ePAG EXCHANGE OF GOOD PRACTICES

18 March 2021
ABOUT ME

Head of Business Development at a med-tech company and the Co-Founder of the rare disease charity The Cure & Action for Tay-Sachs (CATS) Foundation.

I know the challenges we all face in the rare disease world to get research started but utilizing real-world evidence it’s possible to kick start engagement and excitement about your disease community with pharma.
Most small charities are started because of one person. In our case it was Amélie.

THE BEGINNING OF OUR STORY AT THE CATS FOUNDATION
THE CATS FOUNDATION WAS BORN

LOOKED INTO RESEARCH
Investigated what research was being undertaken into Tay-Sachs and Sandhoff disease

RAISED OUR PROFILE WITHIN THE COMMUNITY
We reached out to families across Europe to create a strong community to support each other

RAISED OUR PROFILE WITH RESEARCH
We reached out to all the research teams we found to let them know that we charity existed
Member charities of the European Tay-Sachs and Sandhoff Charity Consortium (ETSCC). Since it was established in 2012 the ETSCC has 5 different member organisations.
To work as a team, patient group, pharma, researchers and patients, can make a difference and really drive forward projects.

Together we can develop a treatment.
RESEARCH & TRIALS – 2011

In 2011 there was only one research project

1. UK
   Gene therapy programme for GM2 Gangliosidosis - The University of Cambridge
Huge progress in 10 years

**Europe**
- Gene therapy programme for GM2 Gangliosidosis — *The University of Cambridge*
- IB1001-202 clinical trial — *Intrabio*
- AMETHIST (substrate reduction therapy) — *Sanofi*
- A new inflammasome complex treatment for GM2 Gangliosidosis — *Seville University*
- Gene transfer in a murine model of Sandhoff using AAV9 — *Institut Necker Enfants Malades*
- Deployment of chaperone treatment for Tay-Sachs — *Laboratory PR Yves Bleriot-Poitier*
- Investigation into gene & cellular therapy treatment for GM2 — *San Raffaele Institute*

**Outside Europe**
- Gene therapy programme — *Taysha*
- Gene therapy programme — *Sio Gene Therapies*

**Upcoming**
- Drug repurposing — *Confidential*
- New drug - Pharma company — *Confidential*
- New drug - Pharma company — *Confidential*
HOW TO DEVELOP GOOD PROMS FOR YOUR OWN DISEASE?

How to start the process? How to collect and bring patient views?
FIRST STEPS

1. TERMINOLOGY
   Understand the different types of data that can be collected

2. DISEASE
   Understand the disease from all stakeholder viewpoints

3. ENGAGEMENT
   Understand how best to engage with all stakeholders

4. COLLABORATION
   Understand how collaborating can make a big difference
WHAT IS WHAT?

- **PROs** (Patient Reported Outcomes)
- **PROMs** (Patient Reported Outcome Measures)
- **COAs** (Clinical Outcome Assessments)
- **PREMs** (Patient Reported Experience Measures)
GOAL SETTING

WHAT IS THE GOAL?
We need to set a goal which is achievable.

HOW DO WE SET THE GOAL?
What is the process at setting the goal.

WHO’S INVOLVED IN THE GOAL?
Defining who is involved in achieving the goal is vital.

HOW DO WE WORK TO THE GOAL?
How do we work towards achieving the goal.
WHO ARE THE STAKEHOLDERS?

- **PHARMA**: The group who can start the process
- **PATIENTS**: The most important member
- **PATIENT GROUPS**: The group who can make things happen
- **CLINICIANS**: A vital member in the process
- **PHARMA**: The group who can start the process
SETTING THE GOAL

LISTEN
It’s important to listen to all stakeholders and hear their views

LEARN
Learn what is important to each group and how it can impact the goal

BE BRAVE
We want to make the biggest impact – so be bold and brave

PATIENT FIRST
The patient SHOULD and MUST be at the forefront of the discussion
The bump on the road is an opportunity to overcome challenge
Relevance is all a matter of perspective.

What is important to you may not be to me and vice versa.
NURTURING THE IDEA AND MAKING IT GROW

PLANT
Your collaborative idea is “planted” together as a group

NUTURE
You can all nurture the idea to make it grow in to reality

REALISE
If you all work together you can realise that goal
THE DIGITAL REVOLUTION

Every patient, like every rare disease is different and the way they manage their condition can vary

What the Doctor sees...

Patient A
Test 1
Patient B
Test 1

Test 2
6 mo. later

What the Patient experiences...

Patient A
Test 1

Patient B
Test 1

Test 2
6 mo. later
The patient voice is becoming more and more important, and it can shape the way that clinical studies can be developed.

Technology can support patient engagement and compliance through ease of use and with a patient focus design.

Studies can potentially be completed much more rapidly due to the amount of data and frequency of assessments, reduced site and patient burden.

Objective, high-frequency data, all captured in real time, to support endpoint validation whilst generating longitudinal data for real world evidence.
WHY IS THIS IMPORTANT?

STEP 1
Identify appropriate PROMs / PROs / COAs

STEP 2
Work with the patient community to develop treatments

STEP 3
Work with pharma to bring treatments to your community
The originally developed TUG assesses mobility, balance, walking ability, and fall risk in older adults.

- Patients wear their regular footwear and can use a walking aid, if needed.
- They begin by having the patient sit back in a standard arm chair and identify a line 3 meters, or 10 feet away, on the floor:
  - From word “Go” the patient is to:
    - Stand up from the chair
    - Walk to the line on the floor at your normal pace.
    - Turn 180 degrees
    - Walk back to the chair at your normal pace
    - Turn to Sit down again
- In the original method, the patient will be timed from the word ‘Go” until they sit down again.
MODIFIED TUG WITH COMPUTER VISION (vTUG)

The vTUG enables high frequency, objective assessment from the comfort of the patient’s home.

