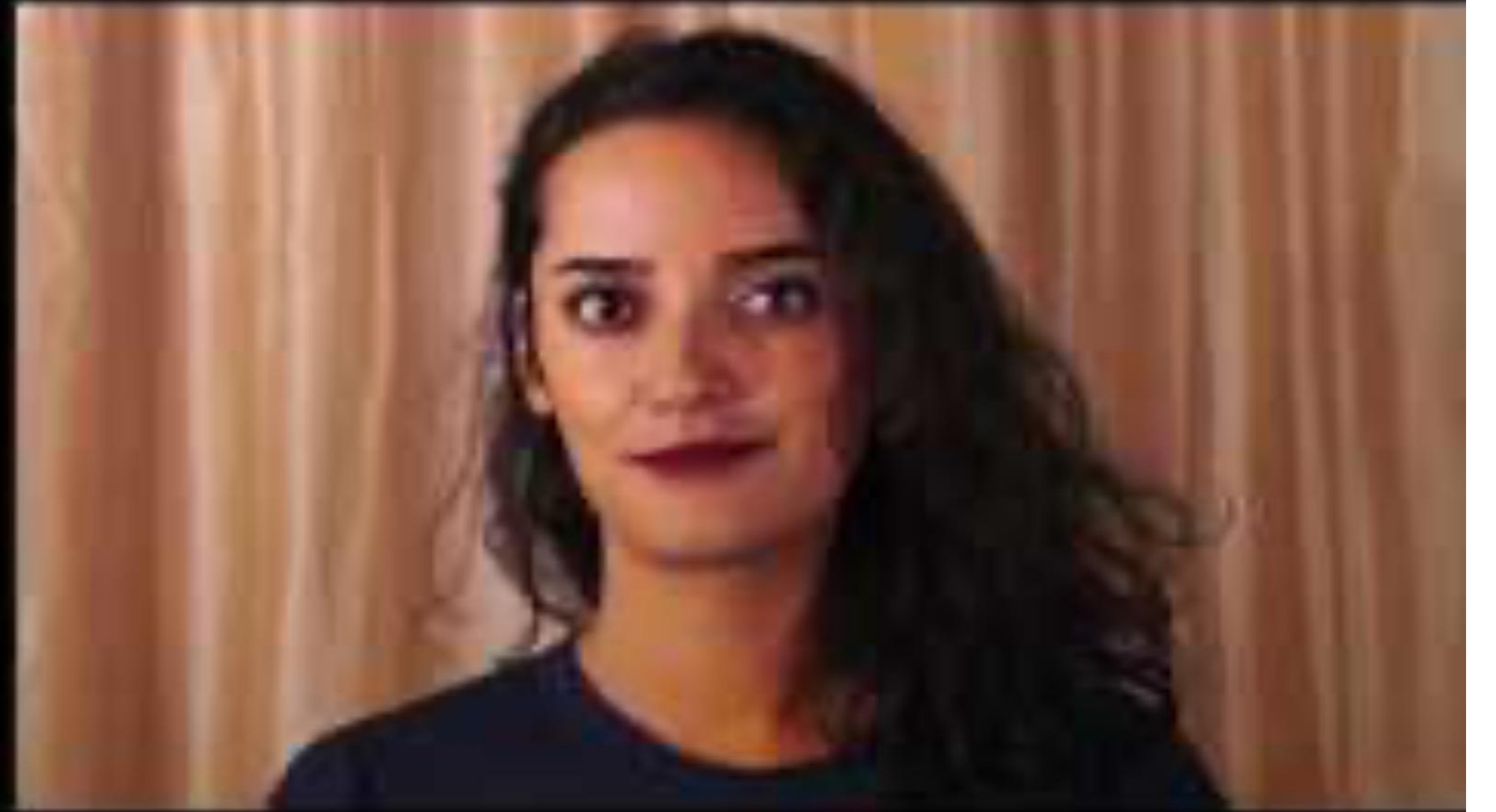


LIVING WITH A HCTD

COPING AND MANAGMENT (OF CHRONIC PAIN)

Sabine Hellemans | 12 11 2024





1. WHAT DOES IT MEAN TO LIVE WITH VEDS?



*"It's **invisible**, you can 't tell. but that does not mean that the person is not suffering and hurting and having problems on the inside..."*

OUTSIDE vs INSIDE

What you see from the outside can be different than what is living inside

1. WHAT DOES IT MEAN TO LIVE WITH VEDS?



*"I really want to play basketball, and **I want to join the team**. But I can't... it 's not safe..."*

NEED TO BELONG

Being part of a group, feeling of belonging, being capable of doing what peers/friends can do

1. WHAT DOES IT MEAN TO LIVE WITH VEDS?



"I know my mom, every time I call her ... it's a bit hesitant."

"Hey mom, everything is fine.."

"It is not just me living with anxiety and fear, but it is also everyone in my family...."

SHARED ANXIETY

A diagnosis affects not one individual, but the entire family

1. WHAT DOES IT MEAN TO LIVE WITH VEDS?



"My brother Mike passed away at 49, my brother Steve at 42 and my brother Tom at 40... It feels like losing friends..."

GRIEF

The task of letting go of loved ones and knowing that more loved ones are at risk

1. WHAT DOES IT MEAN TO LIVE WITH VEDS?



*"My husband will fall asleep, but I am **staying** awake, I am alive today..."*

EXISTENTIAL FEAR

A desire for living, daily confrontation with diagnosis, that you could die...

1. WHAT DOES IT MEAN TO LIVE WITH VEDS?



"I don't know what's gonna happen the next days, the next hours, the next minutes...."

UNPREDICTABILITY

Symptoms might change day to day -> profound impact on one's emotional and physical well-being

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2. A HERITABLE CONNECTIVE TISSUE DISORDER = A CHRONIC DISEASE

A 'chronic condition' is defined as a condition from which there is generally no prospect of full recovery.

A chronic condition is usually associated with pain, psychological distress, functional limitations or other symptoms.

The extent to which people experience discomfort varies from condition to condition and from person to person.

