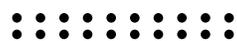




Patient generated data



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

REQUIRE 24 HOUR CARE

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

GENETIC CONDITION

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

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

THREE DISEASE CLASSIFICATIONS

Both diseases have an Infantile, Juvenile and Adult Onset form

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

Charities

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

Awareness

JOINED THE RESEARCH COMMUNITY

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

Registry

DISEASE REGISTRY

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

Research

IDENTIFY WHAT WAS NEEDED

.....



**THE NUMBERS
SPEAK FOR
THEMSELVES,.**

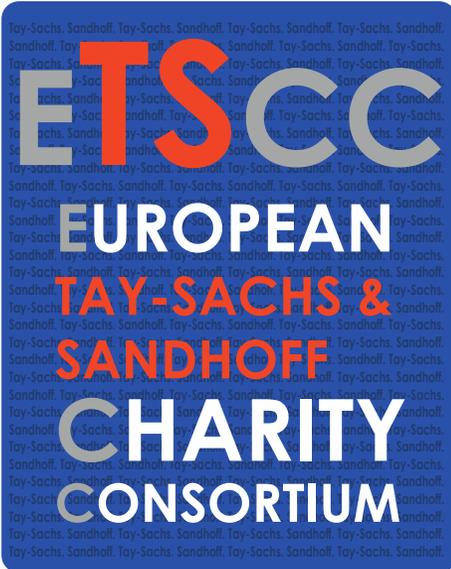
QUOTEHD.COM

Kevin Kalkhoven

**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.



WHO ARE THE MISSING STAKEHOLDERS?

.....



ADVOCACY CAN BRING EVERYONE TOGETHER

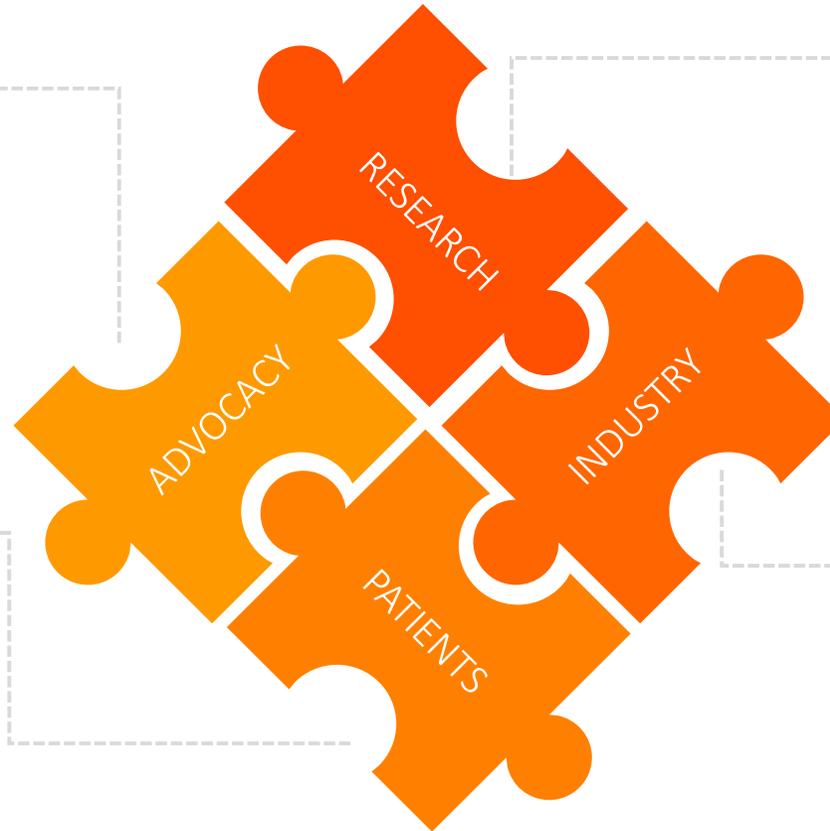
.....

