Patient generated data

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Eurordis Open Academy

June 2025

TODAY

DATA

The why

Its importance to advocacy

We will talk through various data collection topics and how this can impact advocacy

EXPERIENCE

The how

Real examples of data & advocacy

All the topics today will be related back to personal and practical experiences of other advocacy groups

OUR PERSONAL JOURNEY

It is important to understand my experience and why I am speaking today. My family's rare disease journey began after our eldest daughter was born in 2009 as was then diagnosed with Tay-Sachs at 15 months of age.

OUR DIAGNOSIS EXPERIENCE

“

Bla bla bla…your daughter is going to die…*bla bla bla*

…and there is nothing we can do….*bla bla bla.*

”

THE FUTURE

“You probably won’t meet another family with a child affected by the disease.”

THE IMPACT OF TAY-SACHS

It is cruel. It is terminal.

It is important to capture the essence of the disease to drive forward to treatments.






A COMPLEX DISEASE

It is important to understand the complexity of the disease when think about the data collection required for it

TERMINAL DISEASES IN CHILDREN

Tay-Sachs and Sandhoff disease are terminal in children where they have a very short life expectancy

AFFECTS 1 IN 320,000

The carrier rate for the diseases is 1 in 300 and there are approximately 1 in 320,000 affected children born a year

REQUIRE 24 HOUR CARE

Children lose the ability to function independently, have regular seizures and require 24 hour care

THREE DISEASE CLASSIFICATIONS

Both diseases have an Infantile, Juvenile and Adult Onset form

GENETIC CONDITION

They are both genetic conditions which are passed onto a child through autosomal inheritance

AVERAGE LIFE EXPECTANCY OF 5 YEARS OF AGE

In the infantile form,the life expectancy is only 5 years of age whilst adults can live much longer but with support

THE CATS FOUNDATION AND OUR GOALS

FOUNDED IN 2011

Set-up after finding there was no charity providing support to families affected by Tay-Sachs and Sandhoff in the UK

RAISED OUR PROFILE WITHIN THE COMMUNITY

We reached out to families across Europe to create a strong community to support each other

LOOKED INTO RESEARCH

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

OUR GOAL

“Our goal is to save children from Tay-Sachs and Sandhoff disease.
We want to give them an opportunity to have a future.”

RESEARCH & TRIALS – 2011

In 2011 there was only one research project

1 Global | Gene therapy research for GM2 Gangliosidosis

WHAT COULD WE DO?

RAISED AWARENESS

We raised awareness within the pharmaceutical community to make people aware of the diseases

JOINED THE RESEARCH COMMUNITY

We raised awareness of the diseases in the research community so they would be interested in our diseases

SET-UP EUROPEAN CHARITIES

We could see the impact the power of numbers could have so we set-up charities throughout Europe and the ETSCC

DISEASE REGISTRY

We developed the largest disease specific registry for Tay-Sachs and Sandhoff in Europe

IDENTIFY WHAT WAS NEEDED

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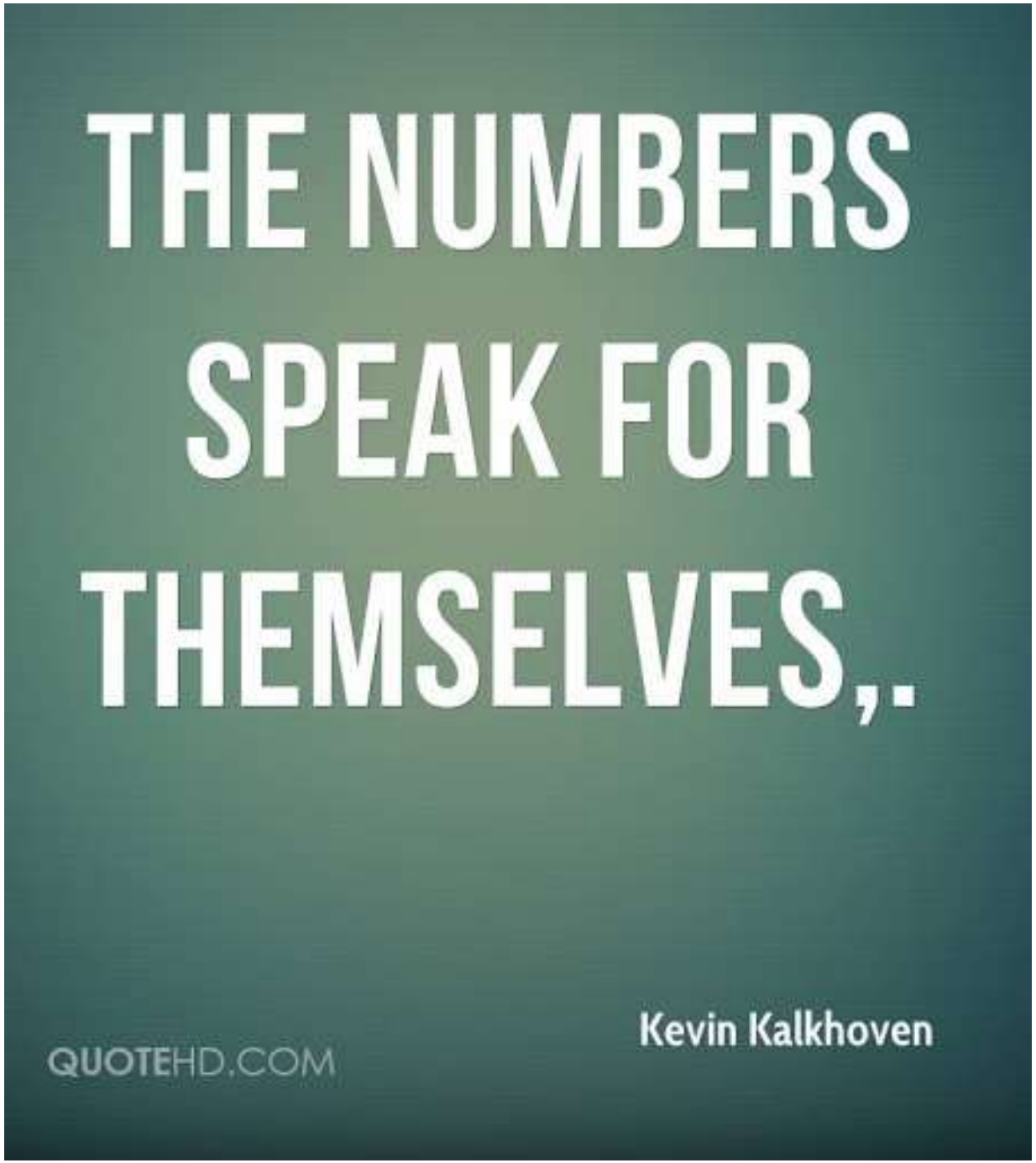

PEOPLE ARE THE KEY

YOU NEED TO SHOW THAT A DIFFERENCE CAN BE MADE

When you are the family who is 1 in a million (or 1 in 320,000) it hurts when you are told your disease is too rare for research.

