Supplementary material: Whole Genome Analysis of Ovarian Granulosa Cell Tumors Reveals Tumor Heterogeneity and a High-Grade TP53-Specific Subgroup

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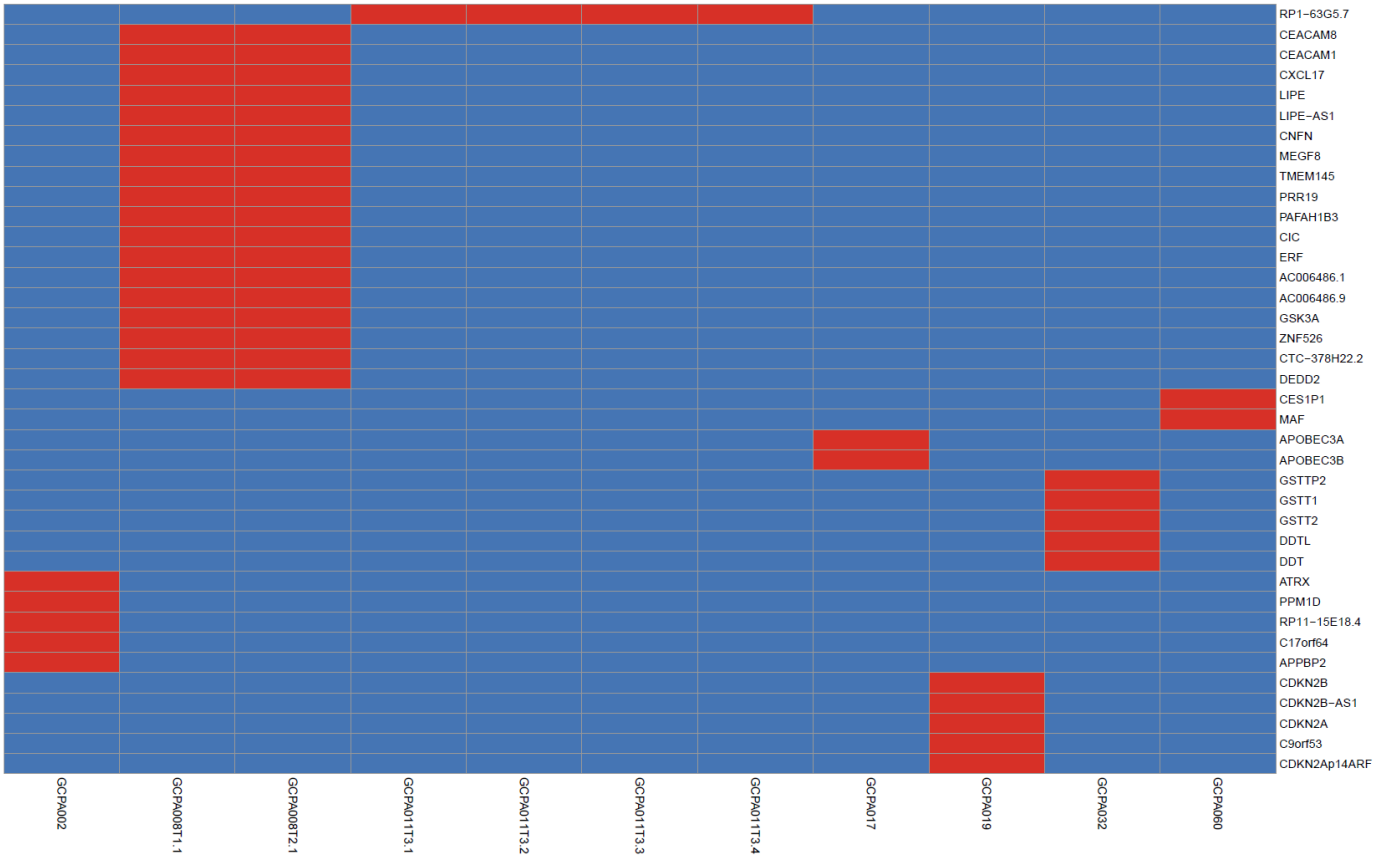
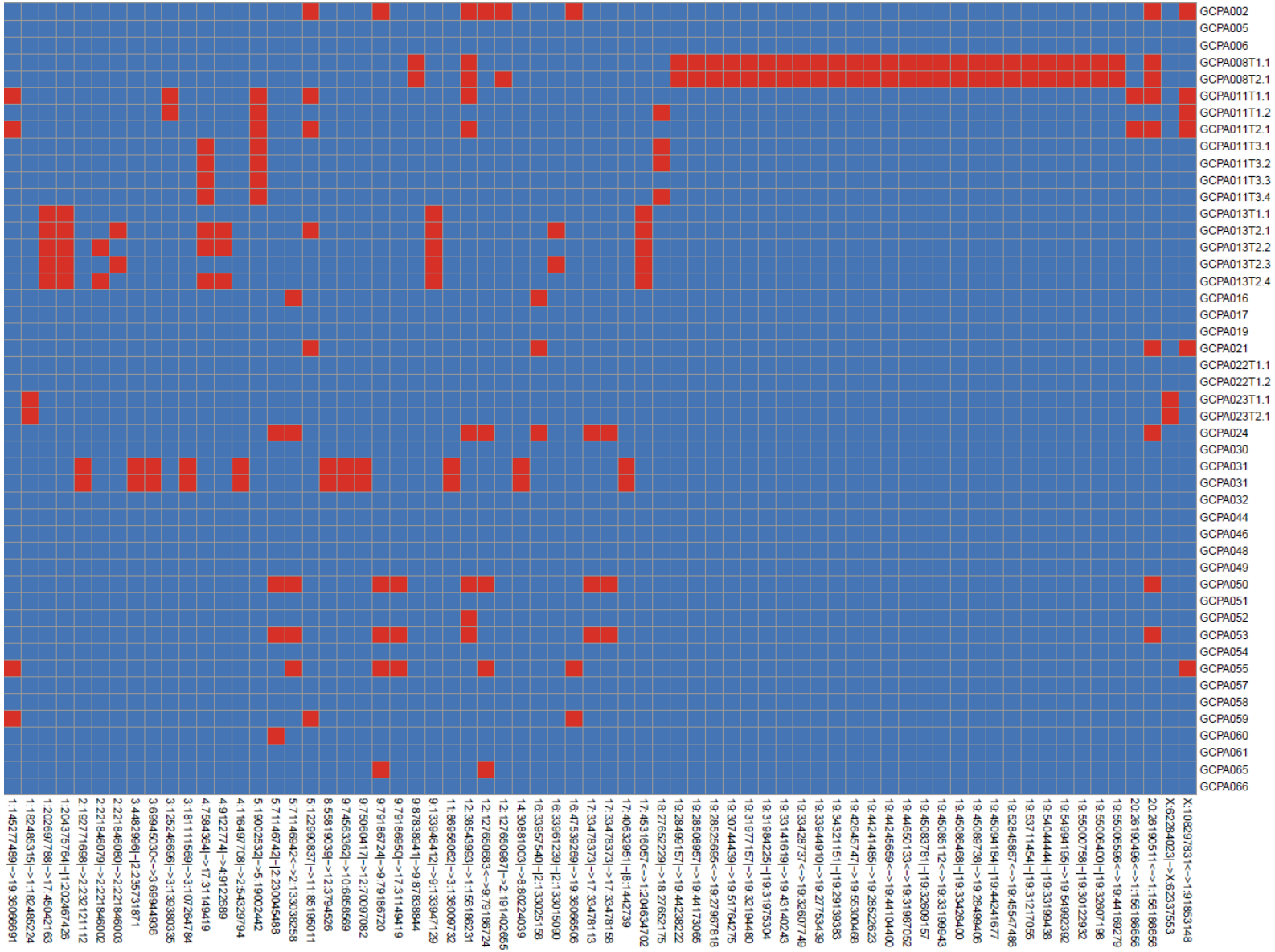


Figure S1. Full gene loss.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GCPA065
GCPA066
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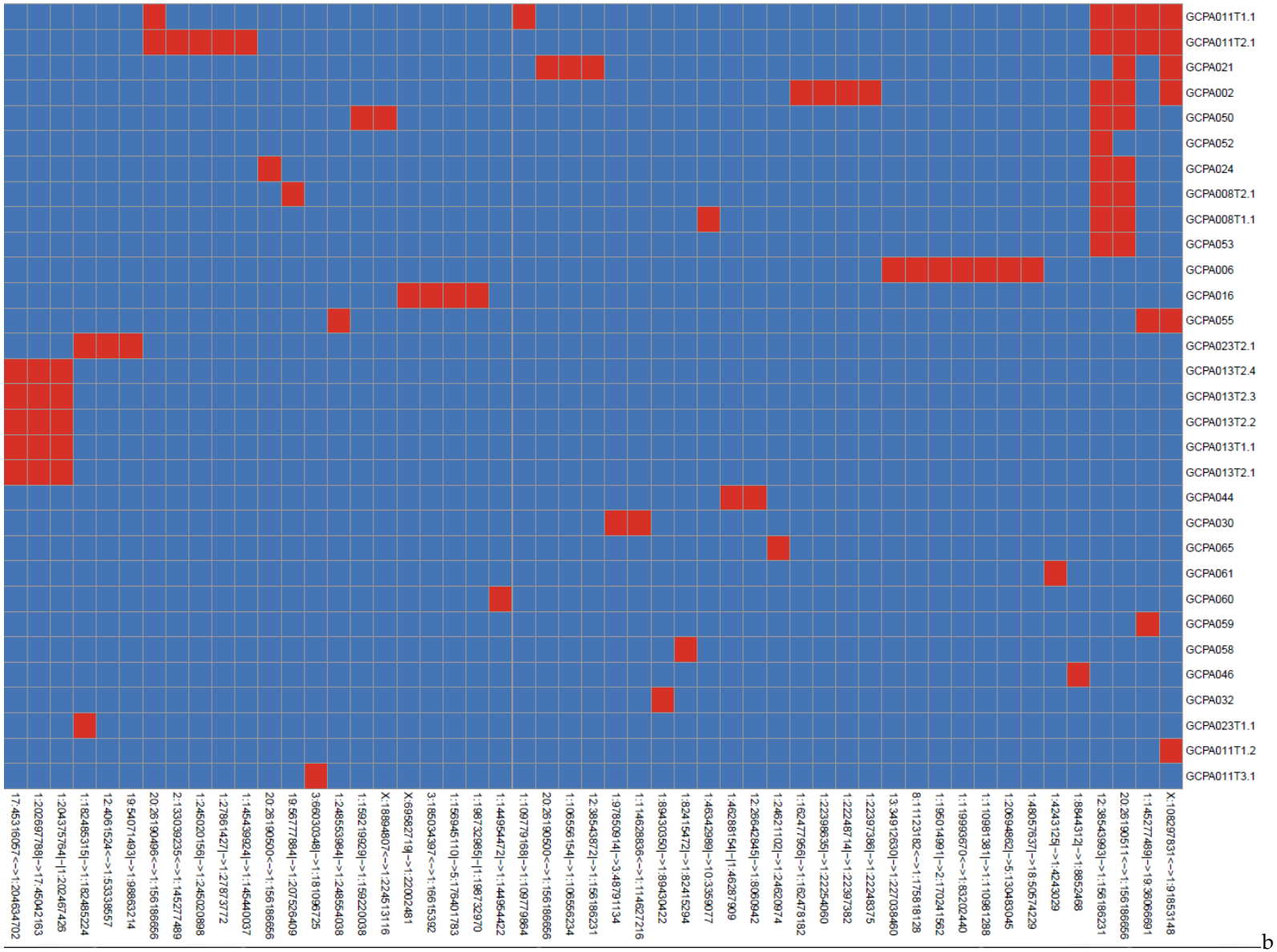
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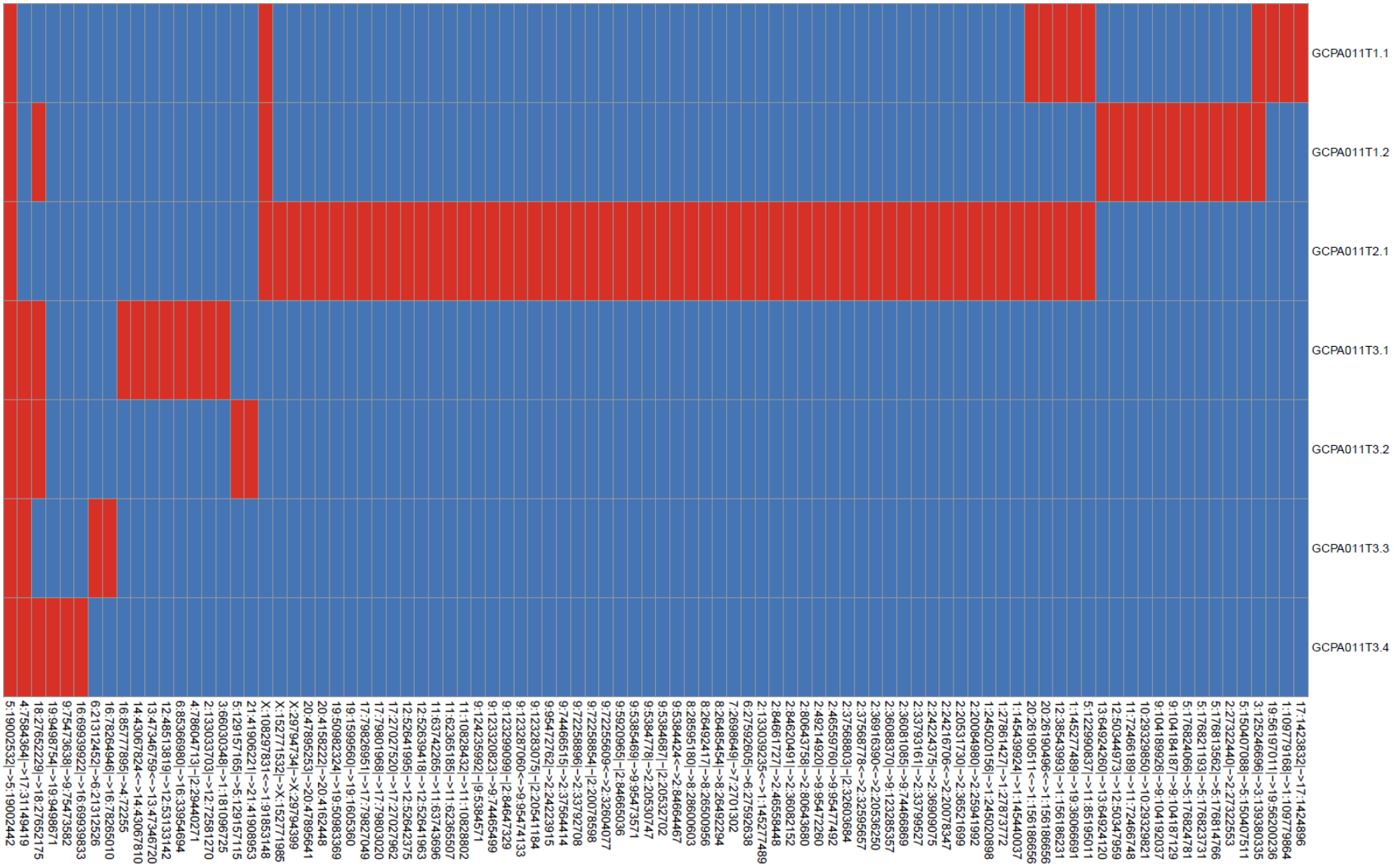