ADVOCACY GROUPS

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

PATIENTS

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



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

RESEARCH & TRIALS – 2011

In 2011 there was only one research project

1

Global

Gene therapy research for GM2 Gangliosidosis



THE DREAM IN 2031

You need to set goals



EUROPE

Multiple treatments for Tay-Sachs and Sandhoff



**TEAMWORK
MAKES**

the

**DREAM
WORK**

made with ♥ by
HOUSE-CURRENT.COM

**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

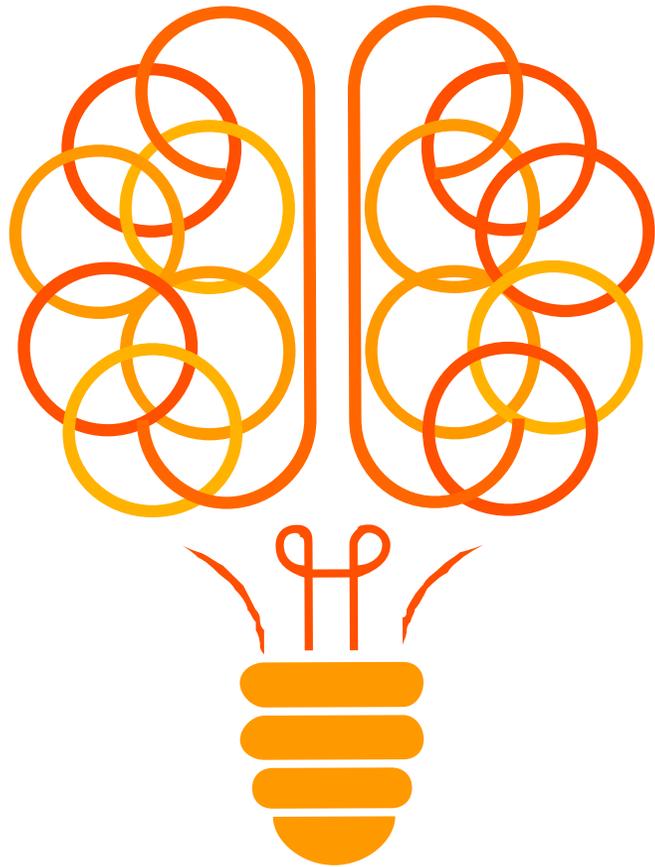


5 MINUTES



WHAT WE WANTED TO ACHIEVE WITH DATA

.....



What did we want to collect?