OUR GLOBAL COMMUNITY

.....

Together, we are working to ensure that families have access to all the support they need. No family should feel isolated and without information about these devastating diseases.

ETSCC EUROPEAN TAY-SACHS & SANDHOFF CHARITY CONSORTIUM

GLOBAL GM1 & GM2 ALLIANCE

CATS CURE & ACTION FOR TAY-SACHS FOUNDATION

ACTAYS ACCIÓN Y CURA PARA TAY-SACHS

NTSAD National Tay-Sachs & Allied Diseases Association

CTSF Cure Tay-Sachs Foundation

DOCE DIVULGAR E ORIENTAR PARA COMBATER E ENFRENTAR A TAY-SACHS E OUTRAS GANGLIOSIDOSES ASSOCIAÇÃO NACIONAL

HAND in HAND gegen TAY-SACHS und SANDHOFF

THE MATHEW FORBES ROMER FOUNDATION SINCE 1998 EDUCATION & RESEARCH OF CHILDREN'S GENETIC DISEASES OF THE BRAIN

Rare Find FOUNDATION Tay-Sachs and Sandhoff Diseases

BLU GENES FOUNDATION

25 ans VML VAINCRE LES MALADIES LYSOSOMALES RARE MAIS PAS SEUL !

CATS CURE & ACTION FOR TAY-SACHS FOUNDATION

WHO ARE THE MISSING STAKEHOLDERS?

RESEARCH

The research teams are spread across the world and are all focusing on different treatment options

INDUSTRY

Industry can move forward the research and start clinical trials for a community to support an unmet need

PATIENTS

The most important part of all the discussions are the patients who are supported by the charities

ADVOCACY CAN BRING EVERYONE TOGETHER

ADVOCACY GROUPS

We represent the patients on a national and international level where we provide support to families

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RESEARCH & TRIALS – 2011

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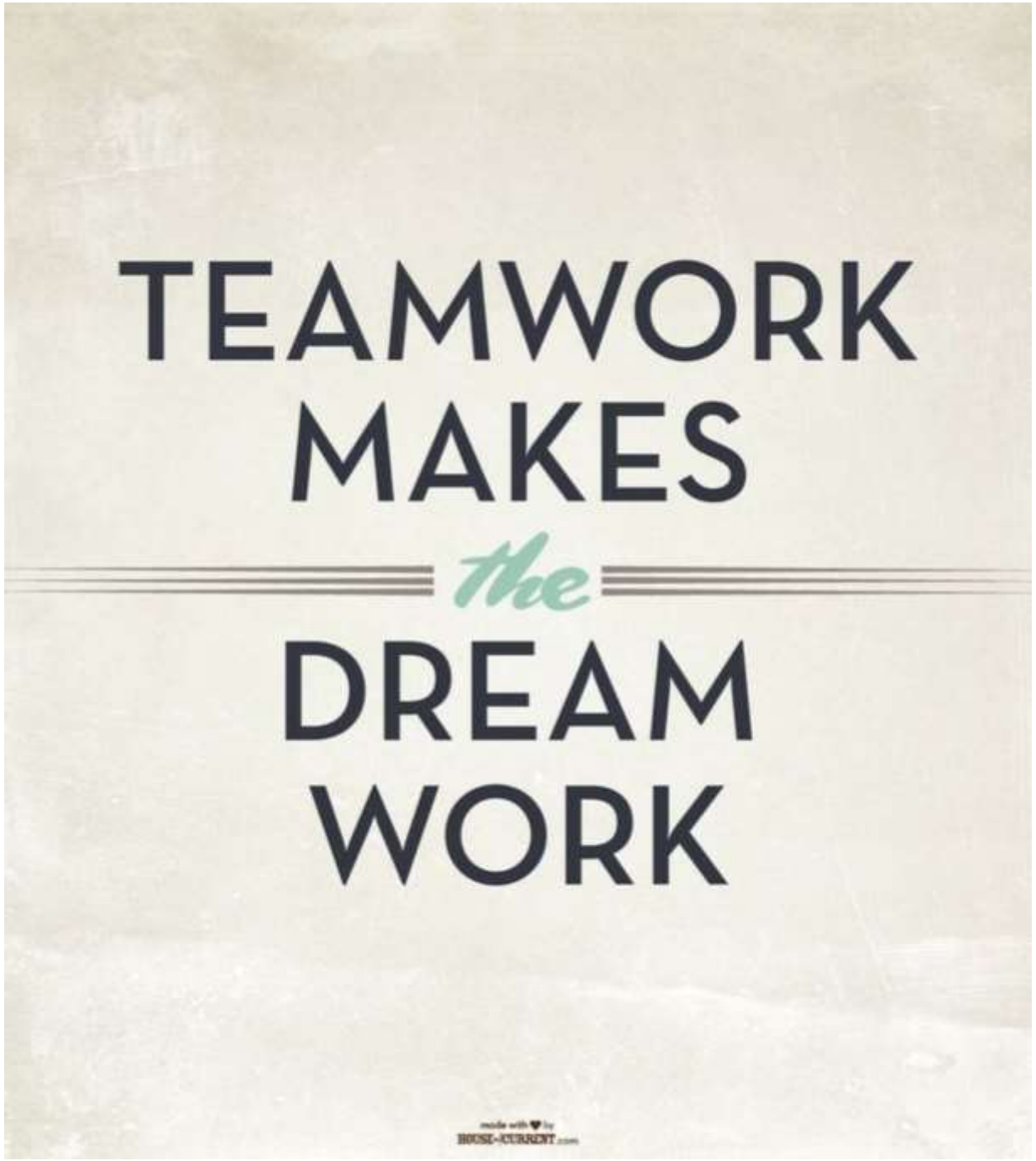
THE DREAM IN 2031

You need to set goals

! EUROPE | Multiple treatments for Tay-Sachs and Sandhoff



COLLABORATION IS KEY

TOGETHER WE CAN
DEVELOP A TREATMENT

Working as a team, advocacy, pharma, researchers and patients, can make a difference and really drive forward projects.

