Center,
Congenital Adrenal
Hyperplasia, Cooks
Children's Hospital



Dr. David Weinstein
US, Clinical
Development, Passage
Bio, former director of
the Glycogen Storage
Disease Program,
Connecticut Children's
Hospital



Dr. Pratik Shah
UK, Consultant in
Paediatric
Endocrinology, Clinician
and Researcher in
Hyperinsulinism and
Ketotic Hypoglycemia,
The Royal London
Children's Hospital



Dr. Terry Derks,
The Netherlands,
Consultant in Paediatric
inherited metabolic
diseases, UMCG,
associate professor,
principle investigator,
University of Groningen,
University Medical
Center Groningen,



Dr. Mary-Elizabeth Patti
US, Investigator and Staff
Physician
Director, Hypoglycemia
Clinic
Co-Director, Molecular
Phenotyping and
Genotyping Core
Associate Professor of
Medicine, Harvard Medical
School



Dr. Joseph Wolfsdorf,
US, Director, Attending
Physician, Division of
Endocrinology, Diabetes
program, Boston
Children's Hospital,
Professor of Pediatrics,
Harvard Medical School

Ketotic Hypoglycemia
International aims to establish a
greater understanding of
idiopathic ketotic hypoglycemia in
the hopes to facilitate improved
treatment options and greater
quality of life for individuals
suffering from **idiopathic ketotic
hypoglycemia**

OUR GOAL





"A NORMAL VARIATION OF CHILDHOOD"

Families being dismissed due to the outdated perception that low blood sugar and high ketones is just a normal variation of childhood



PATHOLOGICAL VS. PHYSIOLOGICAL

Before Ketotic Hypoglycemia International, IKH was not defined into the subtypes: pathological and physiological KH.



MONITORING

The perception that blood glucose and ketone monitors are only for diabetic patients can be life-threatening to patients suffering from pathological ketotic hypoglycemia

Identifying the issues



UNITED PATIENT GROUP.

Over 1800 families from across 89 countries united in an online support group.



SCIENTIFIC ADVISORY BOARD.

