RARE DISEASES

SERBIA KEEPING PACE WITH THE WORLD: ADVANCES IN THE SCIENCE AND HEALTHCARE OF FRAGILE X SYNDROME

Dragana Protić,[1,2]

[1]*University of Belgrade, Faculty of Medicine, Department of Clinical Pharmacology and Toxicology, Belgrade, Serbia*

[2]*Special Hospital for Cerebral Palsy and Developmental Neurology, Belgrade, Serbia*

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Background: Fragile X syndrome (FXS) is a rare disease with prevalence 1/4000 males and 1/7000 females. It is a neurodevelopmental disorder caused by a full mutation in the *FMR1* gene. FXS is the leading cause of inherited intellectual disabilities and the most commonly known genetic cause of autism spectrum disorder. Children with FXS experience behavioural and sleep problems, anxiety, inattention, learning difficulties, and speech and language delays. There are no approved medications for FXS; however, there are several interventions and treatments aimed at managing the symptoms and improving the quality of life of individuals with FXS.

Methods and Objectives: This presentation aims to summarize all healthcare and scientific activities in the field of fragile X over the past decade in the Republic of Serbia. The study utilizes a comprehensive review of presented results.

Results: The results will include following: (i) Research studies have been conducted to understand the genetic and molecular basis of FXS. Collaborative research with international institutions has helped advance the understanding of FXS in Serbia; (ii) Healthcare initiatives: Development and implementation of specialized healthcare programs for individuals with Fragile X. Introduction of programs for early detection and diagnosis of FXS; (iii) Enhanced training for healthcare professionals to improve diagnosis, management, and care for patients with FXS; (iv) Support and advocacy: Establishment of support group and networks for families affected by fragile X; (v) International collaboration: Participation in international consortia and research projects to align with global standards and practices in fragile X research and care in Serbia.

Conclusions: Over the past decade, Serbia has emerged as a regional leader in the field of Fragile X. Through pioneering research and comprehensive healthcare initiatives, supported by international and local institutions, Serbia serves as a model for best practices and innovative approaches to managing and understanding FXS worldwide.

Keywords: fragile X syndrome, rare diseases, Serbia