THE CONNECTION BETWEEN RWD, RWE & RWA

Real-World Data

RWD is:

- A tool
- Used in research

≠

Real-World Evidence

RWE is:

- Analysis
- Used in medicine

≠

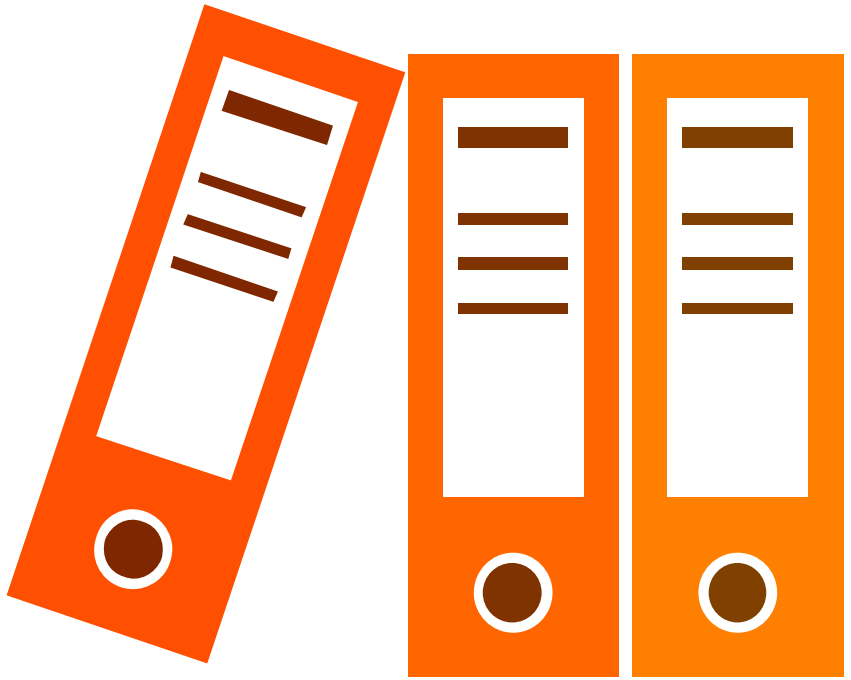
Real-World Answers

RWA is:

- Interpretability
- Used for decisions

Source: Patient advocates in Research (PAIR)

DO YOUR RESEARCH ON DATA



For any data collection you need to have a plan!

1. **What** is needed? Does something already exist?
2. **Why** is it needed? What impact will it have?
3. **How** can you do it? What are the blockers?

ACTIVITY – WHAT, WHY & HOW?

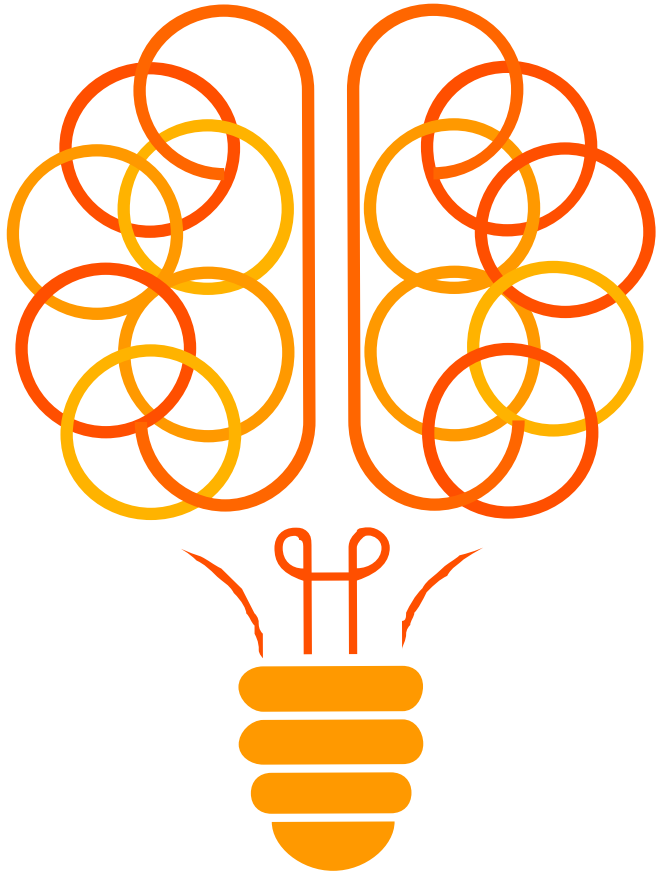
5 MINUTES

What is your community's "**what, why and how**" in relation to a registry?

ACTIVITY – DISCUSSION

5 MINUTES

WHAT WE WANTED TO ACHIEVE WITH DATA



What did we want to collect?

Why did we want to collect it?

How did we collect it?