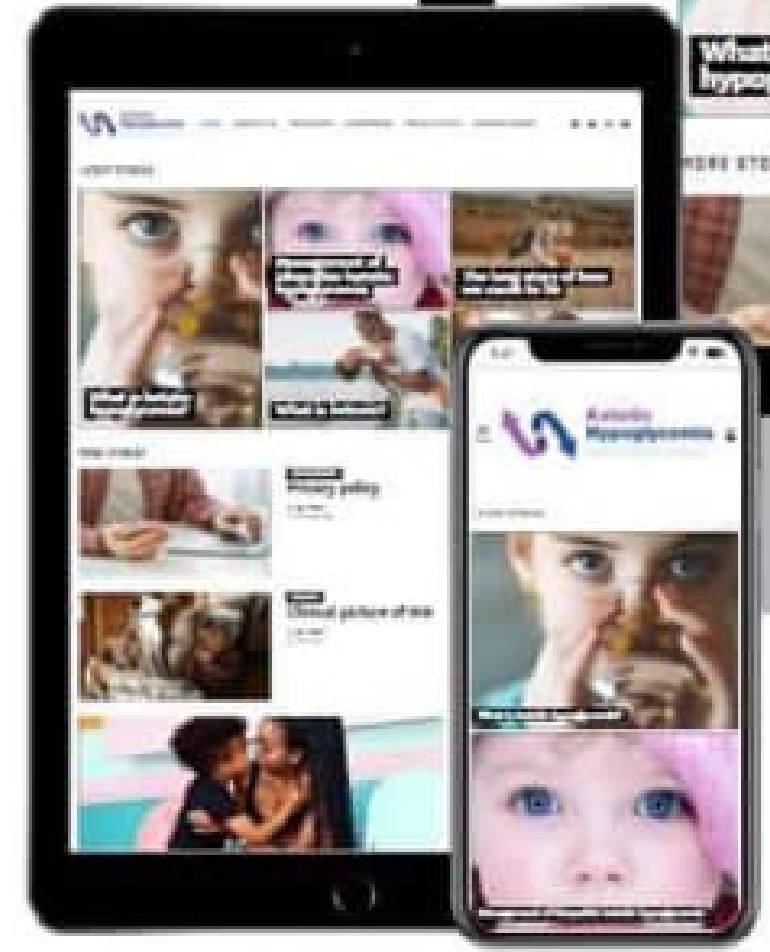
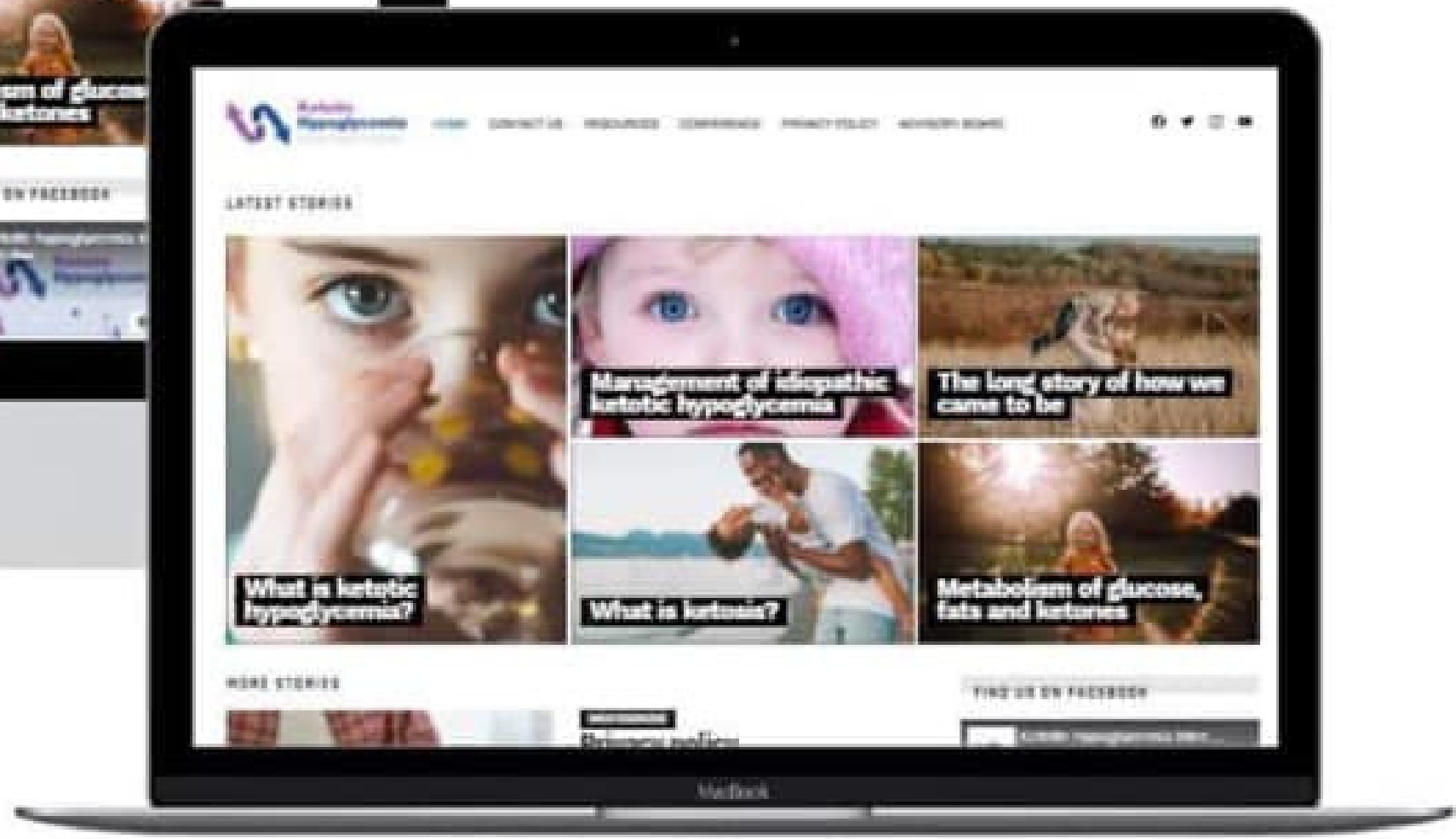