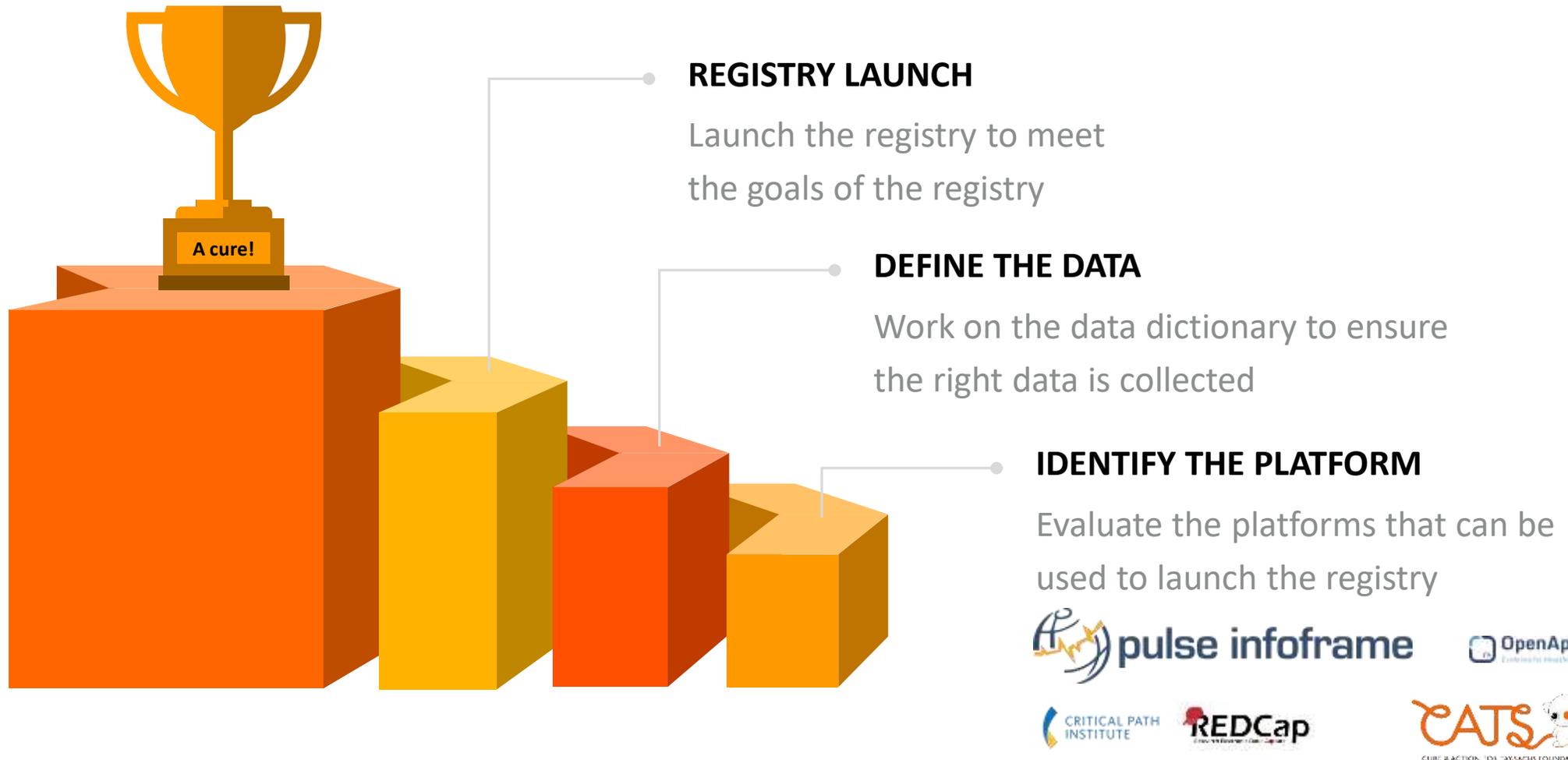
Why did we want to collect it?

