For Rhythm internal use only.

The BEAT

rhythm

Building Engagement & Advocacy Together

• In this issue •

Top 2018 Patient Engagement Priorities

1. Develop and launch repositioning of Rhythm and "Genetic Obesity" lexicon to increase disease education and remove barriers to genetic testing to support Patient Identification
2. Map Pt Advocacy and non-HCP Influencers to develop action plan to build rare genetic disorders of obesity community
3. Develop and execute non-HCP and patient relationship management plan (ongoing communications platform); evolve current website and social media assets
4. Support enrollment of burden of illness questionnaire, and deliver patient and caregiver input into Phase 3 BBS trial
5. Develop strategy and gain feedback on meaningful support programs and access expectations for "patient services"
6. Align and support improvements to GO-ID Genotyping study to increase interest and participation

rhythm

Genetic Disorders UK Leadership Symposium

Genetic Disorders UK Leadership Symposium

Every March, two advocacy groups, the **Genetic Disorders UK** and **Global Genes**, co-host the UK Genetic Disorders Leadership Symposium, which brings together the leaders of UK-based genetic disorder charities and patient groups. The event, which took place on March 9-10 in London, began with a Friday networking reception, and was followed by educational and best-practice-sharing discussions on Saturday. Presenters included experts in areas of public policy and healthcare, genetic research, and patient group activities.

Rhythm sponsored the event for the very first time. At the event, Irene Aquino was able to meet group leaders from **Bardet-Biedl Syndrome UK, Alström Syndrome UK, Prader-Willi UK, Global Genes** and **FindaCure**. **Debbie and Alex Potter**, a mother and son affected by LEPR deficiency, registered and attended the event, to learn and network with those that have started patient groups. Alex was recognized with a **"Champion of Hope"** award and connected with FindaCure, an organization that runs training workshops, peer mentoring, and an online portal, to help empower those to facilitate learning on how to start a patient group.

In future issues, different advocacy groups will be featured. If you'd like to learn more about a certain group, please email Irene Aquino at iaquino@rhythmtx.com

Genetic Disorders UK

Champions of Hope

This year saw the second Champion of Hope awards at the annual Genetic Disorder UK, symposium dinner. The awards recognise the past and future contribution of individuals to the UK genetic disorders community. Rare Revolution were thrilled to be part of this special evening of sharing and celebration

2018 Champion of Hope award for courage to start a new organisation within the UK genetic disorders community

Following on from this tradition which started last year, these awards were given in the form of **knowledge boxes**, to those who are in the process of setting up a genetic disorder charity or patient group.

Attendees of the evening dinner, were invited to offer their considerable experience, advice, guidance and words of support to fledgling charities, presented in beautifully engraved knowledge boxes.

Vanessa Martin Childhood Tumour Trust

Andrew Gibson Noonan UK

Amarpreet Kaur MEDpicture Klippel-Feil Syndrome UK

Alex Potter Rare Genetic Obesity UK

Maria Walters SMA&I Gene Foundation

Carlos Heras-Palou TTR Amyloidosis Patient Ass. UK

EVENTS

Chloe Joyner Usher Kids UK

Oliver Gardiner RJH UK

Howard Don The Haemochromatosis Society

Stephen Dichlberger-Bradshaw Wolf Hirschhorn Syndrome Trust

Sandra Webb HP Support Group

Michael Mason VAP Gene Support Group

2018 Champion of Hope awards for lifetime dedication

These special awards were given to those who have contributed to a genetic disorder charity or patient group for more than 15 years.

It acknowledges long service and dedication of individuals to their chosen charity showing commitment to the field of genetic disorders and the patients they serve.

Key Dates

1. **June 30 – July 1**
   2018 Bardet Biedl Syndrome Family Association Conference
   Salt Lake City, UT

2. **July 20 - 22**
   Obesity Action Coalition Convention and "Your Weight Matters" Expo
   Denver, CO

Meet Katy: Life with POMC Heterozygous Deficiency

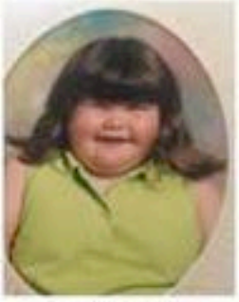
"It causes extreme unrelenting hunger and excessive eating, as a child…the fridge and food was controlled massively…but nobody could understand (obviously at the time I didn't know it was the best thing for me) that I was desperately hungry and just wanted to stop that feeling."

Birth

Katy is a "normal" weight at birth

9 weeks old: rapid weight gain

Childhood



Katy at 3yo

3yo: Bullying begins at nursery school

4yo: Diagnosed with POMC Heterozygous Deficiency Obesity

6yo: Self-isolation and missed school days

8yo: Asthmatic, increased pain and pressure on her knees make play and PE difficult

Teen Years

11yo: Katy is 231 pounds, numbness and agonizing back pain

14yo: Katy is on antidepressants

15yo: Pubertal development is abnormal

Katy at 11yo

Adulthood

Katy at 23yo

Currently ~450 lbs. (over twice what she weighed at 16yo)

Sleep apnea; some cardiac issues; insulin resistance

Acanthosis nigricans—cracked and bleeding skin

As of 05/10/18, Katy was not on any treatments for her obesity related to POMC heterozygous deficiency

rhythm

Unmet Need: Patient-Friendly Education

Back in December 2017, Katy reached out to Rhythm. Despite her diagnosis of POMC deficiency, and knowing that her father also had the same condition, Katy sought to learn more. She has since identified her mutation, and has become a strong advocate for herself.

There is an unmet need for patient-friendly education about POMC and LEPR deficiencies. Many who have a diagnosis are met with highly clinical information, or ones about rat and canine genetics.

Rhythm is leading the effort to help those who may feel there is something different about their obesity—something that caused it to start earlier and seem harder to manage than anyone else's. Over the next few weeks, we will be developing the educational materials to help those that may be on their journey to a diagnosis of a rare disorder of obesity, or seeking to learn more about their conditions.

Advocacy Groups Engaged

The Obesity Action Coalition
Global Genes
Genetic Disorders UK
FindaCure
EveryLife Foundation
Bardet Biedl Syndrome Foundation and Family Association
Alström Syndrome International
Alström Syndrome UK
Prader-Willi UK
Foundation of Prader-Willi Research
PWSA (USA)
PRISMS (SMS)

---

rhythm