

**S** UNIVERSITÉ DE SHERBROOKE

2014 MDF Annual Conference  
Sept. 12 - 13 Washington, DC



MYOTONIC  
DYSTROPHY  
FOUNDATION



# DM and Emotional Health: Depression and Anxiety

Benjamin Gallais, Ph.D.\*

Postdoctoral Fellow

GRIMN

Université de Sherbrooke  
Sherbrooke, Quebec, Canada

Sarah K. Tighe, M.D.\*

Assistant Professor

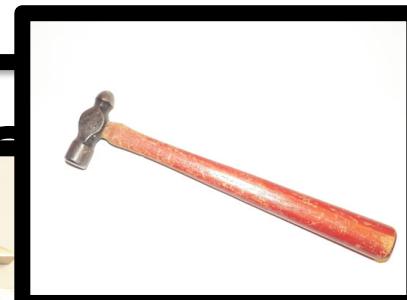
Department of Psychiatry  
University of Iowa  
Iowa City, Iowa, USA

\*No conflicts of interest



## MYOTONIC DYSTROPHY FOUNDATION

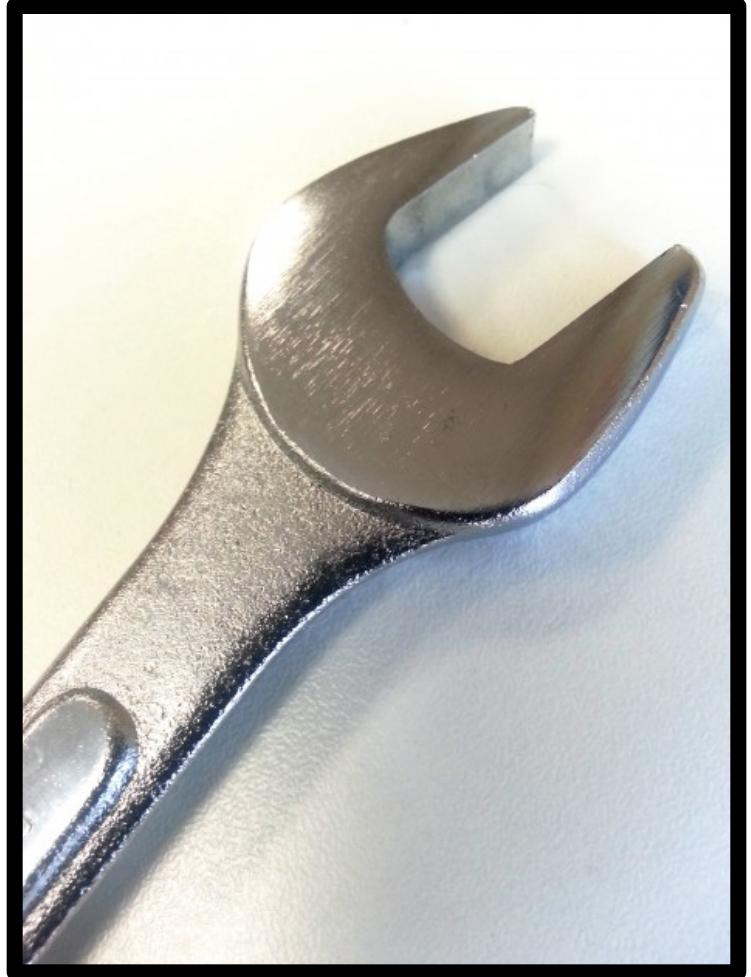
Toolkit



city

# Outline

- Key definitions
- Features of depression and anxiety in DM
  - Frequency
  - Signs and symptoms
  - Impact on daily activities
- Treatment options
  - Role of the caregiver
- Gaps in knowledge



# Important preface

**We believe that all individuals  
with DM deserve happiness  
and a good quality of life.**

**This applies to caregivers of DM patients, too!**

# Key Definitions



- Brief periods of sadness or uneasiness in certain situations are normal
  - Sadness after loss of a loved one
  - Nervous before a public speech
- Depression and anxiety are medical conditions requiring treatment
  - Distressing
  - Persistent
  - Disrupt day-to-day functioning

# Warning signs



	Depression	Anxiety
Mood		
Activity		
Physical signs		
Life outlook		
Function		



# Warning signs

	Depression	Anxiety
Mood	Down “I don’t care” Emotionless	
Activity	Slower Loss of interest in usual activities	
Physical signs	Insomnia Loss of appetite More fatigued/daytime sleepiness	
Life outlook	Useless or burdensome Hopeless Thoughts of death or suicide	
Function	Recent worsening	



# Warning signs

	Depression	Anxiety
Mood	Down “I don’t care” Emotionless	Worried “Keyed up” Uneasy
Activity	Slower Loss of interest in usual activities	Restless Avoid usual activities
Physical signs	Insomnia Loss of appetite More fatigued/daytime sleepiness	Insomnia Loss of appetite More fatigued
Life outlook	Useless or burdensome Hopeless Thoughts of death or suicide	Hopeless Thoughts of death or suicide
Function	Recent worsening	Recent worsening



# Warning signs

	Depression	Anxiety
Mood	Down “I don’t care” Emotionless	Worried “Keyed up” Uneasy
Activity	Slower Loss of interest in usual activities	Restless Avoid usual activities
Physical signs	Insomnia Loss of appetite More fatigued/daytime sleepiness	Insomnia Loss of appetite More fatigued
Life outlook	Useless or burdensome Hopeless Thoughts of death or suicide	Hopeless Thoughts of death or suicide
Function	Recent worsening	Recent worsening

# What is the frequency of depression and anxiety in DM?



The Myotonic Dystrophy  
**Family Registry**



# The Myotonic Dystrophy **Family Registry**

- Developed and maintained by the MDF
- Anonymous data available to DM researchers
  - Identification of potential research participants
  - Patient-reported information about life with DM
    - Demographic information (age, gender, etc.)
    - Type of DM
    - Nature and severity of symptoms
    - Quality of life
- Sign-up at [mytonicregistry.org](http://mytonicregistry.org)

