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DEVELOPMENT OF METHOD FOR SHORT REPEATS EXPANSION CAUSED ATAXIAS DIAGNOSTIC

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There are known many hereditary ataxias caused by short repeats expansions, most prominent of them being Huntington disease, Friedreich ataxia and a number of spinocerebellar ataxias (SCA`s).

These ataxias are very similar with each other being sometimes almost phenocopies, so a molecular-genetic diagnostic method is strictly necessary for differential diagnostics. There is also a well-known effect called genetic anticipation, when some alleles, usually ones with a quite large, but still normal number of repeats may further expand during gametogenesis or first zygote divisions. It puts potential their children of people harboring these alleles at risk of disease development. That's why a screening of affected child's parents and their siblings is necessary.

A number of previously-published papers shows that number of repeats is most frequently directly correlated with age of disease onset and speed of its progression. So, counting repeats number may give not only diagnostic, but prognostic information for affected persons.

In this work we developed a diagnostic approach based on a combination of agarose gel AFLP and capillary electrophoresis-based fragment analysis to detect short repeats expansion and calculate total repeats number for a number of diseases, including different forms of SCA, Huntington disease and Friedreich ataxia. As a test group a selection of patient's DNA with presupposed diagnosis of SCA or Friedreich ataxia stored in LGMU biobank was used. As a control group were used DNA of presumptive healthy individuals from the same biobank.

Keywords: SCA, repeats, Friedreich, Huntington, ataxia.