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

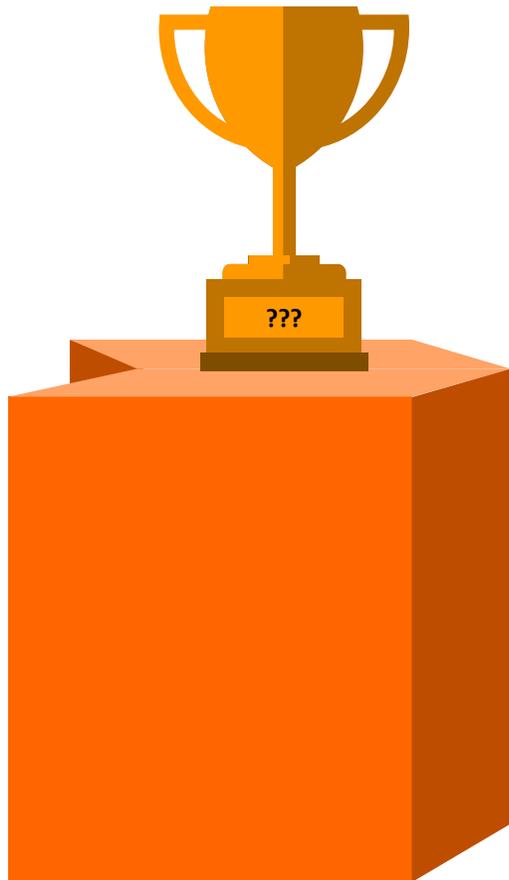
HOW TO GET THERE

There are steps to success to help your community



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

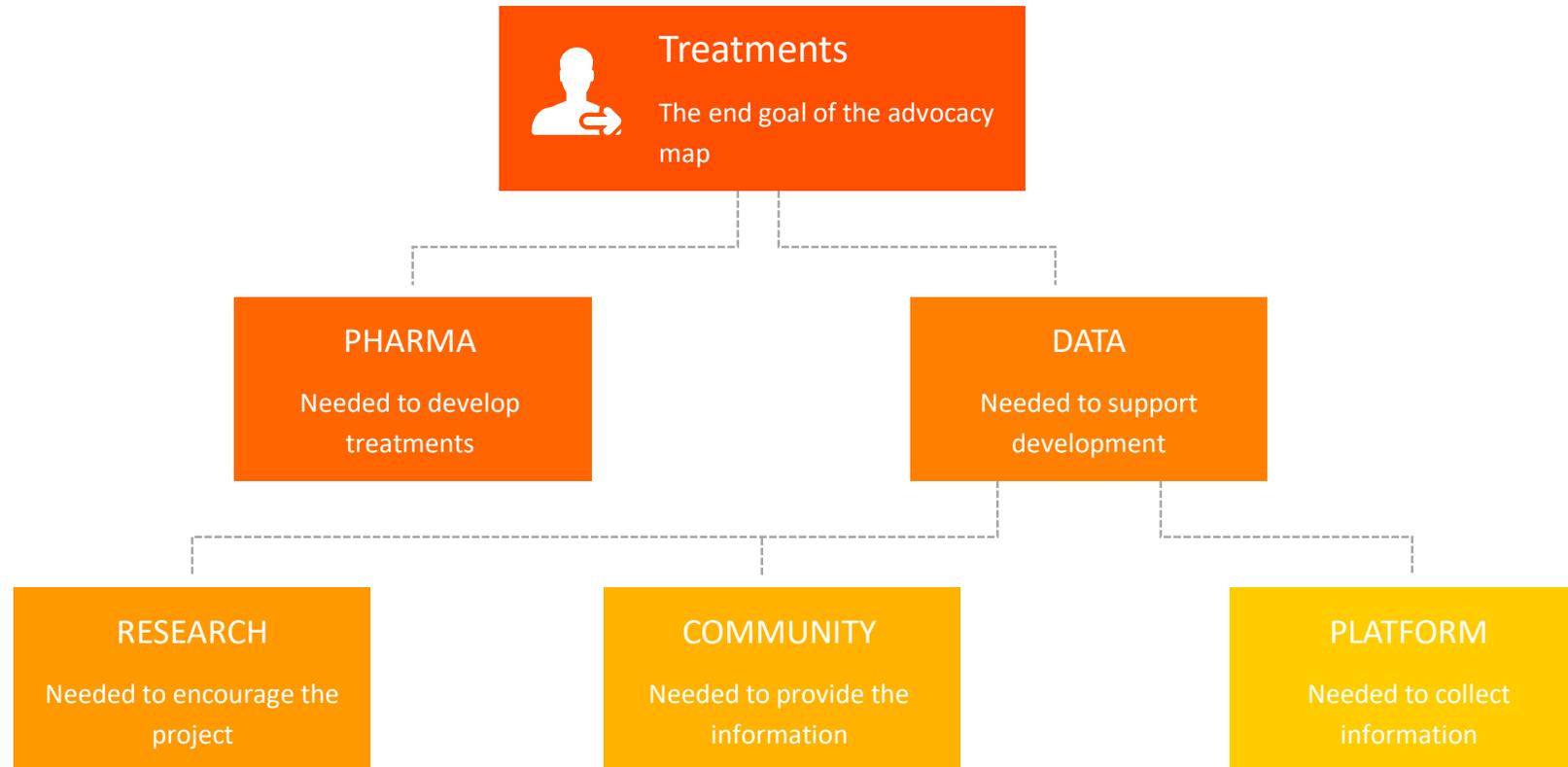
.....

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?