- A Natural history of the disease
- B Advance treatments
- C Support research with pharma
- D Set up a patient reported registry

HOW TO GET THERE

There are steps to success to help your community

REGISTRY LAUNCH

Launch the registry to meet the goals of the registry

DEFINE THE DATA

Work on the data dictionary to ensure the right data is collected

IDENTIFY THE PLATFORM

Evaluate the platforms that can be used to launch the registry

pulse infoframe

OpenApp

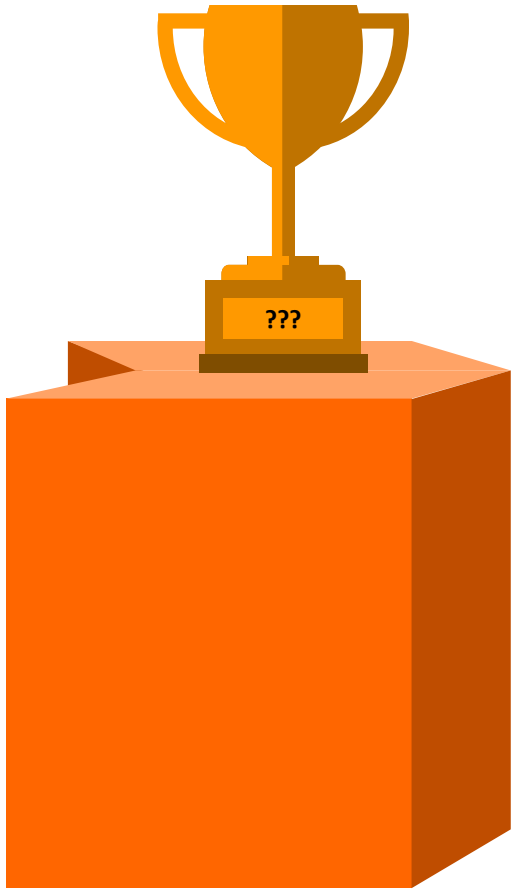
CRITICAL PATH INSTITUTE

REDCap

CATS

WHAT IS THE GOAL?

Define what you want to achieve



01 Treatment for the community?

02 Contact database to build a network?

03 Publications to answer natural history?

04 Clinical database for use by clinicians?

05 Revenue source for the advocacy group?

All you need is the plan, the road map, and the courage to press on to your destination.

(Earl Nightingale)

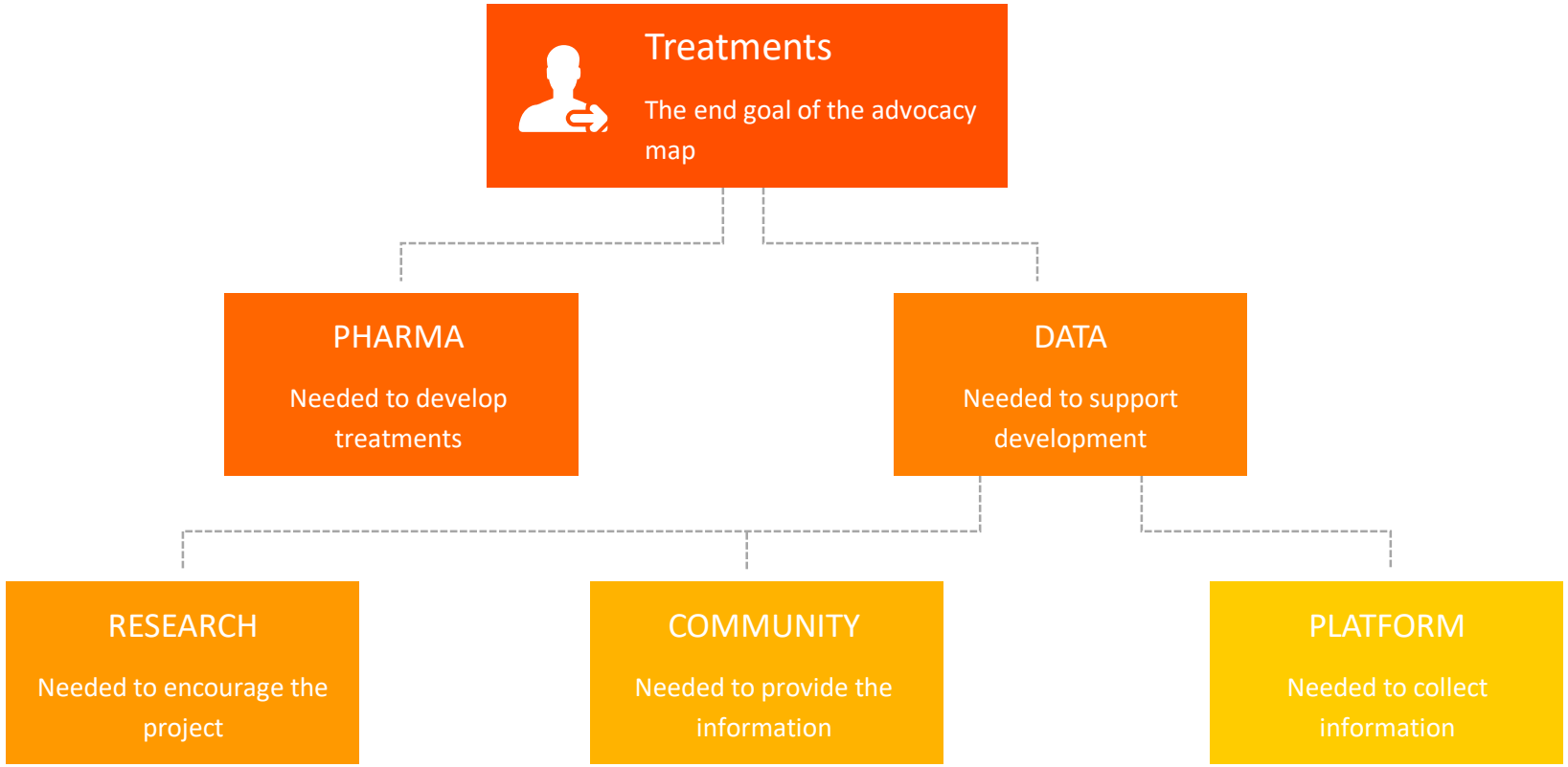
izquotes.com

THE DATA MAP

How do we get to our goal for our community?