HCTD: DIFFERENT CONDITIONS

TYPE	OVERERVING	GEN	EIWIT
classical	AD	COL5A1	type V collageen
		COL5A2	type V collageen
hypermobile	AD	niet gekend	niet gekend
vascular	AD	COL3A1	type III collageen
kyphoscoliosis	AR	PLOD1	lysyl hydroxylase 1
arthrochalasia	AD	COL1A1	type I collageen
		COL1A2	type I collageen
dermatosparaxis	AR	ADAMTS2	type I procollageen N-proteinase

2. ACUTE VS CHRONIC DISEASE

ACUTE DISEASE	CHRONIC DISEASE
sudden onset	gradual onset
cure usual	cure rare
course short	course lengthy
patient passive	patient active
physician dominant	team care, patient included
return to normal likely	return to normal unlikely
future uncertainty rare	future uncertainty common

3. IMPACT OF LIVING WITH A HCTD

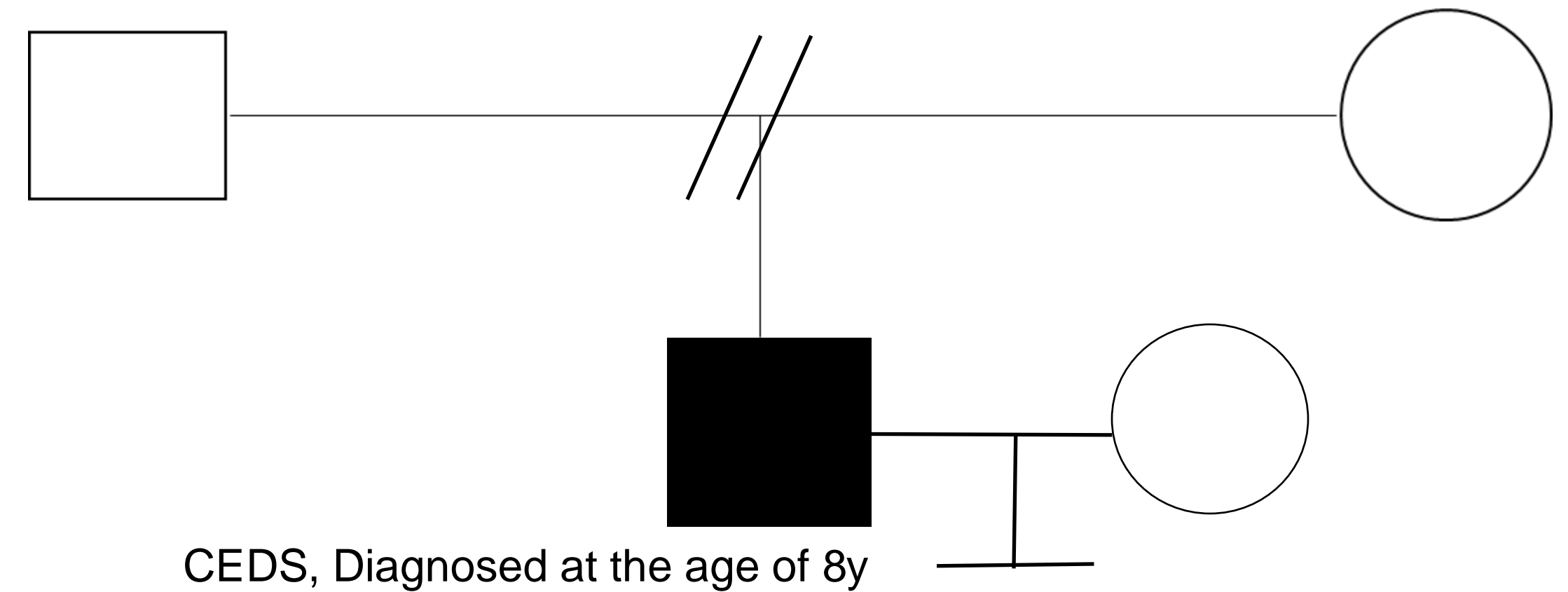
Living with a rare, genetic, chronic condition

- What does it mean for my own life?
- What does it mean for the lives of those around me (partner, children, family members)?



TESTIMONIAL KEVIN

FEELING OF BEING STUCK IN DE MUD....



- Hard working family : "What we can do ourselves, we do ourselves"
- Motocross: riding, repairing,... = a family experience
- Core identity Kevin: getting satisfaction from physical activities <-> cEDS
- "I am a man of 38 years old and my grandfather is in better shape than me"