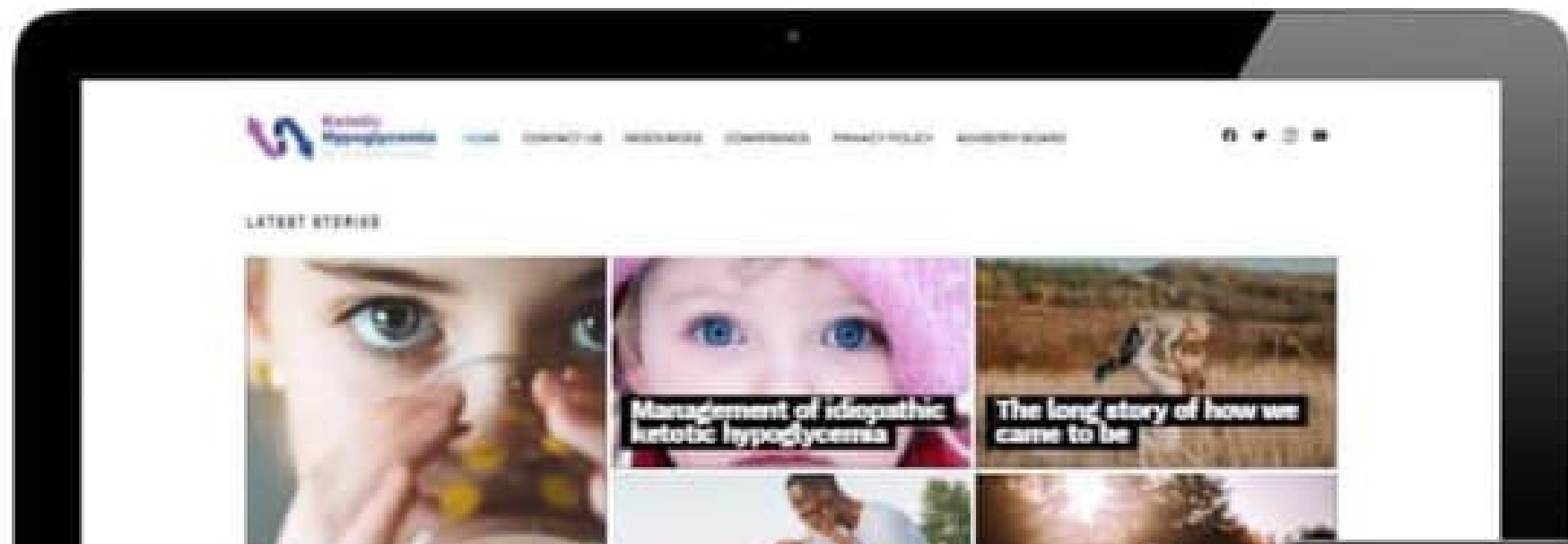
Experts from the fields of endocrinology and metabolism.



