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Implementation and validation of an improved allele specific stutter filtering method for electropherogram interpretation

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Highlights

- Novel implementation of allele specific stutter filtering for electropherograms.
- Combined stutter peaks have additive values for forward and back stutter applied.
- Allele specific model trialed against traditional model of locus specific filters.
- True minor allelic peaks filtered as stutter reduced from 101 to 22.
- True stutter peaks not filtered reduced from 85 to 17.

Abstract

Modern probabilistic genotyping (PG) software is capable of modeling stutter as part of the profile weighting statistic. This allows for peaks in stutter positions to be considered as allelic or stutter or both. However, prior to running any sample through a PG calculator, the examiner must first interpret the sample, considering such things as artifacts and number of contributors (NOC or N).

Stutter can play a major role both during the assignment of the number of contributors, and the assessment of inclusion and exclusion. If stutter peaks are not filtered when they should be, it can lead to the assignment of an additional contributor, causing N contributors to be assigned as N+1. If peaks in the stutter position of a major contributor are filtered using a threshold that is too high, true alleles of minor contributors can be lost.

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