*Prof. Fransiska Malfait
Clinical Geneticist, Belgium*

*Sabine Hellemans
Psychologist, Belgium*

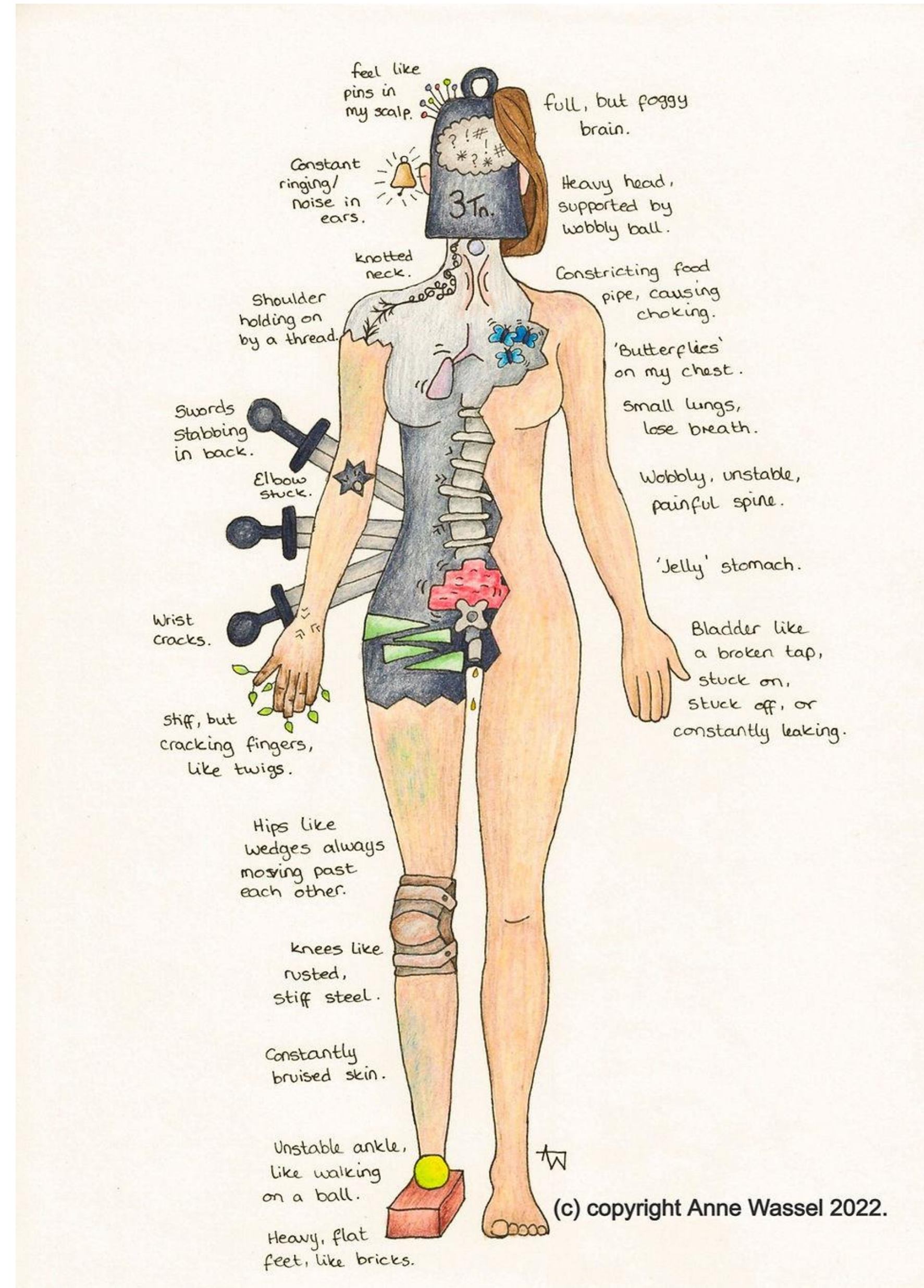
*Kevin
cEDS Patient*

workouts, I haven't done too intense exercises, and

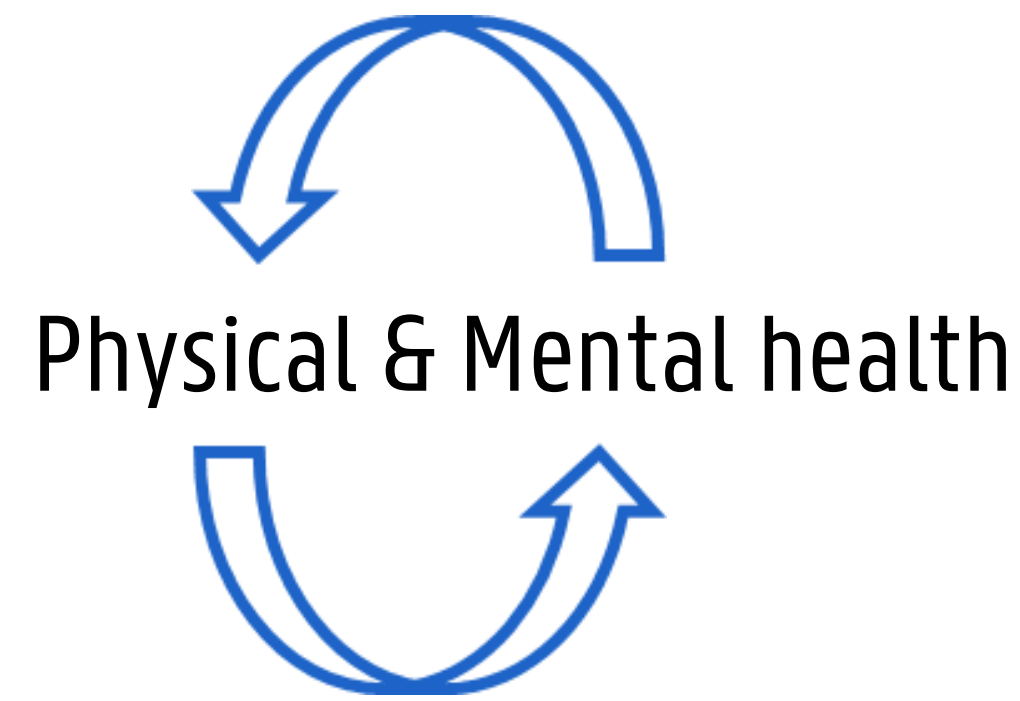
3. IMPACT OF LIVING WITH A HCTD: PHYSICAL HEALTH

Frequently reported physical complaints:

- Fragile skin, slow wound healing
- Joint hypermobility, luxations
- Organ prolapse
- Pain
- Fatigue
- Sleeping problems
- Eating problems
- ...



3. IMPACT OF LIVING WITH A HCTD: PHYSICAL AND MENTAL HEALTH



- Limited freedom due to necessary medical care
- Uncertainty about the course of the disease
- Lack of autonomy
- Difficulty with attention due to pain
- Stress, emotional distress
- Noticing uncertainty among caregivers regarding the diagnosis and treatment plan



3. IMPACT OF LIVING WITH A HCTD: PHYSICAL AND MENTAL HEALTH

Feelings of depression

- Loneliness
- Lack of social support
- Hopelessness
- Lack of (self-)confidence
- Frustration
- Guilt about possibly passing it on to children
- Anxiety from uncertainty



3. IMPACT OF LIVING WITH A HCTD: RELATIONSHIP FUNCTIONING

- Relationship satisfaction
- Sexual intimacy
- Role patterns in the family
- Children / childwish
-





Christel Mols
vEDS Patient



Prof. Fransiska Malfait
Geneticist

the doctors did not want to assist with a risk

Case report and discussion: Pre-implantation genetic diagnosis with surrogacy in vascular Ehlers–Danlos syndrome

Chloe Angwin^{1,2}, Neeti Ghali^{1,2} and Fleur Stephanie van Dijk^{1,2*}

¹London National Ehlers-Danlos Syndrome Service, North West Thames Regional Genetics Service, London North West Healthcare University NHS Trust, Harrow, United Kingdom, ²Department of Metabolism, Digestion and Reproduction, Section of Genetics and Genomics, Imperial College London, London, United Kingdom

Introduction: Vascular Ehlers–Danlos syndrome (vEDS) is an autosomal dominant inherited connective tissue condition, characterized by generalized tissue fragility with an increased risk of arterial dissection and hollow organ rupture. In women with vEDS, pregnancy and childbirth carry significant risks of both morbidity and mortality. The Human Fertilisation and Embryology Authority has approved vEDS for pre-implantation genetic diagnosis (PGD), given the potential for life-limiting complications. PGD avoids implantation of embryos that are affected by specific disorders by carrying out genetic testing (either for a familial variant or whole gene) and selecting unaffected embryos prior to implantation.