KHI FAMILY VOLUNTEERS.

Dedicated family members working voluntarily for our organization.

The People Power





Ketotic Hypoglycemia INTERNATIONAL

11th ANNUAL VIRTUAL CONGRESS

DIOPATHIC KETOTIC HYPOLYCEMIA

11-12 JUNE 2022



ketotichypoglycemia.org

SCIENTIFIC ADVISORY BOARD

-  **Dr. Henrik Christesen**
Clinical professor
McGill 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 Diagnostics
Royal Children's Hospital
-  **Dr. Paul Thornton**
Medical Director
Great Ormond Street Children's Hospital
-  **Dr. Mary-Elizabeth**
Medical Director
Seattle Children's Hospital
-  **Dr. Joselyn**
Clinical Director
Boston Children's Hospital

Register now:
ketotichypoglycemia.org/register



Our Greatest Achievements So Far

MEDICAL BASELINE PAPER PUBLISHED

Collaboration between experts and patients

DOWN SYNDROME & KH PAPER PUBLISHED

Identifying new potential correlations

KHI ANNUAL CONFERENCES

Over 1500 registrations from countries



The Ambition

**Better Diagnosis.
Better Management.
Better Treatment.**



The mom



The PhD
Student

How do we work - and how can you work?

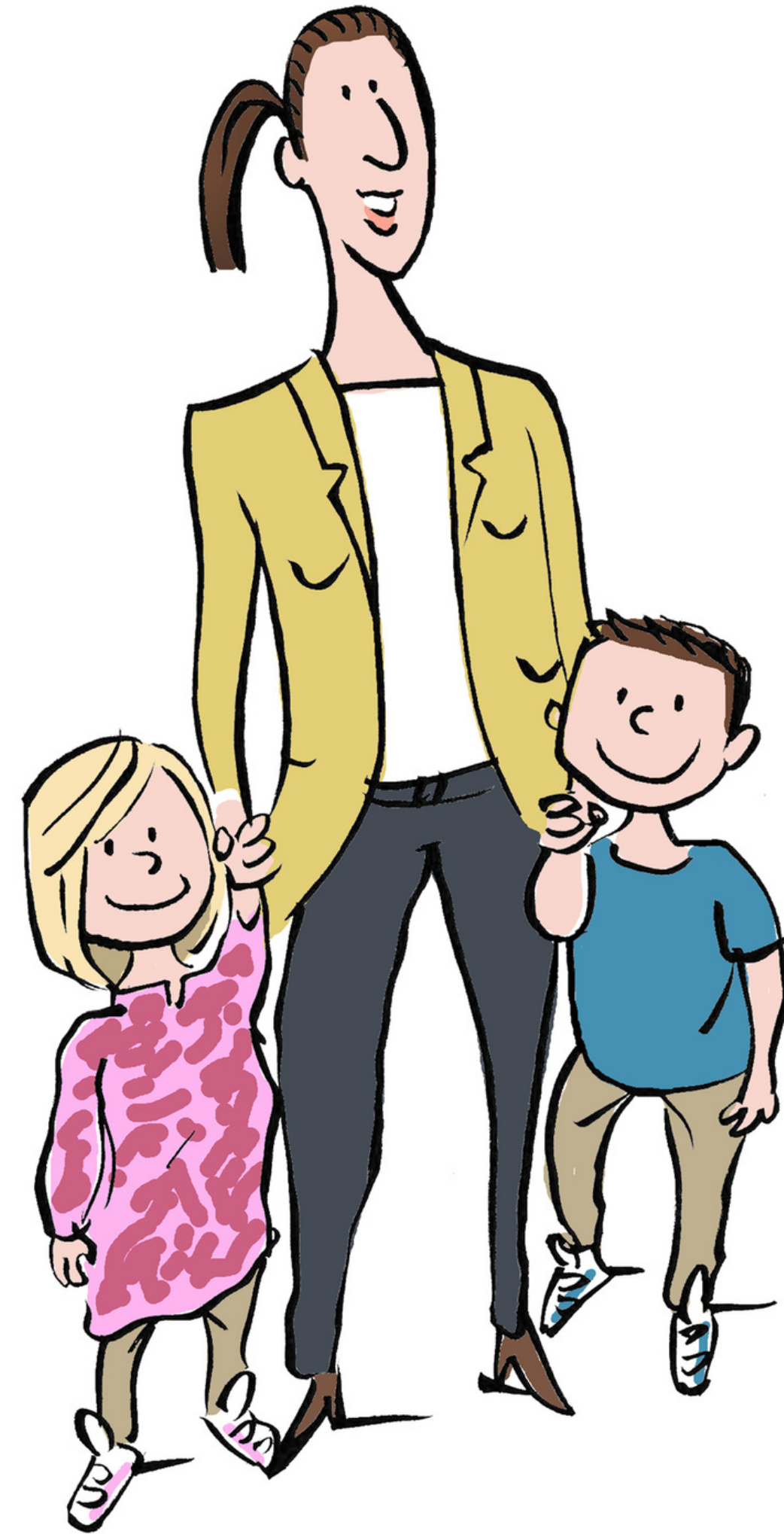
PATIENTS, CAREGIVERS AND
SCIENTIFIC ADVISORY BOARD

HOW?

