Summary report from the MAP-RSeq workflow. Report in .html format which summarizes the study design, alignment and expression statistics per sample, links to pre- and post-QC plots as well as to the resulting files on gene and exon expression, fusion transcripts and SNVs identified per sample.
V. Results Summary:

- QC steps - FastQC Report
  FastQC aims to provide a simple way to do some quality control checks on your sequence data coming from high throughput sequencing pipelines. It provides a modular set of analyses which you can use to give a quick impression of whether your data has any problems of which you should be aware before doing any further analysis.

- Statistics based on per Sample Analysis (Column Description)
  Analysis is carried out using fastqc sequence files as input and generates output tables. For paired-end runs, the tables contain counts for each sample combined from both reads.

<table>
<thead>
<tr>
<th>SAMPLE</th>
<th>TOTAL READS</th>
<th>EXON READS</th>
<th>SPliced READS</th>
<th>SPliced READS (unspliced)</th>
<th>SPliced READS (junction)</th>
<th>TOTAL READ COUNT</th>
<th>EXON READ COUNT</th>
<th>SPliced READ COUNT</th>
<th>STRanded READ COUNT</th>
</tr>
</thead>
<tbody>
<tr>
<td>s_AB</td>
<td>254,028,830</td>
<td>249,583,823</td>
<td>242,922,294 (95.9%)</td>
<td>236,569,802 (91.5%)</td>
<td>21,722,442 (8.5%)</td>
<td>162,745,496 (60.7%)</td>
<td>180,605,197</td>
<td>292,827</td>
<td></td>
</tr>
<tr>
<td>s_CD</td>
<td>367,407,544</td>
<td>356,409,715</td>
<td>350,734,037 (99.4%)</td>
<td>341,056,109 (96.2%)</td>
<td>34,077,946 (9.3%)</td>
<td>156,550,950 (55.3%)</td>
<td>156,985,171</td>
<td>383,190</td>
<td></td>
</tr>
</tbody>
</table>

VI. Results Delivered

The following three sets of tables are delivered and column description is available in Appendix:

- Exon table: contains counts for the number of times an exon has been detected
  count: (read count) * sum of exon read counts

- Gene table: contains counts for the number of times a gene copy has been detected
  count: (read count) * sum of exon read counts, with an exception that if reads start in different exons of the same gene locus, these are counted only once for the gene

- SNV reports: contains Single Nucleotide Variants (SNV) called using GATK software
  sample gets SNP + rare (SV) calls for each sample

- KHiGHC PLOTS:

<table>
<thead>
<tr>
<th>SAMPLE</th>
<th>Junction Saturation</th>
<th>Splicing Junction</th>
<th>Splicing Event</th>
<th>InrER distance</th>
<th>Read Duplication</th>
</tr>
</thead>
<tbody>
<tr>
<td>s_AB</td>
<td><img src="image" alt="Junction Saturation" /></td>
<td><img src="image" alt="Splicing Junction" /></td>
<td><img src="image" alt="Splicing Event" /></td>
<td><img src="image" alt="InrER distance" /></td>
<td><img src="image" alt="Read Duplication" /></td>
</tr>
<tr>
<td>s_CD</td>
<td><img src="image" alt="Junction Saturation" /></td>
<td><img src="image" alt="Splicing Junction" /></td>
<td><img src="image" alt="Splicing Event" /></td>
<td><img src="image" alt="InrER distance" /></td>
<td><img src="image" alt="Read Duplication" /></td>
</tr>
</tbody>
</table>

- Toprall fusion (circle plot)
  Tab delimited fusion results are available [here](link)
  Fusion fusion report in vcf, format is available [here](link)

- Statistics based on per Sample Analysis are recorded in the tab delimited file [details](link)

- GATK Visualization
  The SVR and ABC, annotation reports (both standard and filtered) include visualization links to IGV to enable a realistic view of the variants. Please follow steps in the following link to setup IGV (takes less than 5 minutes) and utilise this feature for variant visualization

VII. Useful Links

- Toprall 2
- Toprall Fusion
- HiSeq
- IHiSeq
- GATK
- UCSC Genome Browser

VIII. Appendix

Full Length CEA Sequencing (mRNA-seq) results delivery format [appendix]

Authorship Consideration

Advancing scientific research is a primary mission of all bioinformatics and acknowledgment of contribution through authorship on manuscripts arising from this analysis is one way our work is assessed and attributed. We require to be considered for authorship on any manuscripts using the analysis results provided if you believe we have made substantive intellectual contributions to the study.

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