Summary Part 1

- Location => Look out for Feature
- Object creation from Adaptor or from API object
Use case

...  
# Information for variation:  
my $variation_adaptor = $registry->get_adaptor(‘human’, ‘variation’, 
‘variation’);  
my $variation = $variation_adaptor->fetch_by_name(‘rs5571078’);  
print $variation->source(), “\n”;  
my @variation_features = $variation->get_all_VariationFeatures();  
foreach my $vf (@variation_features) {  
  print $vf->seq_region_start, ‘ ’, $vf->seq_region_end, “\n”;  }

my $slice_adaptor = $registry->get_adaptor(‘human’, ‘core’, ‘slice’);  
my $slice = $slice_adaptor->fetch_by_region(‘chromosome’, 13, 
48985833, 48987289);  
my @variation_features_on_slice = $slice->get_all_VariationFeatures();
Structural variations

- Defined as a region of DNA > 1kb in size
- Different types of structural variations:
  - Copy number variation (CNV)
  - Allele type ‘Gain’
  - Inversions
  - Duplication
  - Translocation
### StructuralVariation

- Similar to Variation object

<table>
<thead>
<tr>
<th>Attribute</th>
<th>Return value</th>
<th>Method</th>
</tr>
</thead>
<tbody>
<tr>
<td>Structural variation name</td>
<td>String (esv214236)</td>
<td>$sv-&gt;variation_name()</td>
</tr>
<tr>
<td>Class</td>
<td>String (CNV)</td>
<td>$sv-&gt;var_class()</td>
</tr>
<tr>
<td>Study</td>
<td>Object</td>
<td>$sv-&gt;study()</td>
</tr>
<tr>
<td>Supporting evidence(s)</td>
<td>Listref of objects</td>
<td>$sv-&gt;get_all_SupportingStructuralVariants()</td>
</tr>
</tbody>
</table>
SupportingStructuralVariation

- Supporting evidence <=> Allele for a Variation

Region of the genome that a submitter has defined as containing structural variations.

Actual variant calls that were made within a study.
Example

Structural variation: **nsv428936**

<table>
<thead>
<tr>
<th>Variation class</th>
<th>CNV (SO:0001019)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Source</td>
<td>DGVa – Database of Genomic Variants Archive</td>
</tr>
<tr>
<td>Study</td>
<td>Nstd10 – “The genomic architecture of segmental duplications and associated copy number variants in dogs.”</td>
</tr>
<tr>
<td>Location</td>
<td>Chromosome 2:4626823-4953101 (forward strand)</td>
</tr>
</tbody>
</table>

**Supporting evidence**

<table>
<thead>
<tr>
<th>Supporting evidence</th>
<th>Chr:bp (strand)</th>
<th>Allele type</th>
<th>Sample name</th>
</tr>
</thead>
<tbody>
<tr>
<td>nssv458877</td>
<td>2:4635123-4953101 (+)</td>
<td>Loss</td>
<td>German Shepard</td>
</tr>
<tr>
<td>nssv458855</td>
<td>2:4637149-4653454 (+)</td>
<td>Loss</td>
<td>Basenji</td>
</tr>
<tr>
<td>nssv458888</td>
<td>2:4688172-4723226 (+)</td>
<td>Gain</td>
<td>Golden Retriever</td>
</tr>
</tbody>
</table>

...
**StructuralVariationFeature**

- Fetch by `StructuralVariationFeatureAdaptor` or get SVFs from `StructuralVariation` or `SupportingStructuralVariation` objects
- Does not return alleles, only coordinates
- SV specific coordinates
  - Uncertainty of breakpoint locations:
Exercise 3 – Structural variations

• For structural variation nsv428936 reported in dog get:
  • Structural variation class
  • Study name and description
  • Coordinates (check for breakpoint locations)
• Get names and variation classes of its supporting evidences

• (Do a similar analysis with SV esv234231 in human)