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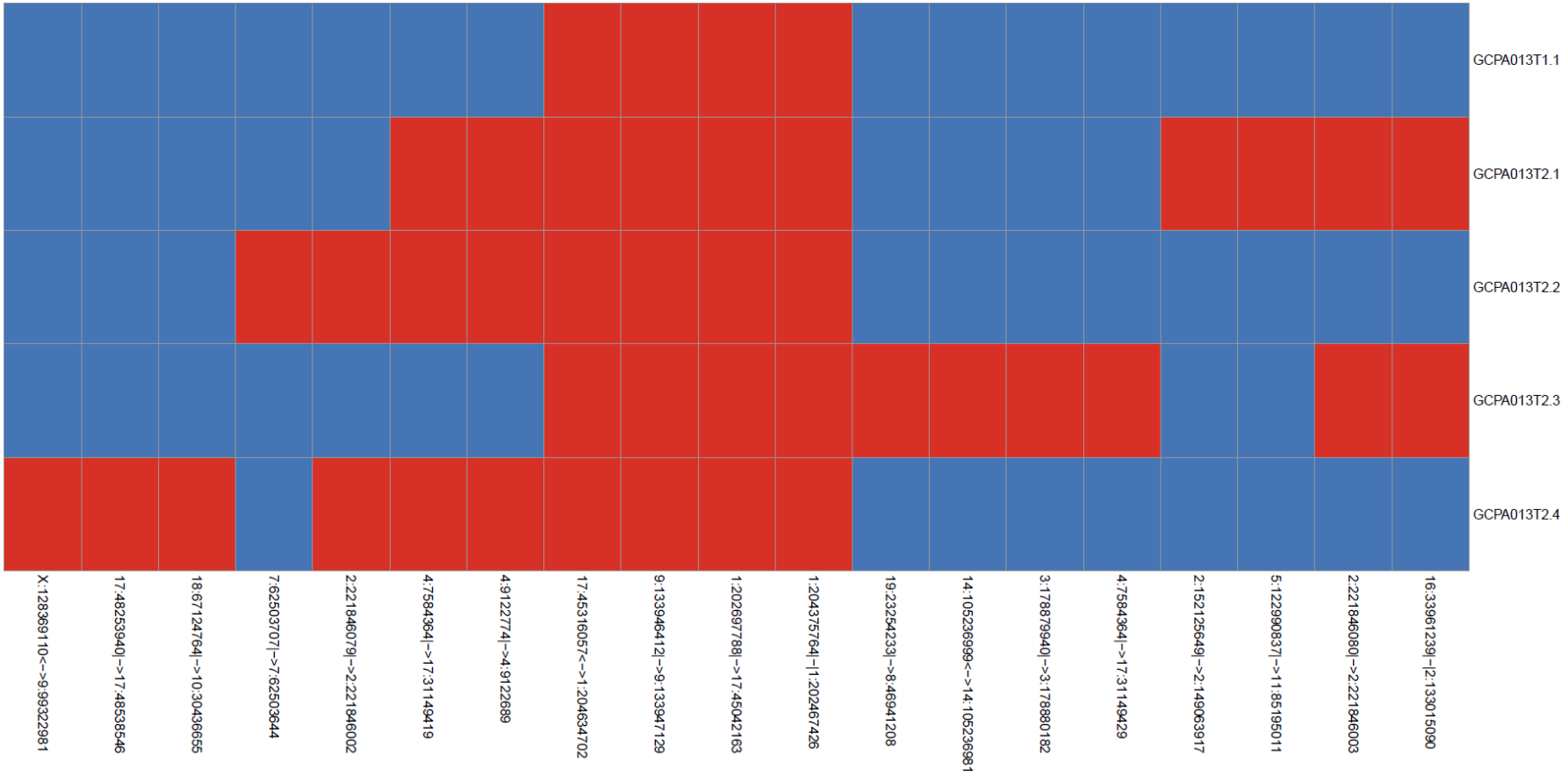
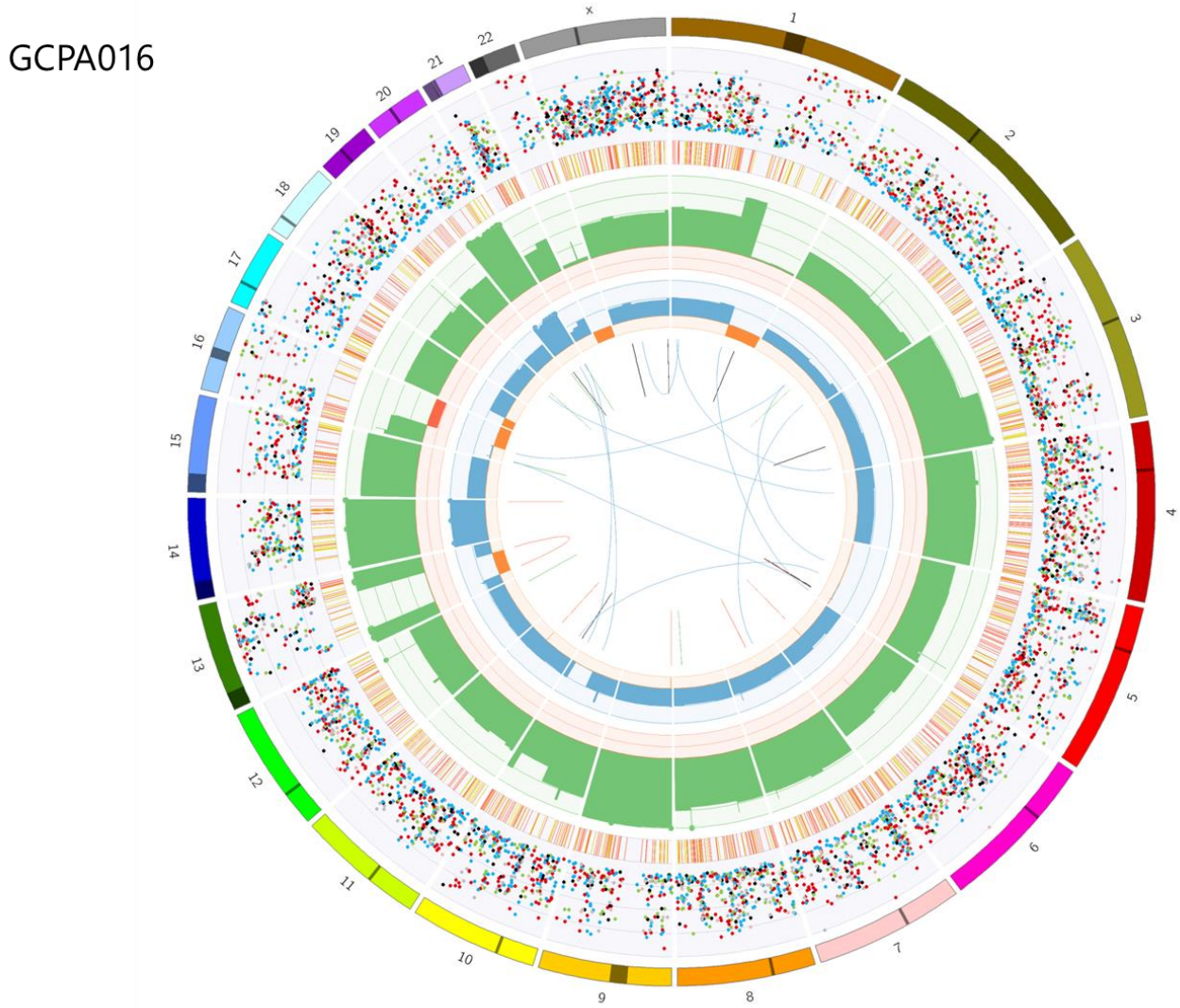


d

Figure S2. Structural Variants. **a**. Structural Variants in more than 1 sample**. b**. Structural Variants in chromosome 1*. **c**. Structural Variants in patient 11**. d**. Structural Variants in patient 13**.** In the labels, "|->" marks a front-to-back breakpoint (preserved direction), "|-|" a back-to-back breakpoint and "<->" a front-to front breakpoint. *The overlapping breakpoint shown in the figure are not exactly on the same position, but detected within a range of 50 base pairs.