Case: We present an essential clinical update to the only published clinical case of a woman with vEDS undergoing PGD with surrogacy, initially through stimulated in vitro fertilization (IVF) and in vitro maturation (IVM) and subsequently through natural IVF.

Discussion: In our experience, a subset of women with vEDS do wish to have biological, unaffected children through PGD despite being aware of the risks of pregnancy and delivery. Given the clinical heterogeneity in vEDS, these women could be considered on a case-by-case basis for PGD. Controlled studies with comprehensive patient monitoring evaluating the safety of PGD are essential to equitable healthcare provision.

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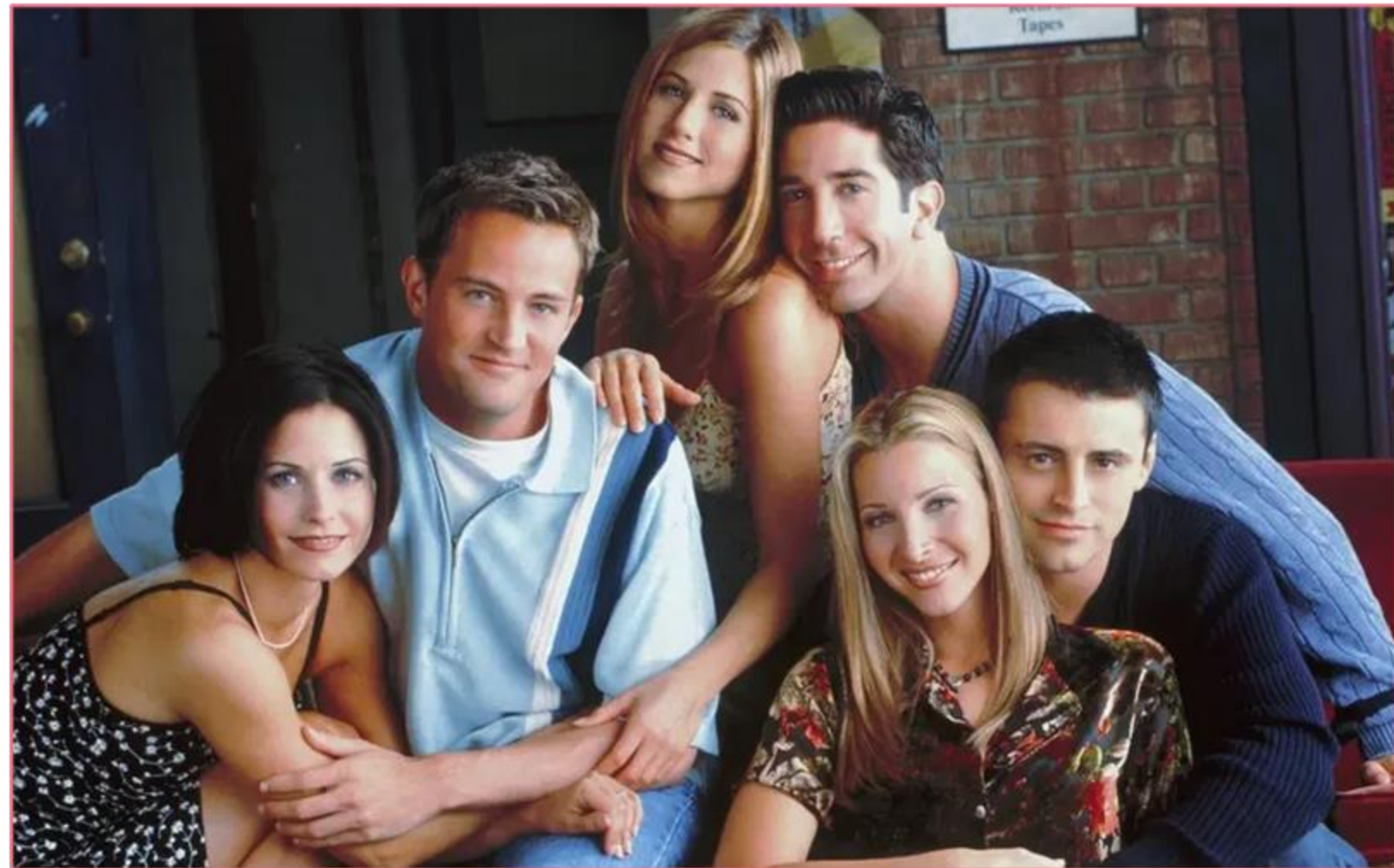
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3. IMPACT OF LIVING WITH A HCTD: SOCIAL FUNCTIONING

- Investing in social activities
- Daily activities require flexibility in planning
- Family and friends
- Work / professional challenges
- Reaction of others (disbelief)
- Financial situation



