ePAG EXCHANGE OF GOOD PRACTICES

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CURE & ACTION FOR TAY-SACHS FOUNDATION

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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.

THE BEGINNING
OF OUR STORY AT
THE CATS FOUNDATION

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Most small charities are started because of one person.
In our case it was Amélie.

THE CATS FOUNDATION WAS BORN

CATS
CURE & ACTION FOR
TAY-SACHS FOUNDATION

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

OUR EUROPEAN CONSORTIUM

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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.

TEAMWORK MAKES *the* DREAM WORK

made with ♥ by
HOUSE of CURRENT.com

COLLABORATION IS KEY

TOGETHER WE CAN
DEVELOP A TREATMENT

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

RESEARCH & TRIALS – 2011

In 2011 there was only one research project

1 UK | Gene therapy programme for GM2 Gangliosidosis - *The University of Cambridge*

RESEARCH & TRIALS – 2021

Huge progress in 10 years

7 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*

2 Outside Europe

Gene therapy programme – *Taysha*

Gene therapy programme – *Sio Gene Therapies*

3 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

TERMINOLOGY

1

Understand the different types of data that can be collected

DISEASE

2

Understand the disease from all stakeholder view points

ENGAGEMENT
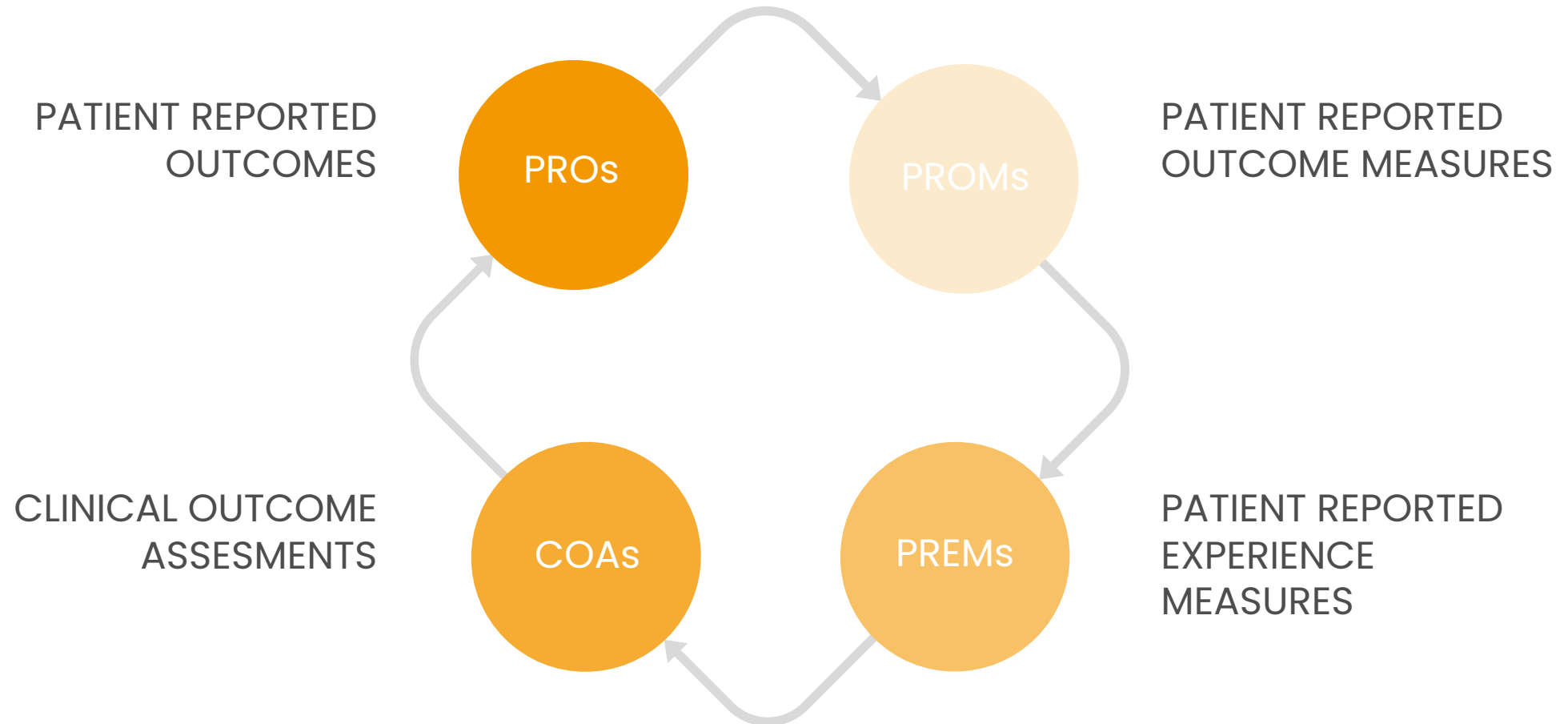
3

Understand how best to engage with all stakeholders

COLLABORATION

4

Understand how collaborating can make a big difference

WHAT IS WHAT?

PATIENT REPORTED
OUTCOMES

PROs

PROMs

PATIENT REPORTED
OUTCOME MEASURES

CLINICAL OUTCOME
ASSESMENTS

COAs

PREMs

PATIENT REPORTED
EXPERIENCE
MEASURES

GOAL SETTING

WHO'S INVOLVED IN THE GOAL?

Defining who is involved in achieving the goal is vital

WHAT IS THE GOAL?