GCPA002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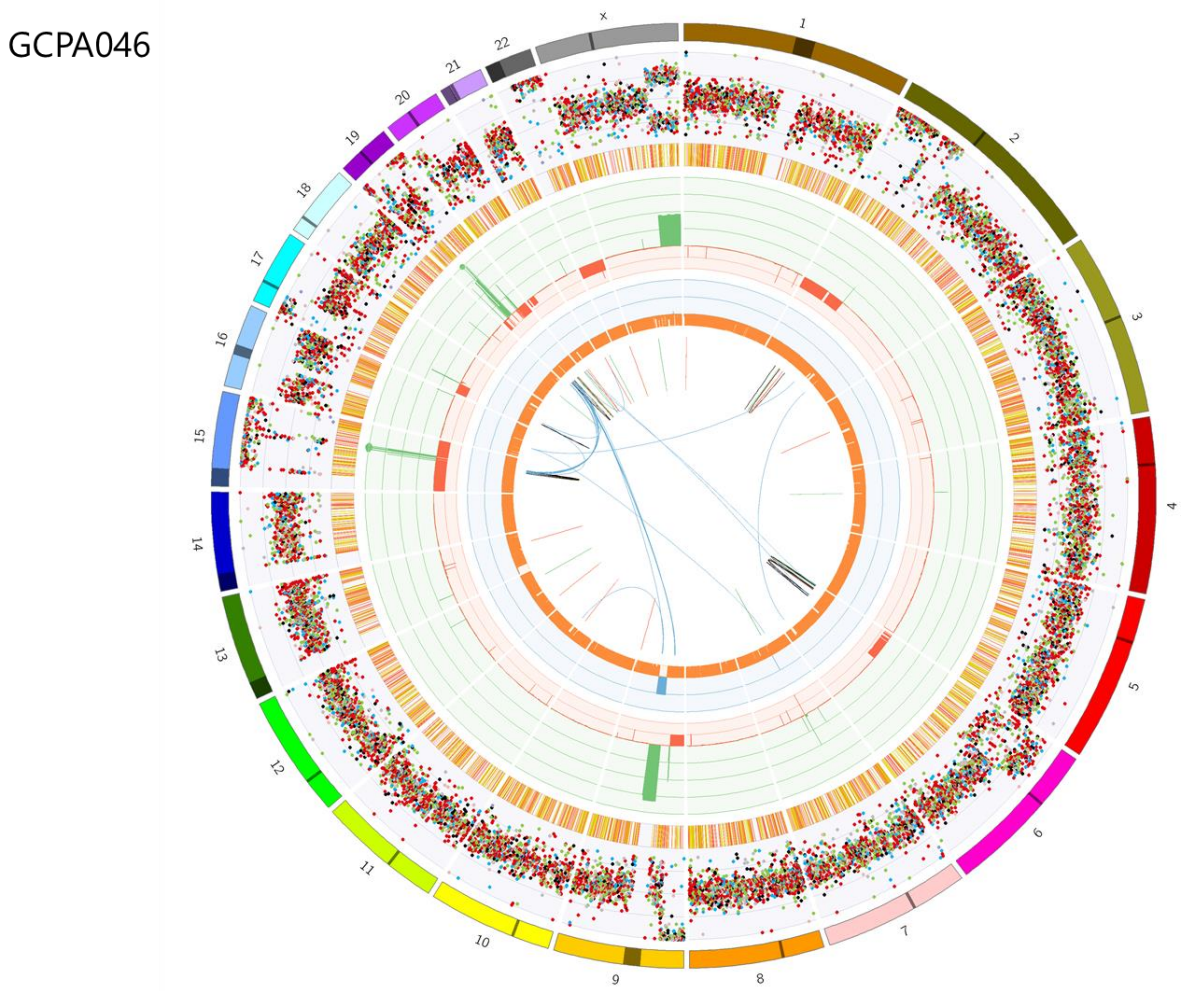
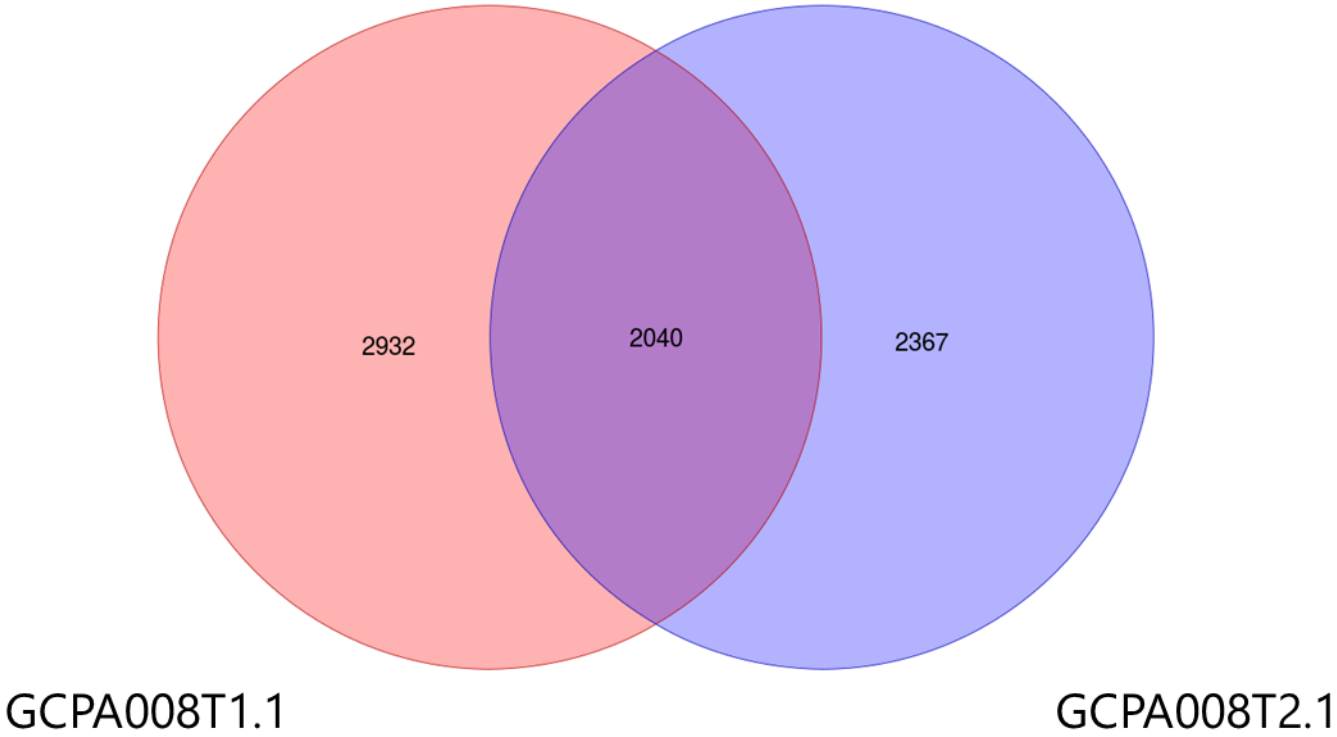
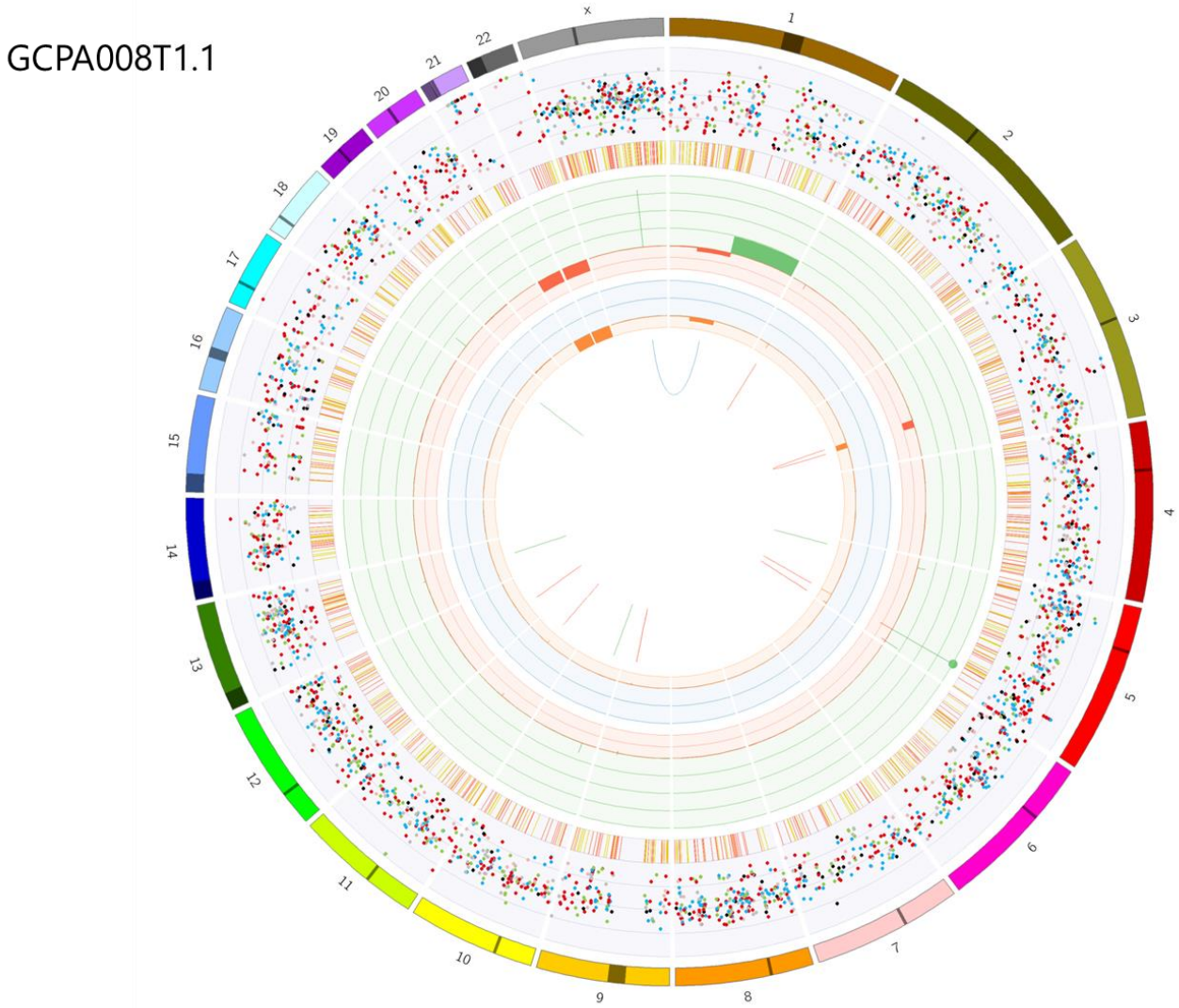


Figure S3. Circos plots and venn diagrams. a. Circos plots of TP53 mutant samples.

Number of overlapping and unique variants

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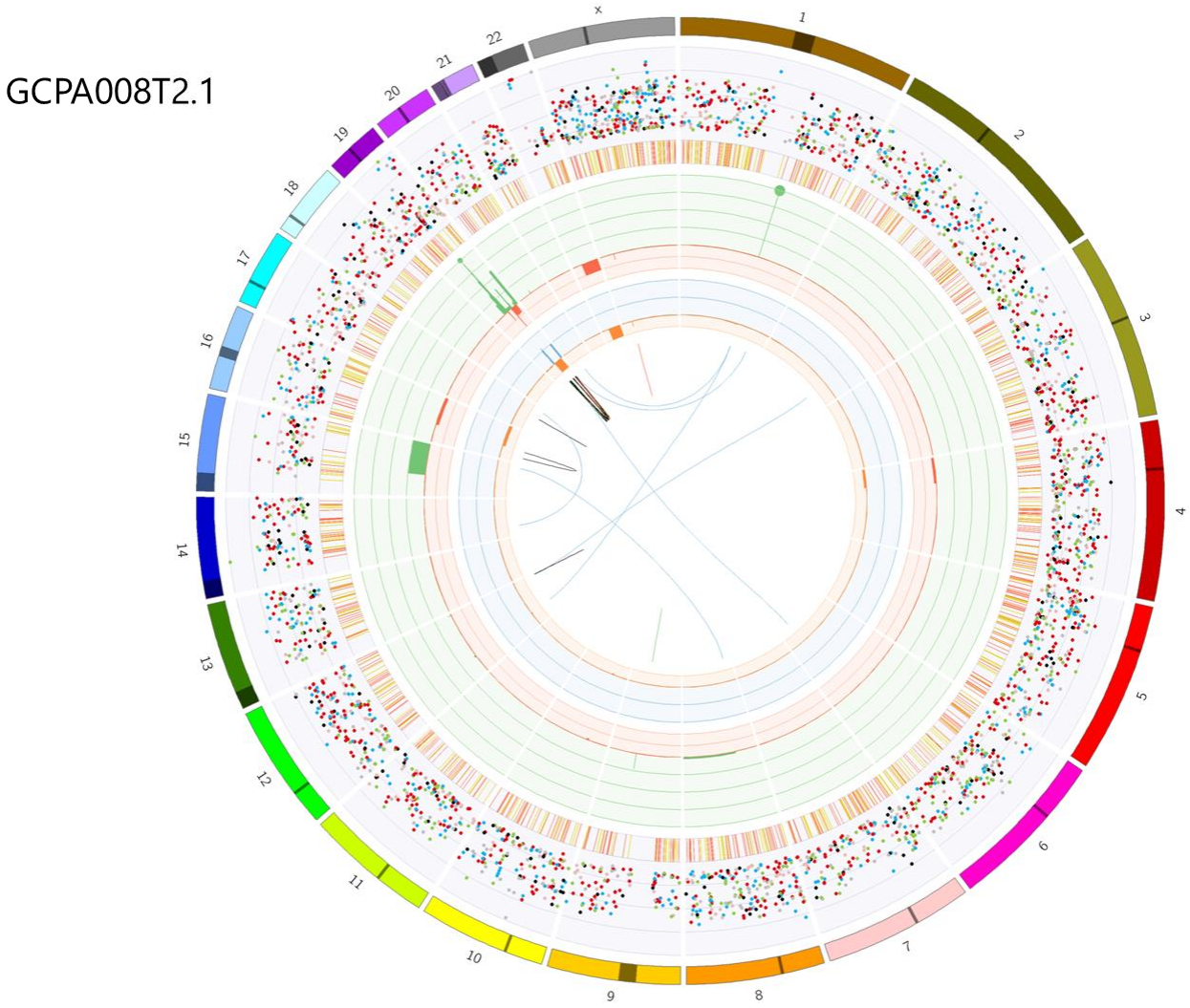
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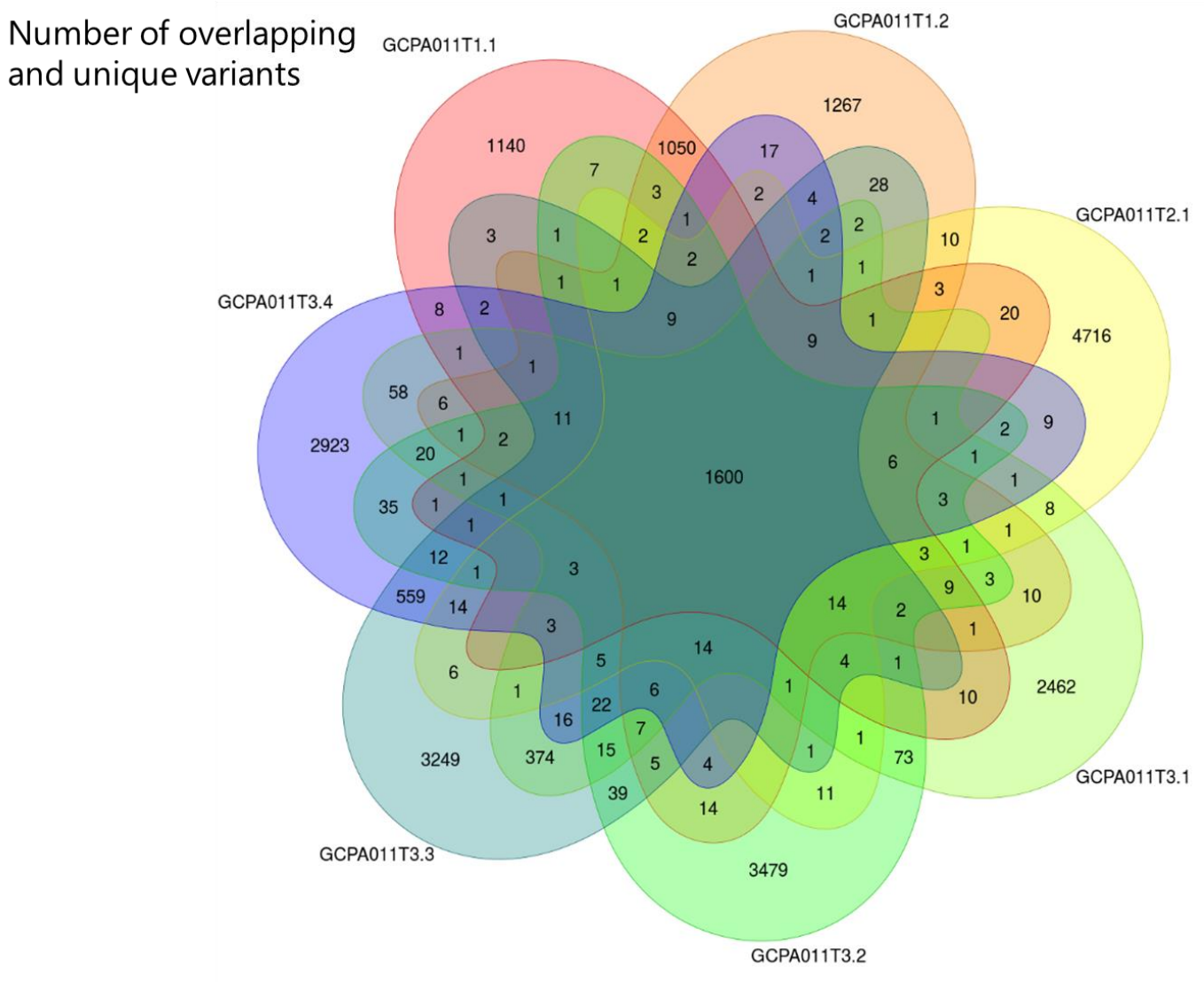
Number of overlapping and unique variants
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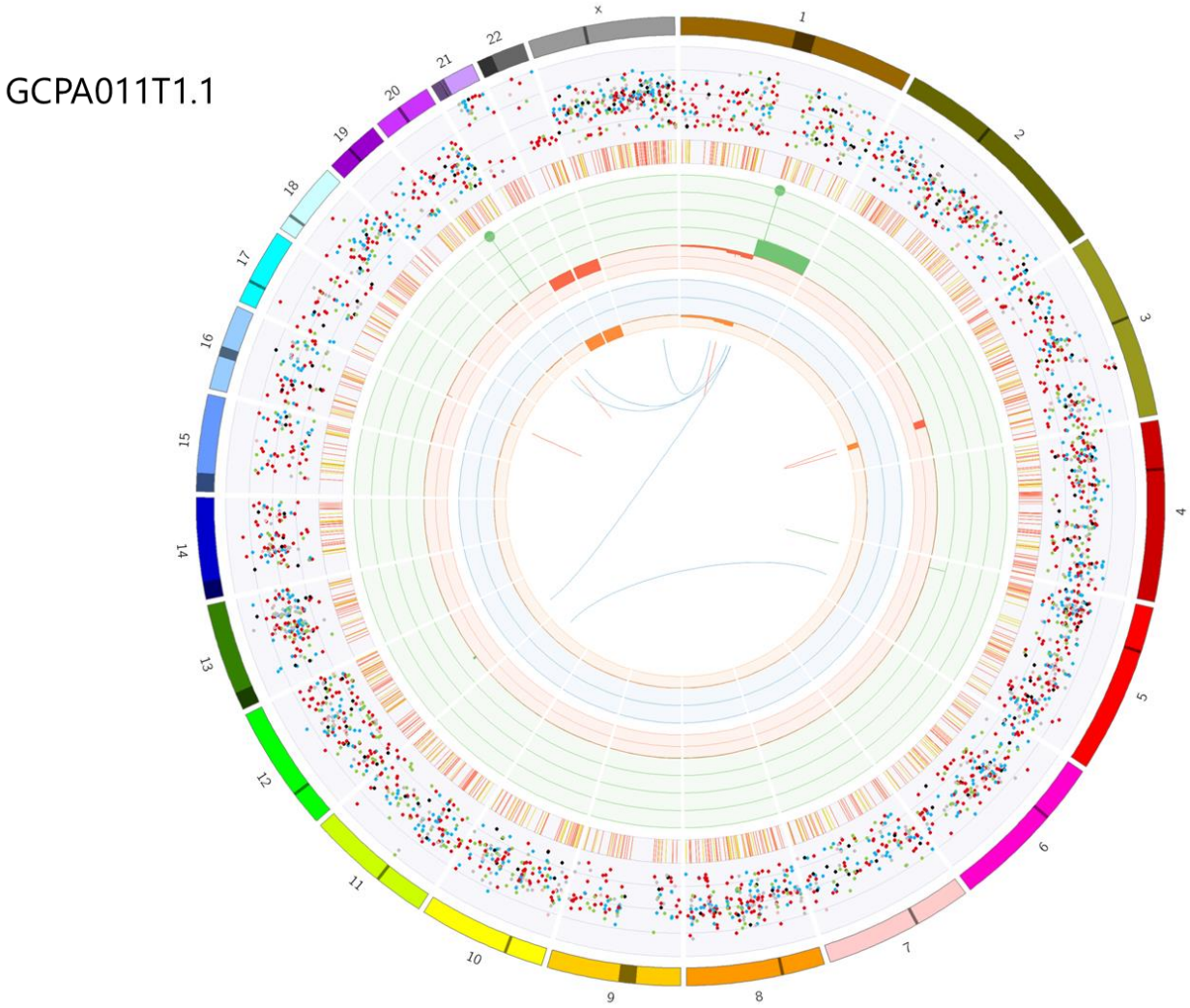
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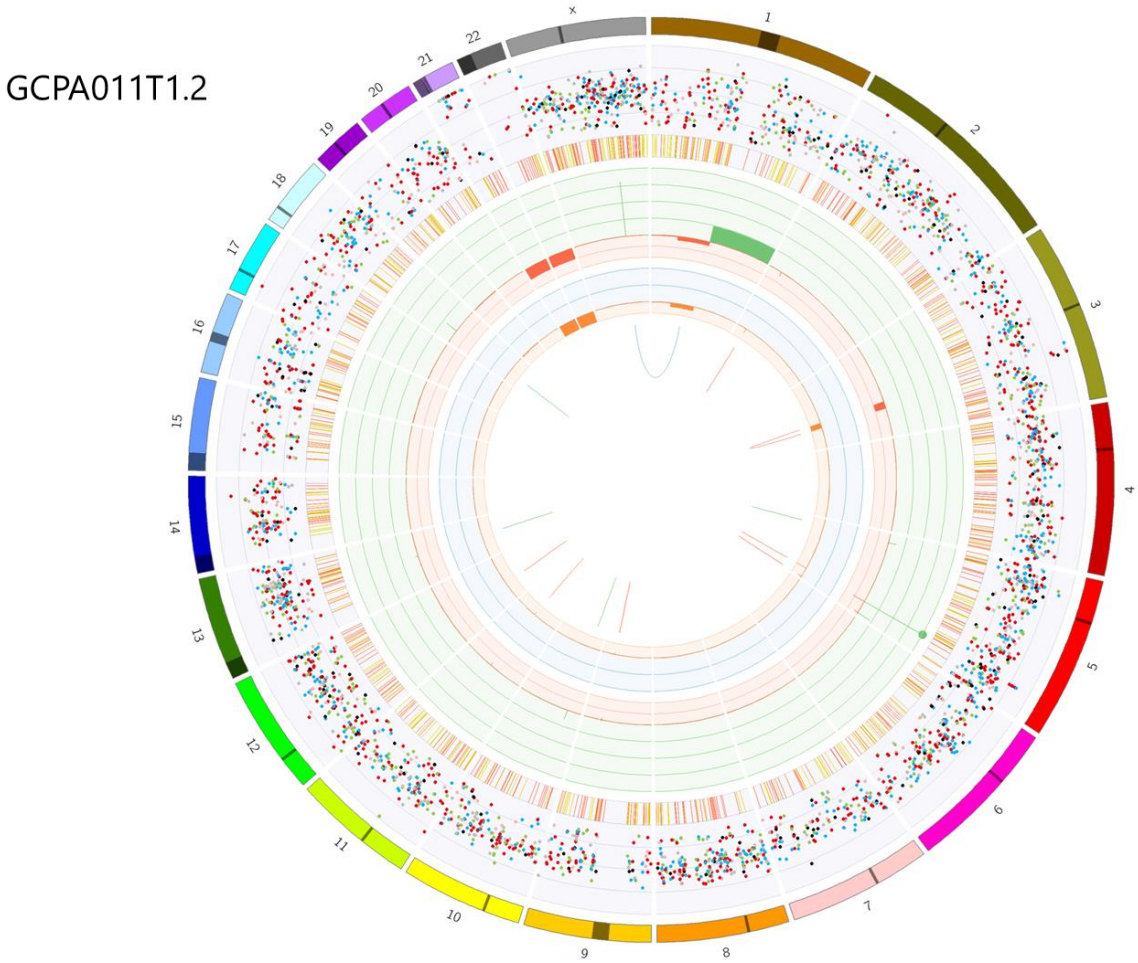
