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REVIEW

**of the dissertation by Ainur Ontalapovna Umurzakova
entitled "Clinical and genetic characteristics of Duchenne muscular dystrophy in the Republic of
Kazakhstan" submitted for the degree of Doctor of Philosophy (PhD)
in the specialty 6D110100 – "Medicine"**

prepared by the international scientific supervisor,
Professor Maciej R. Krawczyński MD, PhD,
Head of the Department of Medical Genetics and Chairman of the Bioethical Committee
at Poznan University of Medical Sciences, Poland.

The dissertation research is dedicated to an urgent and significant issue in modern medicine - the clinical and genetic characterization of Duchenne muscular dystrophy (DMD) first done in the Republic of Kazakhstan. The choice of topic is driven by the high social relevance of the disease, its profound impact on patients' quality of life, and the necessity to improve diagnostic methods and management strategies for this group of patients.

The author has conducted extensive scientific work, encompassing clinical observation, molecular genetic testing, and comprehensive statistical data analysis. The dissertation is characterized by a thorough theoretical exploration of the subject, extensive review of relevant literature, and the use of modern molecular diagnostic methods. The study analyzes the major types of mutations, their distribution across the functional domains of the dystrophin protein, and their correlation with clinical manifestations of the disease. Of particular note is that this study showed the impact of reading frame disruption on disease severity, the rate of motor function decline, and the prognosis for the duration of the ambulatory period.

The scientific novelty of the research lies in the identification of the specific molecular genetic mutation spectrum in the Kazakhstani population, determination of the frequency of inherited and de novo mutations, and the establishment of correlations between genetic characteristics and clinical outcomes. The structure of the dissertation is logical, the presentation of material is clear, consistent, and scientifically justified. The results of the study have been presented at scientific conferences and published in peer-reviewed journals, underscoring the high scientific value of the obtained data.

Thus, the dissertation entitled "Clinical and genetic characteristics of Duchenne muscular dystrophy in the Republic of Kazakhstan" represents a complete original scientific study that meets the requirements for dissertations submitted for the Doctor of Philosophy (PhD) degree. The author fully deserves to be awarded the sought academic degree.



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