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



**SUCCESS COMES
TO THOSE
WHO WORK HARD,
LIFT HEAVY
AND NEVER GIVE UP.**

GYMQUOTES.CO

**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.

Unravelling the Differences Between Infantile Tay Sachs and Sandhoff Disease Using the GM2 Disease Registry

Laura C. Cappuccini, M.D., Richard C. Allen, M.D.

CATL, Heather & Co., London, UK; 1. National Health Discovery, Paris, France; 2. National Health Discovery, New York, NY, USA

INTRODUCTION

GM2 gangliosidosis, including Tay Sachs (TS) and Sandhoff Disease (SD), are neurodegenerative lysosomal storage disorders caused by mutations in HEXA and HEXB. Consequent deficiency in beta-hexosaminidase A (Hex A) or A and B (Hex B) leads to accumulation of GM2 in the neurons of the brain and spinal cord.¹

Classically, TS and SD are debilitating childhood-onset and progressive neurodegenerative and loss of central nervous system function, eventually leading to death.^{2,3}

The variable form in the infant-onset form is characterized by an atypical symptom set of encephalopathy. Most children diagnosed with infantile TS or SD appear healthy at birth but will experience slow developmental decline after a period of normal development. Infantile symptoms appear well within the first year of life and the disease typically progresses rapidly, resulting in significant mental and physical deterioration and ultimately in death.⁴

Currently, treatments are based on symptomatic relief. There are some ongoing clinical trials but, as of yet, there are no approved treatments for use. Incidences of TS and SD are low, with 1:10,000 for TS and one in 363,000 for SD. As with many other rare diseases, more factors need to be effectively investigated from, thus fostering the development of knowledge of disease progression and development of treatment options.

The charity Care & Action for Tay Sachs (CATS) and the Tay Sachs and Sandhoff Care Support Network (TSSN) created the GM2 gangliosidosis disease registry (GM2DR), the first centralized GM2 database, in response to the knowledge and research gaps.

This registry collects information from undiagnosed European TS and SD patients to serve as a research platform of real-world data. Data can be used for epidemiological and translational research and contribute to a better understanding of the longitudinal progression of these diseases.

In this study, we sought to characterize the variable forms of TS and SD and to compare them, using data obtained from the GM2DR.

OBJECTIVES

- To characterize infantile TS and SD and compare outcomes between these two diseases
- To improve the understanding of TS and SD diseases

METHODS

Registry Design

- The United Kingdom (UK), French CATS Foundation and the Spanish Society ACCIBTS, with the support of the European Tay Sachs and Sandhoff Charity Consortium (ETSSC), invited participants, diagnosed the GM2DR using a standardized and collaborative multi-stakeholder approach.
- The registry went live on June 1, 2021; the first patient was enrolled on that day.

Patients and eligibility criteria

- Patients were identified via referrals at diagnostic centers, social media and through the European Tay Sachs and Sandhoff Charity Consortium (ETSSC) organizations.
- Criteria for inclusion in the Registry included being born diagnosed with TS or SD after 0/18 or diagnosed prior to 2/20 but still alive and European nationality or residence.
- Informed consent to participate in the Registry was provided by patients or their caregivers.
- In this study, data from patients diagnosed with the infantile variants of TS and SD were included.

Data collection and management

- Data is collected into a secure web-based platform and is managed, protected and stored by the Cloud App registry system.
- Data have been collected from European countries residing in the following countries: Austria, Belgium, Czechia, France, Germany, Hungary, Italy, Maldives, Poland, Portugal, Slovenia, Spain, Sweden, Switzerland, Turkey, UK, as well as non-EU residents in the United States and Australia.
- Data on demographics and disease-related characteristics including variant of the disease, age at diagnosis, symptoms, feeding dependencies, type of diet, treatments, genetic mutations and eventually date of death, were collected upon enrollment and were transferred.

Statistical analysis

- Chi-squared and t-test were used to evaluate the statistical difference for categorical and numerical variables, respectively between the variable forms of TS and SD.
- Exact tests could have been performed, however the low expectancy of the variable counts of TS and SD.