We need to set a goal which is achievable

HOW DO WE WORK TO THE GOAL?

How do we work towards achieving the goal

HOW DO WE SET THE GOAL?

What is the process at setting the goal

WHO ARE THE STAKEHOLDERS?

CLINICIANS

A vital member in the process

PATIENTS

The most important member

PATIENT GROUPS

The group who can make things happen

PHARMA

The group who can start the process
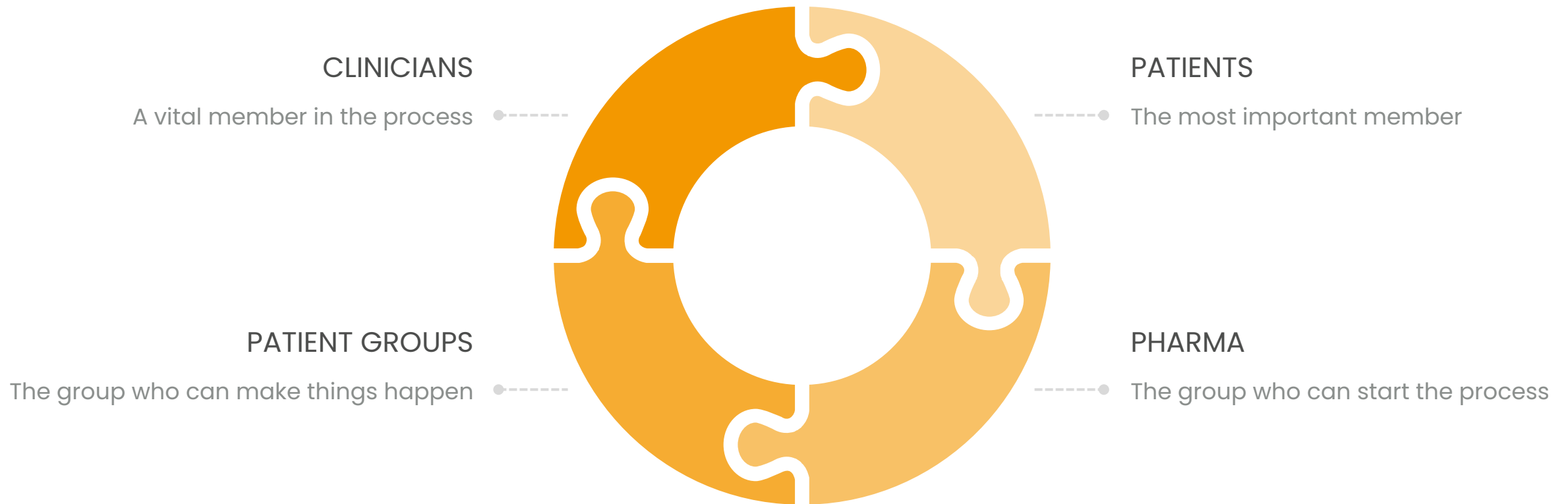
SETTING THE GOAL
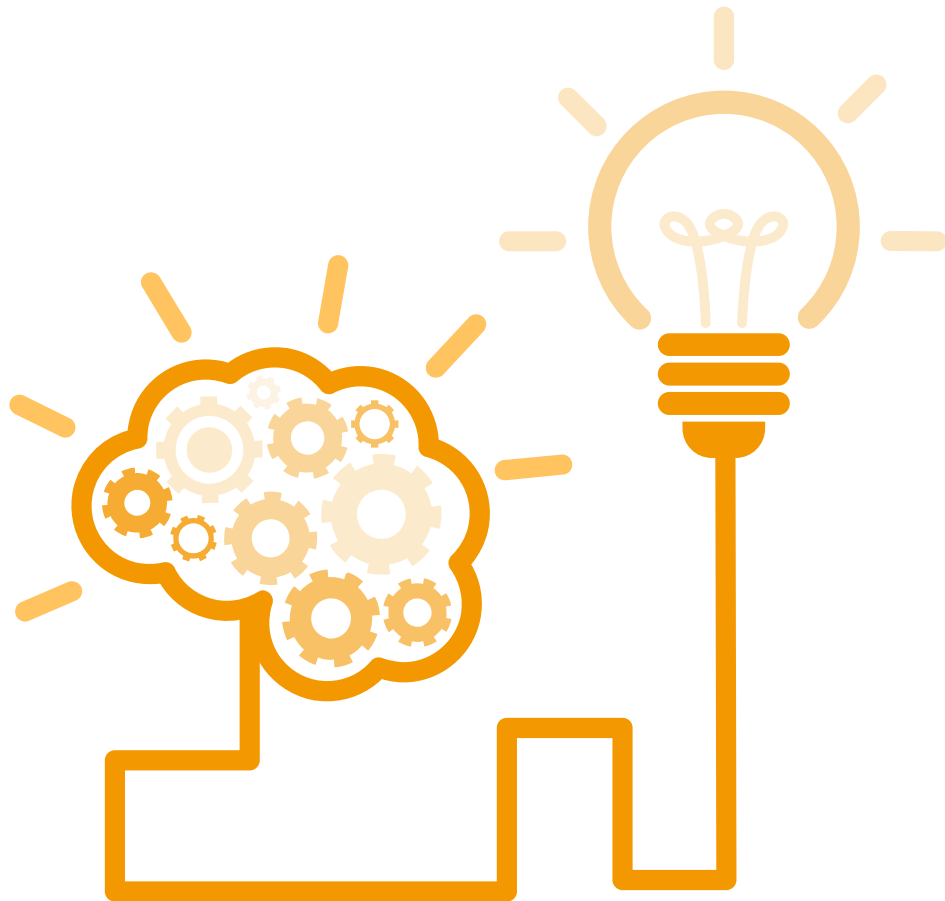
LISTEN

