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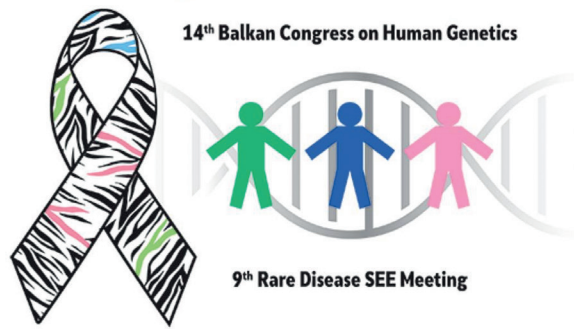


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# ABSTRACT BOOK

14<sup>th</sup> Balkan Congress of Human Genetics  
and 9<sup>th</sup> Rare Disease SEE Meeting

*“Genetic Diseases  
from Diagnostics to Prevention and Therapy”*

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PP-14

## NEW APPROACH IN QUANTITATIVE ESTIMATION OF X CHROMOSOME CENTROMERE INSTABILITY IN ALZHEIMER DISEASE

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**Background:** Chromosome instability (CIN) in Alzheimer's disease has been found in peripheral blood lymphocytes, and also in neurons of affected individuals. CIN is a result of errors in chromosome segregation during mitosis leading to numerical and structural chromosomal abnormalities.

**Material and Methods:** Fluorescent In Situ Hybridization. (FISH) has been a method of choice for the evaluation of CIN in various diseases. We have studied a specific type of X chromosome instability, or premature centromere division (PCD) in interphase nuclei of the hippocampus brain tissue neurons from sporadic AD females. PCD was studied by using a new approach to FISH.

**Results:** Namely, we have established a methodology that allows quantitative analysis of centromere fluorescence intensity, i.e. the size of

individual FISH spots, thus giving a more focused view of the centromere region of interest. By using the micro image system, our results show that quantitative FISH can distinguish PCD positive signals vs. PCD negative signals in cells of brain tissue.

**Conclusion:** This could be a specific biomarker in affected cells in order to verify CIN, thus supporting the established qualitative analysis of FISH spots in the study of centromere and chromosomal alterations in interphase nuclei of patients affected by AD.

**Keywords:** *Chromosome instability, Alzheimer disease, X chromosome, premature centromere division biomarker*

**Topic:** *Neurogenetics and intellectual disability*