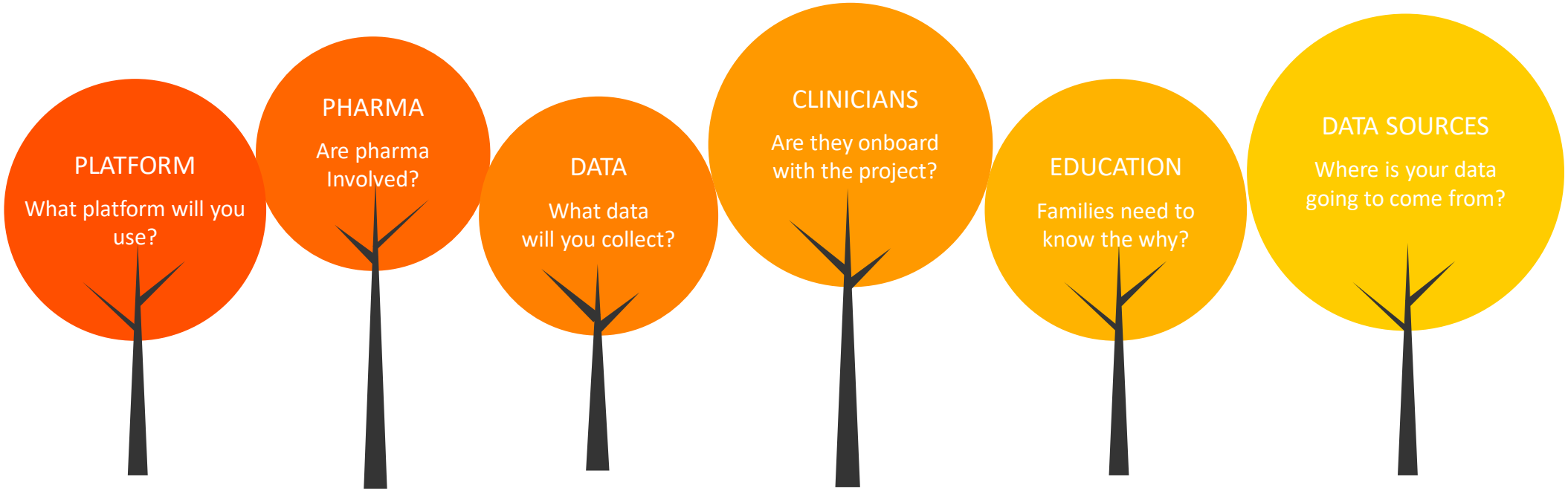
NOW YOU NEED TO MAP IT

A simple map of the key components

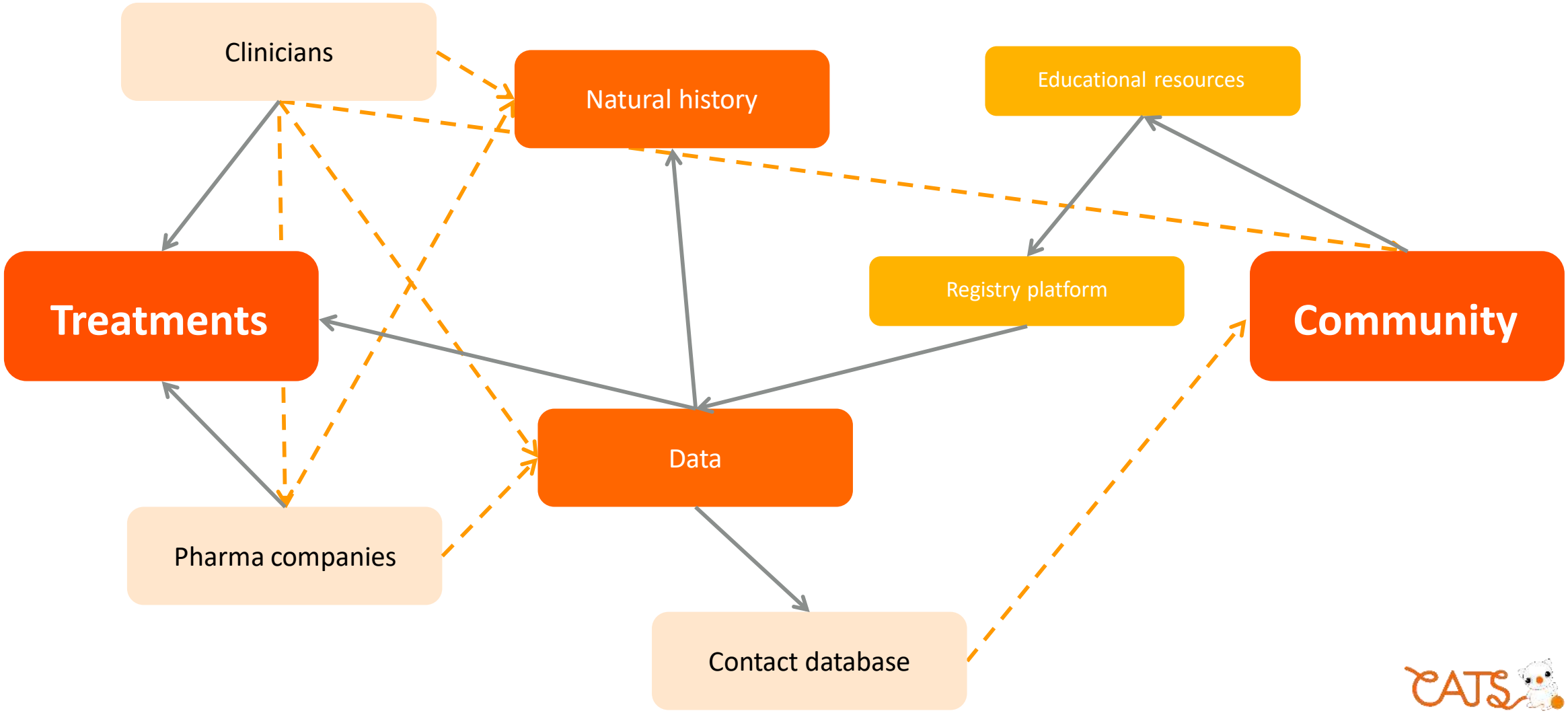


WHAT WILL HELP IT GROW?

What needs to be considered to grow the registry? Who needs to be engaged and at what point can they help? How are they connected?