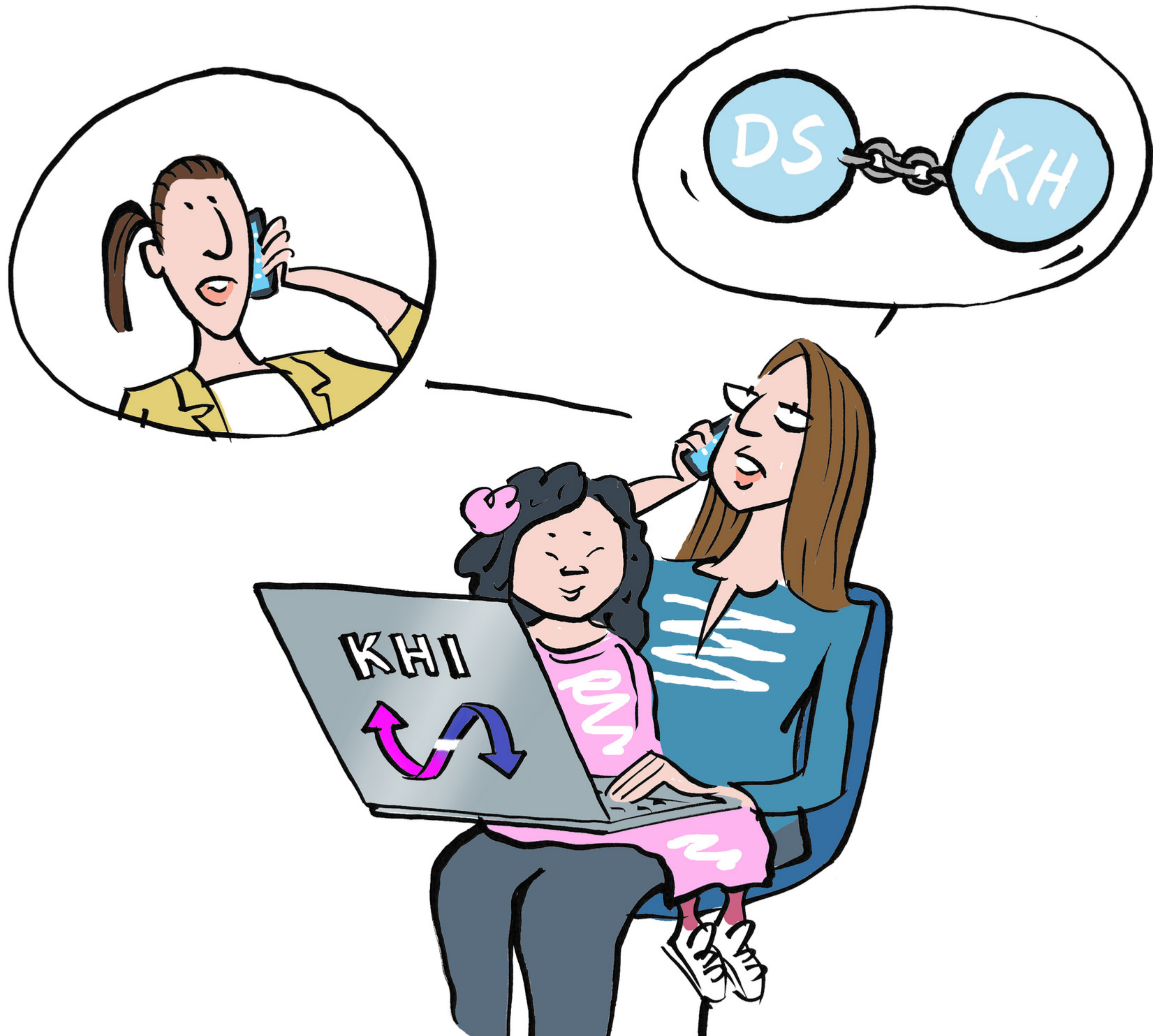


An example from KHI

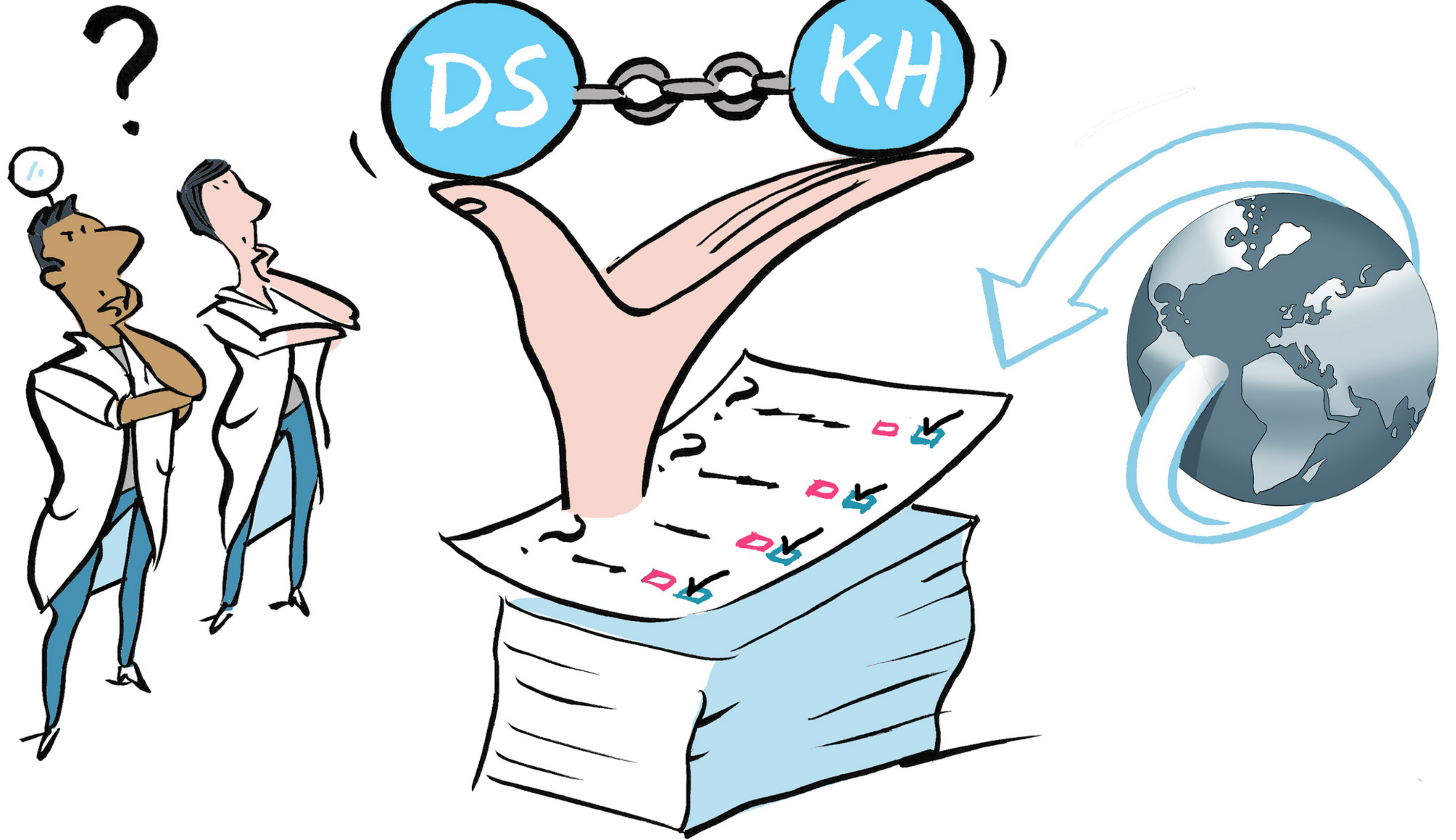
KETOTIC HYPOGLYCEMIA IN