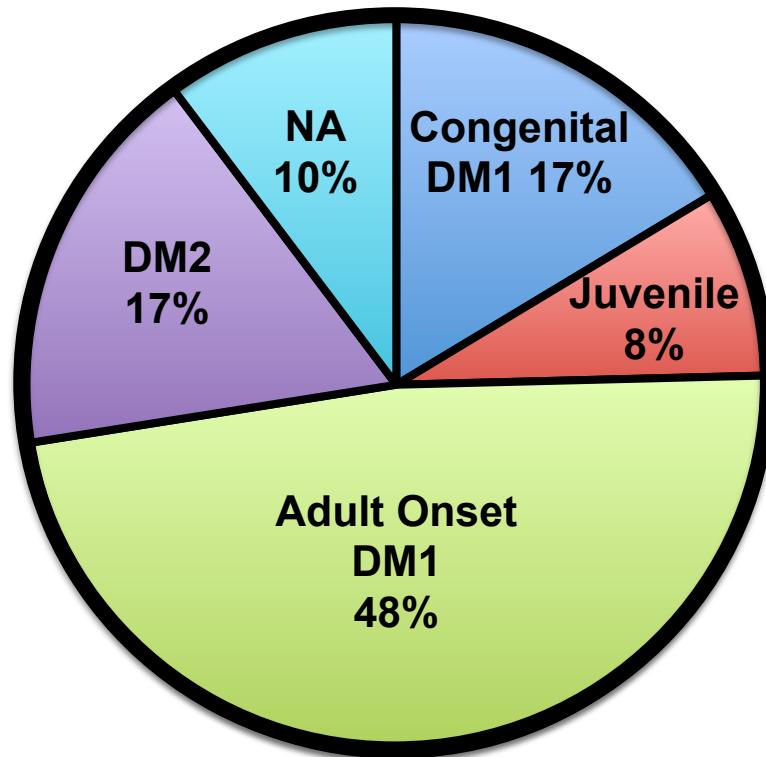


# The Myotonic Dystrophy **Family Registry**

## Frequency of DM diagnoses in the Registry\*

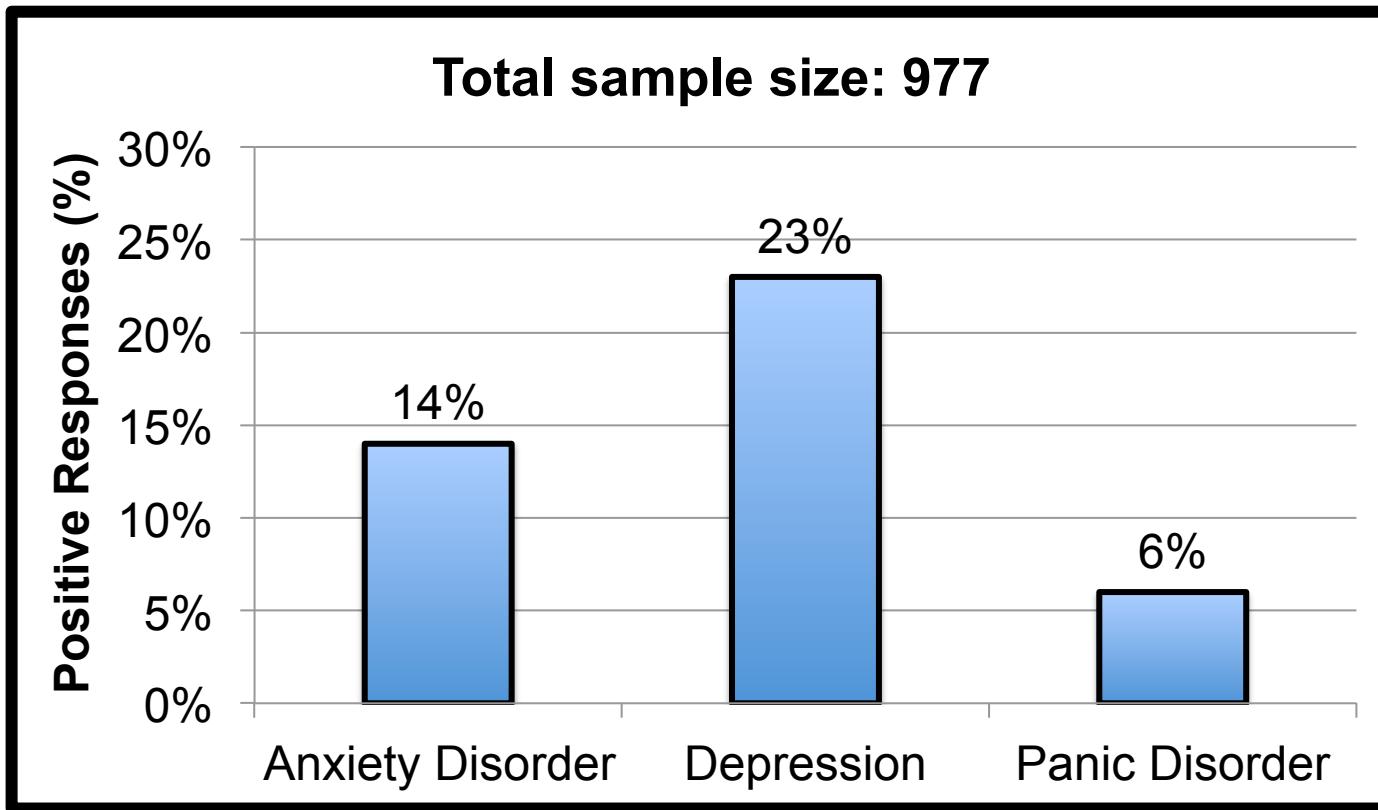
NA: Not available

Sample size: 1069



\*Data courtesy of the Myotonic Dystrophy Family Registry,  
[myotonicregistry.org](http://myotonicregistry.org) (accessed 9/7/14)

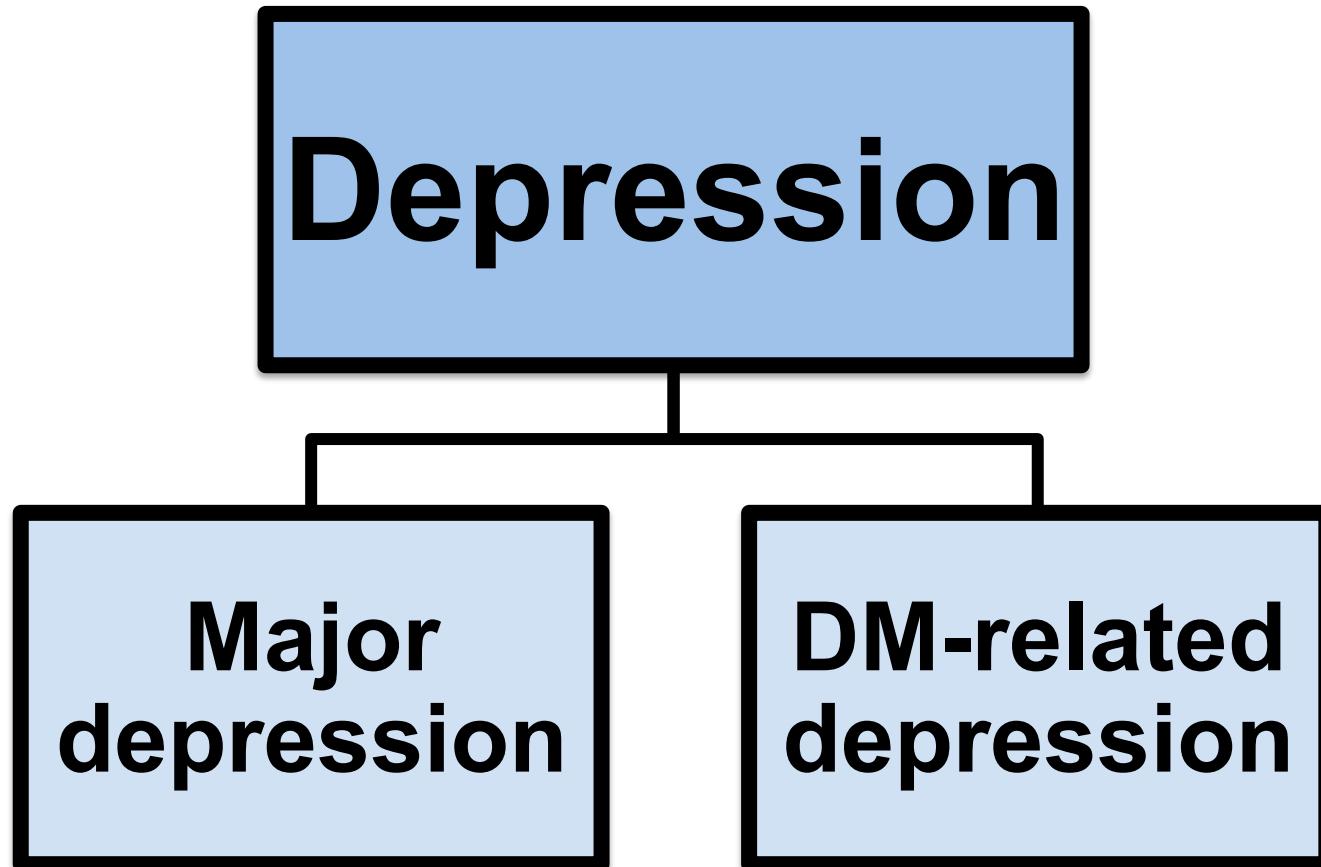
# What is the frequency of depression and anxiety in DM?\*,\*\*



\* Has a doctor diagnosed you with an anxiety disorder, panic attacks, or depression?

\*\* Data courtesy of the Myotonic Dystrophy Family Registry, [myotonicregistry.org](http://myotonicregistry.org) (accessed 9/7/14)

# What does depression look like in DM?



# Major depression

- Episodic nature
  - Occurs mostly in relation to specific situations related to the condition
    - Diagnosis of DM
      - In patients themselves or a relative
    - Evolution of the disease
      - Diagnosis of a new problem (e.g. cardiac, respiratory, walking, etc.)
- Major day-to-day issues
  - Problems at school or work
  - Difficulties in finding a partner
  - Physical changes/other people's looks

# **DM-related depression**

- An invisible handicap that mimics major depression
  - Chronic state
  - Thought to be brain-related (directly caused by DM)
- Features
  - Lack of emotion (negative and positive)
  - Apathy
  - Fatigue
  - Daytime sleepiness
  - Thinking problems: concentration, memory, etc.