RESULTS

In the registry, 55 TS and 21 SD patients were identified on having the infantile variants of the diseases and included in this analysis. Approximately half were female (51.8% TS and 52.4% SD). (See Figure 1).

Figure 1. Gender



As of April 2022, 43.6% (n=20) of the infantile TS patients, and 52.4% (n=11) of the infantile SD registered patients were still alive. (See Figure 2).

Figure 2. Alive



The average age of the first symptoms onset was significantly lower in infantile SD patients (3.5(±1.4) years) in infantile TS patients (5.5(±1.1), p<0.001). (See Figure 3a).

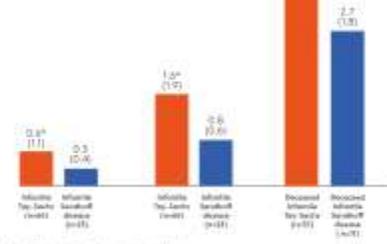
Similarly, patients with infantile SD were diagnosed at an earlier age (3.5(±1.4), ranging from 4 months to 50 months) and older than those with infantile TS (5.5(±1.1), ranging from 1 month to 71 months) (p<0.001). (See Figure 3b).

The average age at death amongst those patients that had died prior to April 2022, was lower in infantile SD patients (1.7(±0.9) years) in patients with infantile TS (4.0(±1.0), p<0.001). (See Figure 3c).

Figure 3a. Average age of first symptoms onset (in years, SD)

Figure 3b. Average age of diagnosis (in years, SD)

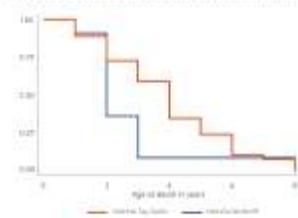
Figure 3c. Average age of death (in years, SD)



* Indicates significant difference (p<0.05)

This is confirmed by Kaplan-Meier curves where the infantic form of the infantile TS is longer than that with the infantile variant of SD. (See Figure 4).

Figure 4. Kaplan-Meier estimates of survival by infantile forms of TS and SD



The prevalence of symptoms reported by the patients was similar between the two diseases for all of them: autistic symptoms, epilepsy, seizures, dyspraxia, dysphagia, and psychosis. Frequency of these clinical signs also varies between infantile TS and SD.

In terms of physical impairment, patients with infantile TS were similar to those with SD in regard to infant, at the evaluation of the variable reflex. A higher proportion of patients with infantile TS than patients with infantile SD reported normal reflex (56.4% vs. 52.4%, p=0.02). (See Figure 5).

Figure 5. Prevalence of startle reflex



CONCLUSIONS

This study contributes to the current knowledge of the infantile variants of TS and SD. These results suggest that the infantile variant of SD typically has an earlier clinical onset and patients can stay well up to an earlier age. This is especially relevant for patients with the infantile variant of TS.

The significant differences are due to the heterogeneity of the disease of the infantile reflex which is reported by a higher proportion of patients with infantile TS.

The results presented differences between SD and TS, despite very similar clinical symptoms, which could be important for the diagnosis and management of the disease.

Disease registries such as the GM2DR may therefore have a crucial role in the discovery of the longitudinal progression of rare diseases and in identifying phenotypic differences between similar diseases or variants of a disease in patient subgroups. A deeper knowledge can potentially open new avenues for the development of new therapies.

REFERENCES

1. Hagerman, J. H. & Gill, T. S. (1999) Tay Sachs disease: A review of the clinical course and management of the previously reported literature. *PLoS ONE*, 14(12), e0243111.
2. Allen, R. C., Sunzel, L., Wang, A. J., & Gokhale, M. B. (2019) Tay Sachs Disease: Current perspectives and management. *PLoS ONE*, 14(12), e0243111.
3. Tay Sachs Disease. (2021) National Organization for Rare Diseases (NORD). <https://rarediseases.org/rare-diseases/tay-sachs-disease/>
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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.