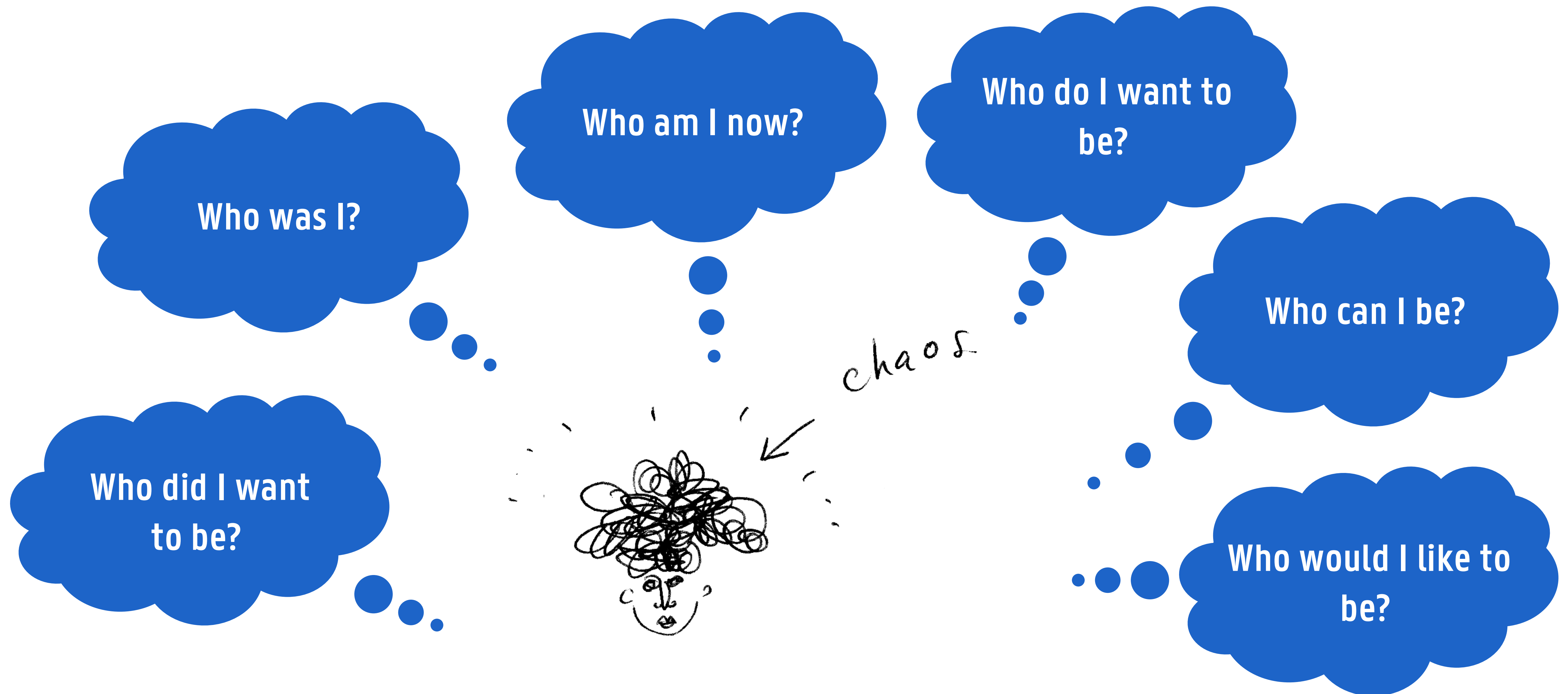
3. IMPACT = WIDESPREAD



3. IMPACT OF A CHRONIC DISEASE ON IDENTITY: LONELINESS...



3. IMPACT OF A CHRONIC DISEASE ON IDENTITY



4. CHALLENGES IN CHRONIC PAIN PATIENTS

1. Perception as 'Difficult Patients'

- Pain is often invisible and cannot be measured
- Patients may feel disbelieved or that their pain is imagined

2. Challenges of Discussing Psychological Factors

- Psychological influences on pain can be hard to address.
- There is a fear that acknowledging psychology implies pain is “not real.”

4. CHALLENGES IN CHRONIC PAIN PATIENTS

3. Persistent Mind-Body Dualism

- Pain is often seen as either physical or mental.
- Many patients are hesitant to seek psychological support

4. Patient Expectations vs. Healthcare Perception

- Patients want medical answers and relief
- Behaviors are sometimes viewed as demanding or complaining by caregivers
- This mismatch can increase patients' feelings of misunderstanding

4. FREQUENTLY USED COPING STRATEGIES

- "Living day by day"
- Experiencing ups and downs but continuing to try after a crisis
- Trying to lead as normal a life as possible
- Not looking toward the future
- Keeping the diagnosis secret out of fear of misunderstanding by others
- Informing and educating others about the condition
- Learning self-management skills
- Seeking adapted education or work



Patients who experience positive effects from medical treatment manage to see more perspective in other areas of life

4. EXPERIENCES WITH THE HEALTHCARE SYSTEM

Lack of knowledge among healthcare providers



Many patients have experiences with healthcare providers who have insufficient knowledge about the diagnosis,
Primarily at the local level — not in specialized settings.

Impact on patients:

- Delayed diagnosis, incorrect treatment, lack of access to additional support
- Patients feel responsible for providing information to their healthcare providers about the diagnosis
- Distrust in the healthcare system
- Feelings of fear and uncertainty