EVERYTHING IS CONNECTED



ACTIVITY - DRAW YOUR MAP

5 MINUTES

Draw a map of the key stakeholders and how they are all connected

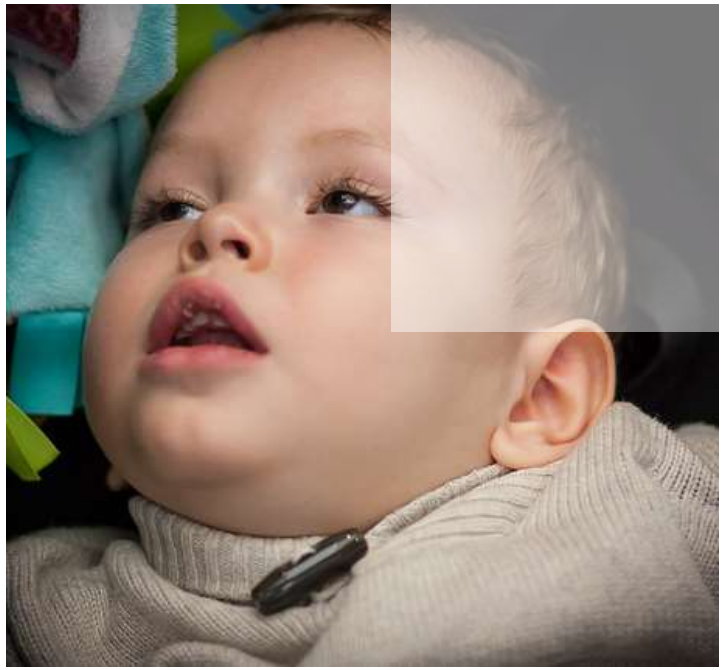
ACTIVITY – PRESENT YOUR MAP

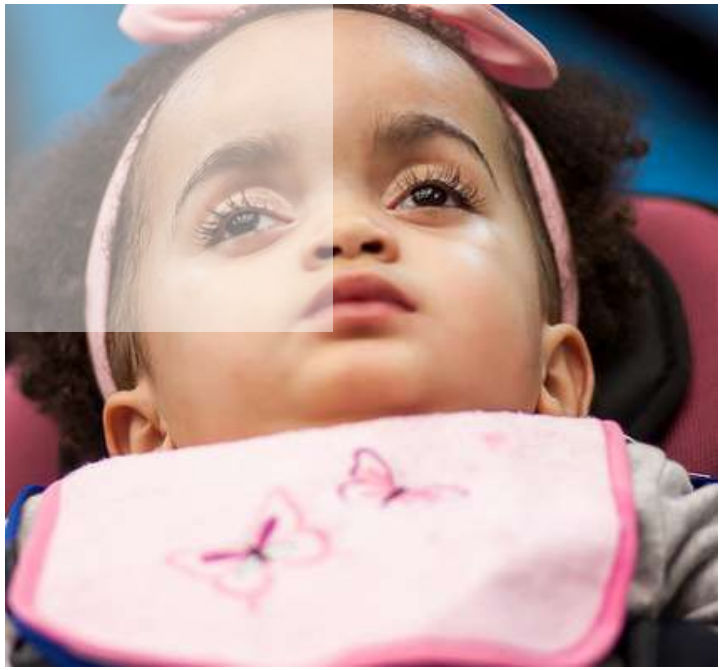
5 MINUTES

SOMEONE NEEDS TO DO THE WORK

THERE IS NO SUBSTITUTE
FOR HARD WORK

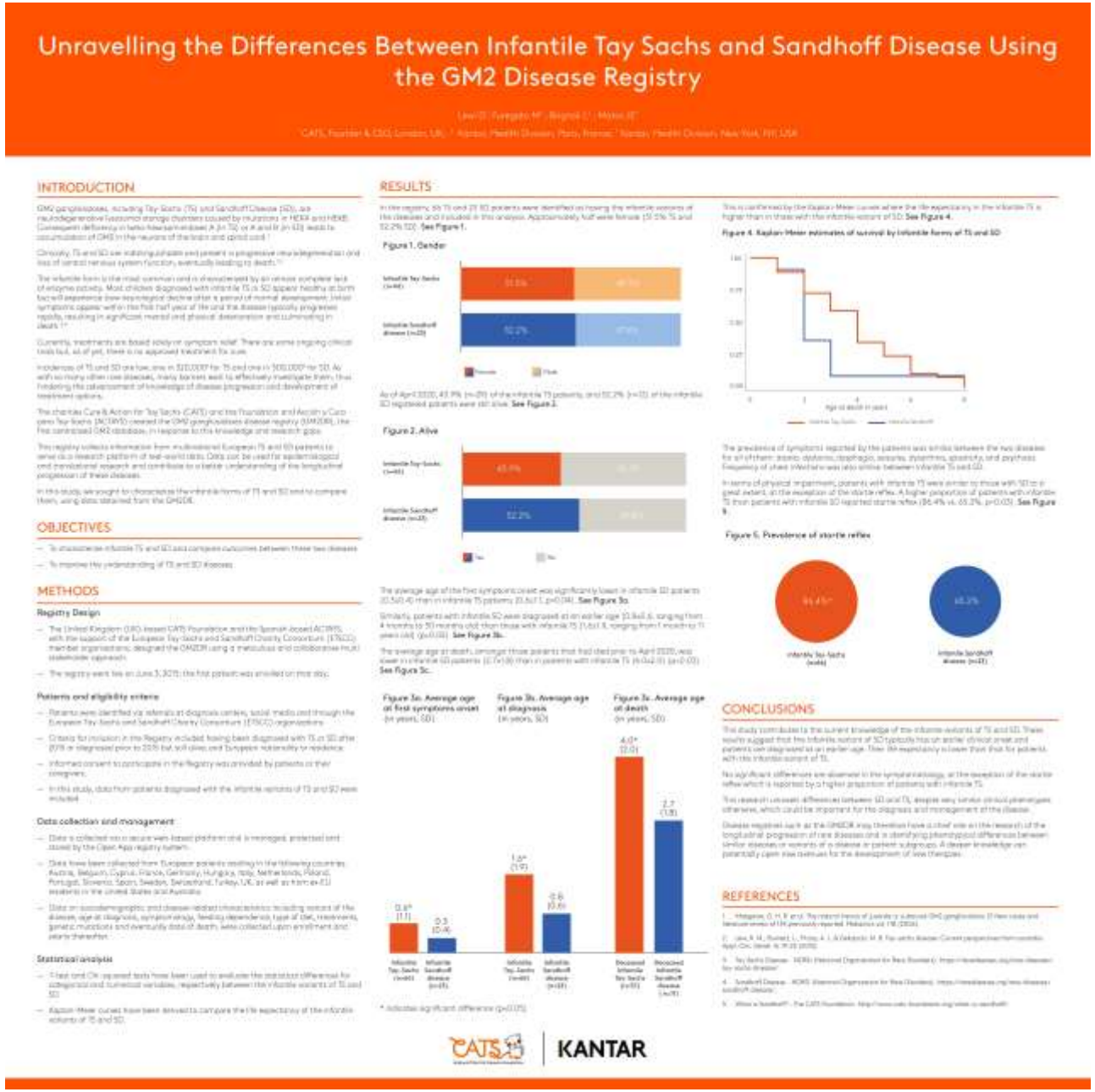
Unfortunately, data and collecting it does not happen by chance. It requires a lot of hard work, but the results are very rewarding and can make a huge difference to your community.