DOWN SYNDROME

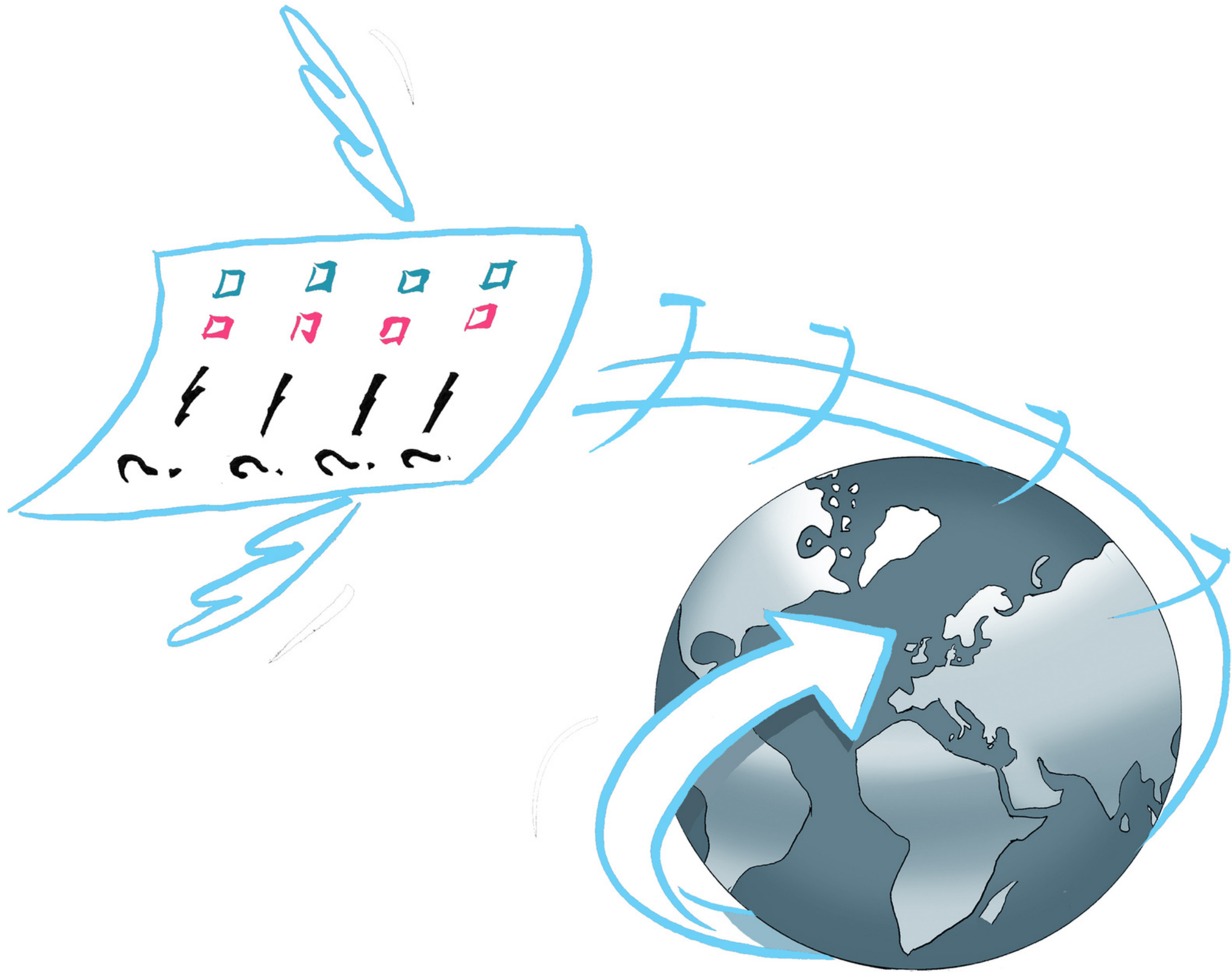






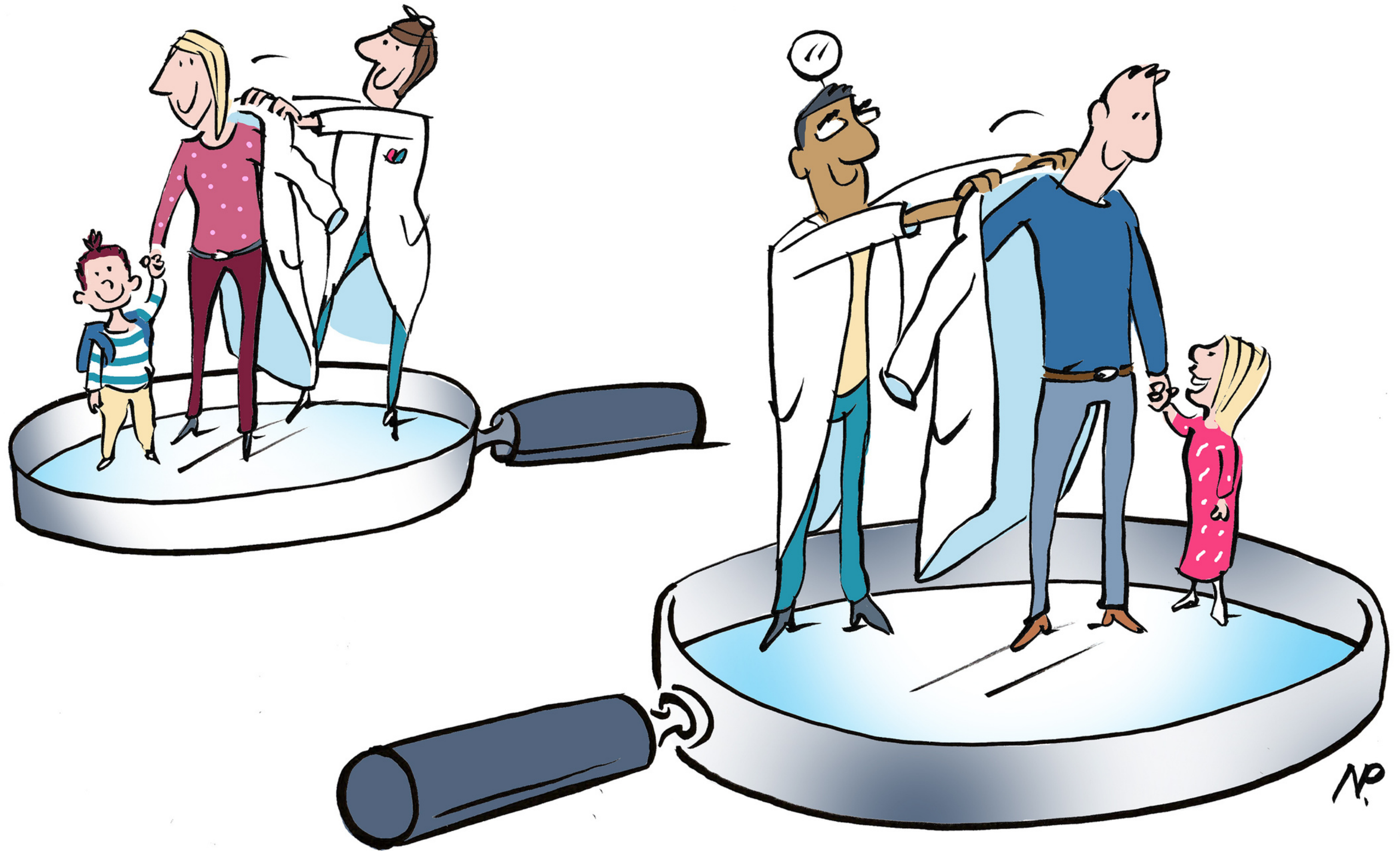