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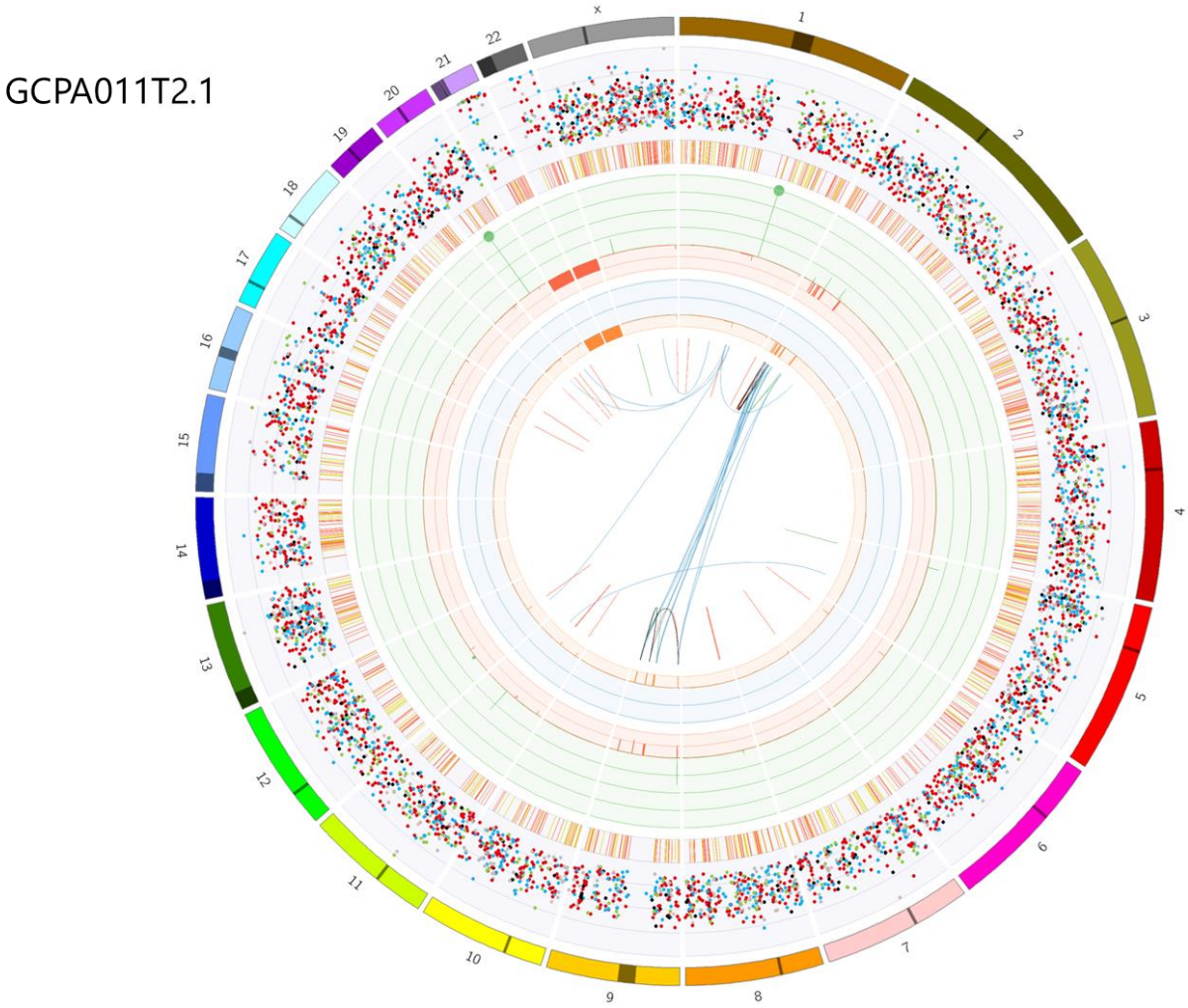
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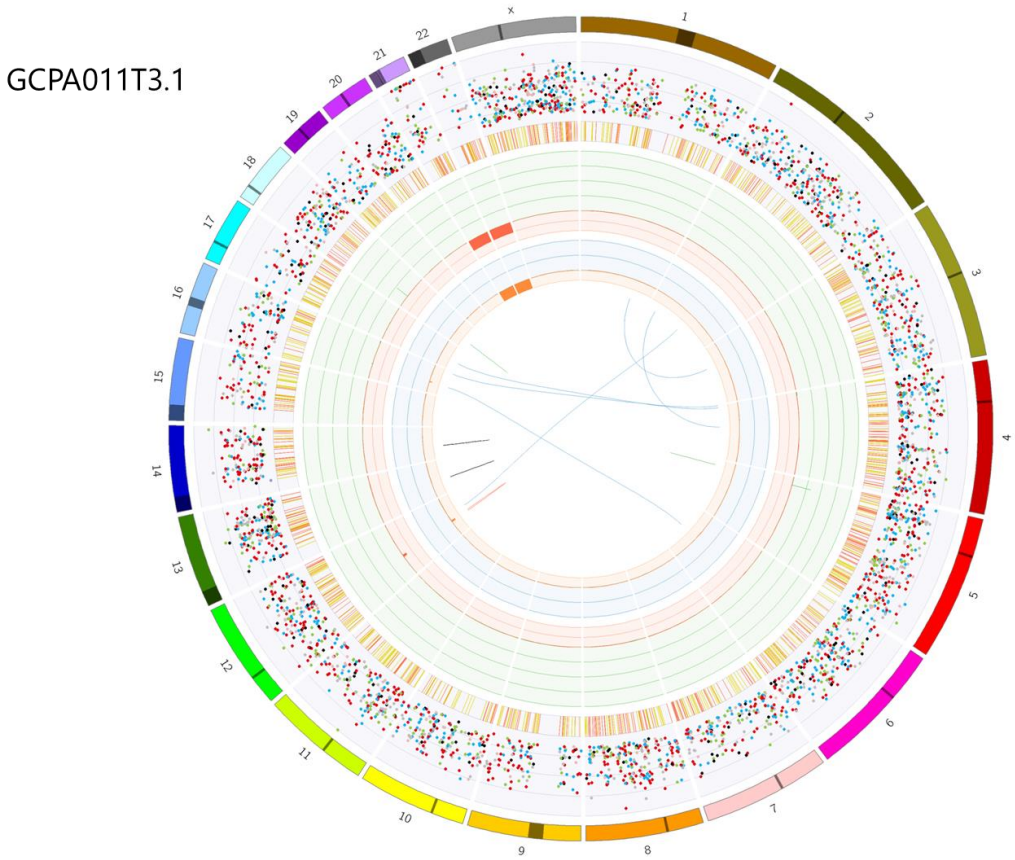
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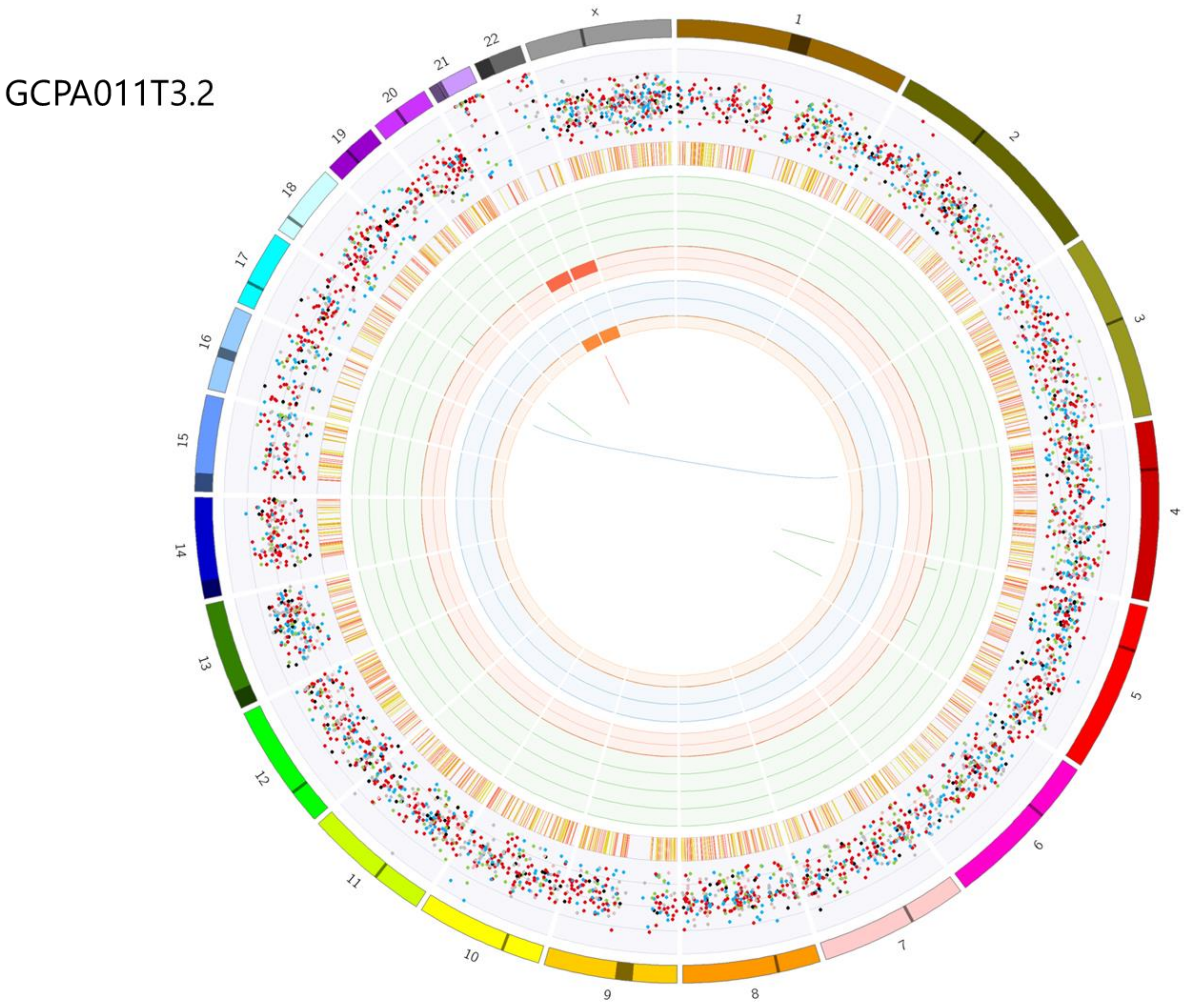
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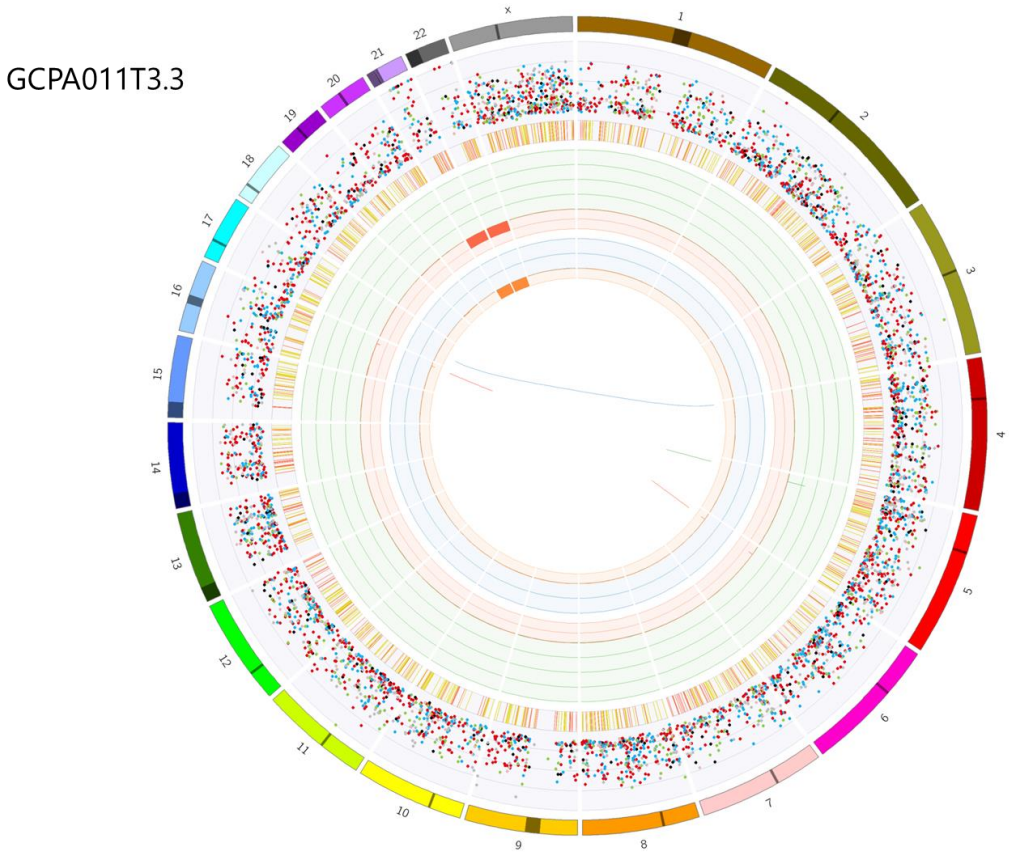
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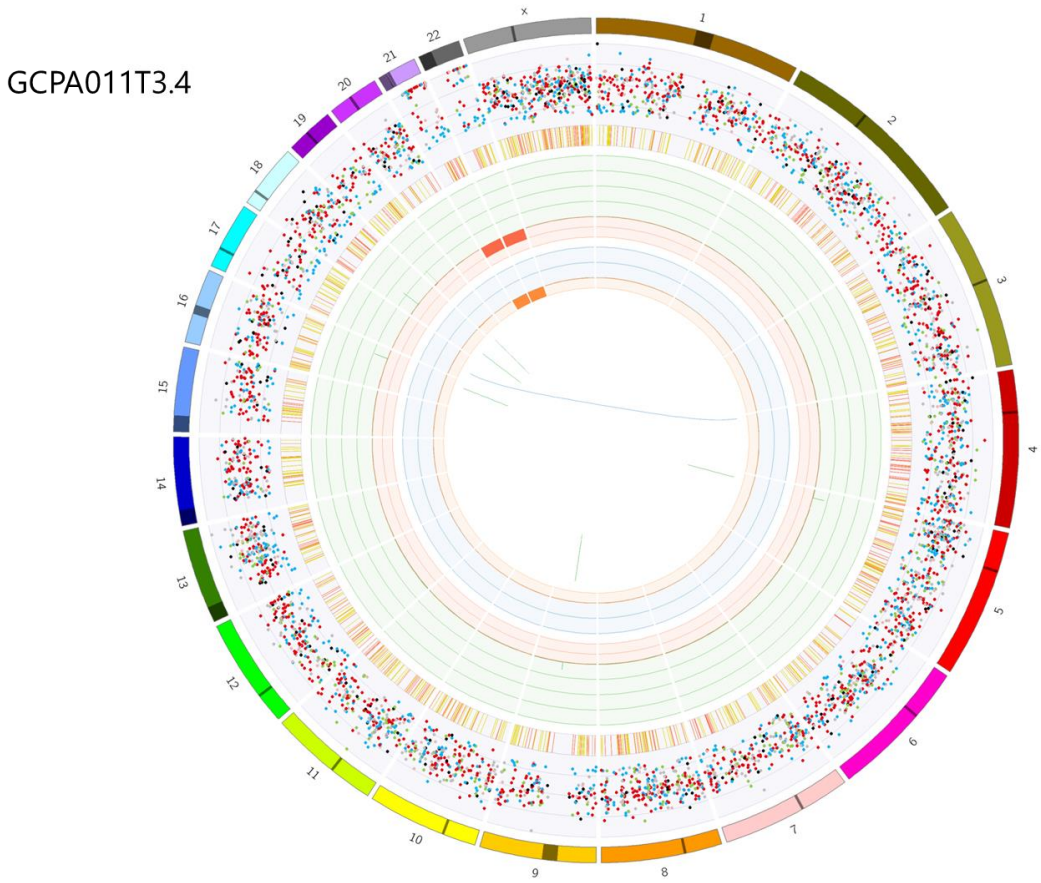
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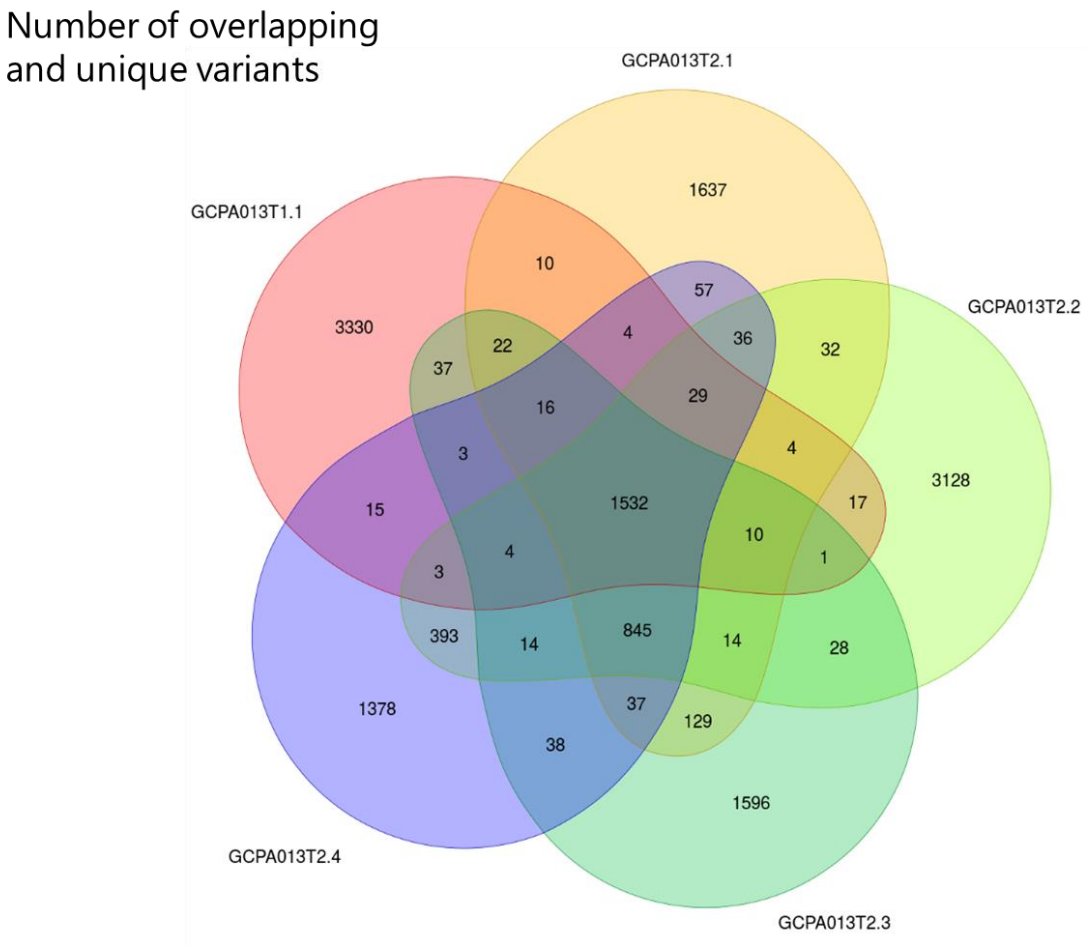
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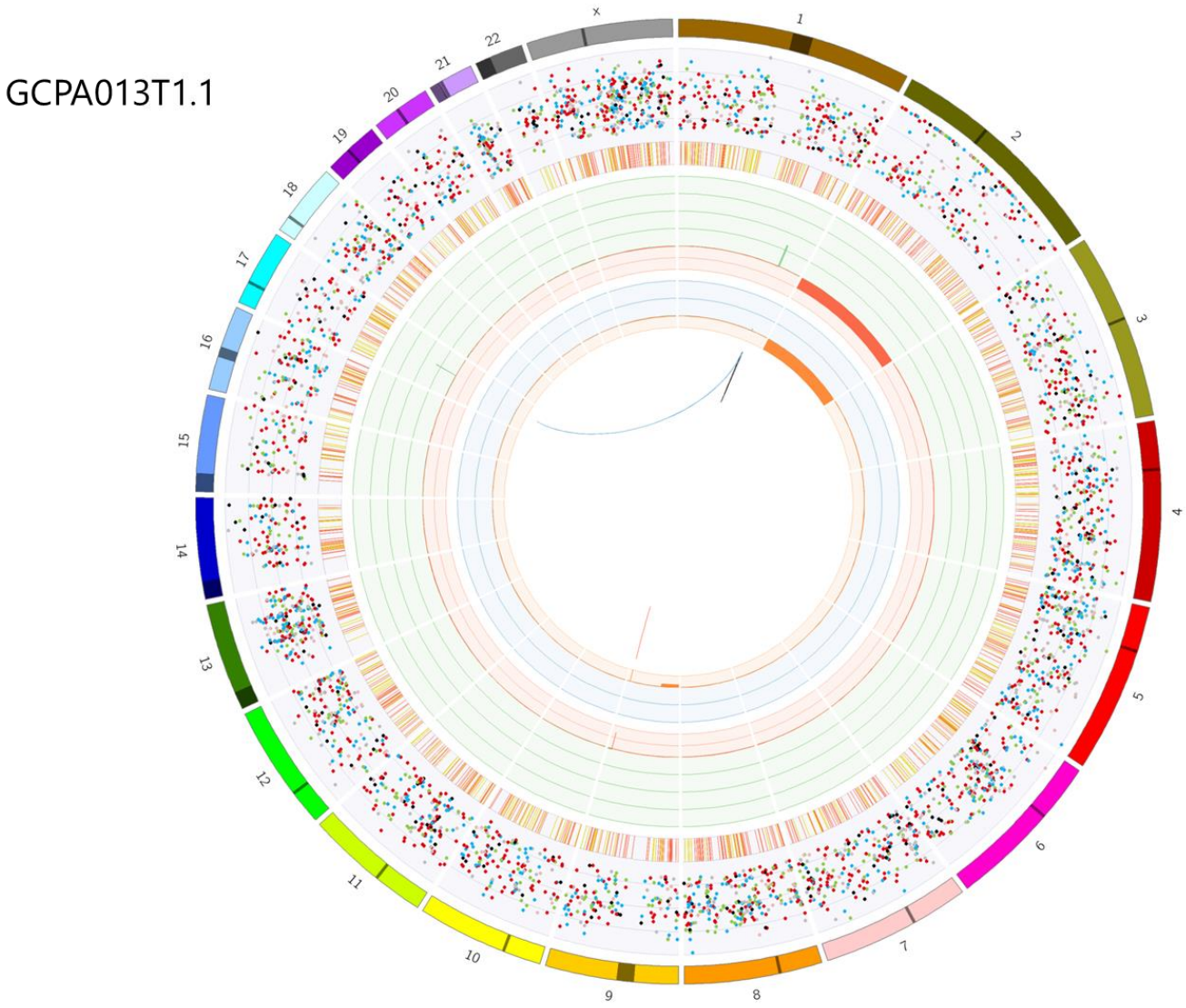
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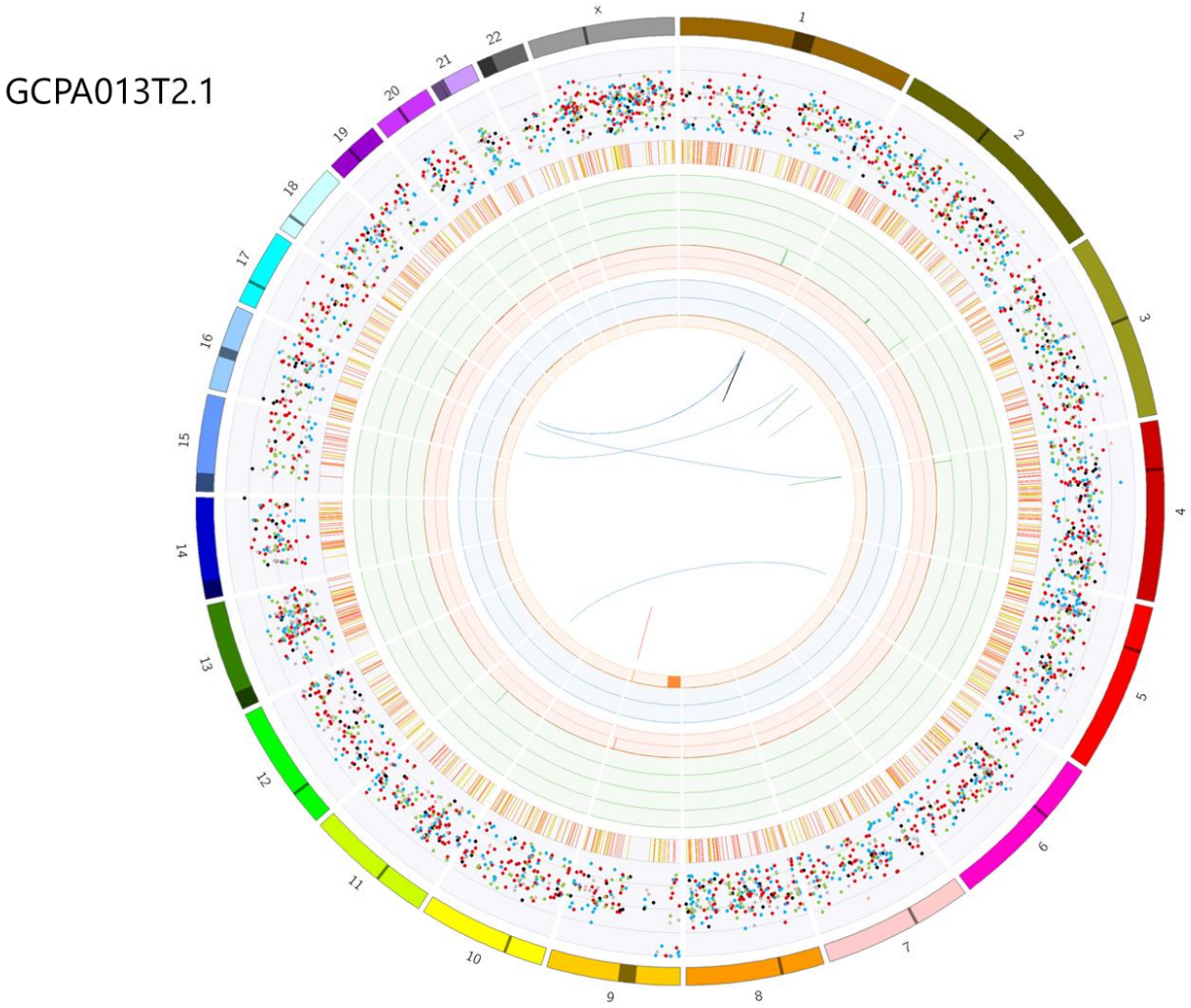