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
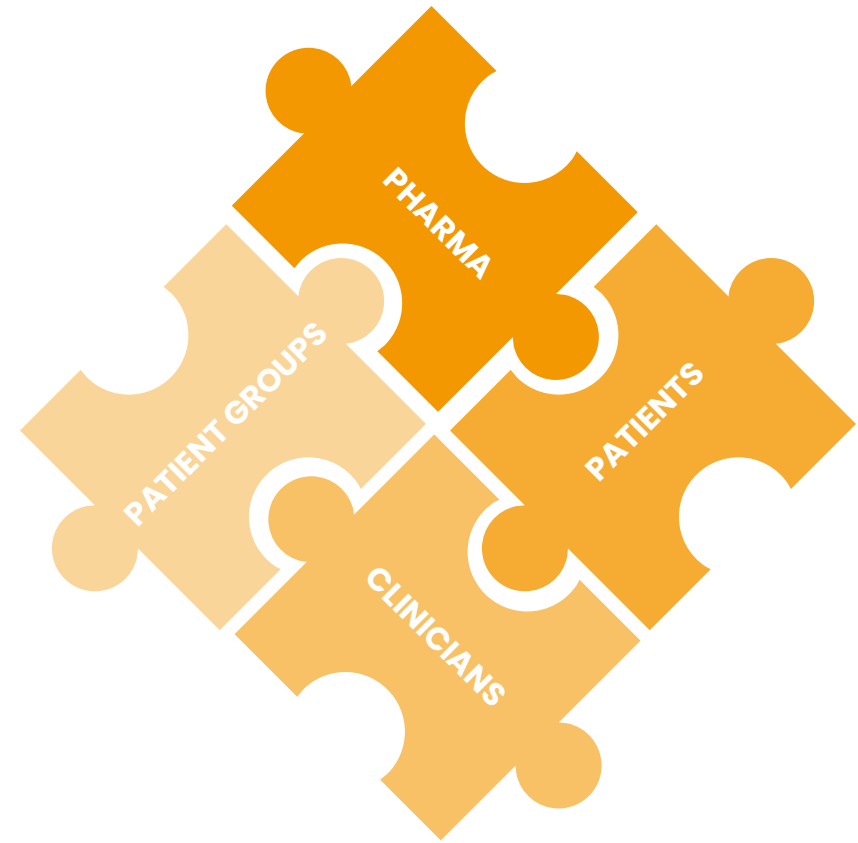
BE BRAVE

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

HOW DO WE WORK TOGETHER?

The bump on the road is an opportunity to overcome challenge

www.silviarossi.com

PHARMA

PATIENT GROUPS

PATIENTS

CLINICIANS

COLLABORATION

Relevance is all a matter of perspective.

What is important to you may not be to me and vice versa.

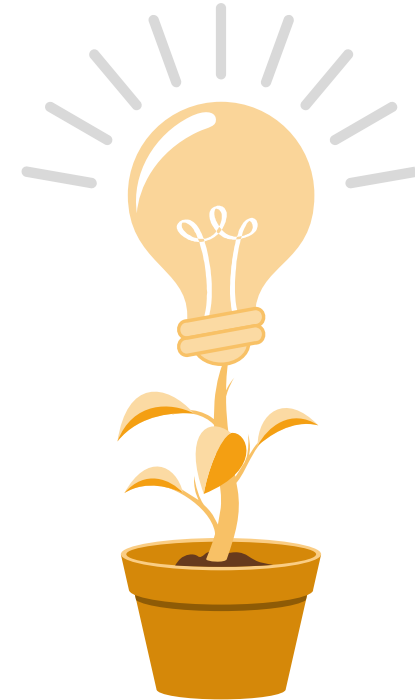# NURTURING THE IDEA AND MAKING IT GROW

PLANT

Your collaborative idea is "planted" together as a group

NURTURE

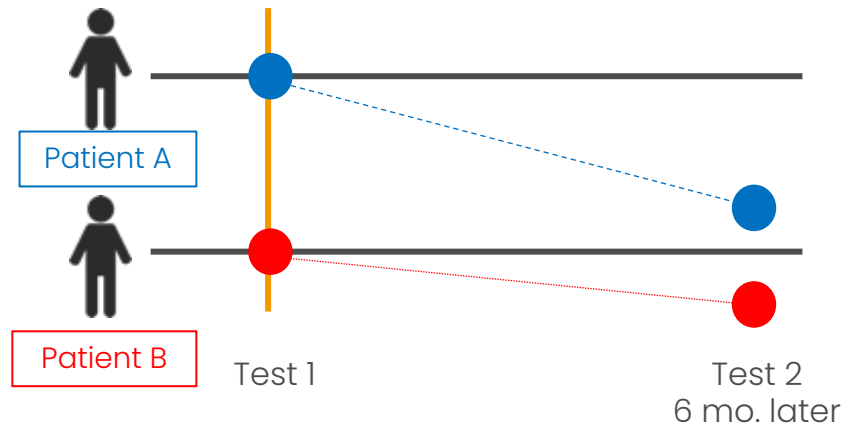You can all nurture the idea to make it grow in to reality