The Future of Assessing Disease Progression

By identifying key-points on the physical feature being measured in each frame, pattern recognition analysis can be performed via video analytics.

The software locates specific points on what is being measured and provides a confidence value associated with each point.

The analysis can compute the speed and trajectory of each movement, which can be used to explore smoothness of motion and erratic patterns.
MODIFIED TUG WITH COMPUTER VISION (VTUG)

Conducting a video recorded TUG (vTUG) as home and utilizing computer vision capabilities is expected to be a more sensitive way to measure disease presentation.

Computer vision techniques can be applied to extract parameters of interest such as postural angles, movement within a plane, gait measurements etc.

Automatic identification of the TUG components or gait phases are then feasible through machine learning classification algorithms.
The vertical location of the heel and toe during the walking section of the video is then reflected by patterns of movement - heel above toe, followed by toe above heel etc. and one can see the results to be evaluated in the graph.
ARM MOVEMENT
ALIGNMENT

Hip - knee - ankle

Angle (degrees)

Frame
MODIFIED TUG WITH COMPUTER VISION (vTUG)

Measuring disease progression in a way that is clinically meaningful is of utmost importance and this technology potentially provides a novel mechanism to support this.

Technology can be used to digitally collected data and computer vision software to update/modify the way assessments are used to calculate progression with the aim to provide more subjective, highly sensitive, frequent results.
QUESTIONS
WHAT CAN ERNS DO IN THE AREA OF QOL MEASUREMENT?

How to bring them in the context of the ERNs?
WHAT IS QUALITY OF LIFE

• Quality of life is determined by:

  • The extent to which hopes, and ambitions are matched by experience
  • Individuals’ perceptions of their position in life taken in the context of the culture and value systems where they live and in relation to their goals, expectations, standards, and concerns
  • Appraisal of one’s current state against some ideal
  • The things people regard as important in their lives

'WHAT DOES IT SHOW?

Do they simply describe a patient’s health in terms of what health professionals or society believe constitutes quality of life for people who are ill?

Do they capture patients’ perspectives of their disease and treatment, their perceived need for health care, and their preferences for treatment and outcomes?
WHAT ARE YOU ASKING?

- Patient centric
- Range of questions
- Innovative
- Disease specific
- Easy to answer

QoL QUESTIONNAIRE
BUILDING A PICTURE OF THE DISEASE IMPACT

ASKING THE PATIENT
Asking the patient means that you can build an accurate picture of the disease.

CREATING A MESSAGE
A QoL can build a message about the disease that can help improve an understanding of it.

FINDING A SOLUTION
We are all trying to find a treatment/cure for our disease and an effective QoL can help.
EQ-5D-5L – PAPER VERSION

Under each heading, please tick the ONE box that best describes your health TODAY.

MOBILITY
- I have no problems in walking about
- I have slight problems in walking about
- I have moderate problems in walking about
- I have severe problems in walking about
- I am unable to walk about

SELF-CARE
- I have no problems washing or dressing myself
- I have slight problems washing or dressing myself
- I have moderate problems washing or dressing myself
- I have severe problems washing or dressing myself
- I am unable to wash or dress myself

USUAL ACTIVITIES (e.g. work, study, housework, family or leisure activities)
- I have no problems doing my usual activities
- I have slight problems doing my usual activities
- I have moderate problems doing my usual activities
- I have severe problems doing my usual activities
- I am unable to do my usual activities

PAIN/DISCOMFORT
- I have no pain or discomfort
- I have slight pain or discomfort
- I have moderate pain or discomfort
- I have severe pain or discomfort
- I have extreme pain or discomfort

ANXIETY/DEPRESSION
- I am not anxious or depressed
- I am slightly anxious or depressed
- I am moderately anxious or depressed
- I am severely anxious or depressed
- I am extremely anxious or depressed

The next month’s health rating is:

- We would like to know how good or bad your health is TODAY.
- This scale is numbered from 0 to 100.
- 100 means the best health you can imagine.
- 0 means the worst health you can imagine.
- Please mark an X on the scale to indicate your health is TODAY.

Now, write the number you marked on the scale in the box below:

YOUR HEALTH TODAY =
THINGS HAVE CHANGED A LOT

The landscape for a patient and parent / carer has changed dramatically in the last 10 years

**Technology**

We have access to so much technology that can do so much more than 10 years ago.

**Social media**

Social media has become a big part of our life as we support each other and share information.

**Expectations**

Everyone has much high expectations now that they are better supported.
PAPER IS NOW DIGITAL
DIGITAL IS MORE INCLUSIVE
EQ-5D-5L – DIGITAL VERSION

Under each heading, please tick the ONE box that best describes your health TODAY.

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I have slight problems walking about
I have moderate problems walking about
I have severe problems walking about
I am unable to walk about

SELF-CARE
I have no problems washing or dressing myself
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Be prepared for a lot of work to develop something that on paper shouldn’t be too tough.

It’s not simple. You have to be open minded in your approach.

Teamwork makes the dreamwork – keep repeating this phrase (it will get tough)!

Teamwork is vital. You’re working with multiple stakeholders who all want input.

You can really enforce change in developing something for your community.

You can make a difference. You CAN make change if you are patient!
QUESTIONS

Contact dan@cats-foundation.org