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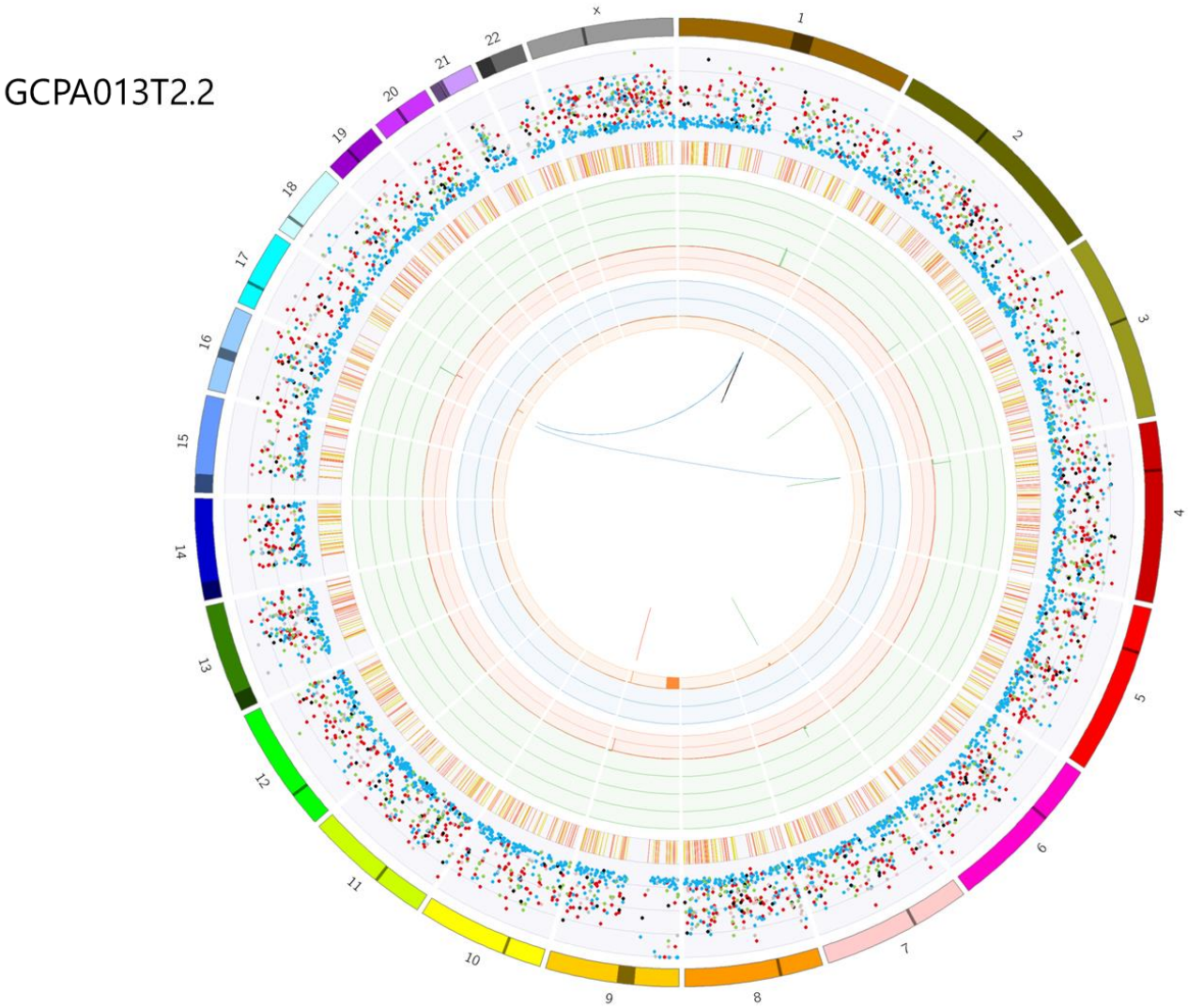
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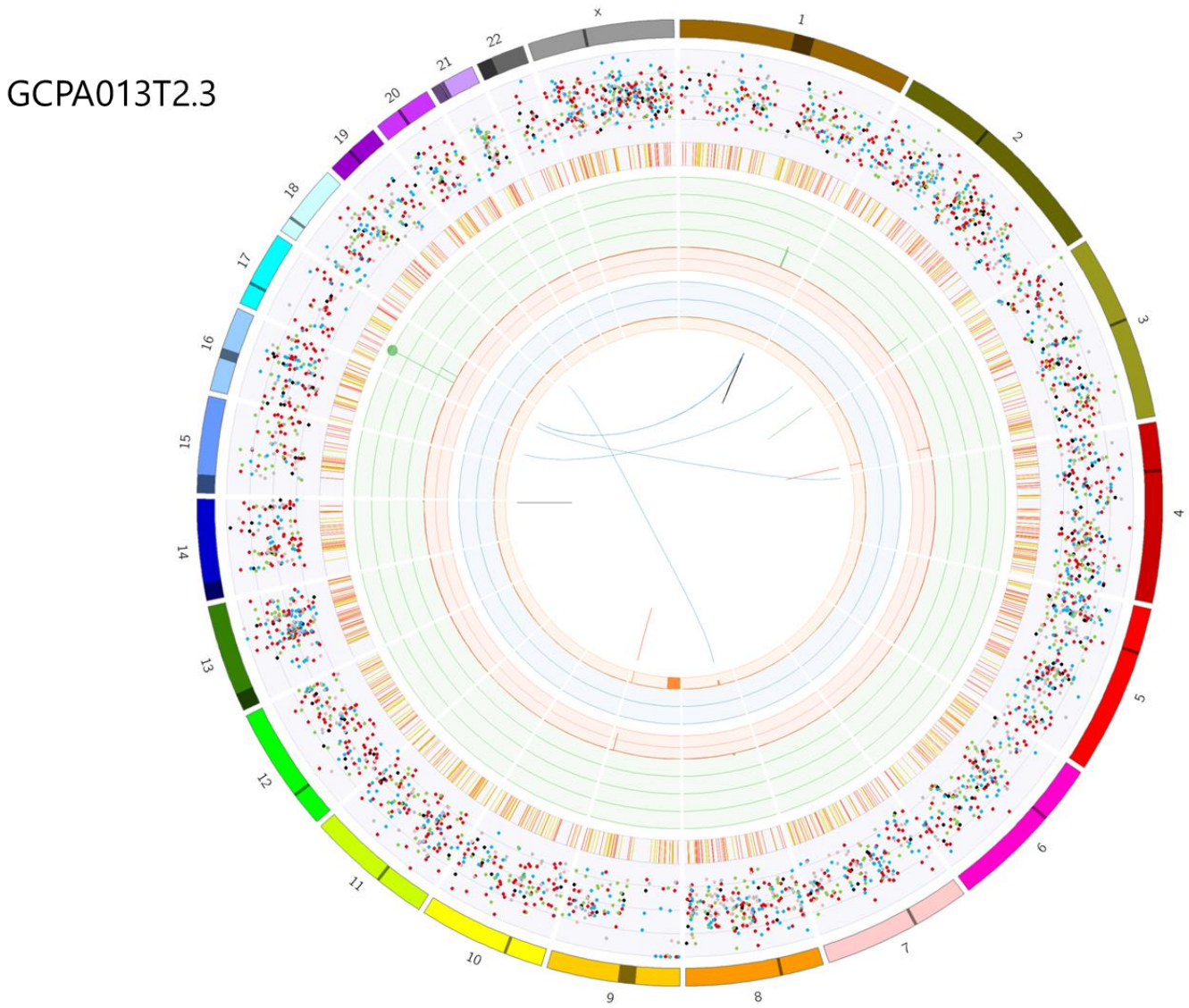
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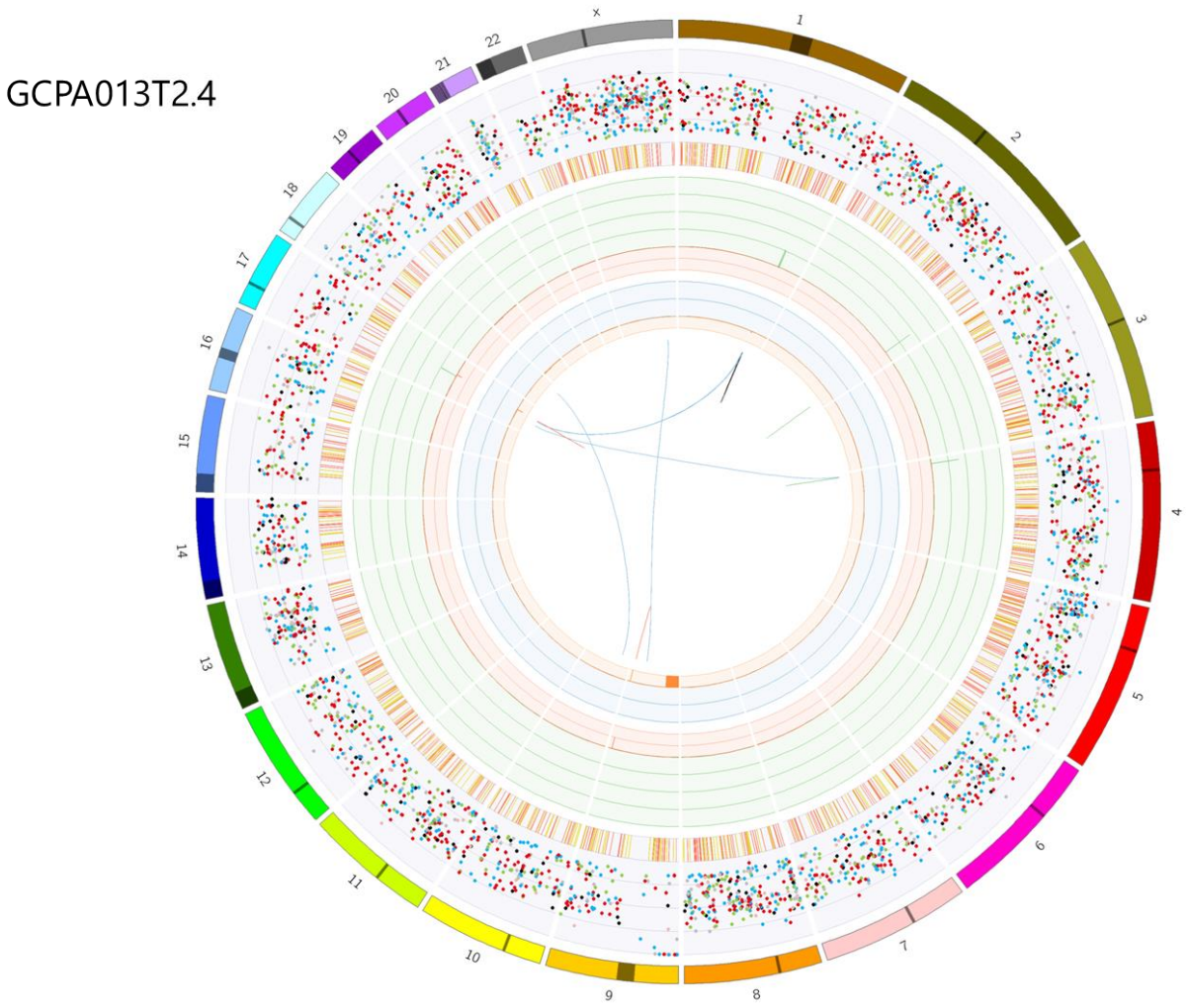
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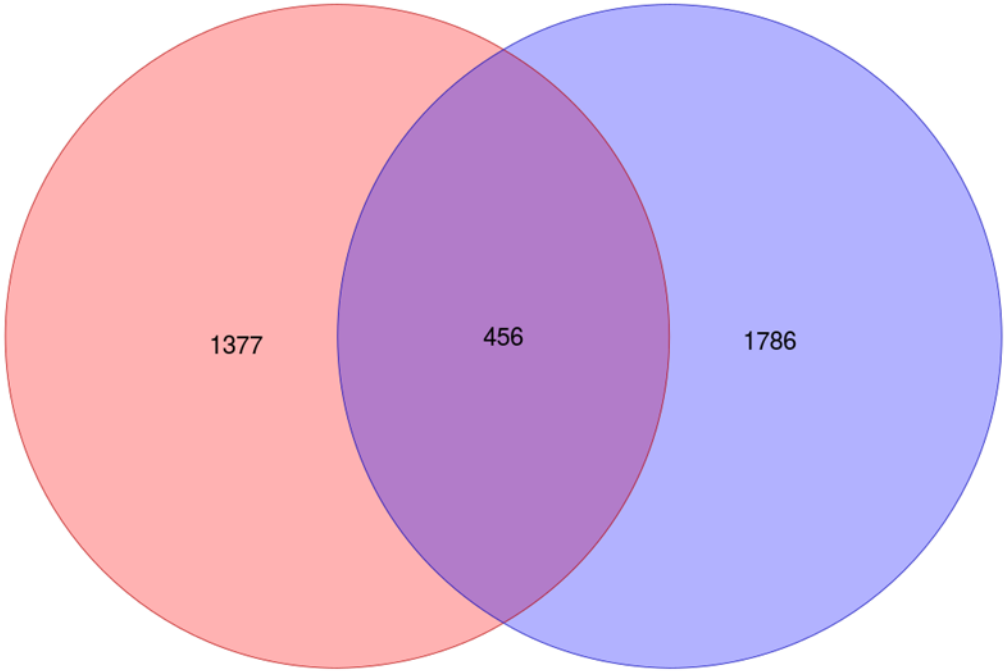
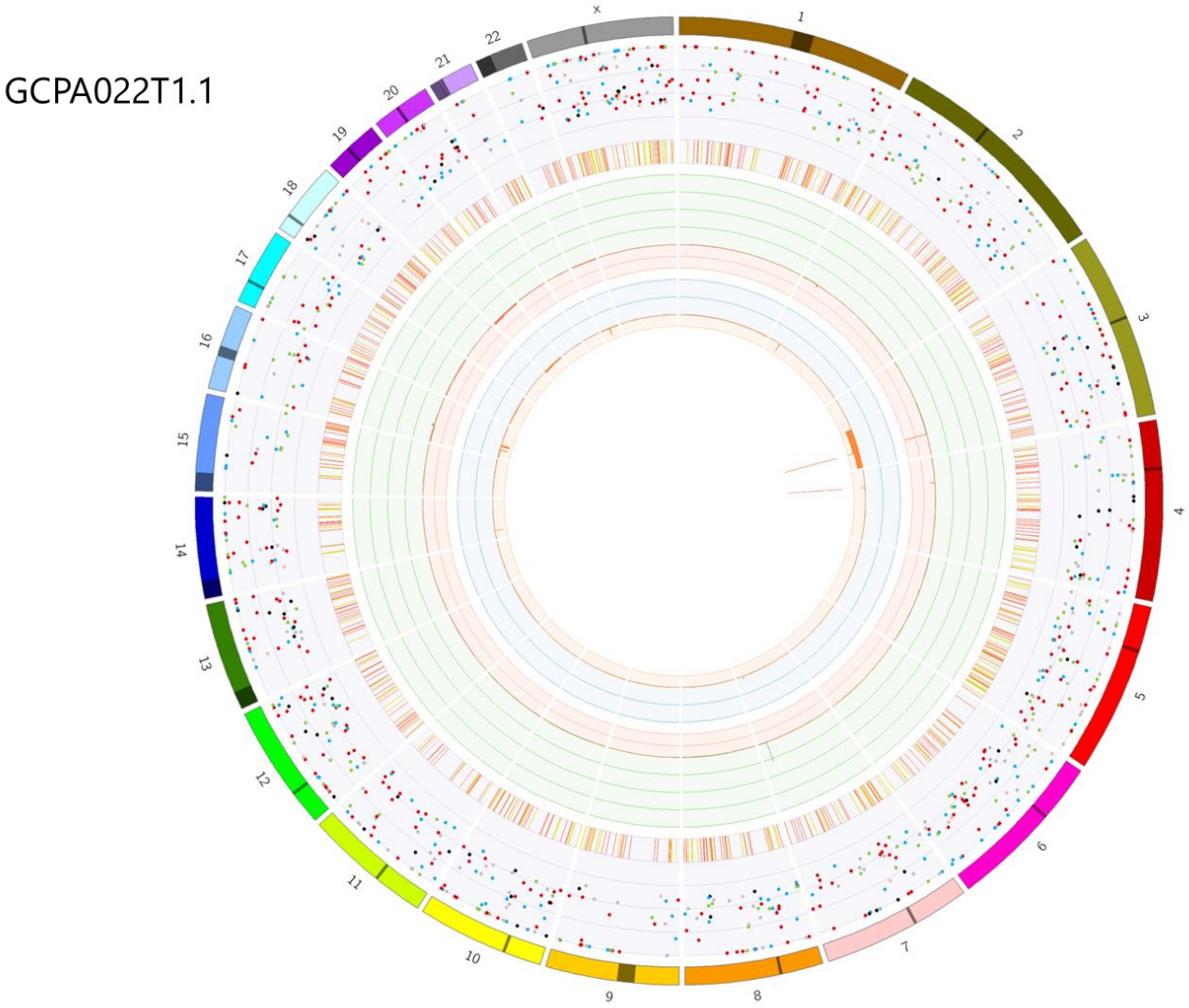
Number of overlapping and unique variants

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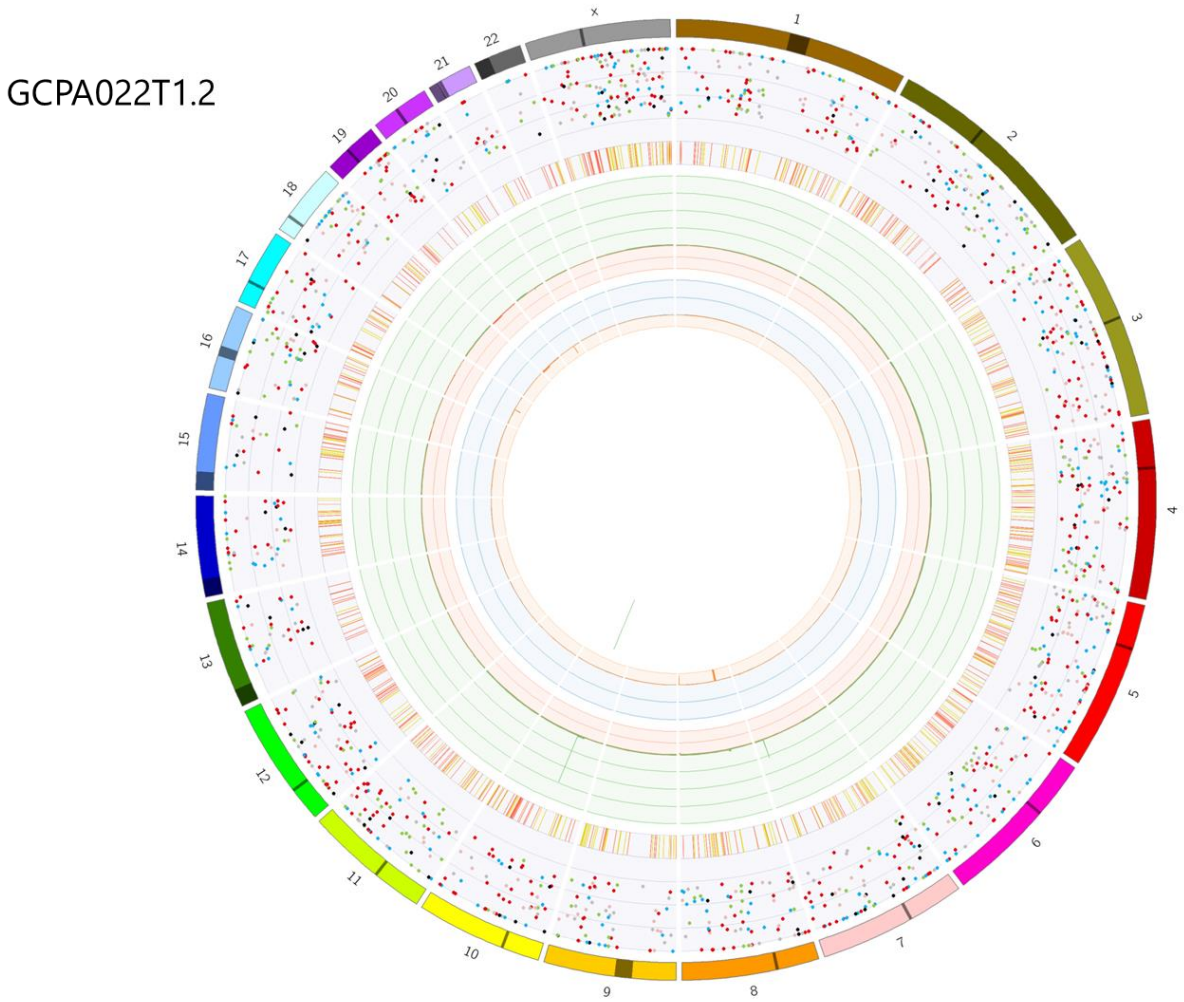
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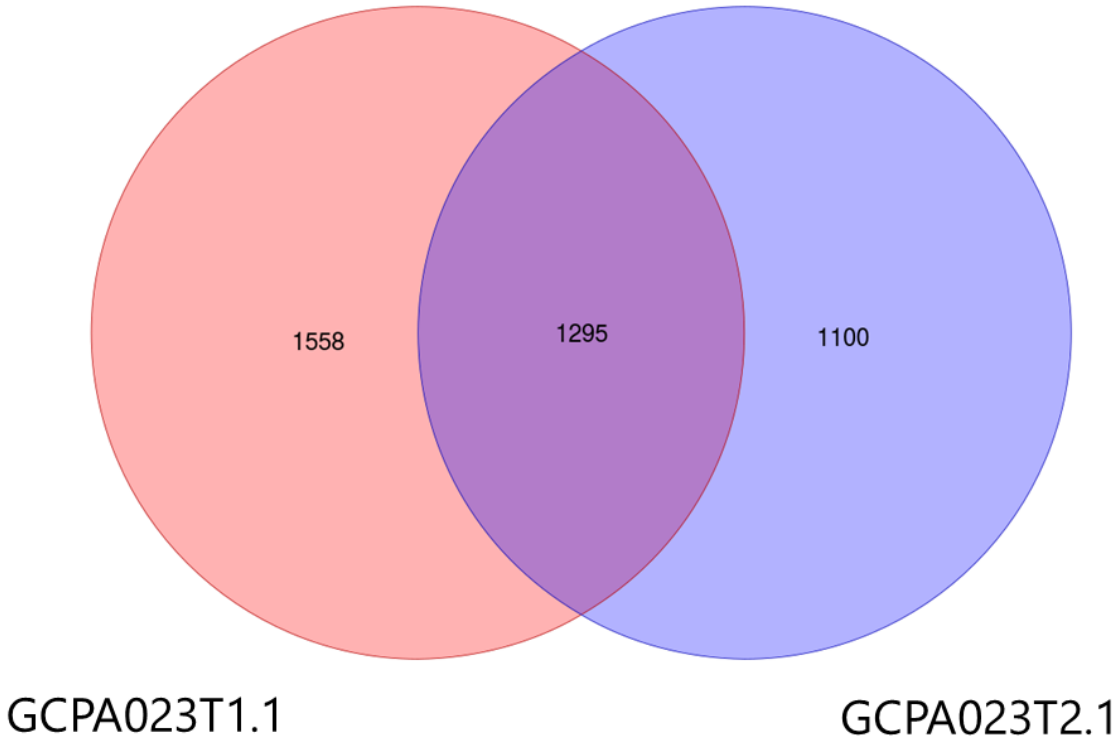
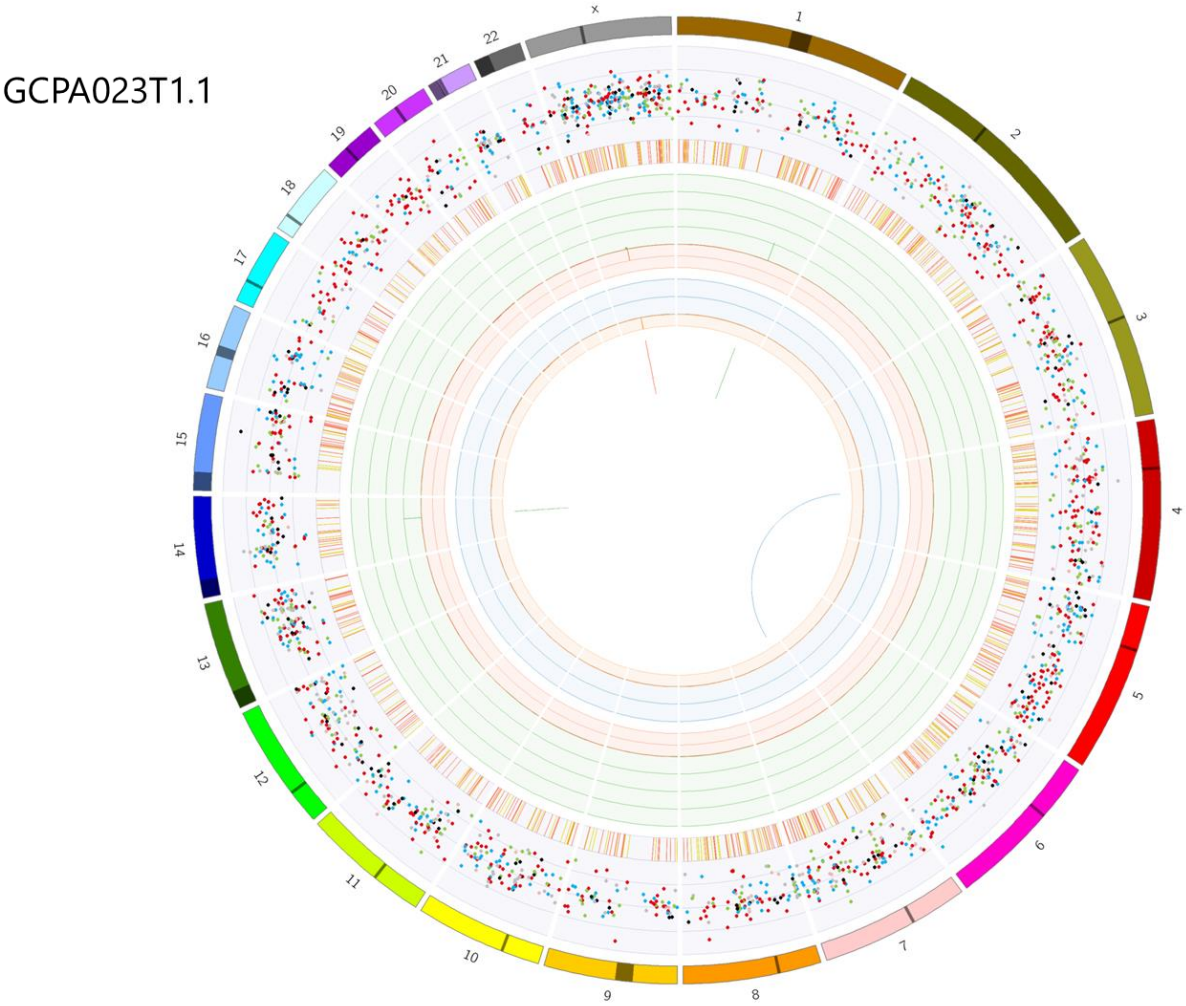