REALISE

If you all work together you can realise that goal

THE DIGITAL REVOLUTION

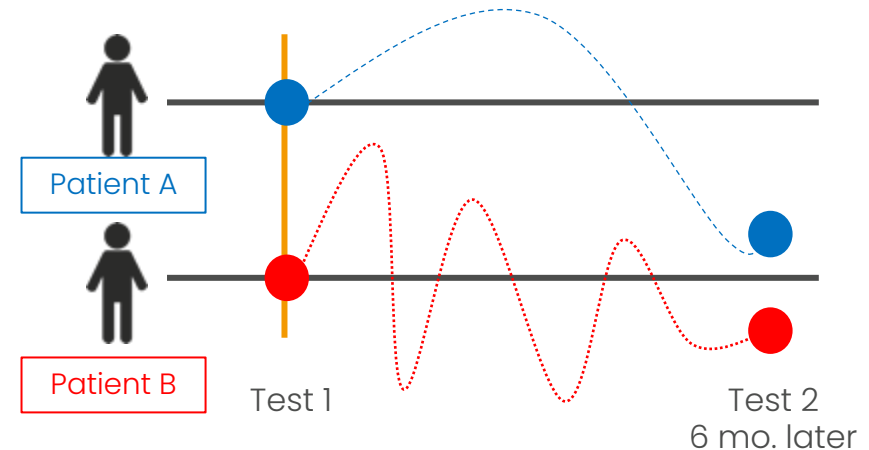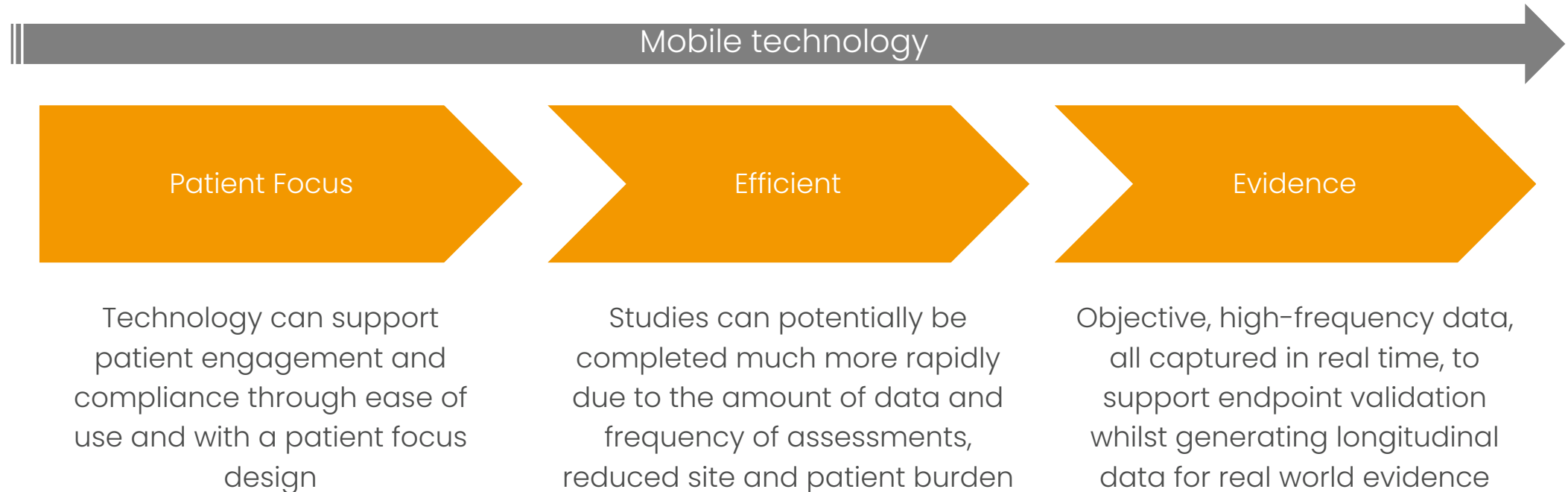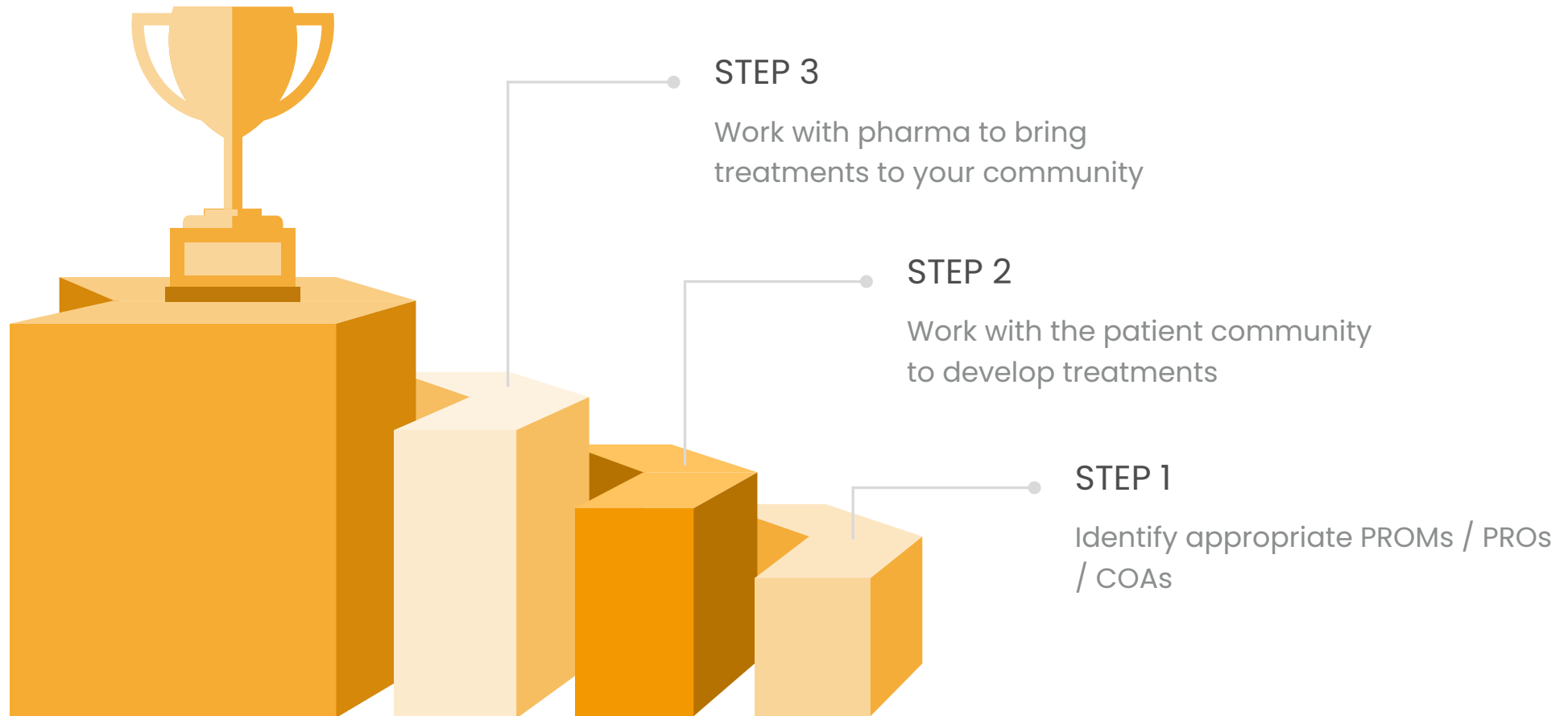Every patient, like every rare disease is different and the way they manage their condition can vary