THESE GUYS ARE VITAL

These people should not be forgotten – they are why you are collecting data in the first place.

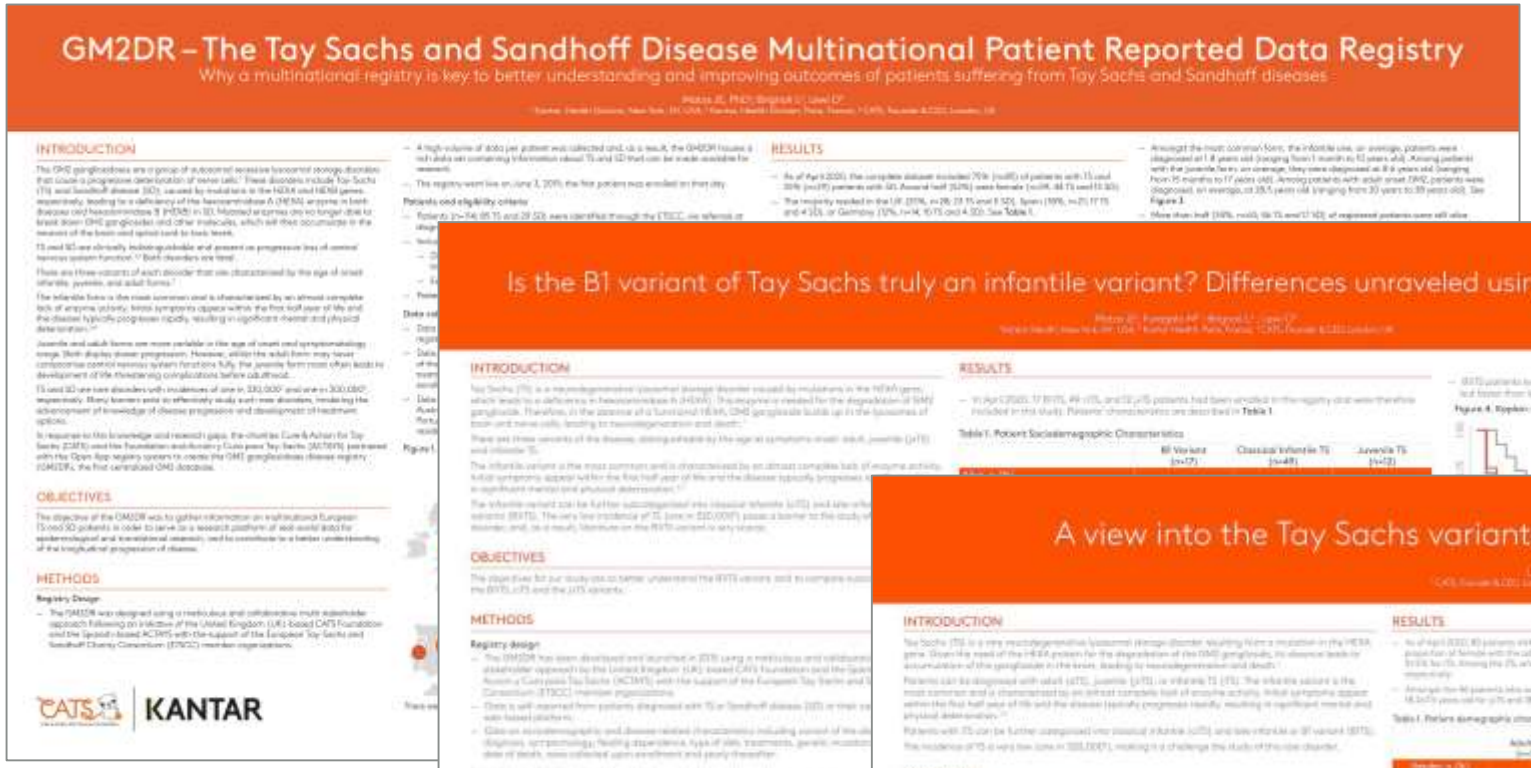


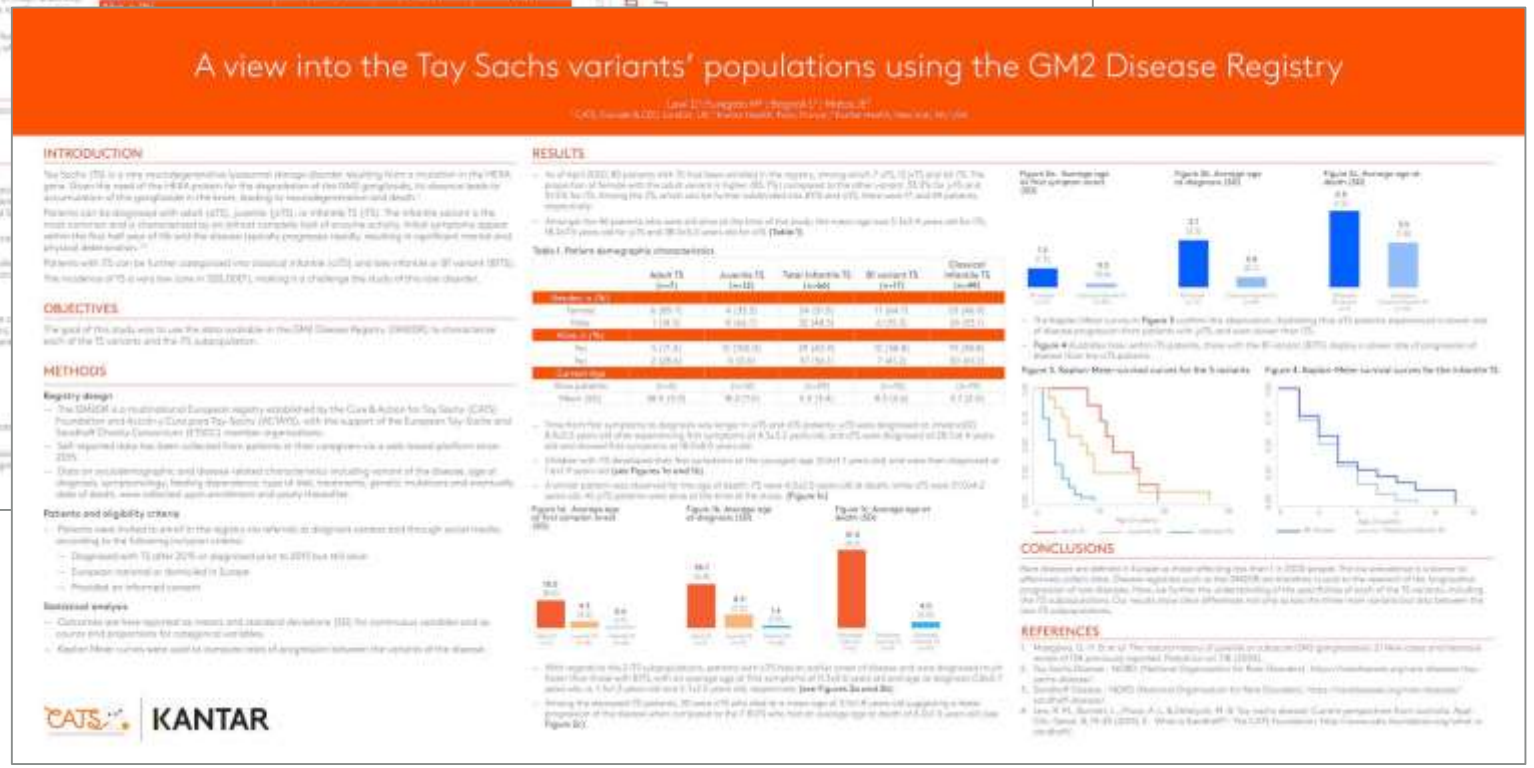
NEW THINGS DISCOVERED

WE CAN UNEARTH UNKNOWN THINGS ABOUT DISEASES

Collecting data is hard work but it can make a huge difference to your disease community if managed efficiently.

* We're Organizing Research on Lysosomal Diseases (WORLD) Conference 2021





National Organization for Rare Disorders (NORD) Conference 2020

International Conference for Pharmacoepidemiology (ICPE) 2021

The Professional Society for Health Economics and Outcomes Research (ISPOR) Conference 2021

CURE & ACTION FOR TAY SACHS FOUNDATION

RESEARCH & TRIALS – 2011

In 2011 there was only one research project

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RESEARCH & TRIALS – beginning 2022

There are now multiple studies ongoing and planned globally

7 Europe

- IB1001 clinical trial – drug repurposing
- AMETHIST trial – substrate reduction
- Gene transfer in a murine model of Sandhoff using AAV9
- Deployment of chaperone treatment for Tay-Sachs
- A new inflammasome complex treatment for GM2 Gangliosidosis
- Investigation into gene & cellular therapy treatment for GM2
- RETRIEVE – Natural History Study

4 Rest of World

- Gene therapy programme for GM2 Gangliosidosis – Taysha Gene Therapies
- Gene therapy programme for GM2 Gangliosidosis – Sio Gene Therapies
- IB1001 clinical trial – drug repurposing
- AMETHIST trial – substrate reduction

6 Upcoming

- Drug repurposing - pre-clinical
- ERT - pre-clinical
- PRONTO – Natural History Study
- REC-3599 for GM2 Gangliosidosis
- Small molecule – pre-clinical
- Stem cell – pre-clinical

azafaros sanofi IntraBio JCR Pharmaceuticals ALLIEVEX Polaryx Therapeutics idorsia TAYSHA GENE THERAPIES SIO GENE THERAPIES Recursion