# What is apathy?

- A lack of
  - Initiative
  - Interests in activities, emotions, care (daily care and care related to DM)
- Disrupts healthcare
  - Promotes treatment non-compliance
- Source of distress among couples
  - Burdensome for partners
- Affects about 40% of adults with DM1\*

\*Gallais unpublished data

# Quotes about apathy

Man with DM1:

*“If my wife suggests shall we go on holiday? Then I say okay, you say where, because I don’t care. She does not like that. She says you can think of what you like. Then I say I enjoy everything, I am very easy.”*

Man with DM1:

*“Because you are at home most of the time, your world becomes smaller, which is very annoying for me...”*

Woman partner:

*“What can be postponed was postponed.  
It completely irritated me!”*

# Fatigue in DM

- Occurs in up to 91% of those with DM1<sup>1-3</sup>
  - 2<sup>nd</sup> most important complaint
  - Had the greatest effect on DM1 patients lives!!



1. Kalkman et al., 2005.
2. Heatwole, et al., 2012.
3. Smith et al., et al., 2014.

# Excessive daytime sleepiness (EDS) in DM

- In up to 88% of individuals with DM<sup>1-4</sup>
- Fatigue versus EDS
  - Fatigue: complex and overwhelming feeling that one is lacking energy
    - Physical, intellectual and/or emotional energy
  - EDS: Urge to fall asleep in situation that is inappropriate
    - For example, falling asleep in class or social events

1. Romigi et al., 2011
2. Laberge et al., 2004 & 2009
3. Hilton-Jones, 1997
4. Heatwole et al., 2012

# Impact of excessive daytime sleepiness and fatigue in DM

- Emotional distress
  - Lower quality of life
  - Worse social and occupational engagement
  - Individual with DM1 about fatigue:

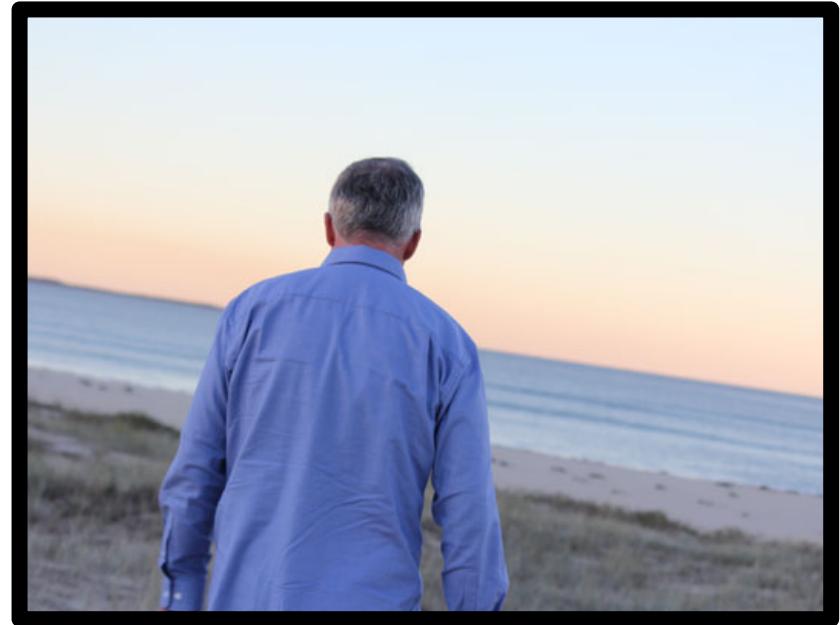
*“It is like a roller-coaster, some days I am as in older days, the next I am not able to do anything, and then I can feel depressed”*
  - Important source of unpredictability in daily life
    - Can “freeze” plans or spontaneity
1. Laberge et al., 2009 & 2011
  2. Gagnon et al., 2008
  3. Laberge & Dauvilliers, 2011
  4. Gallais, 2010
  5. Geirdal et al., 2014

# Impact of excessive daytime sleepiness and fatigue in DM

- Individual with DM1:

*“When I walk, I’m dead tired. I walk as little as possible”*

- The fatigue is real and onerous
  - Can lead to diminished activity or even avoidance of activities



# What does anxiety look like in DM?

- Irritability
- Poor self-esteem
- Fearfulness → avoidant behavior
  - Example: Fear of falling → avoid stairs → diminished walking → inactivity
- Excessive physical preoccupations (hypochondria)
- Social inhibition
- Fear of the future

# Factors unique to DM contribute to these emotional disturbances

Themes of lack of satisfaction in individuals with DM1<sup>1</sup>

- 1) Psychological distress and reduced quality of life (QoL) related to the gap between **expectations about life** and the reality
  - To not have children, or if so, not being able to follow up in activities;
  - Reduced travels;
  - To quit work;
  - Social isolation (+ feeling of guilt because it bothers more the partner than the affected)

# Factors unique to DM contribute to these emotional disturbances

Lack of satisfaction in individuals with DM<sup>1</sup>

Individuals with DM1:

*“My wishes and expectations crumbled”*

*“I do not fear the future, but I can feel depressed because of the situation”*

*“The reduced social relations are worse for my spouse than me.”*

1. Geirdal et al., 2014

# Factors unique to DM contribute to these emotional disturbances

Themes of lack of satisfaction in individuals with DM<sup>1</sup>

- 2) **Misunderstandings**

- Patients seen as uncommitted, or even lazy
  - Not believed on invisible symptoms (fatigue, memory...)

*“I am seen as uninterested and lazy; I have to fight all the time and I try to do small things, but I am not believed when I say that I am not able to perform these tasks anymore. It is hard”*

→ Feelings of sadness and hopelessness

- But diagnoses and **repeated information** to partners and caregivers about the disease can help:

*“The diagnosis gave an answer, it was not only my lack of will or laziness”*

→ apathy ≠ lack of will

# What are the consequences of emotional distress?

- Worse view of the future, worse general wellbeing, more anxiety and depression associated with **need for help** (= physical dependency)<sup>1</sup>
  - Demoralization, hopelessness
  - Give up, postpone, avoid... activities, social life
- Sometimes avoidance may be an adapted strategy, and demoralization is supportable... sometimes it is too distressing and a provider is needed
- Anyway, when quality of life is impaired, help can be provided → sorrow, excessive inactivity, or social withdraw are not written in stone!!

# What are the consequences of emotional distress?

Individuals with DM1<sup>1</sup>:

*“When I walk, I am dead tired. Thus, I walk as little as possible”*

→ Avoidance strictly related to physical impairment

*“I get very annoyed when I’m in a shop and cannot open a bag to put tomatoes in it or something. Then I have to ask to keep the bag open. It may be false modesty or shame, but I find that terrible. That’s why I have quitted doing the shopping”*

→ Avoidance related to both physical and emotional issues

# What's the impact on caregivers?

- Worse general wellbeing and more anxiety associated with a **lack of initiative of the patient** and less marital satisfaction<sup>1</sup>
- Increasing burden<sup>2,3</sup>  
*“You feel that you are on your own and have to do it all. The idea that it will never change, can be an enormous burden.”*
- Change in family responsibilities; partner is in charge
- Social isolation:  
*“We have become isolated and lonely”*
- Marital dissatisfaction  
*“Because everything has to be planned very carefully, we can never do anything spontaneous”*
- Hopelessness and anxiety about the future:  
*“How will I face it in 5, 10, 15 years...?”*

