Congenica

Enabling genomic medicine

Take the leap from GRCh37 to GRCh38

Enabling better NGS analysis

Aligning your NGS data to the GRCh38 reference genome provides clear advantages. GRCh38 is of higher quality, having corrected numerous mistakes and assembly issues present in GRCh37. As a result, working with the GRCh38 reference genome makes it possible to detect variants that would otherwise be missed and to avoid the pitfall of identifying false positive variants present in GRCh37 that have since been determined not to be real. Plus, as many data sources such as Decipher are now moving soley to GRCh38 and not supporting GRCh37, migration is required to ensure continued access to new knowledge.

Despite such clear advantages, it can be challenging for laboratories to migrate to using the GRCh38 reference genome. Most labs maintain a historic repository of variants that they've analysed and reported on, which informs their analysis of new cases. This data is often stored in isolated databases, spreadsheets and/or text files and must be converted from GRCh37 to GRCh38 coordinates, or even transcript-based coordinates, before it can be applied in the new environment.

Congenica can help with this challenge. Through our professional services, we will transform your variant repository into GRCh38 coordinates, enabling you to continue to leverage your historic data while accessing the benefits of working with the more accurate reference genome.

Overview of the lift over process:

1. Meet with a Congenica data analyst to review your legacy data and desired outcome
2. Congenica performs an initial review of the data and provides a statement of the work to be done
3. A Congenica data analyst prepares the data and performs lift over to GRCh38 coordinates
4. Transformed data is uploaded into the Congenica platform as a custom Curated Variant List (CVL) or delivered in a structured file format

Additional reanalysis services

Previously processed samples with BAM files can be realigned to GRCh38 with new variant calls produced against the new alignment, providing the possibility of identifying previously undetected variants. This approach is equivalent to performing a new analysis, with no loss of data.

Previously processed samples with VCF files can be lifted over to GRCh38 coordinates and rerun through the platform. As this service relies on standard coordinate mappings between genomes some loss of data is anticipated, but is expected to be outweighed by gaining access to the latest variant annotations.

Discover Congenica today

There's no need to persist in using an outdated GRCh37 reference genome for your analyses. Reach out to us today – we're happy to discuss the specifics of your data conversion needs.

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