4. EXPERIENCES WITH THE HEALTHCARE SYSTEM

Contact with Healthcare Providers



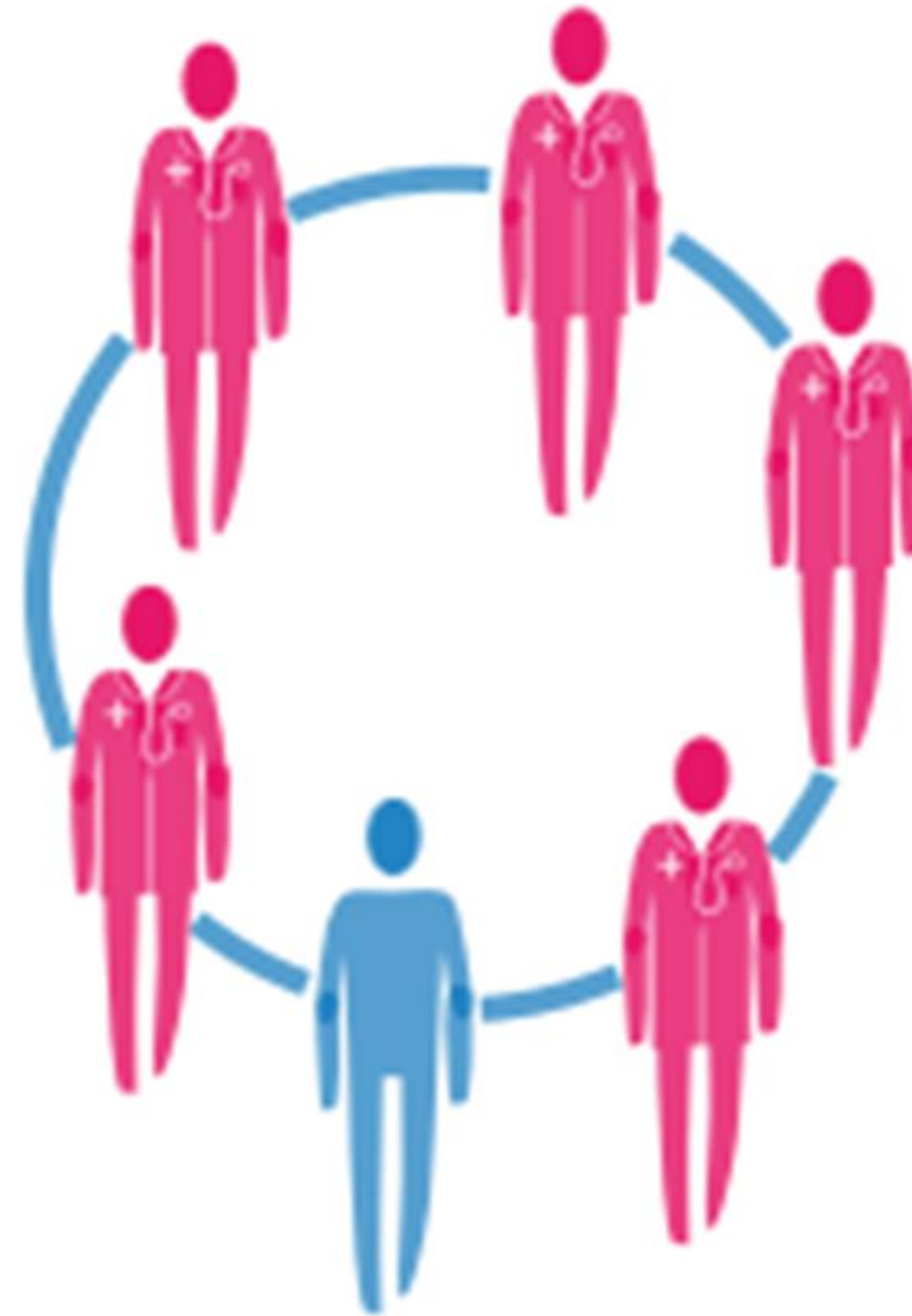
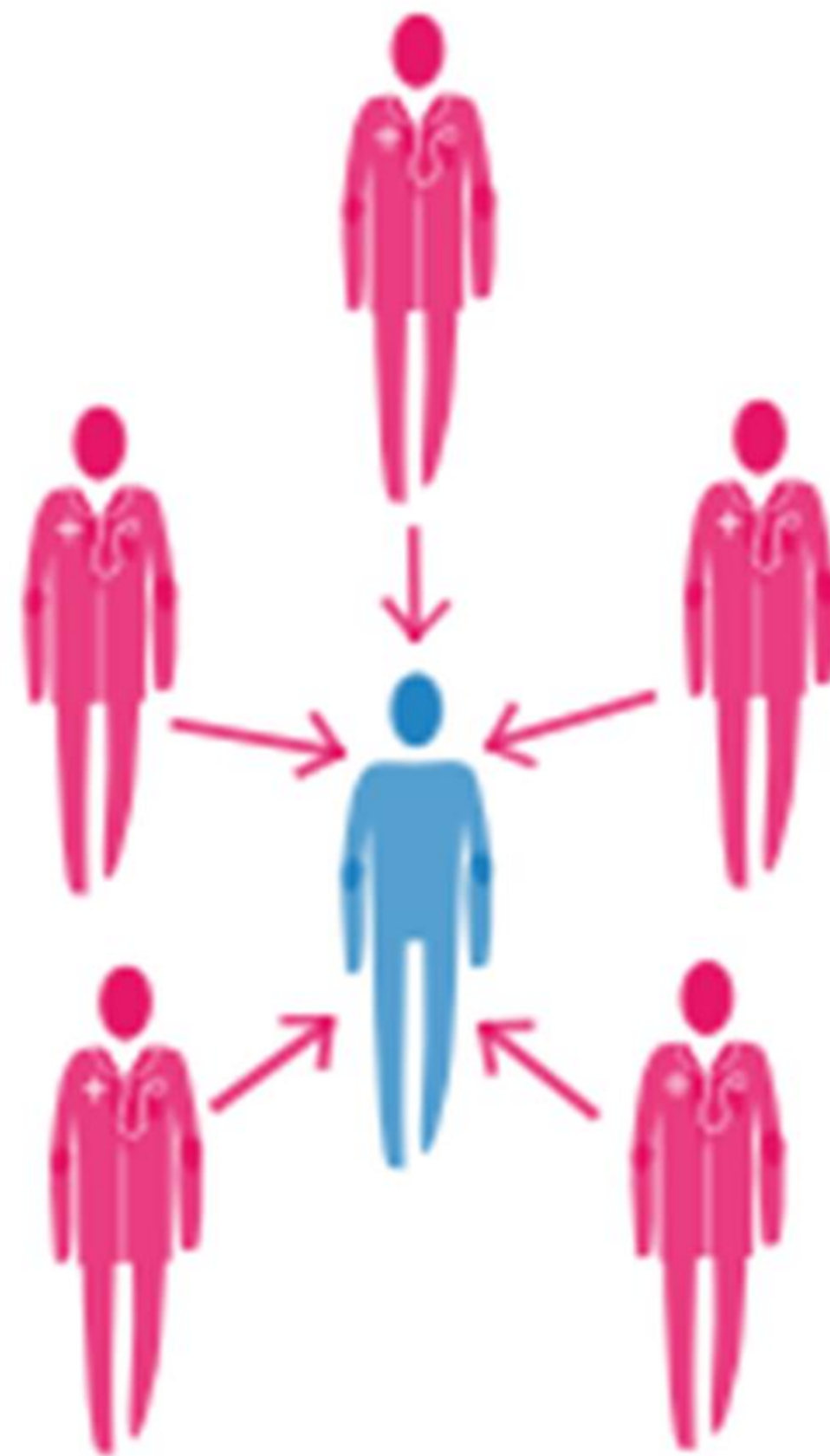
Patients have a strong need for a holistic approach!

- A coordinated approach
- Change of healthcare providers is experienced as challenging: “I have to start all over again.”
- Clear and understandable information is essential
- Viewing the person as a whole: “I am more than my diagnosis.”

4. EXPERIENCES WITH THE HEALTHCARE SYSTEM



FROM DOING THINGS
"TO" THE PATIENT...



... TO DOING THINGS
WITH THE PATIENT!

