

Appendix A

SURVEY

Please fill in this survey. The survey is anonymous. The collected data would serve exclusively for statistical analysis, and it would be published only in a summary form as a group to establish a baseline of knowledge related to Fragile X syndrome.

The Fragile X Syndrome (FXS) is the most common cause of inherited intellectual disability, and the most common cause of single gene mutation caused autism spectrum disorders (ASD). It is the result of a full mutation of the *FMR1* gene. The FXS is a rare disease, with the prevalence of 1:4,000 in males, and 1:8,000 in females. It is known that up to 60% of those with FXS are comorbid with ASD. Therefore, according to the international guidelines, individuals diagnosed with ASD of unknown cause and/or intellectual disability, should be tested for the *FMR1* gene mutation.

Gender: M F Age: _____ Academic year of medical studies: _____

Section I: General knowledge of FXS

1. The symptoms of the Fragile X Syndrome (FXS) can first be seen:
 - a. during intrauterine development.
 - b. in the early childhood (during the first few years of life).
 - c. during puberty.
 - d. in the adulthood.

2. Early treatment of the FXS is based on:
 - a. non-pharmacological methods.
 - b. pharmacotherapy.

3. Pharmacotherapy in children with the FXS is usually started:
 - a. during the first year of life.
 - b. between 2-5 years of age.
 - c. between 5-7 years of age.
 - d. during puberty.
 - e. in the adulthood.

4. According to the results of studies, the frequency of pharmacotherapy in individuals with FXS is:
 - a. 15-25% in adults
 - b. 25-40% in newborns
 - c. 40-55% in children aged 1-5 years
 - d. More than 60% in children aged 10 years and over.

5. Pharmacotherapy in patients with FXS is most commonly:
 - a. causal/targeted therapy.
 - b. symptomatic therapy.

6. Pharmacotherapy in individuals with FXS can modify:
 - a. behavioral disorders.
 - b. attention disorders.
 - c. hyperactivity.

- d. sleeping disorders.
 - e. All listed answers are correct.
7. Preclinical studies on animal models of FXS (e. g. experimental mice):
- a. cannot be conducted
 - b. can be conducted, but the results are not predictive for humans.
 - c. can be conducted, but they are expensive, slow and inadequate.
 - d. are conducted with great success.
8. Clinical studies, that would help develop new pharmacological agents in treating the fragile X syndrome:
- a. have never been conducted, and they are not planned to start.
 - b. have not been conducted yet, but they are going to start soon.
 - c. are currently being conducted, and some have already finished.
 - d. have been stopped due to negative results and severe side effects, and they will not be conducted again.

Section II: Knowledge of pharmacotherapy of FXS

1. Attention Deficit Hyperactivity Disorder (ADHD) is frequently seen in individuals with FXS. What should be used in treating this disorder?
- a. Anxiolytics.
 - b. Mood stabilizers.
 - c. Antidepressants.
 - d. Psychostimulants.
2. What should be used to treat sleeping disorders in individuals with FXS (regardless of the presence of autism spectrum disorders, ASD)?
- a. Anxiolytics.
 - b. Antihistamines.
 - c. Melatonin.
 - d. Low doses of antiepileptics.
3. Alpha-adrenergic agonist (e. g. clonidine) can be used in individuals with FXS to treat which of the following conditions?
- a. Mild to moderate hypertension.
 - b. Aggressiveness.
 - c. Anxiety.
 - d. Sleeping disorders.
4. Guanfacine, that is used to treat sleeping disorders and ADHD in individuals with FXS, is:
- a. Histamine receptors antagonist.
 - b. Serotonin receptors antagonist.
 - c. Alpha-adrenergic receptors agonist
 - d. Beta-adrenergic receptors agonist.
 - e. Muscarinic acetylcholine receptors antagonist.
5. In individuals with FXS, Selective Serotonin Reuptake Inhibitors (SSRI) should be used to treat which of the following conditions?
- a. Mood disorders
 - b. Anxiety

- c. Attention deficit
- d. Hyperactivity

6. Anxiety is a common symptom in individuals with FXS. What would you use to treat mild to moderate anxiety in these patients?

- a. Diazepam or lorazepam
- b. Clomipramine
- c. Sertraline
- d. Low doses of haloperidol

7. Which of the following drugs should not be used to treat aggressiveness, that is a common symptom of FXS?

- a. Risperidone
- b. Aripiprazole
- c. Olanzapine
- d. Methylphenidate

Section III: Self-assessment of education in neurodevelopmental disorders during medical studies

1. During studies:

- a. I have never heard about FXS
- b. I have heard about FXS, but don't know much about FXS
- c. I gained basic knowledge of FXS
- d. I learned about FXS in detail.

2. Do you think that, during studies, you gained sufficient knowledge about pharmacotherapy of neurodevelopment disorders, including FXS and ASD?

- a. Yes
- b. No

3. Do you think that education in the field of pharmacotherapy of neurodevelopment disorders should be more intense?

- a. Yes
- b. No