GM2DR – The Tay Sachs and Sandhoff Disease Multinational Patient Data Registry

Why a multinational registry is key to better understanding and improving outcomes of patients suffering from Tay Sachs and Sandhoff diseases

PROF. DR. FRANK LANGER, MD, PhD, Director, GM2DR
 Prof. Dr. Frank Langer, MD, PhD, Director, GM2DR, University Hospital, Vienna, Austria

INTRODUCTION

The GM2 gangliosidosis are a group of autosomal recessive lysosomal storage disorders that cause a progressive deterioration of nerve cells. These disorders include Tay Sachs (TS) and Sandhoff disease (SD), caused by mutations in the HEXA and HEXB genes, respectively, leading to a deficiency of the hexosaminidase A (HEXA) enzyme in both disorders and hexosaminidase B (HEXB) in SD. Milder variants are no longer able to break down GM2 ganglioside and other molecules, which will then accumulate in the neurons of the brain and spinal cord to toxic levels.

TS and SD are clinically indistinguishable at onset on progressive loss of central nervous system function. Both disorders are fatal.

There are three variants of each disorder that are distinguished by the age of onset: infantile, juvenile, and adult forms.

The infantile form is the most common and is characterized by an almost complete lack of enzyme activity. Initial symptoms appear within the first half year of life and the disease typically progresses rapidly, resulting in significant mental and physical deterioration.

Juvenile and adult forms are more variable in the age of onset and symptomatology range. Both diseases progress more slowly. However, within the adult form may occur a progressive motor neuron system dysfunction. The juvenile form may occur after birth or development of the respiratory complications before adulthood.

TS and SD are rare disorders with incidences of one in 300,000 and one in 300,000, respectively. Many families seek to identify early on-set rare disorders, resulting in the advancement of knowledge of disease progression and development of treatment options.

In response to this knowledge and research gaps, the Chair for Care & Action for Tay Sachs (CATS) and the European Association for Care for Tay Sachs (EACTS) partnered with the Open App registry sponsors to create the GM2 gangliosidosis disease registry (GM2DR), the first centralized GM2 disorders.

OBJECTIVES

The objective of the GM2DR was to gather information on multinational European GM2DR patients in order to have a research platform of well-validated and fully epidemiological and translational of research, used to contribute to a better understanding of the long-term progression of diseases.

METHODS

Registry Design

- The GM2DR was designed using a multi-step and collaborative multi-submitter approach following an initiative of the United Kingdom (UK) based CATS Foundation and the Spanish-based ACERTS with the support of the European Tay Sachs and Sandhoff Charity Consortium (ETSCC) member organizations.



RESULTS

- A high volume of 8000 per patient was collected and as a result, the GM2DR has a rich data set containing information about TS and SD that can be widely used for research.
- The registry was live in June 1, 2021, the first patient was enrolled in the study.

Patients and eligibility criteria

- Patients with TS and SD were identified through the ETSCC or referred to the registry.
- Patients with TS and SD were included in the registry if they met the following criteria:

- Age of onset (SD) 18 years or older, or TS 18 years or older
- Diagnosis confirmed by genetic testing or enzyme assay
- European residence or domicile in Europe
- Provided an informed consent

RESULTS

- As of April 2022, the complete dataset included 706 (54%) of patients with TS and 306 (39%) patients with SD. Annual total (2021) cases female (48, 68 TS and 133 SD).
- The majority resided in the UK (37%, n=26, 23 TS and 3 SD), Spain (19%, n=21) IT (17) and 4 (2%), or Germany (12%, n=14, 10 TS and 4 SD, see Table 1).
- More than half (54%, n=473) of registered patients were still alive.

OBJECTIVES

The objective of this study was to use the data available in the GM2DR to characterize the TS variants and the SD subpopulation.

METHODS

The goal of this study was to use the data available in the GM2DR to characterize the TS variants and the SD subpopulation.

RESULTS

The infantile variant is the most common and is characterized by an almost complete lack of enzyme activity. Initial symptoms appear within the first half year of life and the disease typically progresses rapidly, resulting in significant mental and physical deterioration.