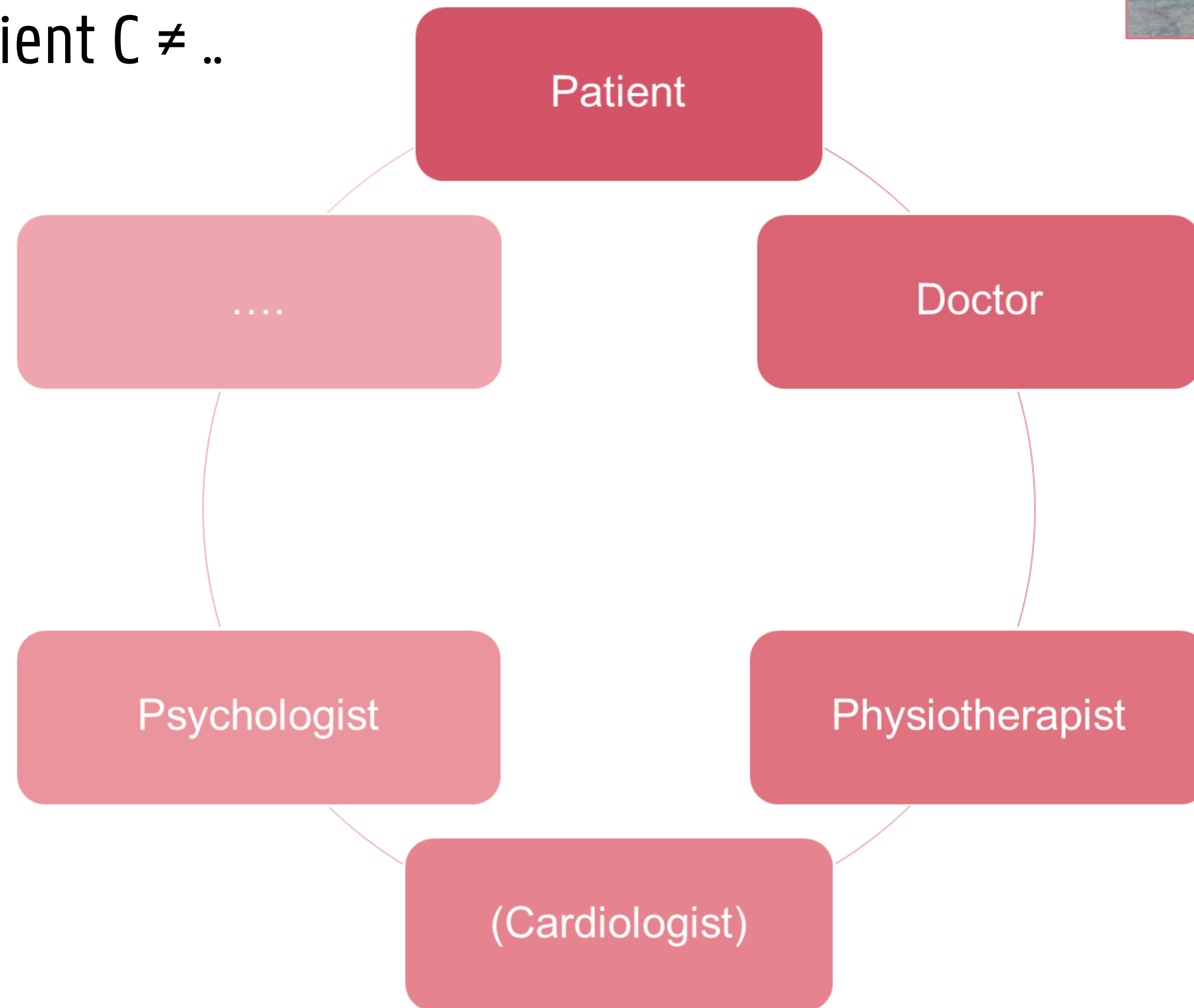
Which role can I take on in the healthcare system?

4. EXPERIENCES WITH THE HEALTHCARE SYSTEM

What do I expect from my caregivers?

What is my question? What do I need in order to move forward?

Patient A \neq Patient B \neq Patient C \neq ..



5. PSYCHOTHERAPY



Conversations with people experiencing chronic pain can be multifaceted and cover much more than just pain.



How is the **social context around pain** important, and how can **social connectedness**, sharing the pain together instead of alone, empower someone?



5. DISEASE VS. BEING ILL

DISEASE	BEING ILL
Physical, biological	Feelings and thoughts related to the disease
Abnormal condition of the body	A social reality
	Being ill is a personal experience

"Who am I (still)?" "Can I still be the person I have always been?" "Will people accept my illness?" "How do I explain that today I am unable to do anything, yet yesterday I was washing the windows?" "Do they think I am taking advantage?" "I am exhausted, I feel so useless." "Is it all in my head?"

FIRST THINGS FIRST: MEDICAL COMPLAINTS -> PSYCHOLOGICAL IMPACT

- 1. RECOGNISE THAT THE PROBLEM IS REAL AND IMPORTANT _**
- 2. FOCUS FIRST ON PHYSICAL COMPLAINTS AND SYMPTOMS**
- 3. IN A SECOND STEP EMOTIONS, THOUGHTS, BELIEFS, RELATIONAL CHALLENGES, ...**

It is essential to find the right balance between addressing the physical and psychological aspects of a patient's well-being.

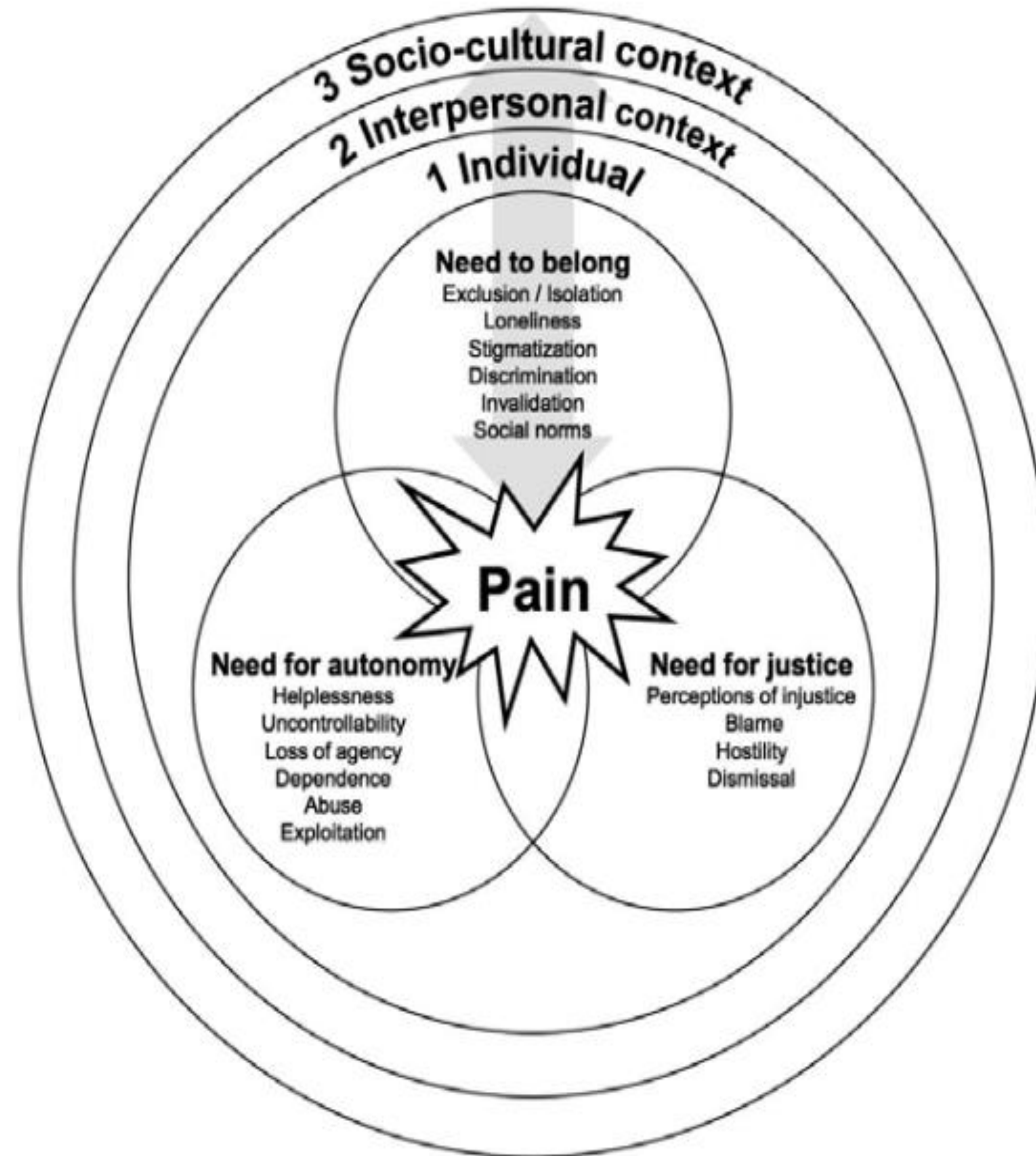
Switching too quickly to psychological topics can provoke resistance, especially if the patient feels unheard or misunderstood.

