

Repeat Expansion Analysis in GeneMarker® software: Streamlined workflow for custom or commercial chemistries of tri- and hexa- nucleotide repeat data, including Huntington's Disease (HTT), Amyotrophic Lateral Sclerosis/Frontotemporal Dementia (ALS, C9ORF72) and Dystrophia Myotonia Protein Kinase (DMPK)

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Kayla Hendricks, Teresa Snyder-Leiby, Ning Wan SoftGenetics, LLC State College PA

Introduction

Expansions of simple sequence repeats, mainly but not limited to tri-nucleotide repeats, are responsible for over 40 human diseases.¹ In general, an increasing number of repeats results in more severe phenotype and the number of repeats increase (expand) as the disease gene is inherited.²

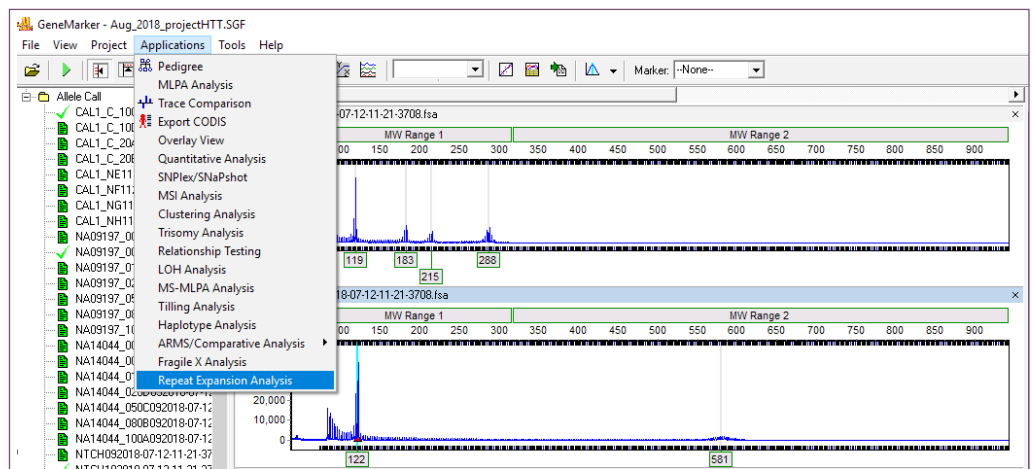
GeneMarker is a user-friendly tool for rapid and accurate genotyping of repeat expansion data (Figure 1). The new linked Repeat Expansion Application which

- avoids the potentially error prone step of data transfer.
- provides a straight forward user interface to lock in analysis templates that conform to laboratories' standard operating procedures.
- performs the repetitive calculations for converting fragment size to repeat length (Figures 2 and 3).
- print or save final reports with customized header (Figure 4).

Procedure

1. Import raw data files, make size and allele calls and select Applications – Repeat Expansion Analysis (no need to export sized data to a second analysis software).

Figure 1: Link directly to the Repeat Expansion Analysis application from the sized data.



2. Select from a list of analysis templates, or create/modify existing templates

Figure 2.

Select a template or create one by entering a descriptive template title (Huntingtons, C9orf72, DMPK....)

Enter the appropriate Report category, Repeat Cutoff values _____ and if desired, Highlighting color.

Calculate the C0 and M0 or enter values from lab validation studies. _____