TECHNOLOGY AND THE PATIENT

The patient voice is becoming more and more important, and it can shape the way that clinical studies can be developed

Mobile technology

Patient Focus

Efficient

Evidence

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 3

Work with pharma to bring treatments to your community

STEP 2

Work with the patient community to develop treatments

STEP 1

Identify appropriate PROMs / PROs / COAs

TIMED UP AND GO (TUG)

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

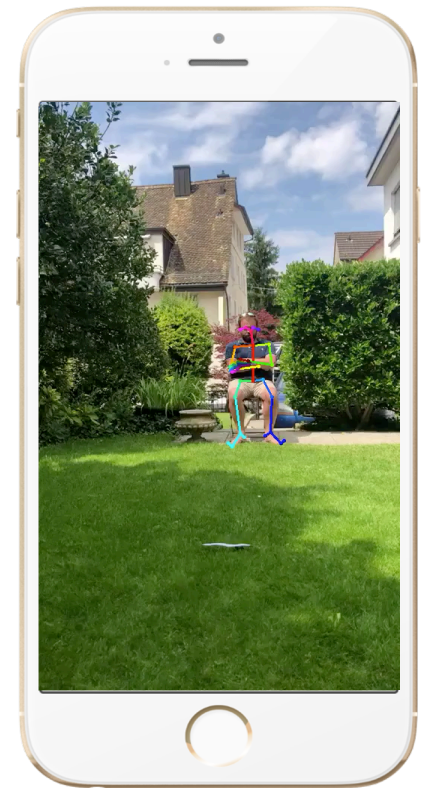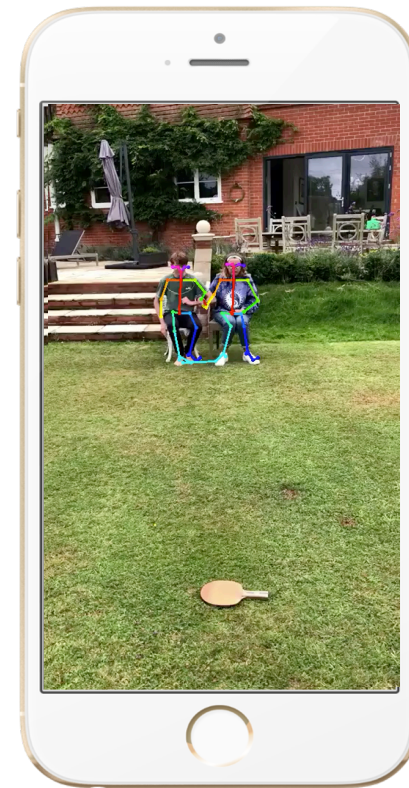# MODIFIED TUG WITH COMPUTER VISION (vTUG)