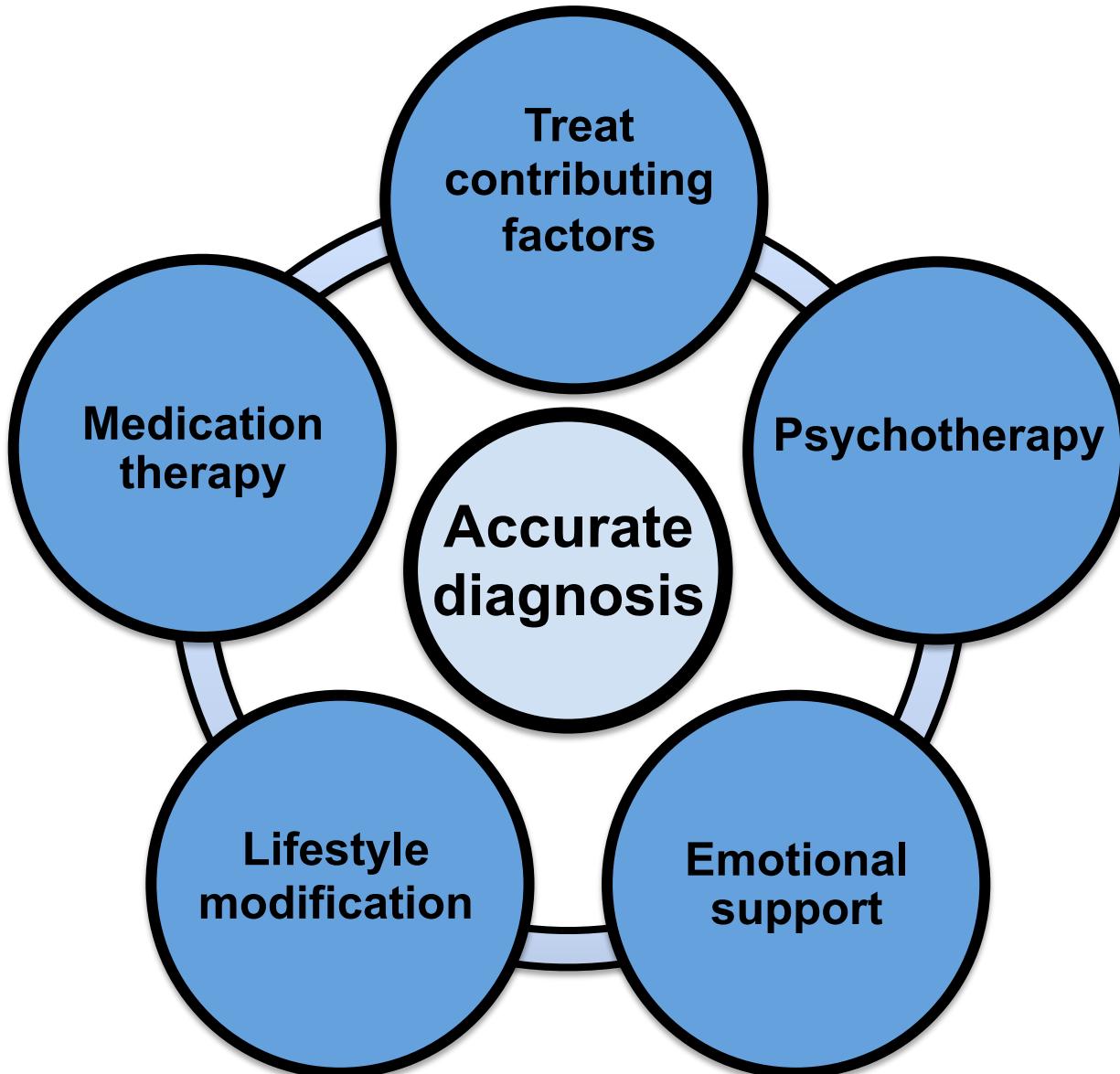
1. Timman et al., 2010; 2. Cup et al., 2011; 3. Geirdal et al., 2014

# Principles of treatment

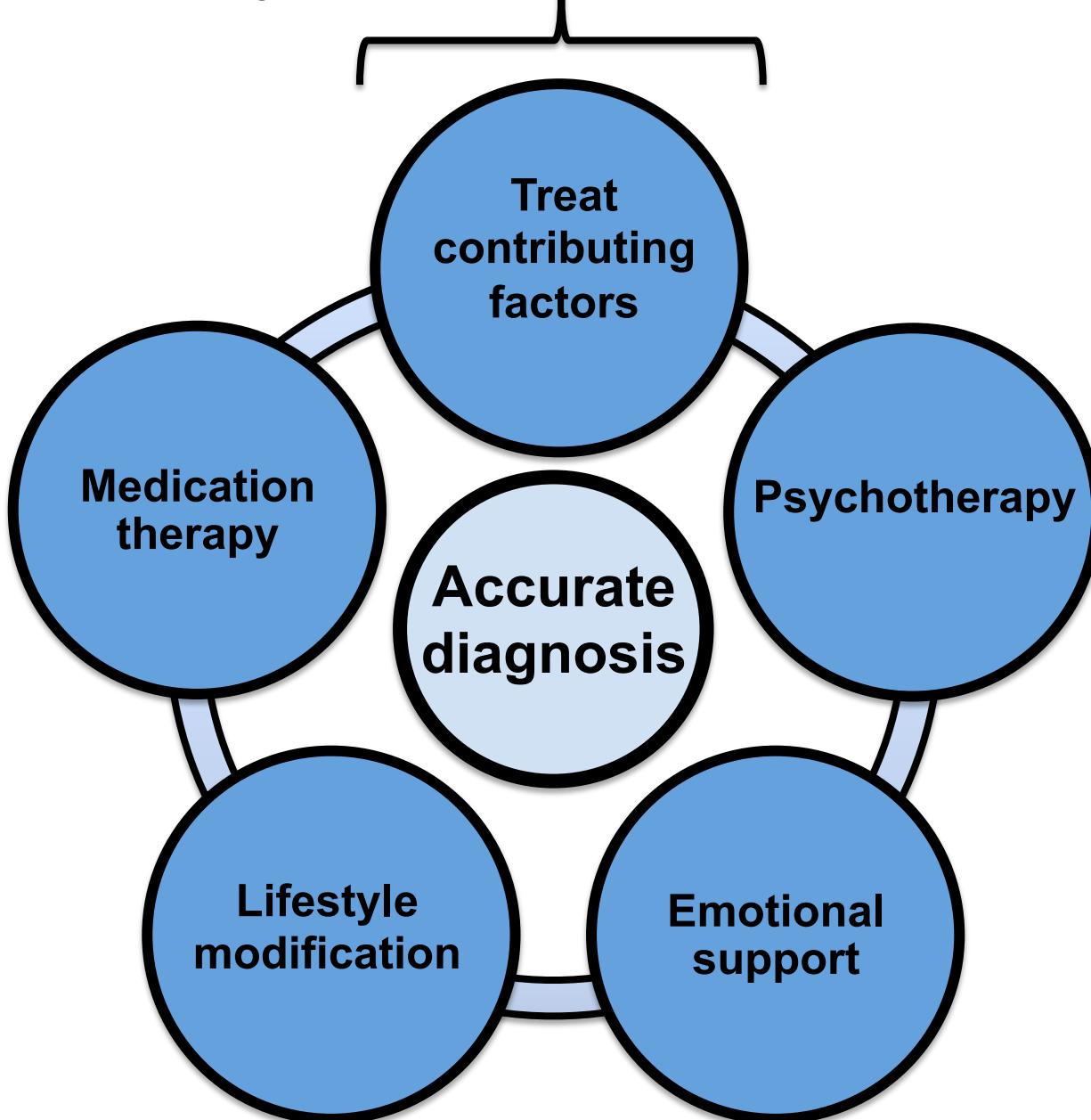
1. Depression and anxiety are treatable conditions
2. Safety comes first
  - Get help immediately for thoughts of suicide or plan for self harm
3. Accurate diagnosis is critical
4. Team approach
  - Close coordination among providers
  - Involve caregiver
5. Must be individualized