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Is the B1 variant of Tay Sachs truly an infantile variant? Differences unraveled using the GM2 Disease Registry

PROF. DR. FRANK LANGER, MD, PhD, Director, GM2DR
 Prof. Dr. Frank Langer, MD, PhD, Director, GM2DR, University Hospital, Vienna, Austria

INTRODUCTION

Tay Sachs (TS) is a neurodegenerative lysosomal storage disorder caused by mutations in the HEXA gene, which leads to a deficiency in hexosaminidase A (HEXA). It is categorized into three variants: infantile, juvenile, and adult. However, the presence of a functional HEXA, GM2 gangliosidosis (GM2) up to the symptoms of their infantile variant, leading to neurodegeneration and death.

There are three variants of the disease, distinguished by the age at symptom onset: infantile (IT) and juvenile (JT).

The infantile variant is the most common and is characterized by an almost complete lack of enzyme activity. Initial symptoms appear within the first half year of life and the disease typically progresses rapidly, resulting in significant mental and physical deterioration.

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A view into the Tay Sachs variants' populations using the GM2 Disease Registry

PROF. DR. FRANK LANGER, MD, PhD, Director, GM2DR
 Prof. Dr. Frank Langer, MD, PhD, Director, GM2DR, University Hospital, Vienna, Austria

INTRODUCTION

Tay Sachs (TS) is a neurodegenerative lysosomal storage disorder resulting from a mutation in the HEXA gene. Over the years, the HEXA protein has been identified in the GM2 ganglioside, its absence leads to accumulation of the ganglioside in the brain, leading to neurodegeneration and death.

Patients can be diagnosed with infantile (IT), juvenile (JT) or infantile TS (IT). The infantile variant is the most common and is characterized by an almost complete lack of enzyme activity. Initial symptoms appear within the first half year of life and the disease typically progresses rapidly, resulting in significant mental and physical deterioration.

Patients with IT can be further categorized into classical infantile (IT) and variant (ITV). The incidence of ITV is very low (around 10%), making it a challenge to study the true extent of the disease.

OBJECTIVES

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RESULTS

As of April 2022, 8000 patients with TS had been enrolled in the registry, among which 7,473 (93%) and 527 (7%), the infantile variant (IT) and the variant (ITV) respectively. The majority of patients with ITV were registered in the UK (37%, n=26, 23 IT and 3 ITV), Spain (19%, n=21) IT (17) and 4 (2%), or Germany (12%, n=14, 10 IT and 4 ITV, see Table 1).

Table 1. Patient Sociodemographic Characteristics

	IT Variant (n=7473)	Classical Infantile IT (n=448)	Infantile IT (n=2925)
Female (%)	48.2	48.2	48.2
Male (%)	51.8	51.8	51.8
Age at onset (years)	1.2	1.2	1.2
Survival (%)	54.0	54.0	54.0

Figure 1. Kaplan-Meier survival curves for the Infantile IT



Figure 2. Kaplan-Meier survival curves for the Variant IT



Figure 3. Kaplan-Meier survival curves for the Juvenile IT



Figure 4. Kaplan-Meier survival curves for the Adult IT



Figure 5. Kaplan-Meier survival curves for the Infantile IT



Figure 6. Kaplan-Meier survival curves for the Variant IT



Figure 7. Kaplan-Meier survival curves for the Juvenile IT



Figure 8. Kaplan-Meier survival curves for the Adult IT



Figure 9. Kaplan-Meier survival curves for the Infantile IT



Figure 10. Kaplan-Meier survival curves for the Variant IT



Figure 11. Kaplan-Meier survival curves for the Juvenile IT



Figure 12. Kaplan-Meier survival curves for the Adult IT

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



RESEARCH & TRIALS – 2011

In 2011 there was only one research project

1

Global

Gene therapy research for GM2 Gangliosidosis



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

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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





RESEARCH & TRIALS – 2025

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

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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

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Rest of World

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

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Upcoming

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





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



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