Number of overlapping and unique variants

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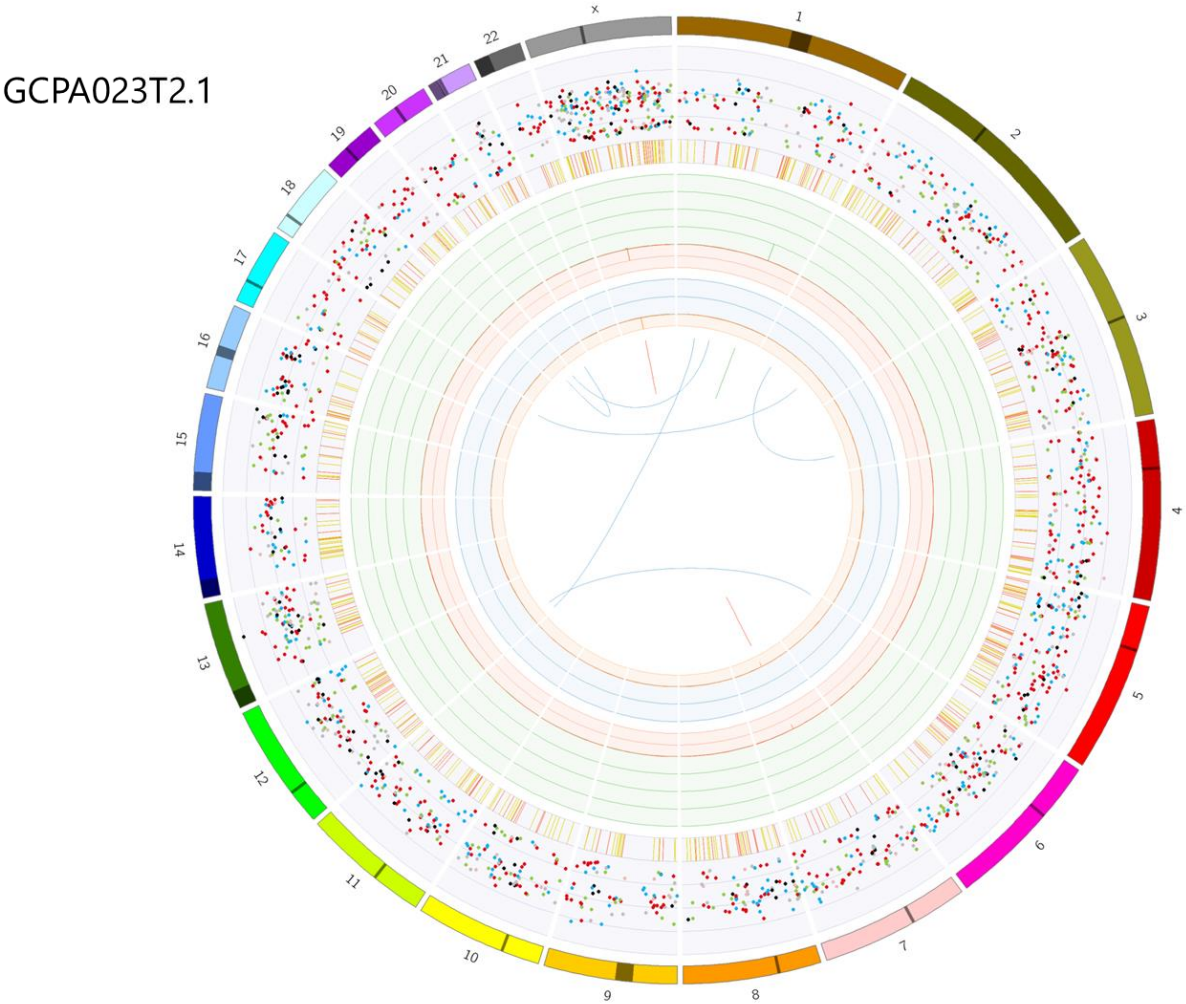

Figure S3b. Circos plots and venn diagrams from all intra-patient comparisons.

Details on CIRCOS plot

- The outer first circle shows the chromosomes. The darker shaded areas represent large gaps in the human reference genome: i.e. regions of centromeres, heterochromatin & missing short arms.
- The second circle shows the somatic variants (incl. exon, intron and intergenic regions). Somatic variants are further divided into an outer ring of single nucleotide polymorphism (SNP) allele frequencies and an inner ring of short insertion/deletion (INDEL) locations. SNP allele frequencies have been corrected for tumor purity and scale from 0 to 100%. Each dot represents a single somatic variant. SNPs are colored according to the type of base change (e.g. C>T/G>A in red) and are in concordance with the coloring used in Alexandrov et al. 2013 Nature paper that describes the use of mutational signatures. INDELs are colored yellow and red for insertions and deletions respectively.
- The third circle shows all observed tumor purity adjusted copy number changes, including both focal and chromosomal somatic events. Copy number losses are indicated in red, green shows regions of copy number gain. The scale ranges from 0 (complete loss) to 6 (high level gains). If the absolute copy number is > 6 it is shown as 6 with a green dot on the diagram.
- The fourth circle represents the observed 'minor allele copy numbers' across the chromosome. The range of the chart is from 0 to 3. The expected normal minor allele copy number is 1, and anything below 1 is shown as a loss (orange) and represents a LOH event. Minor allele copy numbers above 1 (blue) indicate amplification events of both A and B alleles at the indicated locations.
- The innermost circle displays the observed structural variants within or between the chromosomes. Translocations are indicated in blue, deletions in red, insertions in yellow, tandem duplications in green and inversions in black.

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