# Approach to treatment

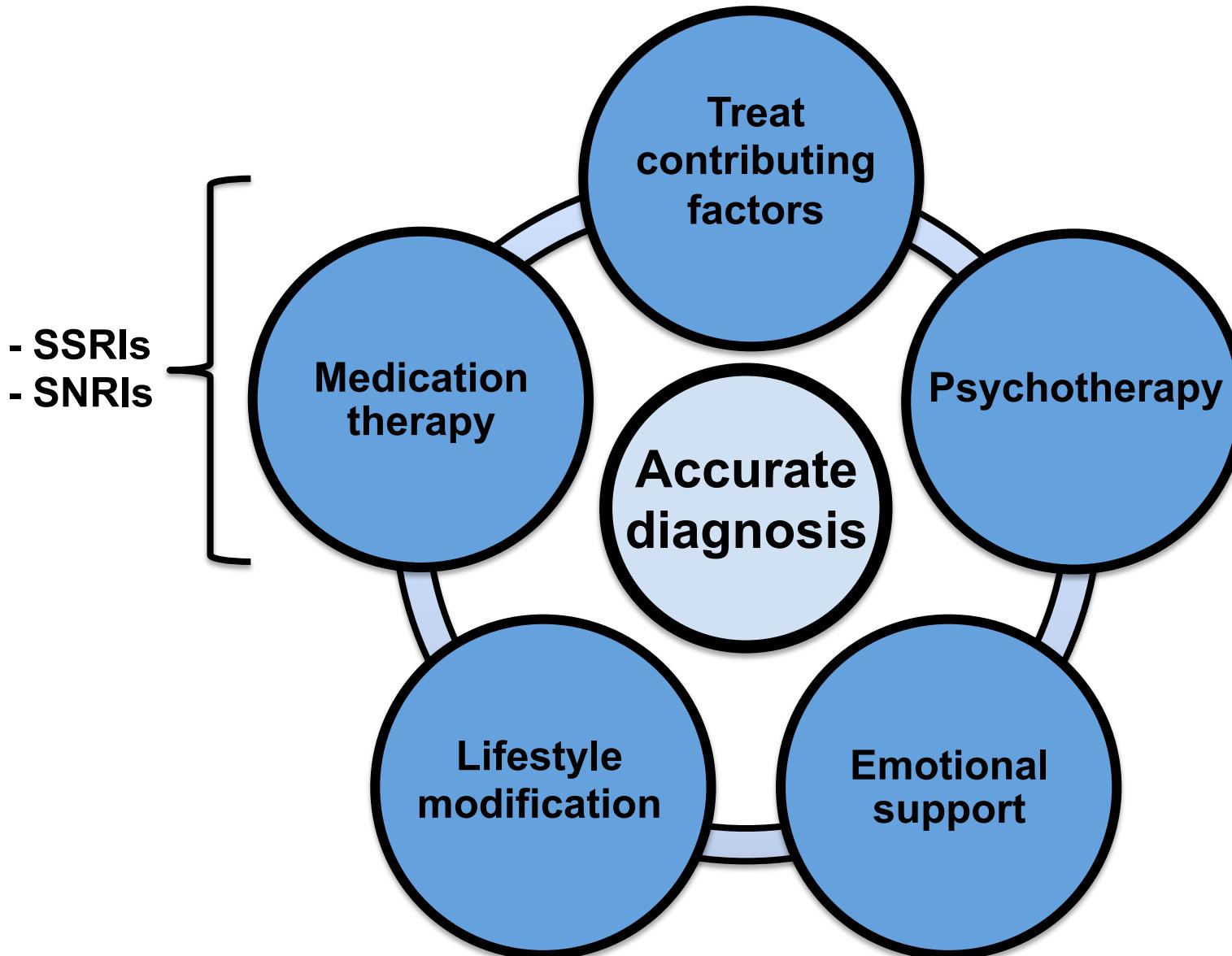


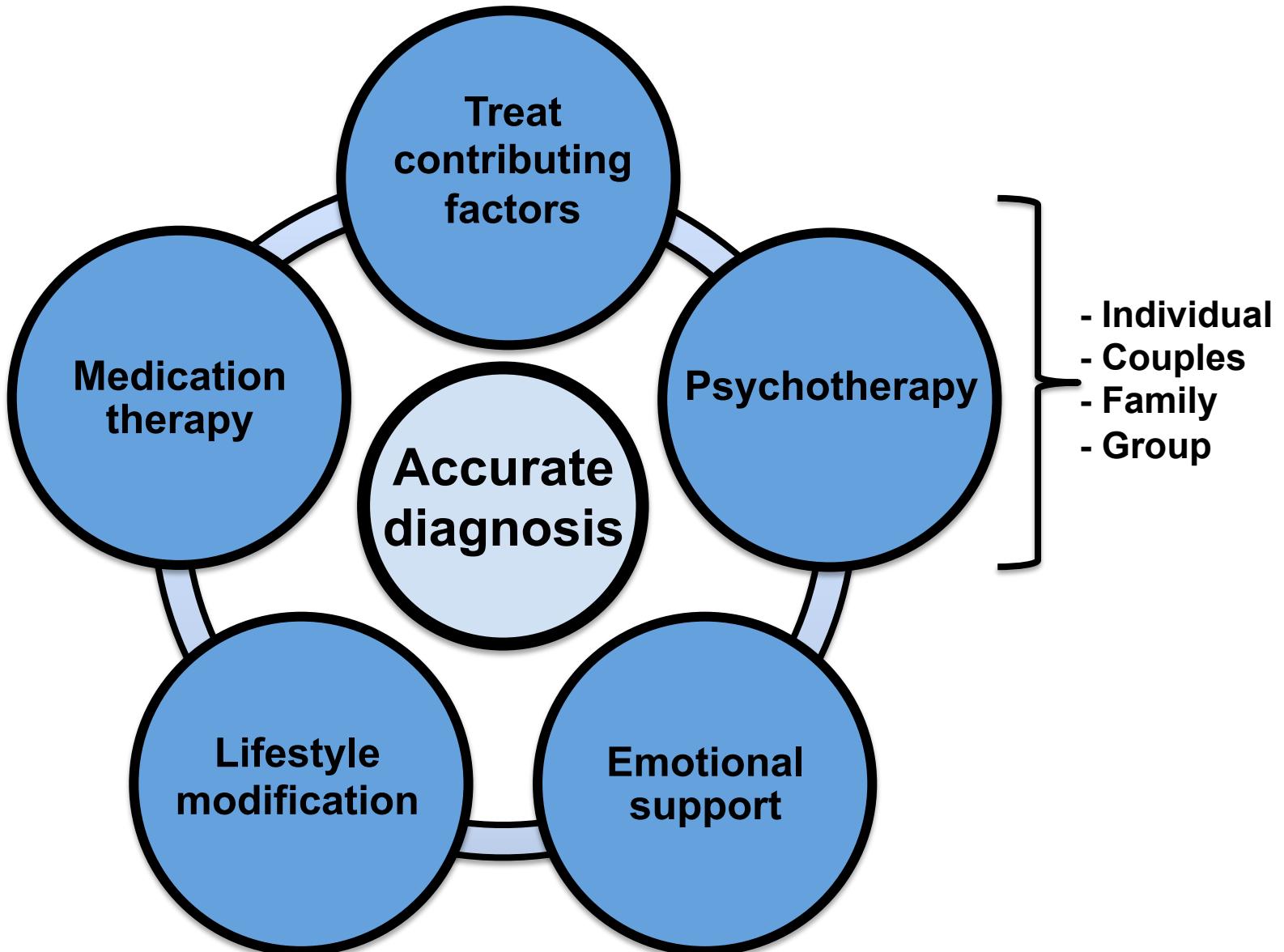
- Optimize DM treatment
  - Endocrine dysfunction
- Nutritional deficiency
  - Medication side effects