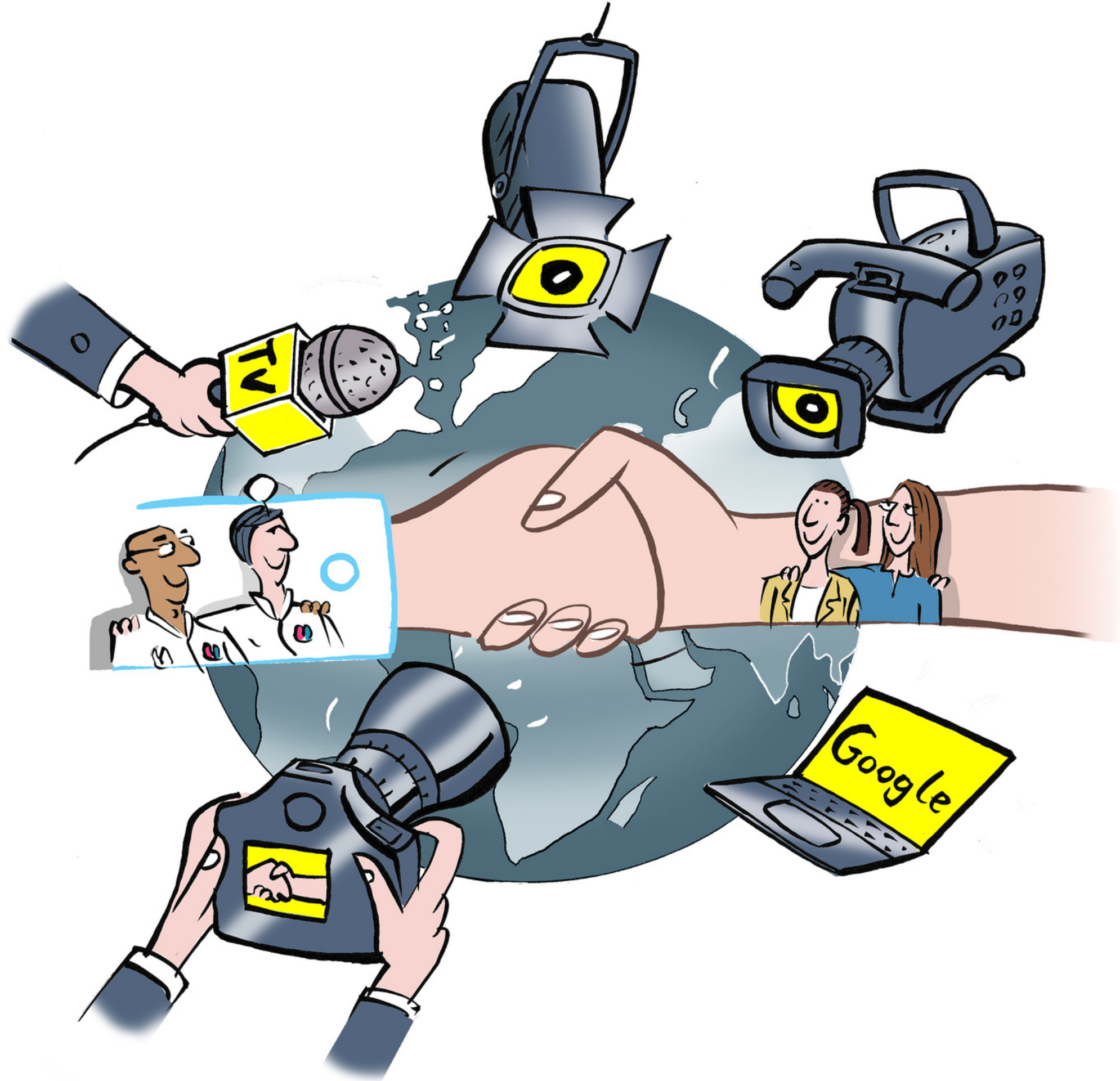












Ketotic hypoglycemia in patients with Down syndrome: an example of extreme citizen science in biomedicine

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Questions?

THANKS FOR LISTENING

THANK YOU