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
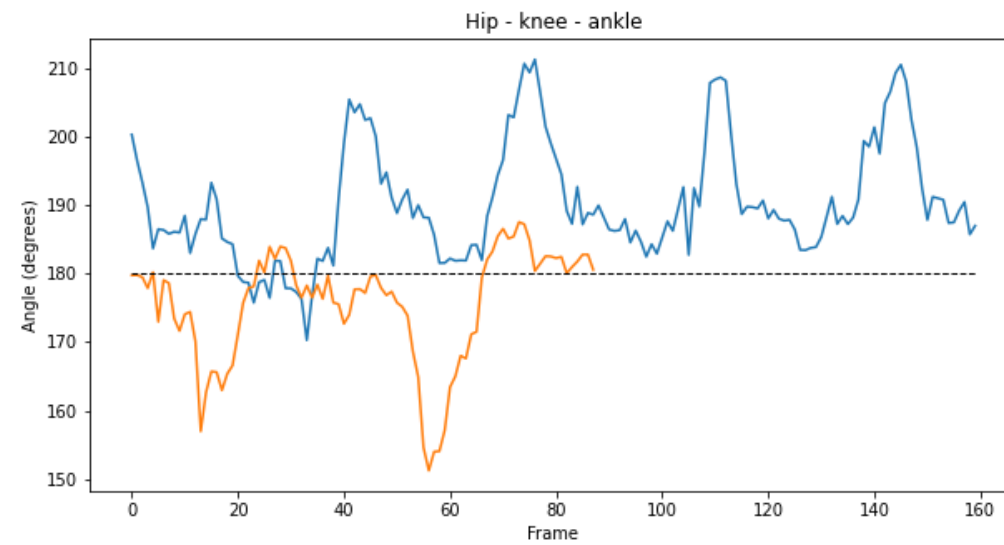
HEEL-TOE ALIGNMENT
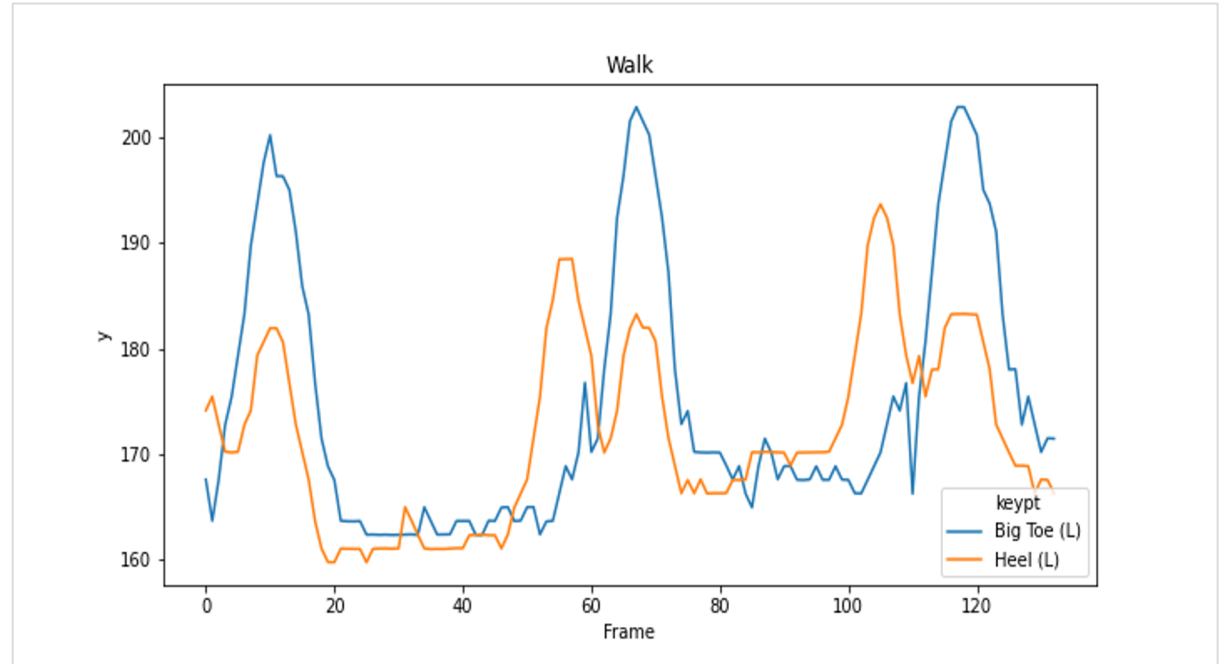
The vertical location of the heel and toe during the walking section of the video is then reflected by patterns of movement - heel above toe, followed by toe above heel etc. and one can see the results to be evaluated in the graph