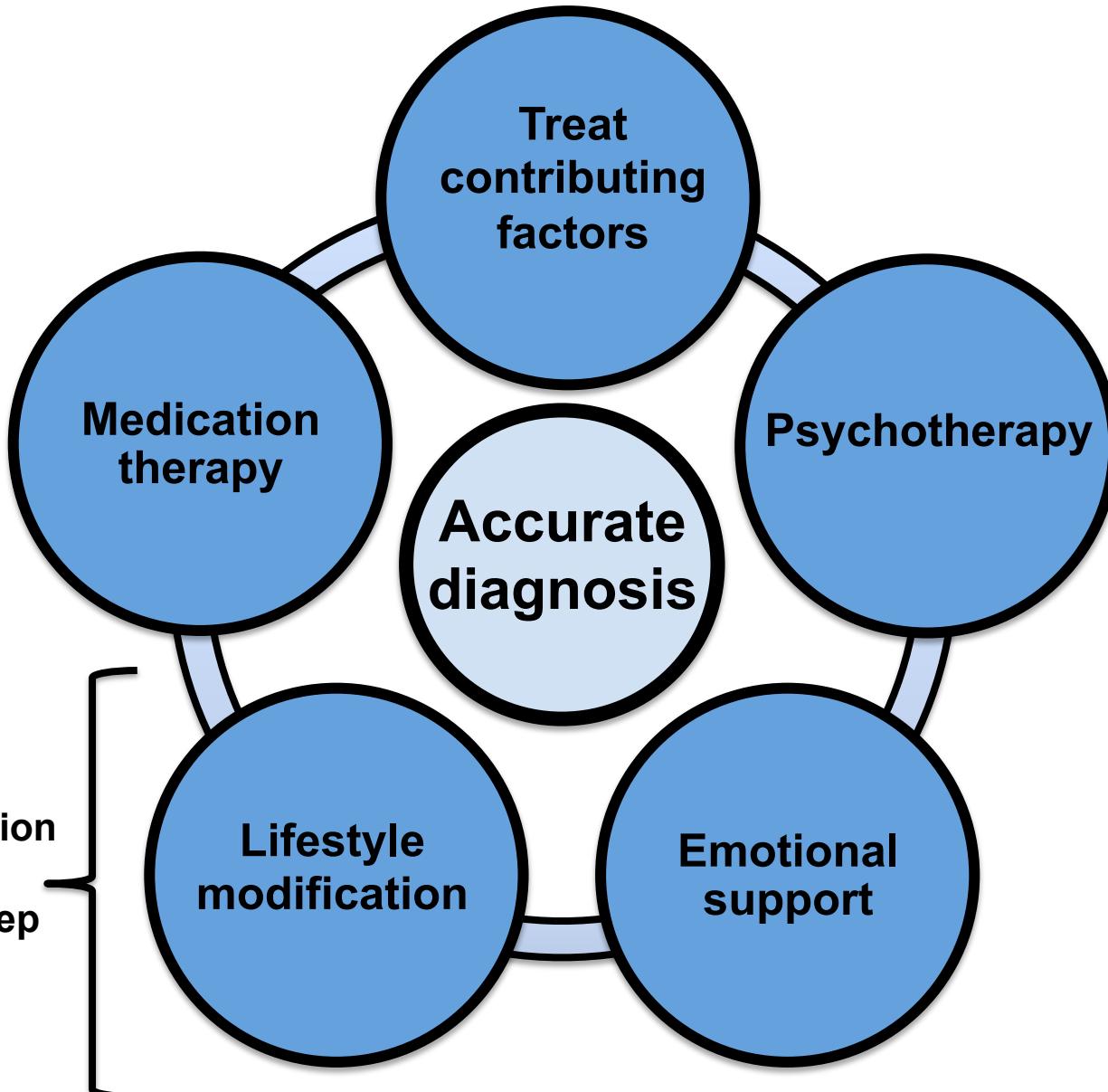


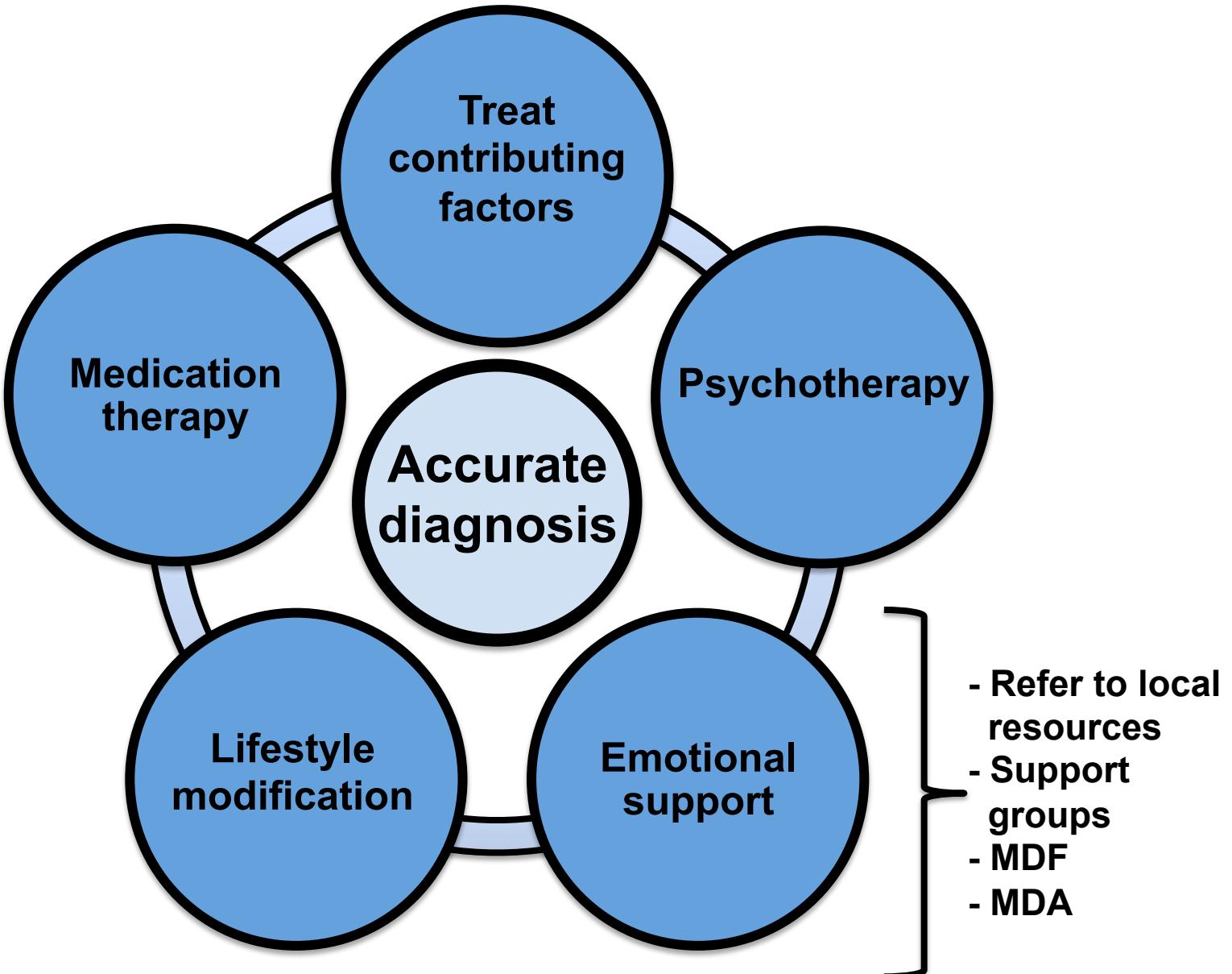
## **Classes of most commonly used:**

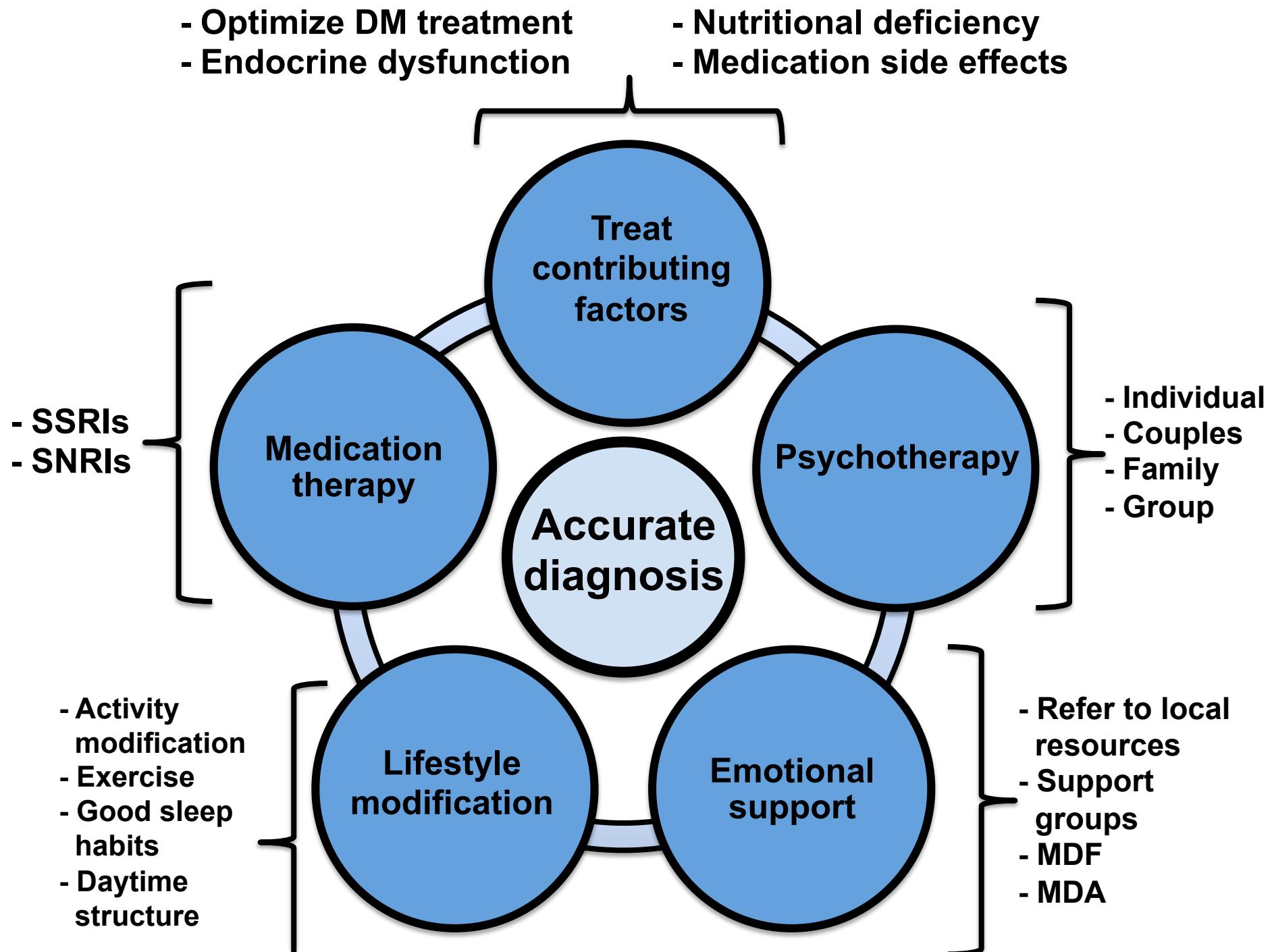
- SSRIs: selective serotonin reuptake inhibitors
- SNRIs: selective norepinephrine reuptake inhibitors











# Tips for caregivers



- Attend appointments if patient allows
  - Bring up concerns about patient's emotional health
  - Describe what symptoms you have observed
    - How long have they been present?
    - When did they occur?
- Take good care of yourself
  - Alone time is imperative
  - Get help if feeling overwhelmed
  - Inquire about community resources to assist you

*“Freedom is important, because at times it is quite heavy. A source of energy, whether it is sports or something else, gives you new energy, as long as you have something for yourself.”*

# Tips for caregivers



## Finding the right balance

- This is hard!
  - Helping versus doing in place of (partner)!!
  - Taking some risks versus giving up (person with DM1)!!

### Man partner:

*“I think it is important that you do it together. The one with the disease should not give up too easily, but should try to do what is possible. You have to find a good mode together.”*

- Two important qualities for effective partners
  - A high level of communication
  - Mutual trust and respect

# Tips for caregivers

- Reminders, lists (help organization in DM1)
- Telephone calls during day to prompt actions
- Informational meetings
  - Reduce misunderstandings
- Treatment in groups
  - Address common problems, challenges and solutions in groups led by professionals



# Gaps in knowledge

- Evidence needed to guide treatment in DM
  - Which medications are most helpful?
  - What type of psychotherapy works best?
  - Do educational interventions help?
- How does underlying biology of DM contribute to depression and anxiety?
  - How do genetics of DM correlate with occurrence of major depression?
