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Review of Doctoral Thesis

*“Clinical-epidemiological characteristics of mucopolysaccharidoses in children in the Republic of Kazakhstan”*  
by Tulebayeva Assel Kairatovna  
Specialty: 6D110100 (Medicine)

PhD candidate Assel Tulebayeva has undertaken an important and burning problem concerning the prevalence, diagnosis and treatment of patients with mucopolysaccharidoses (MPS) type I, II, VI and IV in the Republic of Kazakhstan.

In my opinion this is the first such detailed study, covering clinical, biochemical, and molecular characterization of the mentioned mucopolysaccharidoses, including the effects of enzyme replacement therapy currently available.

PhD candidate Assel Tulebayeva made a retrospective analysis (from 1998 to 2016) of the prevalence of these diseases in the Republic of Kazakhstan by estimating their incidence at 0.75 per 100 000 live births. She also found the mutual ratio of the incidence of particular types of mucopolysaccharidoses showing that the most common is MPS II. These results provide a more reliable estimation of the incidence of MPS. What is notably important for prospective decisions regarding diagnostics including newborn screening and treatment of MPS patients in the Republic of Kazakhstan.

In cooperation with foreign centres, a number of new mutations, not described so far, have been identified in the Kazakh population. This allowed to establish the molecular database for the Kazakh population, but also to make a significant contribution to the general knowledge of the correlation between genotype and phenotype in these mucopolysaccharidoses. What is interesting is that in the study population PhD candidate did not find correlation between the genotype and phenotype, showing that similar genotypes may lead to different phenotypic expression.