ARM MOVEMENT

ALIGNMENT

Hip - knee - ankle
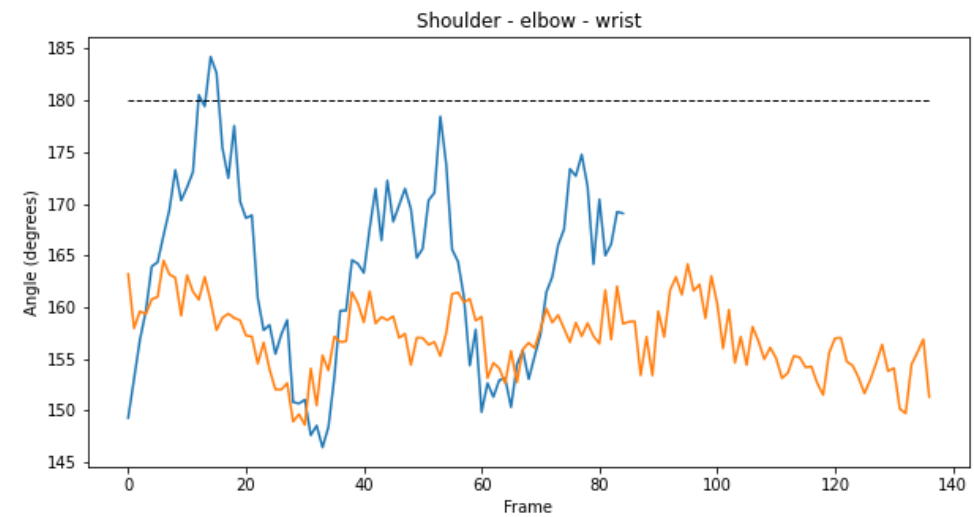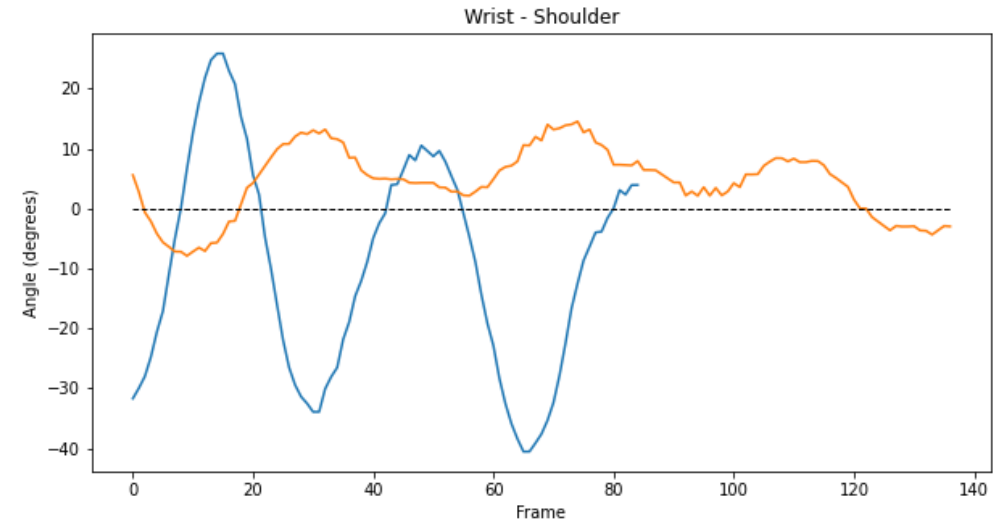
MODIFIED TUG WITH COMPUTER VISION (vTUG)

Measuring disease progression in a way that is clinically meaningful is of utmost of 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

Carr AJ, Higginson IJ. Are quality of life measures patient centred?. *BMJ*. 2001;322(7298):1357–1360. doi:10.1136/bmj.322.7298.1357
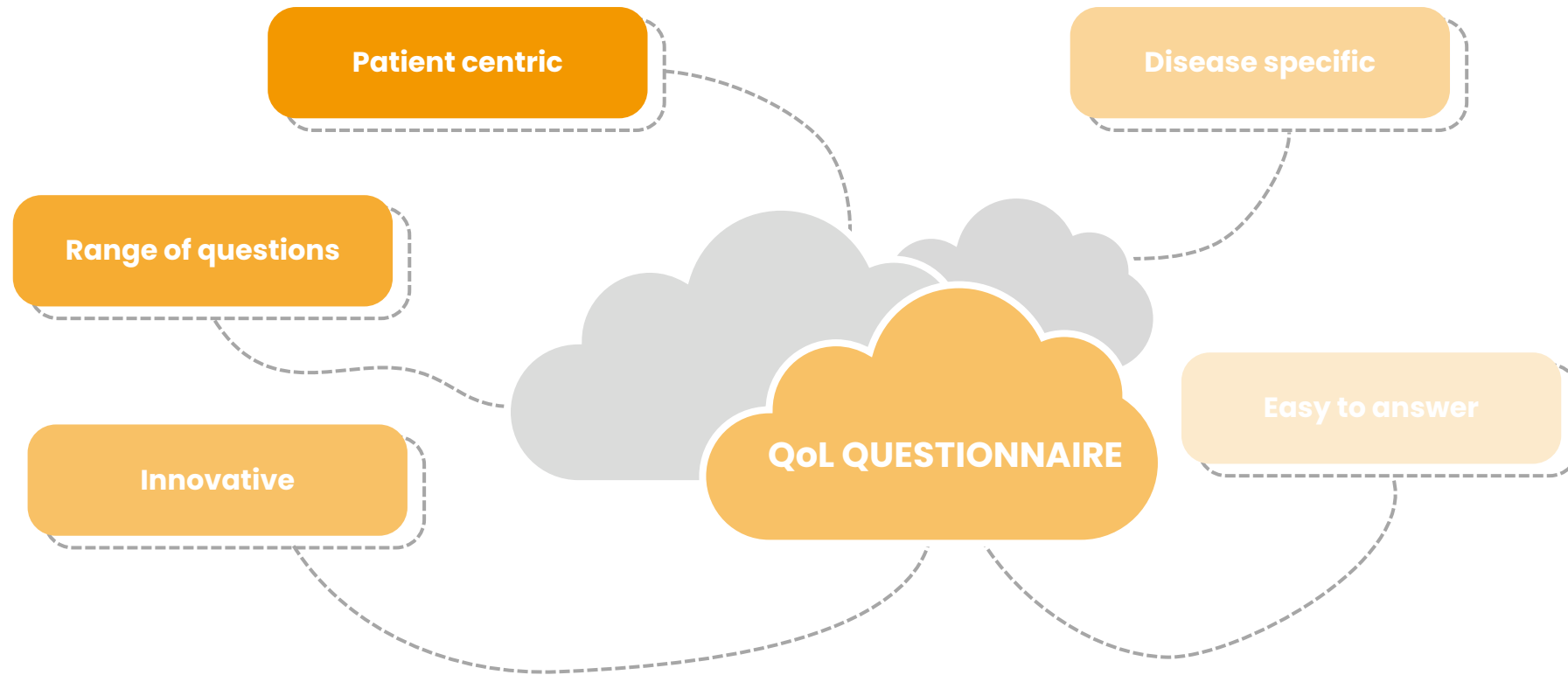
WHAT DOES IT SHOW?