  - Are there noticeable differences in brain structure in anxious DM individuals versus those who are not anxious?

# **OPTIMISTIC:**

# **A promising study**

- OPTIMISTIC study
  - A TREAT-NMD protocol
  - Coordinator: Baziel van Engelen, Nijmegen, Netherlands
- Focus on improving clinical practice in the management of individuals with DM1
- Aim: to improve QoL standards
- Targets: apathy & fatigue management

# Useful Resources

- Myotonic Dystrophy Foundation (MDF)
  - [www.myotonic.org](http://www.myotonic.org)
- Muscular Dystrophy Association (MDA) Support Groups
  - [www.mda.org/services/finding-support/support-groups](http://www.mda.org/services/finding-support/support-groups)
- American Psychiatric Association Provider Locator
  - [www.psychiatry.org/mental-health/key-topics/finding-help](http://www.psychiatry.org/mental-health/key-topics/finding-help)
- National Alliance on Mental Illness (NAMI)
  - [www.nami.org](http://www.nami.org)
- The National Suicide Prevention Lifeline
  - Trained telephone counselors available 24/7
  - 1-800-273-TALK (8255)

# Acknowledgments

- Our patients and research participants
- Myotonic Dystrophy Foundation
- Myotonic Dystrophy Family Registry
- Association française contre les myopathies
- Our mentors and colleagues:



- Cynthia Gagnon, Ph.D.
- Luc Laberge, Ph.D.
- Louis Richer, Ph.D.



- Eric Axelson
- Stephen Cross
- Laurie Gutmann, M.D.
- Maria Ly
- David Moser, Ph.D.
- Peg Nopoulos, M.D.

**Thank you!**



# Impacts of excessive daytime sleepiness / fatigue in DM1



**Psychostimulants** are very helpful for many individuals with DM1. When Modafinil© was been withdrawn in UK:

*“Without Modafinil my husband cannot stay awake. He needs it to be able to drive, work and interact with us. When he doesn’t take it, he sleeps everywhere”*

*“To be blunt, my wife would change from a happy, healthy fully functional woman to someone barely capable of sustaining any form of employment, social structure or anything else regarded by the rest of us as a normal life”*