It's important to create a safe and supportive environment, respect the patient's pace, and introduce psychological aspects into the conversation at the right time. This can lead to more effective collaboration and a better therapeutic relationship.

5. PAIN IS A THREAT TO THE SOCIAL SELF

Pain hinders 3 interpersonal needs:

1. Need for autonomy
2. Need for justice, fairness
3. Need to belong



(1) NEED FOR AUTONOMY

Definition of Autonomy

- A sense of control over one's own actions and environment (sense of agency).
- Despite being social beings, individuals want to predict and influence their own lives.

Impact of Chronic Pain on Autonomy

- Pain can undermine autonomy, creating a disconnect with one's own body.
- Doubts arise about personal abilities and independence.

NEED FOR AUTONOMY

Shift in Control and Reliance on Others

- Increased reliance on family, friends, and healthcare providers.
- Important to help patients explore ways to regain control and predictability in their lives.

Goal in Therapy:

- Support patients in finding a new balance of autonomy and support.
- Facilitate a healthy relationship with pain management and social support

(2) NEED FOR JUSTICE, FAIRNESS

Connection to Meaning-Making

- Patients with chronic pain often question: “How can I find meaning despite my pain?”
- Struggle to reconcile life changes and different needs with a sense of purpose.

Feelings of Unfairness and Loss

- Chronic pain can bring about emotions of unfairness, grief, and loss.
- Especially difficult when pain is caused by an external event (e.g., accident or negligence) or genetic default.

NEED FOR JUSTICE, FAIRNESS

Beliefs about Fairness

- Pain may challenge fundamental beliefs about justice
 - > “Why me?”
 - > “What did I do to deserve this?”
- Important to support patients in exploring these existential questions.

Therapeutic Goal:

Facilitate patients' search for meaning and assist in the process of accepting and finding purpose despite pain.

(3) NEED TO BELONG

Striving for Meaningful Relationships

- Pain impacts the ability to enjoy social activities (work, leisure).
- Chronic pain leads to feelings of exclusion, affecting social belonging.

•Stigma and Negative Judgments

- People with chronic pain may face negative social judgments (e.g., perceived as less warm or competent).
- Especially true when pain is unexplained or lacks a clear diagnosis.

Social Exclusion and Pain Perception

- Social disconnection is linked to increased stress, weakened immunity.
- Social exclusion increases the experience of physical pain, and vice versa.

(3) NEED TO BELONG

Importance of Social Support Networks

- A strong social network helps improve pain tolerance and coping.
- Healthcare providers should assess social connections, roles, and societal pressures.

Therapeutic Focus:

- Conversations about chronic pain should explore not just physical pain but the social context.
- Facilitating social connectedness and shared experiences of pain can help patients cope.

SOLUTION-FOCUSED QUESTIONING

START OF A CONVERSATION

- "What would be useful for you to discuss?"
- "How would you know that this conversation has been helpful for you?"
- "What brings you here today?"
- "What have you brought with you today?" (or: "What's on your mind today?")
- "What is your purpose for coming here today?"
- "What would make this conversation worthwhile for you?"
- "Before we start, may I ask: what's already going a little better?"
- "What are you satisfied with? What's going well in your life?"
- "What would you like to keep from your current situation?"

SOLUTION-FOCUSED QUESTIONING

CLARIFYING THE NEED FOR CHANGE

- "What would you like to be different?"
- "What would you like to change?"
- "What should change as a result of our conversations?"
- "How can we best discuss this?"
- "How important is it for you to make this change?"
- "What are you willing to invest in this change?"
- "What would you like to see improve?"
- "What do you want to achieve?"

SOLUTION-FOCUSED QUESTIONING

COMPLIMENTS

- "It's admirable how you've been able to do your work under such difficult circumstances. How do you do it?"
- "What helps you keep going?"
- "How did you manage to keep going before things became too much?"
- "How did you manage to accomplish...?"
- "What positive qualities do you have?"
- "What makes you so good at...?"
- "When do you feel proud and satisfied?"
- "What strengths did you use when...?"

SOLUTION-FOCUSED QUESTIONING

SEARCHING FOR EXCEPTIONS

- "What is already going well?"
- "What has helped so far?"
- "What could you do more of that is already working?"
- "When does it go better?"
- "When is the problem less present?"
- "What do others say works?"

SOLUTION-FOCUSED QUESTIONING

PROGRESS

- "What is already getting better?"
- "What else?"
- "Where do you stand now on the scale?"
- "What did you notice that could help you?"
- "How did you manage to achieve that improvement?"
- "Of the things you've already tried, what worked, even if just a little?"
- "What did the other person think went better?"
- "What else could you do?"

UPS & DOWNS... It goes in waves...



→
TIME

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