Human Genome version 38 FAQ

General Questions

1. Is hg38 the same genome version as GRCh Build 38?

Yes, they are the same version of the human genome. GRCh Build 38 stands for "Genome Reference Consortium Human Reference 38" and it is the primary genome assembly in GenBank; hg38 is the ID used for GRCh Build 38 in the context of the UCSC Genome Browser.

2. Essentially, how is GRCh Build 38 different from hg19?

The hg19 build is a single representation of multiple genomes. The GRCh Build 38 build provides alternate sequences ("alt_sequences") for some genomic regions for which their variability prevents adequate representation by one single reference. There are some other differences such as changes in the centromere representation, the mitochondrial genome, updated sequences and an "Analysis Set" (FASTA format sequences in a package convenient for use by various Next Generation Sequence read alignment pipelines).

3. When are you making the change?

We are gradually making the transition over the next month or so. Ion Reporter™ software has already transitioned, and Primer Designer™, Applied Biosystems® Analysis Module Variant Analysis™ software, and Applied Biosystems® Analysis Module Next-Generation Confirmation™ software will transition on November 11th.

4. Why are you making this change?

We are moving to GRCh Build 38 because it is a more accurate representation of the human genome and is the most recent genome version being accepted by the human genome community. The change allows us to link out to newer annotations presented in the context of GRCh Build 38. Many of the annotation databases have moved their content to correspond the GRCh Build 38 coordinates.

5. How is the GRCh Build 38 reference used by Thermo Fisher Scientific software different from the references publically available from places like UCSC or NCBI?

- The version used by our software is based on GRCh38.p2 (http://www.ncbi.nlm.nih.gov/assembly/GCF_000001405.28)
- Repeat and SNP locations are soft-masked into lower case letters, while the ambiguous IUPAC bases, duplicated centromeric arrays and chrY PAR regions are hard masked into 'N's.
- It contains chr1-22, chrX, chrY, chrM, and chr22_KI270879v1_alt.
- Contig chr22_KI270879v1_alt is hard masked except region 269814-279356 (1-based).
- Gene GSTT1 is located at chr22_KI270879v1_alt:270308-278486.

6. I know there are many more "alt_sequences", why is that your version of GRCh Build 38 only considers one of those?

Our version of GRCh Build 38 only considers the chr22_KI270879v1_alt. This alt chromosome contains gene GSTT1 that was part of chr22 in hg19.

7. Can I download the GRCh Build 38 files from NCBI and use them directly for my analyses of Ion sequencing data?

We strongly recommend that you download our version of the GRCh Build 38 from our website. This version is the one that is assumed in all of our software applications.

8. Do you have a conversion tool from hg19 coordinates to GRCh Build 38 coordinates?

At this moment we do not offer any conversion tools. There are publicly available tools to convert coordinates, such as UCSC's liftover tool: http://genome.ucsc.edu/cgi-bin/hgLiftOver

9. Can I analyze Ion Ampliseq™ assays or panels that use the hg19 human reference with the new GRCh build 38 human genome reference?

No. The old assays and panels were created using hg19 as a reference and should be analyzed with the tools and analysis pipelines created for hg19.

Primer Designer Transition

10. Can I still order HG19 primers from Primer Designer after the transition?

We removed a small fraction of primers from our inventory because they do not map to GRCh Build 38 anymore. All other primers will have the same assayid and are the same primers, but presented in the context of GRCh Build 38.

The Primer Designer Tool will no longer support ordering of hg19 primers that do not map to hg38 after November 11th. If you know the sequences of your primers, you can order custom primers through this link:

https://www.thermofisher.com/us/en/home/products-and-services/product-types/primers-oligos-nucleotides/invitrogen-custom-dna-oligos.html.

11. If I search by the same coordinates I used for HG19, will I get the same primers?

No, we cannot guarantee that you will get the same primers.

12. When can I buy GRCh Build 38 primers? Do I have to wait?

GRCh Build 38 Primers will be available for purchase through the Primer Designer tool after November 11th. Until then, only hg19 primers are available.

CE Software Analysis

13. How can I analyze my data with HG19 primers after transition?

After the transition, you should use software and analysis pipelines that still support hg19. CE desktop software, such as Variant Reporter™ and SeqScape™, will still continue to support hg19.

You can also perform analysis using the Variant Analysis or Next-Generation Confirmation cloud apps, but only by creating a reference using an hg19 Genbank file. If the hg19 Genbank file has annotations, VA/NGC will report out variants based on your provided annotations. In NGC, you also need to provide the NGS variant file (vcf file) which is generated using hg19 as a reference.

14. Will the Variant Analysis and Next-Generation Confirmation cloud apps support analysis of data using GRCh Build 38 primers?

Yes, the Variant Analysis and Next-Generation Confirmation cloud app will support all primers that map to GRCh Build 38, and you will be informed with an error message if the primer pair does not map to this release.

NGS Transition

15. Can I still design, order and analyze old Ion AmpliSeq panels based on hg19?

Yes. The pipelines and tools for using hg19 as reference for design and analysis will still be available.

16. Can I still annotate my old variants (based on hg19) with Ion Reporter Software?

Yes. The variant calling workflow based on hg19 will be available in Ion Reporter software. If your design was created using GRCh Build 38, then you can also call and annotate variants using Ion Reporter.

17. Can I copy amplicons from a hg19 design to a GRCh Build 38 design (or vice versa)?
No. Amplicons from a custom design can only be copied to another custom design associated with the same reference. It is not possible to copy amplicons to a custom design associated with a different reference.

18. Will there be a new version of the ready to use AmpliSeq Panels based on GRCh Build 38?

Not at this moment. The off-the-shelf panels will still be based on hg19.

19. Will there be a version of the Oncomine™ Panels based on GRCh Build 38?

Not at this moment. The Oncomine panels will still be based on hg19.

20. Are your Ion Reporter Software annotations based on GRCh Build 38 or hg19?

If your AmpliSeq design has been created using GRCh Build 38 as a reference, then you can create and ad-hoc workflow in Ion Reporter for analysis. All analysis and annotations will take in consideration GRCh Build 38 as a reference. However at this moment there are no GRCh Build 38 workflows in Ion Reporter. The tools for analysis and the annotations for hg19 will still be available.