# Clinical care of fatigue (CBT) Propositions



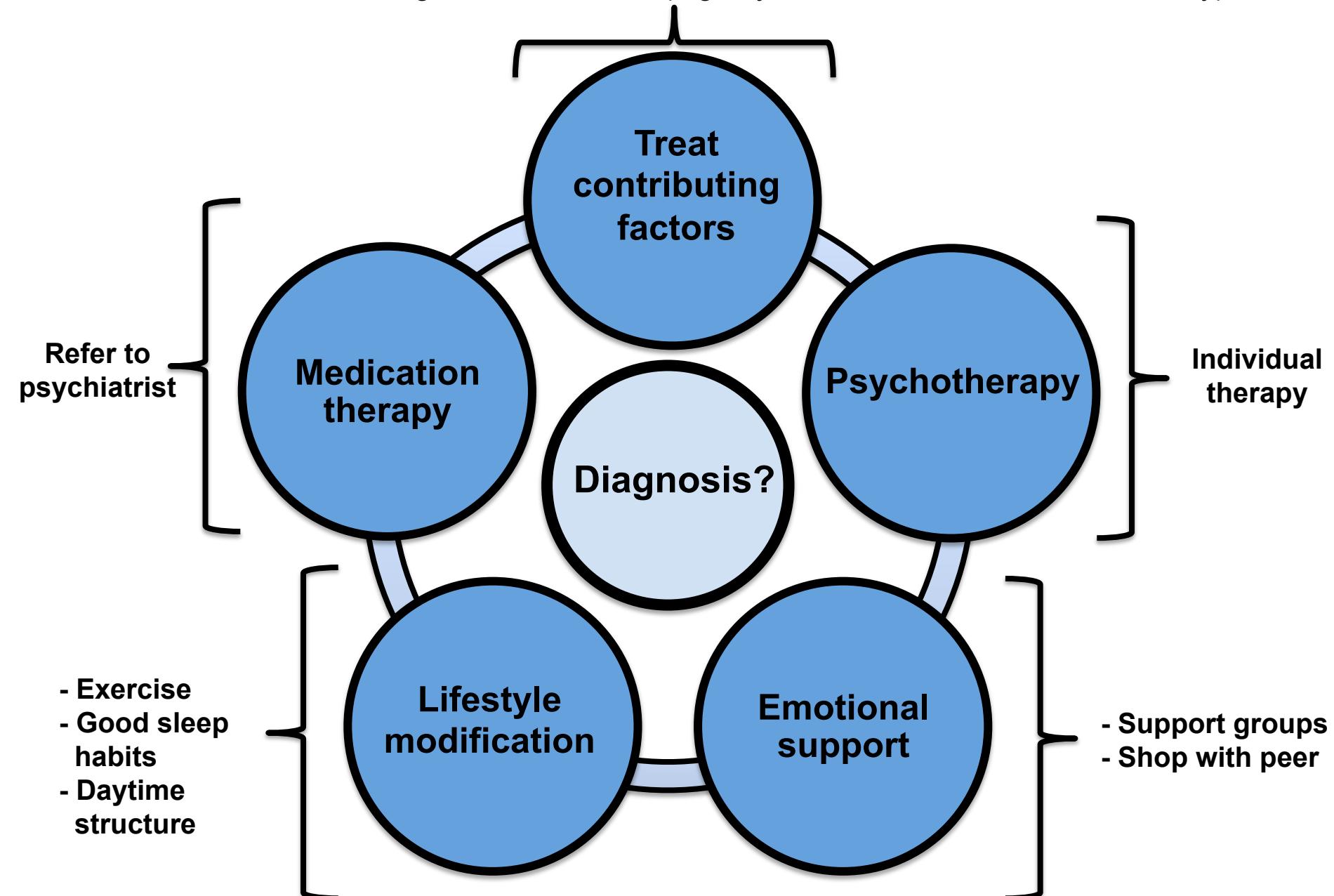
- Progressive effort training: aerobic exercise (swimming-pool, low-resistance bicycle) ≠ deconditioning
- 1<sup>st</sup> axis: EVALUATIONS: Interview about YOUR fatigue (cognitive, physical, subjective) + anxiety, sleepiness, depression scales.
- 2<sup>nd</sup> axis: BEHAVIOURS : sleep & activities agendas (daytime structure)
  - ✓ Detect ineffective and effective behaviours
  - ✓ Define clear, measurable, and patient-focused objectives and steps to success
- 3<sup>rd</sup> axis: COGNITION
  - ✓ Detect thoughts and emotions related to subjective fatigue
  - ✓ Propose other interpretations and develop adapted coping skills
- RELAXATION: permit bodily positive sensations + pain and fatigue management

# Case Discussion

*“When I go shopping now, I am much slower, and it is hard to do routine things like write a check. I have this feeling that others are watching and judging me. It is hard to shake this fear, and at times, I start to feel really panicky. My heart races, and I can’t catch my breath. I break out into a sweat and feel trapped. There have been times when I have abandoned my full grocery cart in the store and left because I felt so badly.”*

## **Coordinate with her neurologist:**

- Can her DM management be optimized in any way?
- Rule out contributing medical disease (e.g. thyroid disease, vitamin D deficiency)



# Case Discussion

*"I look at the other kids in my grade, and they seem so happy. They play on sports teams and have girlfriends. I feel so lonely and worry that this isn't ever going to change for me. Don't get me wrong- I have friends and want to do stuff with them, but I'd rather stay at home than go out with them. That way I don't have to defend myself or worry about getting stared at by other kids. I don't think people understand me or what I'm going through. Sometimes I think it would be so much easier if I was never born or could just die."*

# Principles of treatment

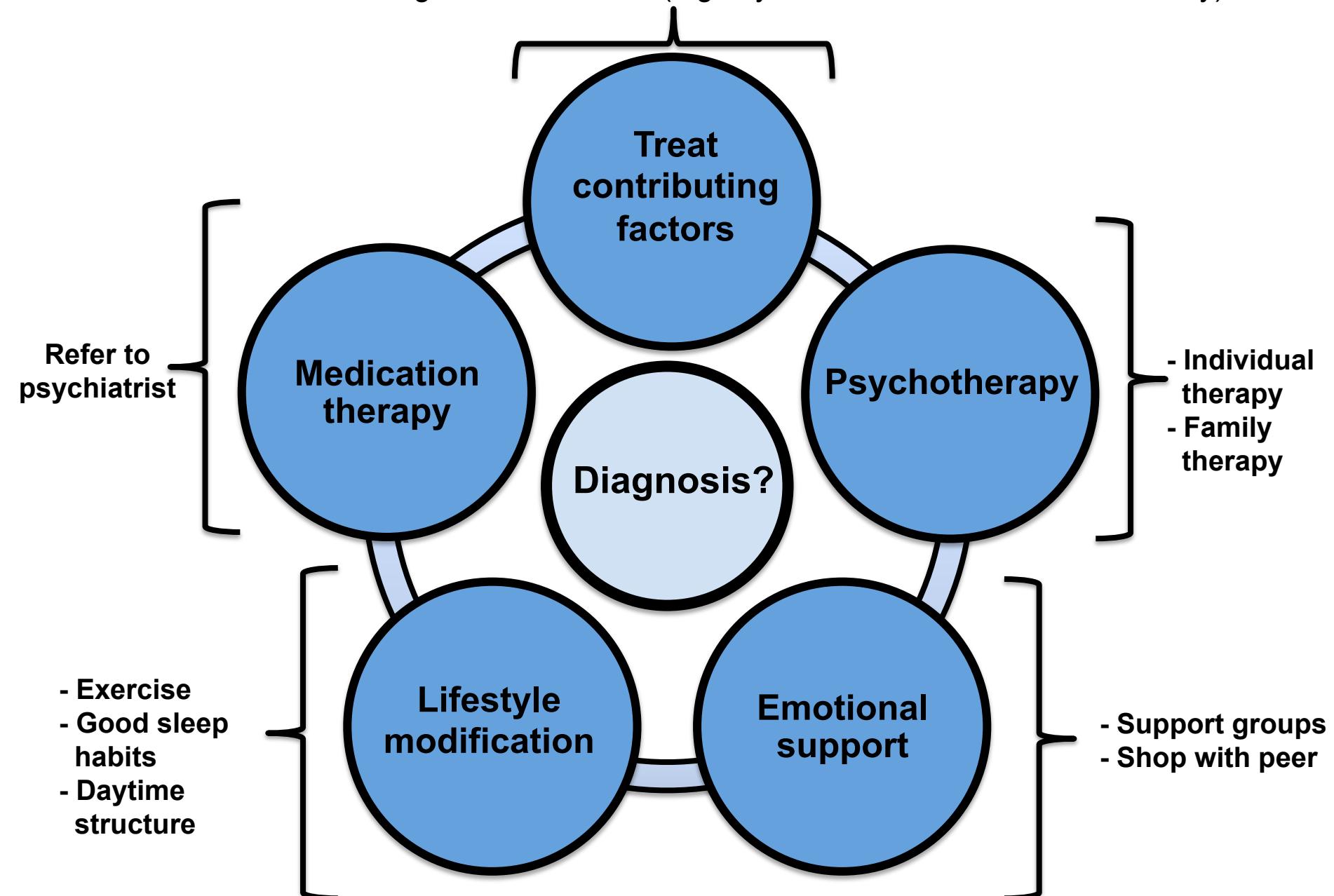


1. Depression and anxiety are treatable conditions
2. Safety comes first
  - Get help immediately for thoughts of suicide or plan for self harm
3. Accurate diagnosis is critical
4. Team approach
  - Close coordination among providers
  - Involve caregiver
5. Must be individualized



## **Coordinate with his neurologist:**

- Can his DM management be optimized in any way?
- Rule out contributing medical disease (e.g. thyroid disease, vitamin D deficiency)



# DM and white matter disruptions