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

Disease specific

Range of questions

Innovative

QoL QUESTIONNAIRE

Easy to answer

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 the 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

EQ-5D-5L

Health Questionnaire

English version for the UK

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 ☐

- We would like to know how good or bad your health is TODAY.

- This scale is numbered from 0 to 100.

- 100 means the <u>best</u> health you can imagine.
  0 means the <u>worst</u> health you can imagine.

- Please mark an X on the scale to indicate how your health is TODAY.

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

YOUR HEALTH TODAY = ☐

The best health
you can imagine

100
95
90
85
80
75
70
65
60
55
50
45
40
35
30
25
20
15
10
5
0
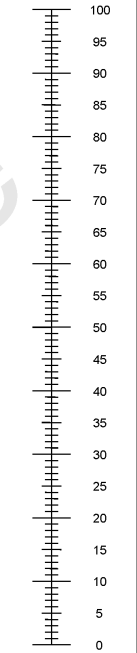
The worst health
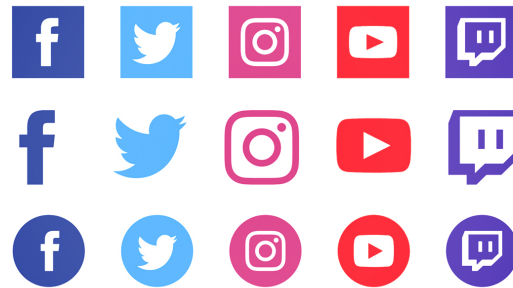you can imagine

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
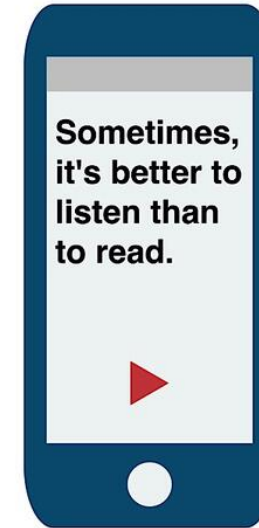
Expectations

Everyone has much high expectations now that they are better supported

PAPER IS NOW DIGITAL

DIGITAL IS MORE INCLUSIVE

EQ-5D-5L – DIGITAL VERSION

EQ-5D-5L

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

MOBILITY
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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

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2

- 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 how your health is TODAY.
- Now, write the number you marked on the scale in the box below.

The best health you can imagine — 100

YOUR HEALTH TODAY =

95
90
85
80
75
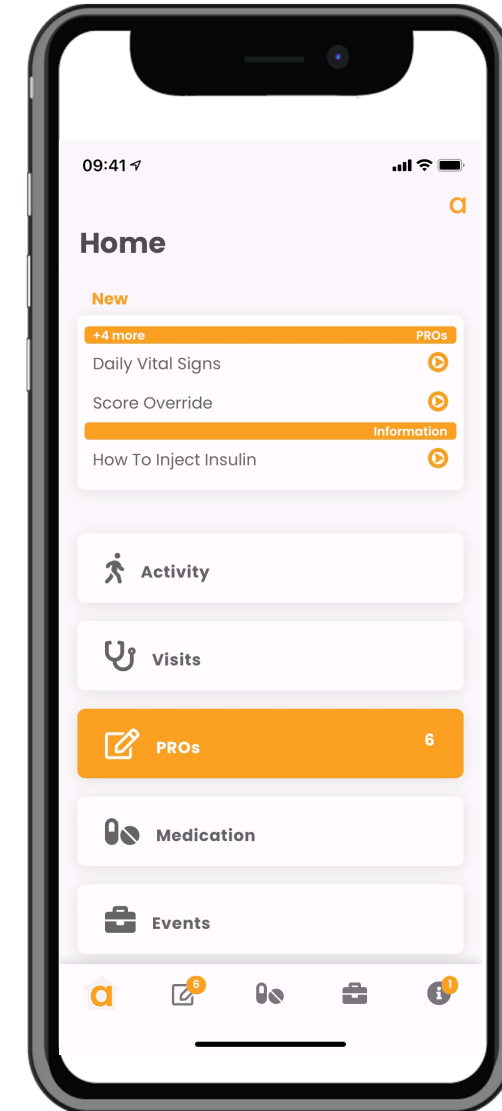70
65
60
55
50
45
40
35
30
25
20
15
10
5
0

The worst health you can imagine

3

Home

09:41

New

+4 more — PROs
Daily Vital Signs
Score Override

Information
How To Inject Insulin

Activity

Visits

PROs — 6

Medication

Events

SUMMARY

Be prepared for a lot of work to develop something that on paper shouldn't be too tough

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

You can really enforce change in developing something for your community

IT'S NOT SIMPLE

You have to be open minded in your approach

TEAMWORK IS VITAL

You're working with multiple stakeholders who all want input

YOU CAN MAKE A DIFFERENCE

You CAN make change if you are PATIENT!

QUESTIONS

Contact dan@cats-foundation.org