RESEARCH & TRIALS – 2025

The rare disease market can be volatile – all the time!

5 Europe

- IB1001 clinical trial – drug repurposing
- Gene transfer in a murine model of Sandhoff using AAV9
- Deployment of chaperone treatment for Tay-Sachs
- A new inflammasome complex treatment for GM2 Gangliosidosis
- PRONTO – Natural History Study
- NAVIGATE – phase III

3 Rest of World

- IB1001 clinical trial – drug repurposing
- PRONTO – Natural History Study
- RAINBOW clinical trial – repurposing study

4 Upcoming

- NAVIGATE – phase III small molecule trial
- Drug repurposing - pre-clinical
- ERT - pre-clinical
- Stem cell – pre-clinical

azafaros JCR Pharmaceuticals IntraBio sanofi ALLIEVEX Ideaya TAYSHA Recursion

DATA CAN GIVE HOPE TO PEOPLE

THERE IS NOW LIGHT
AT THE END OF THE TUNNEL

We are now able to give families hope that there are treatments in the pipeline and that we are working towards giving people with Tay-Sachs and Sandhoff a future.

FINAL THOUGHTS

Things to consider when collecting data

INVOLVE YOUR COMMUNITY

Make sure your community is onboard and understands the project

DEFINE YOUR END GOAL

Define an achievable goal that meets the needs of your community

HOW MUCH DATA TO COLLECT

Too much data is as bad as too little data

MAKE SURE SUPPORT IS PROVIDED

It is vital that you support your community with the project

ASK FOR HELP

There are people who can help you on your journey

DECIDE ON THE RIGHT PLATFORM

The right platform is vital – the wrong platform can set you back

"To succeed, work hard, never give up and above all, cherish a magnificent obsession."

- Walt Disney

DANIEL LEWI

dan@cats-foundation.org

> We will beat these awful diseases so no other family has to go through the experience of losing a child.

CURE & ACTION FOR TAY-SACHS FOUNDATION